

A LITERATURE REVIEW INTO PARENTS' EXPERIENCES OF
RAISING A CHILD WITH A DISABILITY, AND RESEARCH
EXPLORING MOTHERS' EXPERIENCES OF RECEIVING A
DIAGNOSIS OF MONOSOMY 1P36 DELETION SYNDROME
FOR THEIR CHILD

by

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Abstract

Research into parents' experiences of raising a child with a disability is varied, with recurring themes of stress and burden being reported, alongside parents' more positive experiences. This literature review presents a meta-synthesis of qualitative research into parents' experiences of raising a child with a physically or intellectually disabled child, with a view to identifying overarching themes and theory emerging in qualitative research in this field. Nine qualitative papers met the inclusion criteria, of which six also met quality criteria, and a meta-synthesis of these six papers is presented within this review. Overarching themes derived from the papers included two risk factors: Experiences and Challenges; and two protective factors: Strategies for Coping and Support. Each overarching theme had a number of themes and subthemes. These are presented in a visual representation of a parent's journey of raising a child with a disability. The protective factors can be a focus of intervention by psychologists within Learning Disability Community Mental Health Teams and Paediatric services, as these professionals are in a position to encourage families to draw upon their resources, enabling them to employ better coping strategies in the face of adversity throughout the life-cycle of disability.

To Zebidee and Mum

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Firstly, I would like to thank all of the participants who have taken part in this study. They have provided me with a valuable insight into their experiences of living with a child with Monosomy 1p36 Deletion Syndrome. I hope this research will be of benefit to them, and to others who are going through similar experiences.

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OVERVIEW OF THESIS

This thesis is submitted to meet the research requirements for the degree of Doctor of Clinical Psychology (Clin. Psy. D) at The University of Birmingham. It represents both clinical and research components of the course. All identifying information has been changed to ensure participants' confidentiality.

Volume I comprises the research component of the thesis. It consists of two papers which have been prepared for the journal *Social Science and Medicine* (Appendix 1), and an Executive Summary. The first paper is a full length qualitative literature review/meta-synthesis of qualitative research reporting parents experiences of raising a child with a disability. The second paper is a full length qualitative research report of mothers' experiences of receiving a diagnosis of Monosomy 1p36 Deletion Syndrome for their child. The executive summary presents a summary of the two papers, for distribution within the public domain. The final section of Volume I comprises the appendices for all three documents.

Volume II comprises the clinical component of the thesis. It consists of five clinical practice reports which reflect clinical work undertaken during the course. The reports include a behavioural and systemic formulation of a young man with autism demonstrating 'posting' behaviours; a small scale service related project, evaluating a Psychological consultancy service for staff working at a local respite and intensive support service for adults with a learning disability; a case study of a sixty-eight year old lady with anxiety relating to rheumatoid arthritis; a single-case experimental design for a six year old boy displaying aggressive behaviours; and a service evaluation reporting the implementation of outcome scales within a combined adult/older adult specialty.

LITERATURE REVIEW

Introduction

This paper aims to provide a meta-synthetic review of qualitative research into parents' experiences of raising a child with a disability. Research to date on parents' experiences is varied, with two common recurring themes of stress and burden being reported (Dyson, 1997; McLinden, 1990; Ray and Ritchie, 1993), yet studies have also highlighted parents' positive views of raising a child with a learning disability (Hastings, Allen, McDermott and Still, 2002; Olsson and Hwang, 2003). To understand these contrasting findings in more depth, it seems timely and useful to review qualitative studies in this area, since these may clarify our understanding of the complex responses of parents who find themselves in this situation. Quantitative research in relation to stress, indicates that parents of children with a disability report a number of themes, for example worry, fatigue and limited social relationships (McLinden, 1990); but also feelings of depression which are attributed to work overload and having little time left for themselves (Olsson and Hwang, 2003). Olshanksy (1962) described chronic sorrow as a parental response to the loss of a 'perfect child' characterised by waves of grief and sadness throughout their child's life. Recent literature focuses on parental functioning in families with a child with a disability (Cuskelly, Jobling, Chant, Bower and Hayes, 2002; Roach, Orsmond and Barrat, 1999).

Systematic reviews of research that uses quantitative methodologies are conventionally understood to have specific characteristics. They tend to have an explicit protocol, address a pre-specified, highly focused question(s); have explicit methods for searching for studies; appraise the studies to determine their scientific quality; and use explicit methods, including descriptive summary or meta-analysis (where appropriate), to combine the findings across a range of studies (Egger, Smith and O'Rourke, 1995). This type of analysis may result in reductionist and/or standardized models that do not focus on individual variability or the

influence of context (Culpepper and Gilbert, 1999; Forbes and Griffiths, 2002). Ferlie, Wood and Fitzgerald (1999) suggest that evidence-based medicine can be characterised by an abstracted form of pure rationality, often of a meta-analytic nature, with a clear hierarchy of evidence, at the apex of which lies the randomised controlled trial.

However applying the techniques and parameters used to review quantitative studies to qualitative research may raise more questions than solutions. As a result there have been a number of alternative suggestions on how qualitative research could be reviewed. These include: treating the questions which guide the review as ‘a compass, rather than an anchor’ (Eakin and Mykhalovskiy, 2003), and as something which is not settled until the end of the review; or taking an iterative approach which modifies the question in response to search results and findings from retrieved items. Clearly, this could be seen as rather radical as it moves away from one of the principles of conventional systematic review methodology that review questions be precisely specified at the outset (Dixon-Woods, Bonas, Booth, Jones, Miller, Sutton et al., 2006), even though in practice there may often be a period during which a review of quantitative studies is refined and shaped up in response to the initial findings of searches.

Qualitative reviews have taken a number of different approaches when appraising and synthesising the quality of qualitative research, for example Estabrooks, Field and Morse (1994) argue that weak qualitative papers should be excluded and Campbell, Round, Pope, Britten, Pill, Morgan et al. (2003) excluded research that did not meet their quality standards. Attree (2004) graded papers on a scale from A to D, excluded papers graded D, whilst papers graded C were used to provide supporting data and studies A or B were used to identify the

main concepts used in the synthesis. How weak findings should be identified has not yet been made clear, but the NHS Centre for Reviews and Dissemination (2001) recommend that ‘the process whereby conclusions are drawn from study findings are made as transparent as possible and that there are attempts to replicate conclusions’ offers a solution to the difficulties of appraising qualitative research. Sandelowski (1997) suggests that a good meta-synthesis (a term coined to refer to a drawing together of qualitative findings) should focus on a synthesis of findings rather than simply a description of the analysis and findings of previous studies, and in doing so it should provide enough information for readers to follow the decisions made. It is important to recognise that interpretive synthesis is a methodology distinct from systematic review, with distinct benefits, offering the potential for insight, vividness, illumination and reconceptualization of research questions (Dixon-Woods, Bonas, Booth, Jones, Miller, Sutton et al., 2006).

The Public Health Resource Unit offers NHS public health consultancy. Within this organisation lies the area of Public Health Development, who have developed the Critical Appraisal Skills Programme (CASP). CASP has helped to develop an evidence-based approach in health and social care, working with local, national and international groups. Tools were developed by CASP to help with the process of critically appraising qualitative research (2006). These are employed to appraise research studies included in the current review.

The aims of this review are:

- to examine research on parents' experiences of raising a physically or intellectually disabled child, in addition to previous literature focusing on parental functioning
- to incorporate worldwide research, thus allowing for the exploration of cultural differences
- to synthesise these findings, with a view to identifying overarching themes and theory emerging in qualitative research in this field

Method

Inclusion Criteria:

- i) Empirical research papers using qualitative research methods to explore parents' experiences of raising a child with a disability (i.e. excludes professionals' perspectives, research with other family members, and parent-provider relationships)
- ii) Journal papers published in English between January 1998 and June 2008 (i.e. excludes dissertations, books, book chapters and reviews)
- iii) Qualitative research presenting participants' experiences in their own words, thus excluding closed question interviews, i.e. questionnaires
- iv) Qualitative research presenting a number of different accounts, rather than individual accounts (e.g. single case studies or single first person accounts)

Search strategy:

An initial literature search using PsychINFO, PubMed, Web of Knowledge (WOK),

EMBASE, MEDLINE and CINAHL was completed during June 2008. This search used the

terms 'child disability', 'children with disability', 'parents AND children with disability', 'disabled child', 'parent 'experience'. 164 articles were retrieved, and were searched by hand to identify a further 35 articles.

Exclusion stage 1

Papers were excluded in the first instance if the abstracts revealed that they clearly did not fit the inclusion criteria. This resulted in thirteen relevant articles which were obtained.

Exclusion stage 2

Reading the full texts revealed that a further four papers did not fit the inclusion criteria. The papers that were excluded at this stage are listed in Appendix 2.

A final total of nine papers were found to meet all of the above criteria and were included in this review.

Review methodology

Dixon et al. (2004) outline two types of syntheses that can be used to summarise qualitative data: aggregative syntheses, and interpretive syntheses. Aggregative syntheses focus on summarising data, either through a meta-analysis, or less formally through providing a descriptive account of the data. By contrast, an interpretive synthesis is concerned with the development of concepts, and with the development and specification of theories that integrate these concepts. Thus, interpretive syntheses avoid specifying concepts in advance of the synthesis and fixing the meaning of these concepts at an early stage. Interpretative synthesis involves considering the result of qualitative studies in order to identify common

concepts. Having an overview of the studies, this process may bring out concepts that were not actually included in the original studies, but which help to characterise the data as a whole. It then incorporates the identified concepts into a tentative theoretical structure. It is important that an interpretative synthesis of primary studies be grounded in the data reported, verifiable and plausible, with reflexivity being a paramount requirement.

This review attempts to provide an interpretive synthesis of findings.

Process of review

Consideration of quality issues

The following articles were appraised using the proforma developed by Critical Appraisal Skills Programme (CASP) Appraisal Tool for qualitative research (Public Health Resource Unit, 2006). A copy of this proforma can be found under Appendix 3. A table was then developed using the headings from this proforma (see Table 1) and this was used as a way of considering and summarising the way in which each qualitative study met the CASP criteria for good qualitative research.

Identifying themes and developing preliminary synthesis

Papers were then considered according to their stated aims, themes, and any outcomes and implications for professionals. In accordance with Dixon et al. (2004) the identified concepts and themes were incorporated into more inclusive overarching themes.

This review therefore firstly presents an overview of the quality of the research reported in the identified studies, before going on to present a meta-synthesis of the studies that are found to be of adequate quality.

Table 1: Table of Papers Reviewed: Process Issues

Key:

<i>Quality issues</i>	<i>Transferability</i>
<i>T = lack of Transparency in presentation of analysis</i>	<i>X = Does not seem transferable to other settings</i>
<i>S = Does not appear to be systematic analysis of data</i>	<i>I = Insufficient contextual detail to estimate transferability to another setting</i>
<i>D = Does not appear to be grounded in data</i>	<i>A = Transferability issues not explicitly addressed</i>
<i>E = Some data appears to have been excluded</i>	
<i>Q = Quotes not identified</i>	
<i>C = Credibility issues not addressed</i>	

Author, Year & Country	Sample	Method of Data Collection	Method of Data Analysis	Coherent Epistemological Position – narrative constructionist	Theoretical Perspective	Reflexivity	Quality Issues	Transferability
Täänila, Syrjälä, kokkonen and Järvelin (2002) Finland	Parents of 8 children, aged 8-10 years, with physical, intellectual, or both physical and intellectual disability. Ethnicity not known, social classes inclusive of I-IV	Interview	Grounded Theory – constant comparison method	Social constructivism	Not clearly stated	Not openly discussed	No issues	No issues
Smith, Brewer, Eatough, Stanley, Glendinning & Quarrell (2006) UK	8 individual parents, 2 couples Ethnicity and social class not known.	Semi structured interview	Interpretative Phenomenological Analysis	Not clear	Not clearly stated	Not openly discussed	C	No issues

Author, Year & Country	Sample	Method of Data Collection	Method of Data Analysis	Coherent Epistemological Position – narrative constructionist	Theoretical Perspective	Reflexivity	Quality Issues	Transferability
Mackey & Goddard (2006) Australia	5 women living in a rural Australian city. Ethnicity and social class not known.	Semi-structured individual interviews	Heideggerian/ interpretive phenomenology	Not clear	Not based around particular theory	Not openly discussed	No issues	No issues
Kearney & Griffin (2001) Australia	6 parents of children with developmental disability (2 couples, 2 single mothers). Ethnicity and social class not known.	Individual interviews. Couples interviewed together subsequently over 18 months. Clarification post interview in some cases.	Van Manen's phenomenological reflection, interpretation and writing	Not clear.	Olshansky's (1962) chronic sorrow	Attempts made to ensure that first author's prior contact did not influence this piece of work.	No issues	No issues
Hartley, Okwang, Baguwemu, Ddamulira, Chavuta (2005) Uganda	51 parents/caregivers (29 mothers, 7 fathers, siblings and 4 guardians) (8 children with hearing impairment, 7 visual, 12 physical, 10 mental (inc epilepsy) and 14 multiple) Ethnicity and social class not known	Semi-structured interviews with carers of child concerned and non-participatory observations. (13 = both interview and observation, 4 = observation alone)	Framework approach (Ritchie & Spencer, 1994)	Not clear	Model of coping (Lazarus & Folkman, 1984; Burr & Klein, 1994)	Triangulation	E	No issues

Author, Year & Country	Sample	Method of Data Collection	Method of Data Analysis	Coherent Epistemological Position – narrative constructionist	Theoretical Perspective	Reflexivity	Quality Issues	Transferability
Fisher & Goodley (2007)UK	25 families Ethnicity and social class not known.	Interviews with families, and focus groups with medical and social care professionals (latter not reported)	Not clearly identified	Enlightenment and modernism	Mention of model of disability (see Oliver 1996)	Clear about impact of participants insight on researchers engagement.	C	No issues
Ashencaen Crabtree (2007) UAE	15 mothers. Ethnicity: Arab Muslim. Social class not known.	Interviews over 10 months, some participants revisited.	Not clearly identified.	Not clear.	Sociological explanations offered?	Awareness of impact of researcher's different cultural background , and the use of 'disability'.	T S D E C	No issues
Mei-Ying Chang & Li-Ling Hsu (2007) Taiwan	117 parents. Ethnicity and social class not known.	Semi-structured interviews.	Adapted from Glaser and Strauss's GT approach and other works of content analysis	Not clear.	Not clearly stated	Not openly discussed.	T S E C	No issues

Author, Year & Country	Sample	Method of Data Collection	Method of Data Analysis	Coherent Epistemological Position – narrative constructionist	Theoretical Perspective	Reflexivity	Quality Issues	Transferability
Brett (2004) UK	6 parents. Ethnicity and social class not known	Interviews.	Van Manen (1984) 2 steps of thematic analysis and the determination of essential themes	Not clear.	Heidegger's (1962) notion of 'being-in-the-world'.	Triangulation, and discussion of researcher bias.	T S E	X

Findings

Quality Issues

Each article was rated in terms of its standard in relation to a number of quality dimensions to assess whether theoretical perspectives and epistemological positions were offered, whether reflexivity was considered, whether analysis was transparent, systematic and grounded in the data, whether quotes were excluded or not identified, and whether credibility issues were addressed. In addition each article was assessed for its transferability to other settings, and whether there was enough contextual detail to make this judgement.

Overview

The articles within this review were found to vary across these quality dimensions, and there is further variation in the aims of each article as they seek to understand:

- how families cope with disability
- the use of spiritual beliefs to establish meaning
- key features of specific conditions
- the health and wellness of mothers of children with disability
- the experiences of having a child with a disability
- the perceptions of mothers in relation to family acceptance and social inclusion

Theoretical Perspectives

There are very few links made in these articles to specific psychological theories, with four articles not relating findings to any clear model or theory (Mackey and Goddard, 2006; Mei-

Ying Chang and Li-Ling Hsu, 2007; Smith et al., 2006; Täänila et al., 2002). When links are suggested they are vague, for example 'Contemporary sociological models' are offered in discussions, but neither specific models nor references are given (Treloar, 2002). Kearney and Griffin (2001) and Hartley et al. (2005) offer more specific links to Olshansky's (1962) chronic sorrow model, and Lazarus and Folkman's (1984) model of coping, respectively. Burr and Klein's (1984) model of coping is also offered as a theoretical perspective (Hartley et al., 2005).

Epistemological Position

The majority of the articles reviewed did not address their epistemological positions, with the exception of Täänila et al. (2002) who make reference to Social Constructivism, in that their approach emphasises the moulding effect of the spoken language on the action of human beings. Fisher and Goodley (2007) refer to the linear narrative having its roots in enlightenment and modernism, which encourage people to seek out expert help to ensure favourable outcomes in the future.

Reflexivity

Reflexivity refers to the sensitivity with which the researcher addresses the way in which both they and the research process have shaped the collected data, including the role of prior assumptions and experience, which can influence research (Mays and Pope, 2000). Half of these articles do not openly discuss reflexivity and the process of interviewing and analysis, questioning the credibility of these findings. Where reflexivity is discussed, the articles refer to employing strategies such as triangulation, trying to ensure that any prior contact of researchers with participants did not influence the research, having an awareness of their

cultural background, and an awareness of not imposing their own language upon participants, for example not using the word 'disability', unless it is first used by participants.

Methods of analysis

The majority of articles clearly stated the method of analysis used. These included Grounded Theory/constant comparison, Interpretative Phenomenological Analysis, Heideggerian Interpretative phenomenology, Van Manen's phenomenological reflections and interpretations, and Ritchie and Spencer's (1994) Framework Approach. Two articles (Ashencaen Crabtree, 2007; Fisher and Goodley, 2007) do not make it clear which method of analysis they used, calling into question the quality of these particular articles for the purposes of qualitative review.

Transparency and systematic analysis of data

Four of the studies were not transparently reported (Ashencaen Crabtree, 2007; Brett, 2004; Fisher and Goodley 2007; Mei-Ying Chang and Li-Ling Hsu, 2007). These articles do not make it clear how they have set about completing the analysis, and do not make their presentation of analysis clear. There were no concerns relating to the transparency of the other papers.

The lack of transparency identified raises questions about how systematically the analysis was conducted. Ashencaen Crabtree (2007) stated that information obtained was noted in detail, coded into themes and finally analysed, but the paper does not give any indication of how this was done. Mei-Ying Chang and Li-Ling Hsu (2007) stated that they adapted content analysis from Glaser and Strauss' Grounded Theory approach but again they do not state how or why

they have done this, and given that content analysis would not normally be seen as a part of Grounded Theory, it is not clear that the authors have understood the approach. Brett (2004) is not explicit about how themes emerged. Without clear evidence of systematic analysis, it is not possible to accept the validity or trustworthiness of the results and it would be difficult to replicate these studies. Although Fisher and Goodley's (2007) study lacked transparency because the results were not clearly presented, making it difficult to read, it was systematic in reporting how themes emerged.

Credibility

In terms of credibility, five of the studies did not demonstrate that there had been any discussion of the analysis with other researchers (an aspect that is usually undertaken to ensure plausibility and facilitate reflexivity), nor had they triangulated themes with their participants or with other sources of data for further validation.

Use of data

There were a few concerns around the appropriate use of the data during analysis. The three articles that did not show systematic analysis of the data, did not show clear evidence that all of the data were considered in their analysis, e.g. evidencing quotes from the same participants throughout rather than taking them from a range of participants. One of these studies (Mei-Ying and Li-Ling Hsu, 2007) used 117 research participants so it would have been difficult to ensure an accurate representation of all of these participants in terms of developing themes, and evidencing quotes from each participant. The same could be said of Hartley et al.'s (2005) research involving 51 participants. Where the sample is smaller, for example Ashencaen Crabtree (2007) and Brett (2004), it could be argued that a wider range of

the material can and should be considered when reporting themes. However these particular articles did not demonstrate that quotes from each participant were included when presenting example quotes from each theme, calling into question the quality of these studies.

Transferability

Transferability was not an issue for the majority of the studies as they described their sample well, and also the nature of the child's disabilities. It was clear from these studies that the majority of participants were mothers; therefore it is important to bear this in mind when comparing these findings to fathers' experiences. In addition, many of the mothers were full-time caregivers, suggesting that these findings may not be wholly comparable to mothers who work either full- or part-time.

In one of the articles the authors clearly state that they do not believe that their research is transferable to other settings (Brett, 2004) given that they have used Heideggerian techniques of analysis, which view phenomena as "unique and beyond replication, rendering the constructs of generalisability and transferability inappropriate" (Heidegger, 1962, cited in Brett, 2004).

Ethnicity and Social Class

Ethnicity is reported in two articles but is only drawn upon in terms of reflexivity (i.e. the researchers identifying their own ethnicity and class) within one of these. Acknowledgement by the authors is key in enabling the reader to understand the author's own cultural position and the impact of this upon the interviewing and the subsequent analysis. The lack of

information about the ethnicity and social class of the participants also means that conclusions cannot be drawn about whether the findings are transferable across cultures.

Summary of the quality of the studies

In summary, when assessing each article in terms of the above quality dimensions, three of the papers show markedly poor quality in terms of transparency and systematic analysis of the data, and the use of the data (Ashencaen Crabtree, 2007; Brett, 2004; Mei-Ying Chang and Li-Ling Hsu, 2007). Therefore, these three articles, will not be further considered in this review. Brett (2004) has chosen to present only one theme from the data and this therefore provides a biased and incomplete picture of results from her study – this paper is therefore also excluded.

General themes

Table 2 provides a summary of the content issues of each study, highlighting the stated aims of this study and the overarching themes and outcomes.

Table 2: Content Issues

Author, Year & Country	Stated Aims	Themes	Outcomes
Täänilä, Syrjälä, Kokkonen & Järvelin (2002), Finland	To explore how families with physically or intellectually disabled children cope, what kind of coping strategies they use and how the families with good and poor coping capacities differ.	<ol style="list-style-type: none"> 1. Information and acceptance 2. Emotional activities 3. Good family and co-operation 4. Openness 5. Social support 6. Personal resources 	Information and acceptance, good family co-operation and social support were related to the coping strategies most frequently used. Half of the families found successful ways of coping, whereas half had major problems. High and low coping families differed most from each other in the following domains: parents' initial experiences; personal characteristics; effects of the child's disability on family life; acting in everyday life; and social support.
Smith, Brewer, Eatough, Stanley, Glendinning & Quarrell (2006), UK	To understand the key features of Juvenile Huntington's Disease for the child as perceived by the chief carer.	<ol style="list-style-type: none"> 1. First becoming aware something is wrong 2. Physical symptoms 3. Speech and communication difficulties 4. Behavioural problems 5. A slow but relentless process 	Suggests that mothers carry a virtual model of Huntington's Disease, and its signs and symptoms, and call on this when other explanations fail. Parents believed that pain impacted on their child's quality of life, in accordance with findings in cerebral palsy research. Ambiguous aetiology of challenging behaviour caused difficulties for parents, due to its inconsistency and inability for parents to normalise their experiences. Management of behavioural problems alongside an associated condition becomes more complex. Lack of knowledge about JHD leads to the assumption that they are on a continuum of normality. The unrelenting deterioration of a child with JHD can have a serious negative impact on parents.

Author, Year & Country	Stated Aims	Themes	Outcomes
Mackey & Goddard (2006) Australia	To understand the health and wellness of mothers who have a child with an ID aged 0-5yrs	<ol style="list-style-type: none"> 1. Primary work in mothering 2. Being on one's own 3. Being constantly tired 4. Ever vigilant 	<p>Mothers' health is backgrounded in time, space and the physical body, because their horizon of awareness is directed toward the needs of the child with a disability</p> <p>Recommendations for healthcare workers to remember that children with IDs are members of a family and that the health and wellbeing of the whole family must be the concern of the health worker. Importance for healthcare workers to grasp every opportunity to evaluate the health of the family and to advise these mothers regarding the maintenance of their own good health.</p>
Kearney & Griffin (2001) Australia	To explore the experiences of parents who have children with a significant developmental disability	<ol style="list-style-type: none"> 1. Sorrow 2. Joy 	<p>In addition to experiencing anguish and sorrow, the parents also spoke of hope, love, strength and joy. Phenomenological interpretation provides insight and understanding into parents' experiences and has implications for practice, education and research in nursing.</p>
Hartley, Okwang, Baguwemu, Ddamulira, Chavuta (2005) Uganda	<p>To increase knowledge and understanding of how family members cope with their disabled children to provide basis for future service development.</p> <ol style="list-style-type: none"> 1. What are the traditional beliefs and practices relating to children with disabilities in three districts of Uganda? 2. What are the traditional coping strategies of these families? 3. How do these practices relate to current Community Based Rehabilitation (CBR) practice? 4. Could these practices be used as a basis for modifying current service delivery thinking? If so, how? 	<ol style="list-style-type: none"> 1. Challenges faced by families 2. Challenges faced by children with disabilities 3. Identified needs 4. Coping strategies 	<p>Extended family systems break down, and the main burden of caring for a disabled family member generally falls on one, sometimes two, female carers. Suggests that CBR programmes should focus their services on the whole family.</p>

Author, Year & Country	Stated Aims	Themes	Outcomes
Fisher & Goodley (2007 UK)	<ol style="list-style-type: none"> 1. To identify enabling principles of care, primarily from the perspective of parents of disabled babies alongside a consideration of professional perspectives 2. To critically examine professional-client relationships re: empowerment; to investigate how meanings of 'impairment' and 'disability' are negotiated and constructed 3. To consider how these meanings impact on the provision of care 	<ol style="list-style-type: none"> 1. Linear narrative 2. Narrative of challenge 3. Philosophy of the present 	Suggests that parents are developing counter-narratives which, at times, resist linear life models and free parents to enjoy their children as they are. Opportunity for parents to develop stories that are neither linear no heroic but present and becoming.

The themes highlighted in Table 2, which are those identified by the authors of the studies, were examined for commonalities. They were then synthesised as themes and sub-themes, in an attempt to draw them together under overarching themes, sometimes taking the names of these from themes within the studies. The emergence of overarching themes, themes and sub-themes were discussed during supervision with a clinical psychologist who has experience of conducting qualitative research. Also, the order in which the overarching themes are presented below was discussed and altered accordingly. Themes were also discussed with a clinical supervisor who has experience of working with children with disabilities and their families.

The final synthesis is presented in Table 3 and each study is presented in parentheses next to appropriate themes/sub-themes.

Table 3: Categorisation of themes pertaining to experiences of raising a child with a disability

<p>Experiences</p> <ul style="list-style-type: none"> • First becoming aware something is wrong (Smith et al., 2006) • Sorrow v joy (Kearney & Griffin, 2001; Smith et al., 2006) • Being constantly tired (Mackey & Goddard, 2006; Hartley et al., 2005) • Being ever vigilant (Mackey & Goddard, 2006) • A slow but relentless process (Smith et al., 2006) <p>Challenges (Fisher & Goodley, 2007)</p> <ul style="list-style-type: none"> • Problems <ul style="list-style-type: none"> ○ Physical symptoms (Smith et al., 2006) ○ Speech and communication difficulties (Smith et al., 2006; Hartley et al., 2005) ○ Impact of disability (Hartley et al., 2005) ○ Behavioural problems (Smith et al., 2006) ○ Philosophy of the present (Fisher & Goodley, 2007) • Challenges faced by families (Hartley et al., 2005) • Challenges faced by children with disabilities (Hartley et al., 2005) <p>Strategies for Coping (Hartley et al., 2005)</p> <ul style="list-style-type: none"> • Personal resources (Täänila et al., 2002) <ul style="list-style-type: none"> ○ Information and acceptance (Täänila et al., 2002) ○ Openness (Täänila et al., 2002) • Emotional activities (Täänila et al., 2002) • Good family co-operation (Täänila et al., 2002) • Seeking a cure (Hartley et al., 2005) • Attitudes (Hartley et al., 2005) <p>Support (Hartley et al., 2005)</p> <ul style="list-style-type: none"> • Social support (Täänila et al., 2002, Hartley et al., 2005)

Experiences

The first overarching theme in this meta-synthesis is ‘experiences’ and is seen as capturing reports of parents’ experiences at the time of diagnosis and since.

Parents in some of these studies reported comparing the abilities of their child when s/he was younger with other children of the same age, and this prompting them to become aware that something was wrong (Smith et al., 2006). It was difficult to pinpoint exactly what, as the

symptoms were subtle, but parents had a feeling that something was wrong (Smith et al., 2006). Parents in these studies spoke of questioning whether their child would have a reduced quality of life, whilst at the same time experiencing the consequences of behavioural problems impacting on their own psychosocial wellbeing, and their physical health. In a majority of studies, parents reported feeling constantly tired, due to these ongoing difficulties. Mackey and Goddard (2006), for example, reported fatigue and tiredness of parents, due to having to be ever vigilant and alert, and lack of respite from the work of caring for a child with a disability. Smith et al. (2006) reported parents feeling that the experience of raising a child is a slow process, moving at its own pace, despite parents' frustrations.

Kearney and Griffin (2001) found that participants' experiences flipped between the two dimensions of sorrow and joy. Along with Smith et al.'s (2006) findings, they reported the realisation of a child's changed potential brought sorrow. Furthermore, feelings of sorrow were exacerbated when participants reported experiencing the negative reaction of others, who were reported as reacting as if there had been a death in the family. Sorrow was also triggered by experiences of the impact of the health system, which left many parents with a sense that they were on their own, leading them to feel vulnerable and powerless.

By contrast, joy was also described by parents, who reported that this arose from a feeling that they are 'better people' and have been 'strengthened', with this being contributed to by their children not dying; the little things their children do; new perspectives following overwhelming changes in personal beliefs and values; and becoming stronger in the face of adversity. This links with Fisher and Goodley's (2007) thoughts about Davies (1997) and Ezzy's (2000) expression of philosophy of the present. The feelings of joy discussed by

Kearney and Griffin (2001) may allow parents to acknowledge that the future is not always amenable to control, leaving them to enjoy the present for what it is.

Challenges

The second overarching theme that has been drawn out of an overview of the results of these studies is 'challenges'. This was used to describe a cluster of themes concerned with adversities that parents described.

Some of the challenges described by parents raising a disabled child included primary concerns relating to themes of physical symptoms, communication and interaction difficulties and behaviour problems (Hartley et al., 2005; Smith et al., 2006). Also there are challenges faced by the family and the individual with a disability (Hartley et al., 2005). These challenges included the burden of care, poverty, and the impact of the child's disability and communication problems. Challenges faced by children, from the point of view of the parents who were interviewed, included isolation and loneliness due to other peoples' negative attitudes. Fisher and Goodley (2007) developed the idea that parents have narratives of challenge, and develop counter narratives so that they can enjoy their children in the present.

Strategies for Coping

Particularly where the focus of articles was on how parents cope with raising a child with a disability, strategies for coping emerged as one of the themes. This was given prominence in a number of the studies and became the third overarching theme of the meta-synthesis.

Parents would draw on their own personal resources, and ensure they were well-informed about their child's diagnosis, and would actively seek information. In non-Western society

this may take the form of taking their child to various traditional or ayurvedic medicine until they found a cure. Parents were able to cope if they felt like they were doing something to improve or even cure their child's disability. Engaging in emotional activities, for example openly expressing feelings and affections was helpful. Improved family co-operation and functioning, and having an attitude that was accepting of disability also improved parents coping skills.

Support

Along with coping strategies, the importance of formal support systems emerged as a prominent theme. Such systems included family and professionals who have helped parents to cope with the experiences and challenges of raising a child with a disability.

A visual representation of parents' journeys through the experience of raising a child with a disability, developed from the overarching themes extracted from the articles within this review, is shown in Figure 1. This highlights the negative experiences and challenges faced by parents. It also details the positive strategies for coping that parents employed and the support that they have within the systems in which they live and work that enable them to continue their journey with their child and the child's disability. Feedback was received from one of the research supervisors, and also a researcher involved in similar research to ensure this model remained close to the themes derived from this review.

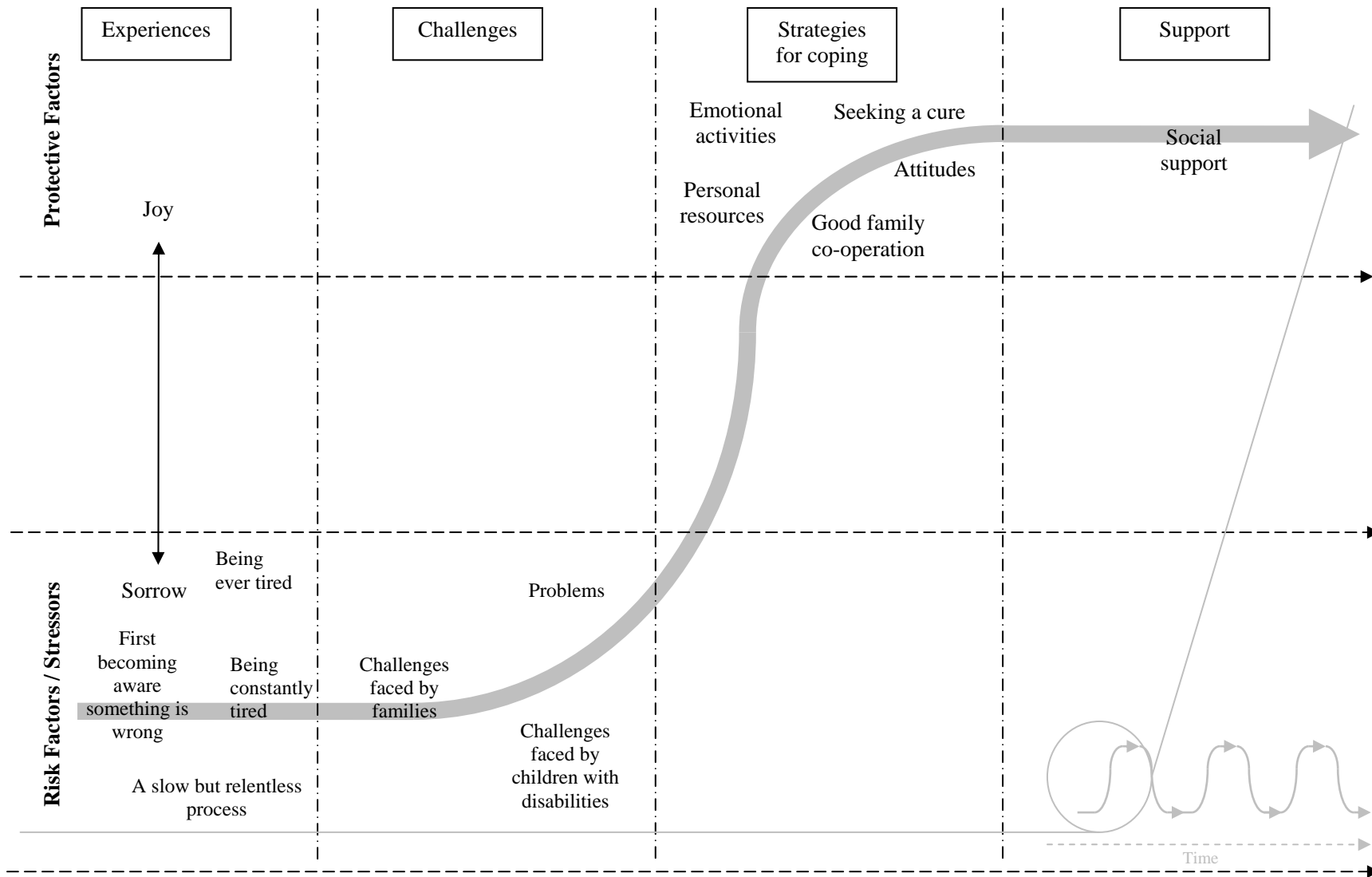


Figure 1 The parent's 'journey' of raising a child with a disability.

Discussion

This review aimed to examine and synthesise research on parents' experiences of raising a child with a physical or intellectual disability with a view to identifying overarching themes and developing a model demonstrating the parents' experience, or 'journey'. One advantage of taking a metasynthetic approach is that a bigger picture can be obtained by drawing together small studies that have had somewhat different foci. In this instance, it is suggested that synthesising studies that focused mainly on problems and challenges, with those that focused on coping and resources, has enabled useful, though tentative, integration of these aspects.

Parents in these studies report that they feel that they go through a number of experiences and challenges prior to receiving a formal diagnosis and these continue to occur throughout the time that they are responsible for the care of their child. Having suitable strategies for coping, and different supportive relationships act as protective factors, and enable parents to enjoy the more positive experiences for what they are, whilst dealing more appropriately with the challenges and stresses that they encounter. As the parents continue their journey with their child, they will be faced with different examples of adversity. This will mean they need to draw upon the protective factors that have helped them before. If these are unsuccessful they will need to re-evaluate their coping strategies and support.

The overarching themes derived from this research, and the subsequent journey that has been suggested correlates with Hill's ABCX family crisis model (1958) and the subsequent expansion made by McCubbin and Patterson (1983) in their Double ABCX model.

McCubbin's Double ABCX model is presented in Figure 2. Hill's model focuses on factors

preceding a crisis that determines a family's capacity to cope with a stressful event, and therefore whether the event develops into a crisis. These factors include family stressors (A) and family crises (X), and protective factors include internal family resources and informal/formal social supports (B) and family perception and parental self-efficacy (C). How families adapt to critical events is shaped by the interaction between family resources and perceptions.

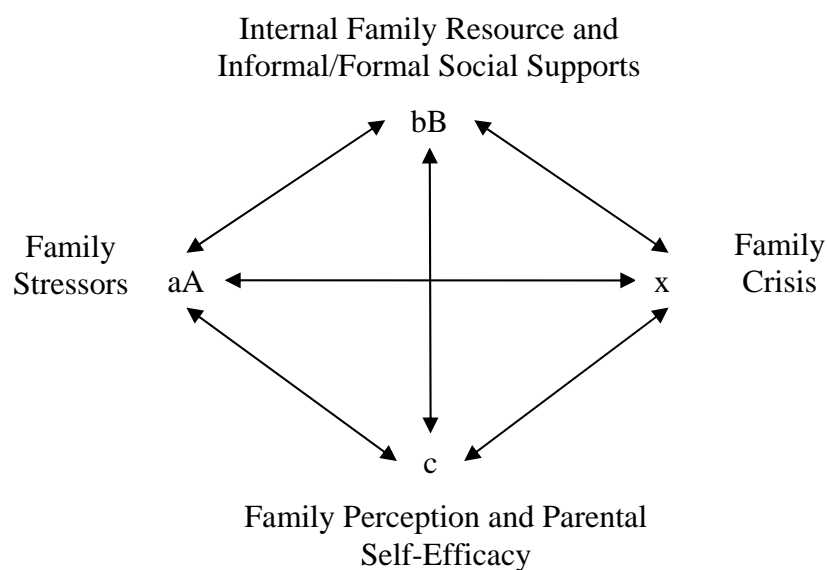


Figure 2. McCubbin's double ABCX model of family stress

McCubbin and Patterson (1983) included four additional factors each corresponding with factors from the original model to develop the Double ABCX Model of family adaptation. These include family crisis (xX), resulting from 'pile-up' of family demands associated with the child and additional life stressors (aA), family adaptive resources applied in order to manage the crisis (bB) and the changes family's make to their definition of the situation in order to understand the situation (cC).

The Double ABCX Model has been influential in the field of developmental disability, suggesting that parents will become stressed as a result of a child's behavioural problems if they do not have the resources to cope. Orr et al. (1991) found that a child's behaviour problems were likely to directly affect parental distress but this effect was partially mediated by parental coping strategies. Pakenham, Samios and Sofronoff (2005) applied the Double ABCX Model to mothers adjusting to caring for a child diagnosed with Asperger Syndrome. They found that better maternal adjustment was related to higher levels of social support and emotional coping; lower levels of child behaviour problems, and a lower number of demands. Bristol (1987) found that mothers of autistic or language impaired children demonstrated positive adaptation depending on the adequacy of social support and active coping strategies. Poorer adaptation was associated with additional family stresses, maternal self-blame and mother's interpreting the disability as a catastrophe. Saloviita, Itälänmäki and Leinonen (2003) used the Double ABCX Model to demonstrate that the most important predictor of stress in parents was a negative perception of the child's disability.

The model proposed in this meta-synthesis, to represent parents' journeys, draws on both risk and protective factors in a similar way to McCubbin et al.'s Double ABCX Model. Parents' journeys include experiences and challenges, which are somewhat similar to McCubbin's aA-xX axis of family stressors and family crisis, whereas strategies for coping and support, are analogous to McCubbin's bB-cC axis of internal family resource and informal/formal social supports, and family perception and parental self-efficacy.

Both of these models take account of factors that may affect the level of stress felt by parents, and account for the continuing array of risk/stress factors experienced by parents whilst fulfilling their care-giving functions.

The parents' journey suggests that parents have a degree of resilience to deal with the experiences and challenges encountered. Patterson (1988) suggests that families engage in active processes to balance family demands and family capabilities. Family demands would include both new stressors and ongoing, insidious tensions, i.e. new behavioural difficulties or the ongoing nature of the disability. Family capabilities include tangible and psychosocial resources and coping behaviours, i.e. what the family has and does.

Limitations of qualitative review

Due to the subjective nature of qualitative research, a review of qualitative research also implies a subjective approach by the researcher. This is moderated by supervision enabling a grounded appraisal of the studies in this review. Another limitation is the small number of studies available for review, which diminished even further due to the quality appraisal.

It is important to note that the majority of studies reviewed do not provide clear information on key variables, for example the age of the child, duration of their diagnosis, and their family composition. This may impact on the quality of each study, due to the different stages that parents may be at within their journey of living with their child's diagnosis. This may also cause problems when making comparisons amongst these studies.

Reflexivity

The notion of completing a qualitative review was a novel idea for the researcher, and as such required background reading into the area. Following this, close communication was sought with a qualitative researcher and Clinical Psychologist who works within an Older Adult Psychology service whilst conducting qualitative research within gerontology. This ensured that throughout this review, the process of developing overarching themes and a subsequent model remained close to the initial studies, and true to the interview data that was available to the researchers.

The model was developed following a synthesis of findings from the studies; however it has been further enhanced by the author's background experiences of working within learning disabilities, and a subsequent awareness of the impact of disability upon parents. Reflecting back on the process of developing the model, it is likely that these experiences have contributed to developing a model that reflects how parents draw upon their protective factors to deal with stressors within their lives in addition to the synthesised themes from this metasynthesis.

Future research

Themes presented in this review, suggest that parents possess both risk factors/stressors and protective factors during the life-cycle of caring for a child with a disability. Much previous research has focused on parents' negative experiences, i.e. the challenges and adversity that they face. It would be interesting for future qualitative research to explore the positive experiences and more protective factors mentioned in this review, for example the

development of personal resources, drawing on family scripts (Byng-Hall, 1998) to find out how these have developed.

Implications for policy and practice

Clinical work within the learning disabilities arena in particular often focuses on assessing families and individuals' objective stressors (behavioural difficulties, activities of daily living, and cognitive impairments), but the research reviewed above highlights that families may also encounter subjective stressors (feelings of exhaustion, loss of intimacy, and being trapped in the caregiver role) and secondary stressors, such as family conflicts. The review also demonstrates that families and individual family members have strengths and resources, yet clinical practice does not often focus upon these.

It would be beneficial for Clinical Psychologists within Learning Disability Community Mental Health Teams, and those with input into paediatric services to encourage families to draw upon their resources to enable better coping strategies in the face of adversity. One approach that may be helpful in this regard would be a systemic family therapy approach that aimed to help family members to mobilise their resources to tackle difficulties, and encouraged them to find constructive ways to help each other (Stratton, 2005). Such approaches aim to make family members more aware of the resources that they have, and how they can employ them to deal with the stressors that they encounter as a result of living with disability. This has the advantage of focusing on the family life-cycle in relation to disability and pays attention to changes over time.

Other opportunities would include providing training for medical staff providing them with the skills to help parents (and other family members) to identify their resources, and support the deployment of these resources in dealing with specific stressors that may arise throughout the life-cycle of their child's disability; or to use a consultation approach within multi-disciplinary teams to encourage psychological mindedness and the identification of resources.

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EMPIRICAL PAPER

**WHAT ARE MOTHERS' EXPERIENCES OF RECEIVING A
DIAGNOSIS OF MONOSOMY 1p36 DELETION
SYNDROME FOR THEIR CHILD?**

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Abstract

This study utilised a qualitative methodology based on Grounded Theory. Data were collected through semi-structured interviews with eight mothers who had received a diagnosis of Monosomy 1p36 Deletion Syndrome for their child. The mothers were asked about their experience of receiving this diagnosis, a rare chromosomal disorder, of which little is known by professionals and parents. An overarching theme of facing uncertainty / reducing uncertainty emerged from the data, and this was connected with five themes: gaining information, being supported / not being supported, being powerless, jumping to the future / living in the here-and-now, and diagnosis making a difference / diagnosis not making a difference. These findings call for more research to be conducted with those who are receiving the diagnosis and/or their carers, whilst ensuring professionals are appropriately skilled to deliver that diagnosis. It is important to draw on the importance placed on delivering a diagnosis within the fields of cancer and dementia, and to highlight the necessity to transfer these skills to professionals delivering a genetic diagnosis.

Introduction

Monosomy 1p36 Deletion Syndrome, or Chromosome 1p36 Deletion Syndrome as it is sometimes referred to, is a terminal deletion of the short arm of chromosome 1. Presenting features of this syndrome include hypotonia and developmental delay, growth abnormalities, craniofacial dysmorphism, cardiac malformations, ventricular dilation, sensorineural hearing loss and ophthalmological anomalies (Slavotinek et al., 1999). The incidence of de novo monosomy 1p36 has been estimated at 1/10,000 newborns (Shapira et al., 1997) making this one of the most common chromosome deletions (Shaffer & Lupski, 2000). In a review of published cases with 1p36 deletions, Slavotinek et al. (1999) found that the most common reasons for chromosome analysis were developmental delay, dysmorphism, seizures, and the investigation of multiple congenital anomalies. Individuals were typically diagnosed between the newborn period or first month of life and 10 years old, with a small minority of patients diagnosed when older than 10 years old. The degree of developmental delay within this review was variable, but it was concluded that intellect is severely compromised in the majority of patients. Due to limited longitudinal research, the prognosis of Monosomy 1p36 Deletion Syndrome is unclear. Information provided by UNIQUE (2008), an organisation for people with rare chromosome disorders, suggests that individuals gradually acquire adaptive behaviours, and can improve their communication skills.

Research into diagnosis focuses on professionals' experiences of giving a diagnosis, or 'breaking bad news', ranging from pre-registration house officers (Schildmann et al., 2005) to consultants (Barnett et al., 2007) rather than patients' experiences of receiving a diagnosis. Findings show that professionals' view giving a diagnosis as a stressful experience, and often feel that they may be blamed by the patient when giving the diagnosis (Buckman, 1992). A

limitation of this research is that the 'breaking bad news' literature focuses on professionals giving a diagnosis of cancer to patients as opposed to other physical health problems and intellectual difficulties.

In spite of the majority of delivering diagnosis literature being centred on the diagnosis or prognosis of cancer, evidence shows that good communication and high satisfaction with an individual's care provider is associated with increased compliance, better emotional adjustment for the patient, and reduced likelihood of litigation (Cameron, 1996; Roberts, Cox, Reintgen et al., 1994). When bad news is delivered poorly, this has a distressing impact on patients' lives (Fallowfield, 1993). As a result of the emphasis on good communication, and the awareness that training is limited amongst health professionals, the UK Cancer Plan (2000) published by the Department of Health stated that 'by 2002 there will be a pre-condition of qualification that (professionals) are able to demonstrate competence in communication with patients. Advanced communication skills training will form part of continuing professional development programmes'. This was an attempt to incorporate communication skills training into medical training, whilst offering the opportunities for medical professionals post qualification to improve and/or develop their communication skills and confidence in delivering a diagnosis.

The bulk of research on delivering diagnoses is concerned with medical staff giving the news to the adult with the illness and there is a lack of research into parents' experiences of receiving a medical diagnosis for their child. It is therefore of little surprise that there is limited research into parents' experience of receiving a diagnosis of genetic disorder for their child. Genetic counselling is available in some geographical areas following genetic testing

and Dinc & Terzioglu (2006) highlighted the importance of genetic counselling both before and after undergoing genetic testing for their child.

Research into genetic syndromes focuses on parents' experiences of raising a child with a genetic syndrome or disability (Abbeduto et al., 2004; Portman, 2004) rather than the process of receiving a diagnosis. In an attempt to address this imbalance, Strehle and Middlemiss (2007) surveyed parents/relatives of children and adults with 4q-syndrome in relation to the medical condition and the interactions between parents and health professionals. Most parents felt distressed at the time of diagnosis, and two thirds of participants complained about the lack of medical information made available to them. Parents spoke of grieving for the loss of their child upon learning of the diagnosis. Some parents felt relief at having an explanation for their child's difficulties, and being able to access services more easily. Participants reported that they would have appreciated it if the diagnosis was given in a personal, sincere, sympathetic and timely fashion.

Cunningham (1984) reported that the way in which parents are told that their child has Down's Syndrome can have a critical effect on their relationship with their child. As a result of their memories of this event, their interactions with professionals are often affected. A report by the Down' Syndrome Association entitled 'He'll never join the army' (Rutter & Seyman, 1999) highlights that although there are guidelines to help medical staff with the difficult task of communicating this diagnosis, 28% of respondents reported a high level of dissatisfaction.

Horwitz & Ellis (2007) aimed to establish the level of support and training available to Paediatric Specialist Registrars in delivering a diagnosis of Down's Syndrome to parents. Of this sample, 15% were aware of guidelines for breaking bad news within their workplace, and 91% had received training. Although individuals' confidence in breaking bad news was high, few had hands-on experience with 21% of respondents having delivered a diagnosis of Down's Syndrome and received feedback on their performance from their seniors. This correlates with research over the last 30 years that has reported a high level of dissatisfaction amongst parents receiving a diagnosis of Down's Syndrome for their child (Pueschel and Murphy, 1976; Murdoch, 1983; Skotko (2005).

Parrington (2002) and Carmichael et al. (1999) explored parents' experiences of receiving a diagnosis of a learning disability and Fragile X for their child, respectively. Both found that the disclosure of a diagnosis continues to be a challenge for health professionals, and a distressing and unsupported experience for the parents. As a result further improvements in communication are paramount if we are to improve parents' experiences of the process of diagnosis.

This research study used a systematic qualitative methodology based on Grounded Theory (Charmaz, 2003; Glaser & Strauss, 1967; Strauss & Corbin, 1998). Given its rarity, parents and professionals are unlikely to have heard about Monosomy 1p36 Deletion Syndrome. In addition, it is an unnamed diagnosis, so parents' and professionals' experiences of the diagnostic process are likely to differ from those involving a diagnosis of Fragile X, Down's Syndrome, Angelman's Syndrome, etc. In light of this, the main aim of the study was to

explore parents' experiences of receiving a diagnosis of Monosomy 1p36 Deletion Syndrome for their child.

Methodology

Design

The study employed a qualitative research design utilising Grounded Theory. Theory is derived from data, systematically gathered and analysed throughout the research process (Strauss & Corbin, 1998). This method of qualitative research ensures a close relationship between data collection, analysis and theory. Theory emerges from the data, and Grounded Theory aims to offer insight, enhance understanding and provide a meaningful guide to action (Strauss and Corbin, 1998). This study employs a constructivist approach in that it draws on the subjective experience of participants to build its theory and as such is not hypothesis driven (Glaser & Strauss, 1967; Henwood, 1996; Willig, 2001) Data were collected through semi-structured interviews with mothers of children diagnosed with Monosomy 1p36 Deletion Syndrome to explore their accounts of their experiences of receiving a diagnosis for their child and the meanings they attached to these experiences. Analysis ran concurrently with the interviews and interview questions altered as theory began to take shape.

Participants

Participant recruitment

Upon receiving ethics approval from the University of Birmingham ethics committee (Appendix 4) participants were identified through UNIQUE, an organisation for people with rare chromosomal disorders. Letters were sent by UNIQUE to 52 parents of children with Monosomy 1p36 deletion syndrome containing an information leaflet about the research and a

consent form (Appendix 5). Parents were invited to take part in the research into developing a phenotype for 1p36, behavioural research and/or this qualitative research.

Out of 48 parents, 24 consented to take part in all aspects of the research, with 19 agreeing to take part upon being contacted. No parents wished to complete the interview only; however, one child was too young to take part in the behavioural research, so his mother completed the interview. Upon reaching the point of saturation, 8 interviews had been completed.

Participant attributes

Interviews were arranged with mothers only, as it was they who offered to take part rather than their husbands or partners. Participants were excluded where English was not their first language, due to financial constraints and the complexities of conducting meaningful Grounded Theory research when translation and interpretation is involved. Table 1 below contains details of each interview participant, and their child.

Table 1: Participant attributes

Participant number	Gender of Child	Location	Age of child at the time of interview	Age of child at the time of diagnosis
1	Female	Married mother of 2, eldest child diagnosed with 1p36, living in a rural area.	3 years old	1 year old
2	Male	Mother of 2 living with partner, eldest child diagnosed with 1p36, living in a urban area	3 years old	10 months old
3	Male	Married mother of 2, youngest child diagnosed with 1p36, living in an urban area	20 months	3 weeks old
4	Male	Married mother of 3, youngest child diagnosed with 1p36, living in a rural area	4 years old	5 months old
5	Female	Mother of 3 living with partner, eldest child diagnosed with 1p36, geographical area unknown	3 years old	10weeks old
6	Female	Divorced mother of 3, middle child diagnosed with 1p36	4 years old	2 years old
7	Female	Married mother with 1 child diagnosed with 1p36, no other children, geographical area unknown	7 years old	3 years old
8	Male	Married mother of 4, youngest child diagnosed with 1p36, living in an urban area	11 years old	3/4 years old

Procedure

Data collection

Pilot interview

The interview technique and schedule (Appendix 6) were refined during a pilot interview with a mother whose son had been diagnosed with Angelman’s Syndrome. The purpose of completing this interview with a parent of a child with a different syndrome was so that if the interview schedule or technique needed refining, then it would not have a direct effect on data that had already been collected. The data from the pilot interview were not used in analysis, however the interview was transcribed (Appendix 7). The pilot interview informed

subsequent interviews by drawing attention to establishing context for the interviews, for example, gaining an impression of what it was like living with 1p36.

Main interview

The interview schedule was semi-structured, so that it enabled 'free-talk' between the interviewer and participants, whilst ensuring that topics deemed relevant were also covered. The specific aim of the interview was to explore parents' experiences of receiving a diagnosis of Monosomy 1p36 Deletion Syndrome, however participants were also asked about their experiences of pre-diagnosis, receiving a diagnosis, and raising a child with 1p36 in order to engage participants and to establish context within the interview in turn allowing 'free-talk'.

Interview procedure

Interviews took place either at the participant's home (participants 1,2,3,4,6 & 8) or their child's school (participant 7), the latter venue being convenient for some as the parent could take part in the research interview whilst their child took part in a linked research project. One parent (participant 5) chose to take part in the interview at a hotel prior to the UNIQUE Annual Conference.

Participants were made aware of their right to withdraw from the research at any point without consequence, and were advised to contact the researcher through Birmingham University should they wish to discuss any concerns.

Interviews lasted between 30 minutes and an hour and a half and were recorded for transcription. An example transcript can be found under Appendix 8.

Data analysis

Analysis was conducted using the qualitative method of Grounded Theory (Charmaz, 1995; Strauss & Corbin, 1998). The eight interviews were transcribed verbatim, and to ensure reflexivity throughout, preliminary analyses were discussed with the supervisors. This informed further data collection, to inform the conceptualisation of an emergent theory, grounded in the experiences of the participants. Analysis of the transcripts occurred using line-by-line and in vivo coding, before progressing to focussed coding. An example of each type of analysis can be found under Appendix 9.

Focussed codes were grouped into themes which were then merged or separated as appropriate, with themes being relabelled throughout this process until they demonstrated the most plausible and useful way of presenting participants' accounts whilst remaining grounded within the data. Grounded Theory ensures a constant comparison of data throughout analysis and allowed the opportunity to return to the data set to further corroborate or negate initial themes. The process of grouping, separating and re-labeling themes was discussed with the research supervisors to enable reflexivity and questioning whilst developing the emerging model.

Researcher's perspective / reflexivity

The researcher was born in England, is engaged to be married, but does not have any children. She became involved in this research due to previous research interests relating to patients' experiences of receiving a diagnosis. Following clinical experiences working with adults and children with learning disabilities, the researcher sought to work with a supervisor whose

research is predominantly within the realm of learning disabilities. This study aims to bring together the researcher's interests and previous academic and clinical experiences.

Reflexivity was ongoing through this study during supervision to challenge any unconscious assumptions and allow creativity to flow. An example of reflexivity can be found under Appendix 10.

Respondent Validation

Respondent Validation can show that there is an agreement between the researcher's analysis and the informant's description. Whilst this can highlight credibility of the research, there are a number of reasons not to include respondent validation. These include whether the informant has enough time to read through the analysis, and the fact that the summary of analysis may not match the account of the individual informant (Bloor, 1997). Also, informants may feel that they are not able to point out anything that they feel is inaccurate. Therefore, it felt important to present the analysis following supervision with a qualitative researcher.

Findings

When exploring parents' experiences of receiving a diagnosis of Monosomy 1p36 deletion syndrome for their child, four main themes emerged from the data (*Wanting information immediately, Jumping to the future / Living in the here and now, Being supported / not being supported, and Being powerless*). Where themes appeared to be dimensional this is demonstrated with the symbol /.

A dimensional overarching theme of *Facing uncertainty / Reducing uncertainty* is present throughout the other main themes and will be discussed in further detail below. A diagrammatic representation of the overarching theme, themes and categories, and possible links is presented in Figure 1.

Given that all the interviews were conducted with mothers, the following findings are related to their accounts of their experience, and their thoughts about what their partners may have been experiencing. All names appearing within the quotations used and the transcripts have been changed to protect the identities of both mother and child.

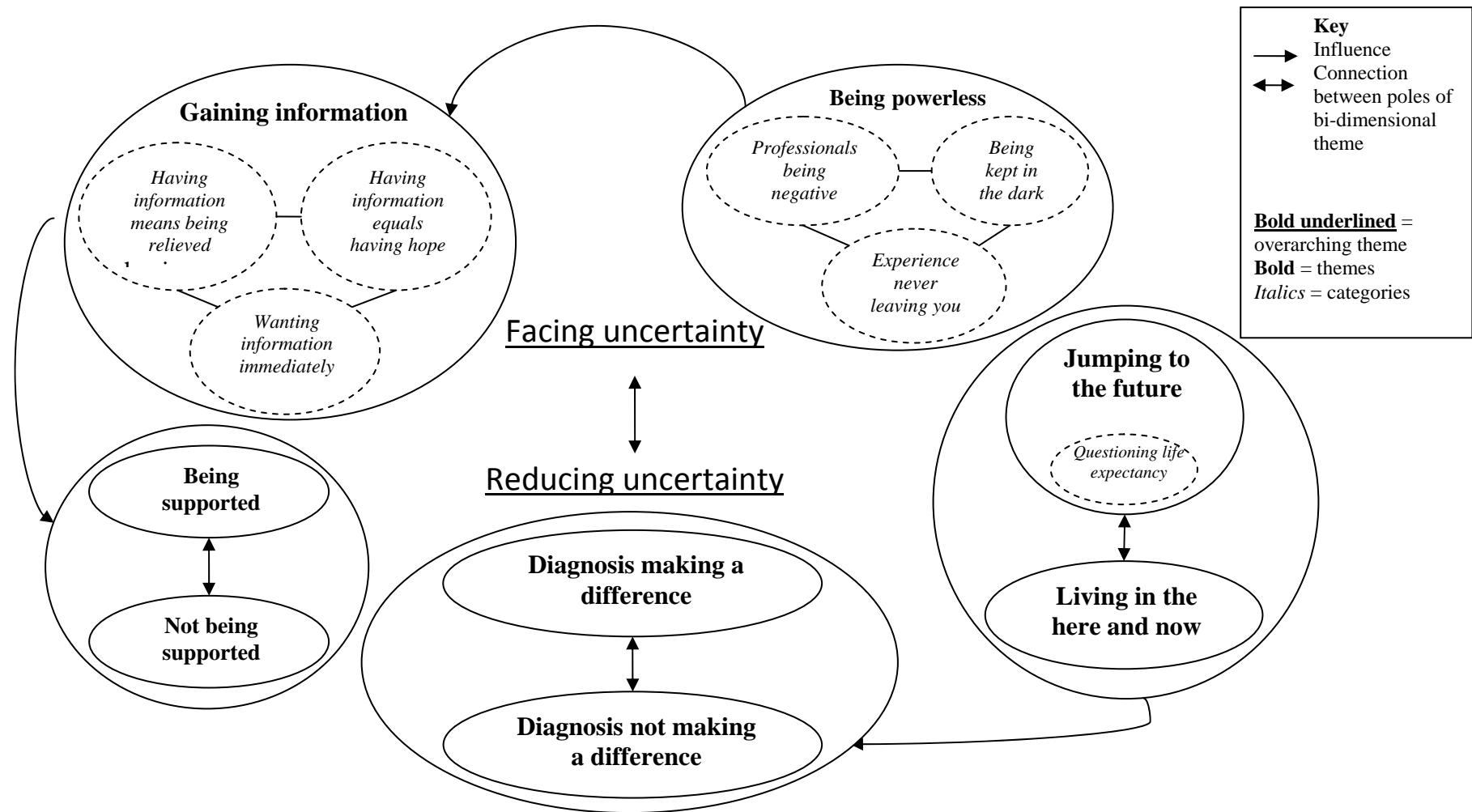


Figure 1. A diagrammatic representation of the overarching theme, themes and categories

Gaining information

Wanting information immediately

The mothers felt that professionals should know what was wrong with their child because of the expert position they held, and as such were looking to them for information at the time they were told about their child's diagnosis. However, participants felt that they were signposted to other sources of information, as a result of professionals' lack of knowledge. They would have preferred for that information to have already been obtained by the professional, so that they could discuss it with them. Due to professionals' lack of knowledge, they were left with a degree of uncertainty about their child's condition and wanted more information at the time of diagnosis.

... Yeah I suppose it was difficult, cos you want, you want the professional to give you all the information [Interview 6: 189-190]

I were a bit scared, sometimes I'd think god, if even these people haven't heard of it and don't know I think god, I feel a bit worried and a bit scared [Interview 5: 544-545]

Having information equals having hope

There was a feeling that whilst participants would have liked more information, receiving the information they did was enough for them to feel that they had some hope in relation to their child. In particular, parents had gone through a period of time of not knowing anything, therefore to know something rather than nothing was a better position to be in:

Having a a label I suppose in some respects helped because it felt like this sort of wilderness we'd sort of been living in for three weeks where we had no idea what was going on ... it felt like we were now going to be able to start to find out a bit more about what was going on [Interview 3: 271-274]

One mother commented that having some information, no matter how negative this information is, would have helped to reduce any uncertainty, again feeling that this would have put her in a better position.

[Yeah] if they'd said anything I think it would have been better than nothing wouldn't it, we were left just wondering ... what .. on earth was going on ... because he did have, it was a very difficult birth I was left there erm and he had to be induced and even though I was induced he still didn't come out ... erm he wasn't it wasn't difficult but I was left on my own in the hospital because there were no staff, everybody was having their babies so I was just left there and erm in the end, she had to rush a doctor in and "what for" and you just wonder, was it that, was it the fact that I was left for so long with no monitors and no I don't know. There's nothing worse than having uncertainty or doubt, it's better to know the worst than it is to know nothing. [Interview 8: 471-478]

Having understood some of the information, participants had something to hold on to and hope that things would be alright for their child.

Yeah so we were constantly clinging onto maybe it was just a sliver, maybe it was just a slice off the top you know, the minutist and we were gonna get away with... you know... [Interview 4:271-274]

Well I suppose you don't have the, again you've got some level of hope haven't you [Interview 7: 184]

This category fits with the dimension of reducing uncertainty in that holding some knowledge was enough to reduce any uncertainty and anxiety about not knowing anything about their child's condition.

Having information means being relieved

There were two aspects to being relieved, in that relief derived from having information in itself, but also this information seemed to help to remove any feelings of self-blame or enable participants to counteract blame from others. Both of these aspects seemed to lead to a further reduction in uncertainty.

I think we were just relieved that we knew, that at least we knew something and some/there was some help out there, rather than he's ... we don't know what's wrong with your child, which is it means everything over [Interview 8: 457-459]

And also relief to know what was actually wrong rather than not knowing [Interview 2: 103-104]

umm, in some respects it was a bit of a relief because you know we knew what it was then [Interview 3: 271-272]

I did worry that maybe the altitude at some point, although I was altitude acclimatised for the whole time before being pregnant I did worry that that might have an effect, and maybe lack of oxygen to the brain and then my husband was worried was it you know, we gave birth in the hospital which was fine, but was there something wrong there, and then my parents were very ... emotionally difficult, I think because they blamed us, they blamed him it's his fault it must be his family that had a problem, so once we'd got the diagnosis and finding out that it was 'de novo' which is out of the blue [... ..] then it gave us the strength it took two of us to turn round to my parents and say actually it's got nothing to do with, we couldn't have done anything else this is just out of the blue, that was then, in some ways it was a relief, erm it gave us a line to draw so that we weren't going to have to take anymore flak from anybody blaming us. [Interview 7:137-146]

Well it helps me, because it clears up... it clears up that it wasn't a birth problem, we knew it was genetic so we was looking through families trying to find, and we had blood tests ourselves, to see if there was anything that was linked to either side of the family [Interview 8: 131-133]

Being supported / Not being supported

Participants reported that whilst more information was not immediately available for them, professionals were supportive in their approach, for example signposting them to

UNIQUE, an organisation for people with rare chromosome disorders, or researching more about the condition.

She gave me a leaflet or told me about the UNIQUE website, errm so sort of said “you know you probably need to contact them, get your information” [Interview 6: 184-186]

Yeah you know the doctor wasn't aware of the actual condition but he did research, he did do it and find some books for me to look at as well [Interview 2: 213-214]

There was also a feeling amongst these mothers that their consultants were willing to make time for them, in order to answer any questions that they may have once they had digested the initial diagnosis, which gave a greater feeling of being supported by professionals involved in their child's care.

She did say if you've got any questions pick up the phone any time and I'll answer you, but we didn't have an actual appointment, to go back [Interview 6:326-327]

We just looked totally stunned I think because we then the consultant said look you know, we left quite quickly, and then he rang me that afternoon he's a really, he's a really nice guy he said “look you know I appreciate you're probably thinking of a million and one questions come back tomorrow and we'll, when you've had time to digest the information and we'll talk” [Interview 1: 115-119]

She said to go home and to make a list of questions for the geneticist she said the geneticist would have more information for us [Interview 3: 313-314]

One parent in particular commented on how they felt supported enough by their consultant to ask what may have come across as controversial questions about their son and his condition.

Fantastic, absolutely brilliant she gave us the impression she had all the time in the world, she answered all of our questions honestly we asked some quite politically incorrect questions like, things like what Josh would look like and she didn't I didn't feel she judged us for asking those questions and she felt that we justified in asking [Interview 2:336-339]

Contrary to being supported, some mothers felt that they had not been supported when they received a diagnosis for their child. They commented on other professionals (student doctors) being present when they learned of the diagnosis and the impact that this had upon them, leaving them feeling subjected to prying eyes, that their child was an object rather than a human being, and that they [mothers] felt invisible during the process.

Yeah I was cross, that he felt, he was able to sit there as though my son was just something to look at, "Oh look at that, see this, look at the bridge of his nose" and he was talking about him like he was this object of interest you know, and I didn't like that very much [Interview 8: 85-87]

I felt really mad with her, I were really mad that there were loads of peop/well it were like three doctors there I couldn't understand why it couldn't have just been the one doctor why she couldn't have just told me do you know, I felt like, I felt like Leanne were not a freak show, that sounds awful but like they all wanted to be in the room, they were student doctors and they never asked me [Interview 5:161-164]

Being powerless

It became apparent that the experience of receiving a diagnosis of 1p36 for their children has been a powerless experience for these mothers, particularly enhanced by the negativity of professionals when disclosing the diagnosis, but also with some parents feeling that they were kept in the dark by professionals, further contributing to parents facing uncertainty.

Professionals being negative

There seemed to be a feeling, amongst the mothers interviewed, that professionals had given up hope on their child in that they painted a bleak picture of the child's future.

She said ... a lot of the children, which I don't know how she means this or maybe this is just what was kept from us, erm, she said these children are institutionalised at a younger age than others, because of their behaviour, which again was ... not the sort of thing you want to hear. [Interview 8: 233-236]

She actually said it in a way ... that made it out that it was going to be a bad thing
[Interview 6: 177-178]

It was quite negative that very first meeting it was all very kind of well he won't do this he won't do that, not really not very specific though but it was very erm, it was very kind of well he won't walk he won't talk [Interview 3: 298-300]

The mothers would have appreciated it if the professionals had been more tentative in explaining what their child's future and quality of life may look like. Some felt that professionals could have handled the situation differently, in particular the way they initially delivered the diagnosis.

Maybe I suppose just the way she said it but then there's no point I think giving people false hope so, but maybe... ... if she hadn't have said it quite like she had in terms of I've got some bad news, maybe if she'd said "Look, we've looked into Alison's chromosomes, yes she has got deletions, it is gonna mean" then maybe that would have been [better] [Interview 6: 317-320]

I think, I suppose it could be a bit nicer to say she might not do this and she might not do that [Interview 5: 649-650]

Furthermore, one mother in particular was left feeling unsupported during the diagnosis period due to professionals' negativity.

As a parent and to have people who are not even positive around you it just, it's so depressing. If the doctor can't even say anything positive you just, you feel so isolated and so "well who do I go to then in terms of help" [Interview 8: 173-176]

Experience never leaving you

This is a category within *professionals being negative*, as it seems that in recalling their experience of receiving a diagnosis these mothers highlighted specific negative comments as those that would remain with them throughout their lives, and the lives of their children.

I just remember the doctor saying to me "She might not errm laugh, she might not hold a spoon, she might not sit up" I just remember, I still think that now, I remember what, cos she does most of that now [Interview 5:188-190]

Errm, I just think like some of the doctors need to know that you have that memory forever don't you and that's whenever, whenever someone asks what is it again, what Leannes's got I always think of that, and I always say to people well they said she'd never do this and they said she wouldn't have any quality/they didn't she wouldn't have any quality of life, but she might not, I always think back to that [Interview 5:641-645]

In addition, some mothers recalled how they knew that what they were going to hear was bad, because they were called to a meeting in a specific room, at a specific time.

Yeah... erm ... we got called into the green room which we'd learnt to dread erm

[Interview 3: 252]

I just remember being sat in that room [Interview 5: 118]

Then we had a telephone call about 10 weeks later from the hospital to say the consultant wanted to speak with us, this was the secretary "the consultant would like to talk to you and we'll book a room" "Book a room, why do you need to 'book a room' just to talk to us" we knew then that, it was a set time, 5 o'clock we'd got to be there at 5 o'clock. Room A that had been booked, we knew then that something, they were gonna tell us something bad. You don't 'book a room'

[Interview 4: 74-79]

Being kept in the dark

Another aspect of being powerless was the idea held by some of the participants that information was being kept from them about what was wrong with their child. Some participants were aware that there was something wrong with their child but recalled feeling that professionals were privy to information and were withholding this from them.

Yeah, well there was I think, I would imagine I'm not the only parent who, the information about 1p36 was kept from me... for a long time [Interview 8: 21-22]

Even now I still think that doctor... it sounds awful actually but that she went to bed that night knowing... and I.. and that night, the night before I were told and that night were like I were totally oblivious, that she carried that, that she had to tell me that next day, does that make sense? [Interview 5:145-147]

Said that they'd found that she'd got this, but I think it must have just been a process of meeting Alison and then taking the pictures and then ... cos it does seem like a long time a year really for them to actually, fathom it out [Interview 6: 149-151]

Jumping to the future / Living in the here and now

This theme also connects with the idea that mothers face uncertainty upon receiving a diagnosis of 1p36 for their child, in that all the mothers talked about questioning the professionals about their child's future and the prognosis of 1p36. There was an immediate question of what they could expect for their child in the future, in terms of his/her abilities, in comparison to a healthy child, and many immediately started to envisage what the long term future might look like.

Errm, that Alison's just gonna be I suppose dependent on me for the rest of her life, errm and that she's gonna ... errm... yeah not lead an independent life and not be able to, you know make friends and not .. not be able to sort of have her own home and have her own family and ... those sorts of things really, so [Interview 6: 208-211]

That, that is really frightening, the thought that we'll be going on holiday in you know in 20 years time with an adult with special needs, and Dave said, my husband "oh well I don't want him to be a burden to his sisters" err... [Interview 4: 660-662]

I think it was just is she ever gonna be... erm... yeah, how's she gonna be, is she gonna be like my son, is she gonna you know do the things she should do and maybe lead an independent life when she's older [Interview 6: 200-202]

Participants commented on how they thought their partners/husbands reacted to the diagnosis of 1p36 in that they thought they immediately jumped to the future.

I remember my husband asking her erm well, you know, my son's five, I think he must have been five at the time, he said, is she ever going to get to the age of like, he is as a five year, and she said "Well it's really difficult to say, because a five year old can actually be quite independent and quite you know" and she said "I don't know that Alison's ever going to get to that stage [Interview 6: 192-196]

Steve was very worried about... well how do we communicate with this child you know and erm how how, very quite long term things like you know, like school and ... lifting all of, and he instantly thought about like when Josh was five or six or whatever [Interview 3: 278-280]

Questioning life expectancy

Little is known about 1p36, in particular relating to life expectancy, and as such several of the mothers reported that both they and their partners or husbands questioned professionals about how long their child would be expected to live for.

Errm we did ask at that first meeting we sort of asked about life expectancy errm and again were told probably twenty something if we're lucky [Interview 3:297-298]

To be fair we initially for some reason and I don't know why, it never clicked with me, that because of this she was going to have a limited life, it just never really entered my head, I don't know why. So immediately you think what's going to happen, certainly think what's happen when I'm not here, that was the first thing that I thought about [Interview 1: 368-372]

Yeah I suppose, yeah I don't think I ever thought about oh is she gonna die early or anything like that [Interview 6: 199-200]

Whilst mothers reported that both they and their partners/husbands had questions about the future, the mothers spoke of living in the here and now as a more helpful strategy, choosing to focus on specific issues as they arose.

You can think well actually we don't know what's going to happen so you don't have to worry about what's round the corner, I've stopped, I must admit, since I've got the diagnosis, when she was very very small, when she was four months

old I stopped thinking more than like a school term ahead or I'm very short term now [Interview 7: 184-188]

In addition, there was the idea that by living in the here and now, this prevented them from experiencing any negative emotions in relation to what the future holds for their child, and they were able to keep hold of the idea that parenting a baby with 1p36 may not be any different to parenting a healthy baby.

Whereas I was probably thinking more here and now, I, and even to this day I don't like to think too far ahead because that just scares me (laughter) [Interview 6: 204-206]

No no, we don't think about that, it's bad enough thinking about what's going to do at school next year (laughter) [Interview 2: 261-262]

I think from my point of view whatever, I think as a mum when you have a baby, you're looking forward to cuddles and feeding and clothes and baths and all of that stuff and none of that's ever been taken away from me, that was still gonna happen you know so I can remember saying to Alison who's the neonatologist, I remember saying "Well you know ok he's gonna have problems, but for the first year of his life is he really gonna be any different to any other baby?" [Interview 3: 280-285]

Diagnosis not making a difference / Diagnosis making a difference

Experiences were different across the interviews with some parents feeling that it did not matter what the diagnosis was because they were able to deal with each aspect of their child's condition without having an overall label for it.

Errm, I suppose it is concerning, but it wasn't the fact that, if you had taken away the title of his condition the doctors would have been able to deal with what he's got [Interview 2: 210-211]

Yes I would say that's probably how, probably how I see things and how I deal with things, that it's the specific issues Alison has, that she's not walking, she's not talking you know, errm [Interview 6:533-534]

In some cases, parents had already lived with the condition for some years prior to receiving an official diagnosis, so felt that it did not make a difference to them.

To me I suppose, I don't class it as 1p36, I suppose to me it's just Alison who's got disability, because Alison errm I don't know how, because it affects children in so different ways, I, it's not that I don't want a label on it or anything [Interview 6: 521-523]

Err, I don't think it made much difference either one way or the other because by then I was mixing with other parents whose children had a diagnosis and whose children were quite different so it was a very... loose term anyway, even if with a

diagnosis, isn't it, autistic doesn't mean they're all going to be/you know, there's such a big range in a way [Interview 8: 121-124]

Two mothers spoke about how they felt that the diagnosis did make a difference, and in both cases this was due to having family members who were pregnant, and having the knowledge that this was not something that would affect them too.

Yes definitely, it was a practical decision and the other thing that hurried it along was my cousin, my first cousin was pregnant and I knew I've got a lot of cousins who are all, we're all a similar age and wanted children so it wasn't just me that because it's often maternal not paternal errm you know there were wider implications for other family members so right... you know, we'll find out now and then we can make that decision [Interview 1: 208-212]

It was nice to know what was wrong, and my sister was pregnant at the time, so it was nice to know obviously it wasn't hereditary you know, was it going to happen again to her [Interview 2:155-156]

Discussion

The aim of this study was to explore parents' experiences of receiving a diagnosis of Monosomy 1p36 Deletion Syndrome for their child. Analysis of interview data using Grounded Theory methods led to a dimensional over-arching theme of Facing Uncertainty / Reducing Uncertainty and five main categories: Gaining Information,

Being Powerless, Being Supported, Jumping to the Future / Living in the Here and Now, and Diagnosis not making a difference / Diagnosis making a difference.

Little is known about Monosomy 1p36 Deletion Syndrome other than the point at which the translocation can be found. This lack of knowledge amongst professionals, and the lack of accessible literature seemed to leave these mothers facing uncertainty upon learning of this terminal condition for their child. They often feel in a powerless position in comparison to health professionals who are delivering the diagnosis. This was only lessened when uncertainty was reduced through gaining information, and power relations were made a little more even when professionals gave the mother the sense of being supported at the time of diagnosis. Participants sought answers to questions about their child's abilities, life expectancy and the prognosis. Uncertainty was exacerbated by mothers' questions about the future for their child, whilst living in the here and now served as a strategy to reduce this uncertainty. Having a diagnosis appears to have had a dimensional effect in that for some it made a difference, meaning they could access services and could obtain more information in relation as to whether it is hereditary, whereas others felt that they were able to deal with each aspect of the condition regardless of the diagnosis.

This study corroborates findings by Starke and Möller (2002) who explored parents' experiences of information given during the diagnosis of Turner Syndrome for their daughters. Twenty three out of forty four parents were dissatisfied with the information provided by the physician. Participants then went on to seek further information, further supporting Seligman and Darling's (1997) finding that a strategy used by parents

to handle their reactions of having a child with a dysfunction is to seek information and gain knowledge about the diagnosis.

‘Tell it Right, Start it Right’ a report by the Down’s Syndrome Association (2009) highlights findings by Down’s Syndrome Association and The Foundation for People with Learning Disabilities (2009). This report revealed huge disparities in the quality of information and support for parents when they receive a diagnosis of Down’s Syndrome for their child. Some parents report supportive and knowledgeable healthcare professionals, whereas other report being given negative and often confusing information. Further findings from Skotko (2005) corroborated this report, highlighting mothers’ reports of factually inaccurate information, the negativity of professionals. Mothers who felt optimistic about the future had received positive information about Down’s syndrome. As this was a longitudinal study, it is important to acknowledge the difference in the use of the term ‘mongoloid’; there was greater use of this term during the earlier stages of this research compared with more recent participants’ more positive experiences. This further highlights the attitudes of health care professionals, who have used derogatory language to describe children with Down’s Syndrome, and the imposition of their opinions regarding mother’s choices to have their child, despite the results of screening tests.

The findings of the current study, corroborate previous research with respect to mothers’ wanting more information about their child’s condition, and feeling powerless as a result of the professionals’ negativity about their child’s diagnosis. Skotko’s (2004) research voiced the requests of parents for clearer guidelines outlining a number

of changes to be made to the process of delivering a diagnosis of Down's Syndrome, adding further weight to the findings of this study, in so much as mother's were requesting more information, the use of sensitive and compassionate language, providing accurate, factual and up-to-date information immediately, and signposting mothers to parenting support groups.

Within the cancer arena, communication has been of paramount importance for some time, with Fallowfield et al. (1998) and Maguire (1999) highlighting the lack of formal training for oncologists. Bruera et al. (2001, cited in Fallowfield et al., 2003) found that poor communication can leave patients feeling uncertain about their diagnosis and prognosis, unsure about further management plans or the therapeutic intent of treatment. Fallowfield et al. (1995) argue that 'honest, positive communication about what is attainable allows the maintenance of hope, whereas avoidance, evasion and secrecy may contribute to a sense of hopelessness' (p.201). Individuals receiving a diagnosis of cancer showed a strong need to receive any information, whether this was positive or negative. This is consistent with the findings from this study in that mothers of children with Monosomy 1p36 Deletion Syndrome sought to gain information about their child's condition during the diagnosis consultation.

Schildmann et al. (2005) reported that pre-registration house officers breaking bad news identified that they found it difficult to deal with the emotions of the patient and of their relatives. These newly qualified doctors found it hard to have all the relevant information available, and thought that they were not the appropriate person to discuss the bad news. They found it difficult to handle the uncertainty in the situation. The

findings of the current study further enhance understanding around the process of giving/receiving a diagnosis.

Current literature around delivering a diagnosis focuses on professionals' experiences from specialist registrars (Schildmann et al., 2005) to consultants (Barnett et al., 2007; Buckman, 1992) as opposed to experiences of those receiving the diagnosis. Over the last fifteen years, research within the area of diagnosis disclosure in dementia has focused on the experiences of carers of people with dementia (Holroyd, Snustad and Chalifoux, 1996; Maguire et al., 1996; Rice and Warner, 1994). There has been a shift within this field, with research exploring the experiences of people with dementia about diagnosis (Husband, 1999; Pratt and Wilkinson, 2001). The main difficulties associated with delivering a diagnosis to an individual with dementia are around whether it is in the individual's best interests not to be informed of the diagnosis and prognosis.

Professionals report the experience of delivering a diagnosis as a stressful time. It seems that parents want empathy and warmth when they receive information to allow them a margin of hope, but professionals seem to be unsure of what they need to say in order to satisfy parents' needs. As a result, parents perceive professionals as being negative, when research implies that actually they are stressed and uncertain about how to manage the situation. When professionals do not give enough information from the mothers' point of view, this leaves them with increased uncertainty, a sense that professionals may be covering up the truth and a sense of urgency for professionals to provide answers to their remaining questions.

The NICE guidelines for cancer and palliative care highlight that health and social care professionals should have the skills to communicate effectively with patients and carers. Those who must communicate particularly complex or distressing information should have enhanced skills or be supported by someone who has those skills. It is important that this information does not stay within communicating bad news in a cancer setting. Findings from extensive research exploring experiences of delivering a diagnosis and receiving a diagnosis should be transferable to other clinical settings, for example delivering a diagnosis following genetic testing. Evidence from Starke and Möller (2002) suggests that parents perceived doctors to be incapable of explaining their child's condition due to having little or no knowledge about the condition. Perhaps if professionals were able to transfer the communication skills outlined in the NICE guidelines for cancer and palliative care, this may removed some of the dissatisfaction associated with mother's sense that professionals know little or nothing about a child's condition, by showing empathy and sensitivity within their communication style.

Limitations of the study

This study aimed to explore parents' experiences of receiving a diagnosis of Monosomy 1p36 Deletion Syndrome for their child, however, no fathers opted to take part in the interviews. Reasons for this could be a result of the health care needs of a child with 1p36. The mothers in this study have opted to take the position of 'primary caregiver' for the child, meaning that fathers may have been the sole earner in the family, and thus not available to complete the interview. Having only one parental perspective can be both a strength and a weakness, however in terms of weaknesses, this meant that

reporting was based on the mothers' ideas about what their partners were experiencing, without an opportunity to corroborate this with fathers of children with 1p36.

All of the participants taking part in this research were of White British origin, thus this research may not be applicable to parents from a minority ethnic background who have a child with 1p36. One father was of Nepalese decent, and another was Asian, however they did not opt to take part in this research, so these possibly culturally different experiences were lost.

It is important to note the length of time that had passed between the initial diagnosis and the interview. The shortest time between diagnosis and interview was approximately 1 year and 8 months, compared to the longest time between diagnosis and interview being 7 or 8 years. Where the child had been diagnosed more than a couple of years ago it brings in to question the accuracy of mothers' memories of the process of receiving a diagnosis for their child i.e. the accuracy of their episodic memory and autobiographical memories.

In addition, in order to engage participants and bring some context to the interviews, the interviews did not solely focus on mothers' experiences of receiving a diagnosis. Other topics covered included the point at which the mothers or health professionals realised that 'there was something wrong' (i.e. prior to diagnosis), what mothers did after receiving the diagnosis, for example accessing the internet, and their experiences of raising a child with 1p36. Due to the focus of this particular piece of research, this has not been reported.

Future research

Given the limitations above it would be beneficial to obtain fathers' experiences of receiving a diagnosis of a rare genetic condition for their child. This is of particular importance because mothers in this study have reported that fathers 'jump to the future', questioning what the diagnosis will mean for them and their son/daughter in the long run. In addition, it would be interesting to see what themes would emerge if interviews were conducted with both parents, either together or separately.

Another possibility would be to undertake trials with parents receiving information and diagnosis. Professionals could receive further training relating to delivering a diagnosis and information giving, and parents' experiences could be followed up with an interview. These experiences could be compared with parents who have received a diagnosis from a professional who had not attended further training.

Implications for practice

NICE guidelines and the UK Cancer Plan highlight the necessity for pre-qualification training in relation to breaking bad news and delivering a diagnosis. This idea should reach across delivering a diagnosis, not just within cancer. Horwitz and Ellis (2007) conducted research with Paediatric Specialist Registrars based within the UK, revealing that only 15% were aware of guidelines within their workplace regarding delivering a diagnosis of Down's Syndrome. The Down's Syndrome Association is currently campaigning to the British Government for clearer guidelines for professionals when delivering a diagnosis to parents. This follows US Legislation stating clear guidelines,

including up-to-date, evidence based written information, and contact information regarding support services.

This Communication is paramount to the psychological well-being and adjustment of parents, and subsequently their children upon learning of a diagnosis. As psychologists, there is scope to be involved in the training and supervision of those delivering a diagnosis. With a greater Clinical Psychology presence within a Paediatric setting, it would be possible to offer training and consultation to ensure staff has opportunities to develop communication skills and the ability to deliver diagnoses to parents and children.

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EXECUTIVE SUMMARY

EXECUTIVE SUMMARY

Research into parents' experiences of raising a child with a disability reports a variety of experiences, most notably recurring themes of stress and burden are reported, alongside parents' more positive experiences. A literature review has been conducted presenting the findings from a series of research projects exploring parents' experiences of raising a child with a physical or intellectual disability. Four main themes emerged from these studies which included both risk factors and protective factors. Risk factors included the experiences and challenges facing parents, and the protective factors represented the strategies for coping and support that they used to face these risk factors. These themes are presented visually as a parent's journey through life raising a disabled child. This journey suggests that parents face instances of adversity throughout the time that they spend raising and caring for their child and this causes them to draw on their resources and show resilience in dealing with these negative experiences and challenges. These findings suggest that professionals need to ensure that they are aware of both the risk factors faced by families and the protective factors they have. Professionals can then enable parents to draw on these resources to deal with negative experiences and challenges faced by parents throughout their child's lifetime. Clinical Psychology can play a part in providing training for professionals working with disabled children and their families, to encourage discussions around resources and other protective factors.

Connected with parents' experiences of raising a child with a disability is parents' experiences of receiving a diagnosis for their child. In this instance parents receiving a diagnosis of Monosomy 1p36 Deletion Syndrome (1p36), a rare genetic condition. A series of interviews were conducted with mothers of child who had been diagnosed with 1p36, of which little is

known. Themes of stress and burden present within the literature review can be linked with findings of this piece of research. The lack of knowledge amongst professionals and the lack of accessible information seemed to leave mothers facing uncertainty upon learning of this terminal condition for their child. It was apparent that how professionals delivered the diagnosis, and subsequent support available impacts on how mothers' felt, with some feeling powerless, and unsupported, whereas others felt that they had the support of the health professional, and the opportunity to ask more questions. Mothers wanted to know about their child's abilities, life expectancy and the prognosis of the condition, much of this being unknown by professionals. In particular, uncertainty was increased through the lack of knowledge about what a child's future would hold as a result of the diagnosis, whilst this uncertainty was reduced by choosing to focus on living in the here and now, a helpful strategy for mothers. Having an official diagnosis for their child had a mixed effect, with some feeling that it made a difference from a practical point of view, i.e. accessing services and obtaining more information, whereas others felt that they were able to deal with each aspect of the condition regardless of the diagnosis. In accordance with suggestions from the literature review, Clinical Psychology can provide training for health professionals and ongoing supervision regarding the experience of delivering a diagnosis, in particular, when little is known about that diagnosis.

Appendices

Appendix 1

Instruction to Authors

Appendices

APPENDIX 2
ARTICLES EXCLUDED AT STAGE 2

J Bromley, DJ Hare, K Davison & Eric Emerson (2004) Mothers supporting children with autistic spectrum disorders *Autism* 8(4): 409-423

QUANTITATIVE QUESTIONNAIRES COMPLETED DURING AN INTERVIEW, not a QUALITATIVE INTERVIEW

A Fitzpatrick & Dowling (2007) Supporting parents caring for a child with a learning disability. *Nursing Standard* 22(14-16): 35-39

A REVIEW OF LITERATURE BUT NOT A SYSTEMATIC REVIEW

J Carr (2005) Families of 30-35-year olds with Down's Syndrome. *Journal of Applied Research in Intellectual Disabilities* 18:75-84.

QUANTITATIVE QUESTIONNAIRES COMPLETED DURING AN INTERVIEW, not a QUALITATIVE INTERVIEW

A McCarthy, M Cuskelly, CE van Kraayenoord, J Cohen (2006). Predictors of stress in mothers and fathers of children with fragile X syndrome. *Research in Developmental Disabilities* 27: 688-704

INTERVIEWS CONDUCTED TO ADMINISTER QUANTITATIVE QUESTIONNAIRES
- NO MENTION OF SEMI/STRUCTURED INTERVIEWS AND NO QUOTES OFFERED

Appendices

APPENDIX 3

CRITICAL APPRAISAL SKILLS PROGRAMME (CASP)

Critical Appraisal Skills Programme (CASP)
making sense of evidence

10 questions to help you make sense of qualitative research

This assessment tool has been developed for those unfamiliar with qualitative research and its theoretical perspectives. This tool presents a number of questions that deal very broadly with some of the principles or assumptions that characterise qualitative research. It is *not a definitive guide* and extensive further reading is recommended.

How to use this appraisal tool

Three broad issues need to be considered when appraising the report of qualitative research:

- **Rigour: has a thorough and appropriate approach been applied to**
- **key research methods in the study?**
- **Credibility: are the findings well presented and meaningful?**
- **Relevance: how useful are the findings to you and your organisation?**

The 10 questions on the following pages are designed to help you think about these issues systematically.

The first two questions are screening questions and can be answered quickly. If the answer to both is “yes”, it is worth proceeding with the remaining questions.

A number of italicised prompts are given after each question. These are designed to remind you why the question is important. Record your reasons for your answers in the spaces provided.

The 10 questions have been developed by the national CASP collaboration for qualitative methodologies.

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Screening Questions

1. Was there a clear statement of the aims of the research? Yes No

Consider:

- what the goal of the research was
- why it is important
- its relevance

2. Is a qualitative methodology appropriate? Yes No

Consider:

- if the research seeks to interpret or illuminate the actions and/or subjective experiences of research participants

Is it worth continuing?

Detailed questions

Appropriate research design

3. Was the research design appropriate to address the aims of the research? Write comments here

Consider:

- if the researcher has justified the research design (e.g. have they discussed how they decided which methods to use?)

Sampling

4. Was the recruitment strategy appropriate to the aims of the research? Write comments here

Consider:

- if the researcher has explained how the participants were selected
- if they explained why the participants they selected were the most appropriate to provide access to the type of knowledge sought by the study
- if there are any discussions around recruitment (e.g. why some people chose not to take part)

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Data collection

5. Were the data collected in a way that addressed the research issue? Write comments here

Consider:

- if the setting for data collection was justified
- if it is clear how data were collected (e.g. focus group, semi-structured interview etc)
- if the researcher has justified the methods chosen
- if the researcher has made the methods explicit (e.g. for interview method, is there an indication of how interviews were conducted, did they use a topic guide?)
- if methods were modified during the study. If so, has the researcher explained how and why?
- if the form of data is clear (e.g. tape recordings, video material, notes etc)
- if the researcher has discussed saturation of data

Reflexivity (research partnership relations/recognition of researcher bias)

6. Has the relationship between researcher and participants been adequately considered? Write comments here

Consider whether it is clear:

- if the researcher critically examined their own role, potential bias and influence during:
 - formulation of research questions
 - data collection, including sample recruitment and choice of location
 - how the researcher responded to events during the study and whether they considered the implications of any changes in the research design

Ethical Issues

7. Have ethical issues been taken into consideration? Write comments here

Consider:

- if there are sufficient details of how the research was explained to participants for the reader to assess whether ethical standards were maintained
- if the researcher has discussed issues raised by the study (e. g. issues around informed consent or confidentiality or how they have handled the effects of the study on the participants during and after the study)
- if approval has been sought from the ethics committee

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Data Analysis

8. Was the data analysis sufficiently rigorous? Write comments here

Consider:

- if there is an in-depth description of the analysis process
- if thematic analysis is used. If so, is it clear how the categories/themes were derived from the data?
- whether the researcher explains how the data presented were selected from the original sample to demonstrate the analysis process
- if sufficient data are presented to support the findings
- to what extent contradictory data are taken into account
- whether the researcher critically examined their own role, potential bias and influence during analysis and selection of data for presentation

Findings

9. Is there a clear statement of findings? Write comments here

Consider:

- if the findings are explicit
- if there is adequate discussion of the evidence both for and against the researcher's arguments
- if the researcher has discussed the credibility of their findings (e.g. triangulation, respondent validation, more than one analyst.)
- if the findings are discussed in relation to the original research questions

Value of the research

10. How valuable is the research? Write comments here

Consider:

- if the researcher discusses the contribution the study makes to existing knowledge or understanding (e.g. do they consider the findings in relation to current practice or policy, or relevant research-based literature?)
- if they identify new areas where research is necessary
- if the researchers have discussed whether or how the findings can be transferred to other populations or considered other ways the research may be used

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Appendices

APPENDIX 4

**LETTER OF APPROVAL FROM RESEARCH ETHICS
COMMITTEE**

August 26, 2008

App 06/08

Dear Fay:

Many thanks for your application to the School of Psychology ethics committee. Members of the committee have now commented on your application and have raised only one minor concern. The study proposes to assess excessive eating/appetite among other behaviours, however, we could not find this referred to in the information sheet

This item is minor and I am happy to provide Chairman's action for approval. Please do bear in mind the following points as you conduct your study:

1. Ethical conduct of the study remains your responsibility. Once investigations begin unexpected issues can arise and you are encouraged to think again about the situation of your participants.
2. You are free to make modifications to your procedures, without further ethical review, so long as you remain confident that your new procedures do not raise any general ethical problems or particular violations of BPS guidelines.
3. If funding is provided by the ESRC then outside review is mandatory from January 1, 2006. A Birmingham wide REC has been created for this purpose and applications can now be accepted by the Chairperson, Suzanne Higgs. There is a dedicated website for submissions.

If you have any questions or further issues arising then do not hesitate to contact me. I wish you the best of luck with your research.

Sincerely yours,



Stuart WG Derbyshire.

Appendices

APPENDIX 5

INFORMATION LETTER AND CONSENT FORM



UNIVERSITY OF BIRMINGHAM RESEARCH INTO CHARACTERISTICS OF INDIVIDUALS WITH MONOSOMY 1P36 DELETION SYNDROME

This booklet should contain:

1. Letter of invitation
2. Professor Chris Oliver's contact details (See letter of invitation)
3. Information sheet
4. Consent forms



Instructions for Returning Consent forms:

1. The consent forms should be completed by the **main caregiver**
2. When you have completed the consent forms, please return them to us in the *freepost* envelope provided.

Thank you for agreeing to participate in this research.



UNIVERSITY OF BIRMINGHAM

School of Psychology
Edgbaston
Birmingham
B15 2TT

Project Director: Professor Chris Oliver
Tel: 0121 414 7206

Date

Dear.....

We would like to invite you and the person you care for to take part in a new research project being carried out at the University of Birmingham in partnership with Unique. The research has the full support of Unique and a good response will provide valuable information for the group and for affected families. Briefly, the research is a study looking at different behaviours and cognitive impairments in children and adults with Monosomy 1p36 Deletion Syndrome, for example memory, attention and concentration problems; and also exploring parents' experience of receiving a diagnosis of Monosomy 1p36 Deletion Syndrome.

There is an information sheet enclosed that gives you more details about why the research is being carried out and what it will involve. If you feel it is appropriate you may wish to discuss the research with the person you care for before a decision is made about taking part.

Please read the information sheet before completing a consent form and the questionnaires and if you are unclear about any aspect of the study or have any questions then contact Professor Chris Oliver at the above address or on 0121 414 7206.

When we have completed the study we will send you a personalised feedback report with information about the person you care for and a report will be written for the Unique newsletter. In addition, the results of this research project may be published in a scientific journal.

If you wish to take part please complete a consent form and return this to us in the envelope provided. Thank you for your time and we look forward to hearing from you.

Yours sincerely,

Signature of Chair
Name of Chair
Information Officer
Unique

Chris Oliver
Professor of Clinical Psychology

Behavioural and Developmental Characteristics of Individuals with Monosomy 1p36 Deletion Syndrome, and parents' experience of receiving a diagnosis for their child

INFORMATION SHEET

Background:

A team at the University of Birmingham is carrying out a study to look at several aspects of behaviour in people with Monosomy 1p36 Deletion Syndrome.

As a research team we would like to investigate the specific behavioural and developmental characteristics of Monosomy 1p36 Deletion Syndrome. This would allow us to describe the behavioural phenotype for the syndrome - that is, the specific and characteristic behavioural repertoire exhibited by individuals with a genetic or chromosomal disorder. Behavioural phenotypes are important as they lead to a greater understanding of behaviour in rare syndromes. This can then lead to an increase in awareness of the potential problems which might arise and in the development and provision of early interventions for these problems.

We would like to investigate some specific problems which have been reported for individuals with Monosomy 1p36 Deletion Syndrome. These are problems with over-eating, self injurious behaviour and sociability. These characteristics can be distressing for parents/guardians as well as professionals, and we would like to gain more insight into these behaviours. This would be done by observing individuals in a range of different situations.

Further, there are many reports of cognitive impairments for individuals with Monosomy 1p36 Deletion Syndrome; however, the literature is not very helpful in telling us exactly what the impairments are. We would therefore like to conduct some simple cognitive assessments with some individuals to try to establish what impairments are present and how severe these are.

In addition, we will be seeking parents'/guardians' experiences of receiving a diagnosis of Monosomy 1p36 Deletion Syndrome for their child. There is currently very little information available to parents when they receive this diagnosis. We would therefore like to conduct interviews with the principal caregiver.

What does the study involve?

You will be invited along to Birmingham University for the day to meet other families with individuals with Monosomy 1p36 Deletion Syndrome. This day will provide an information session for participants, families and Unique about behavioural phenotypes and the research conducted by the University, and a 'get together' for the members of the support group.

Following this day, we would like to visit individuals at their home/day placement/school etc in order to carry out some observations of feeding/eating difficulties, self-injury and sociability. We will visit your child/person you care for at their day placement for the day. During this time, we will carry out short observations of your child/person you care for in different social situations and during a series of games and activities. Video recordings of the observation sessions will be made, as it is necessary for another psychologist at the University of Birmingham to check the accuracy of the observations (additional information on videoing is provided further on in this information sheet). The different social situations and activities will be presented to your child/person you care for by two members of the research team. We will use three different social situations that will last a few minutes each. Firstly we will engage with your child to help them become familiar with the situation and us. We will then let your child play with the toys and games we have but we will not initiate any interaction with your child; but we will interact if the child attempts to initiate interaction. In the next condition we aim to see how your child responds if we do not interact with them socially. We will then see how your child reacts when we ask to join in with them again. Finally we will see how your child acts when we are engaging with someone or something else (talking to the other researcher or reading notes) and there are no toys or distractions for your child.

During our time at the day placement of your child/person you care for we will also carry out some observations to help understand what triggers certain problem behaviours. We will carry out observations as your child takes part in situations where levels of adult attention and demands are varied. The situations include three different conditions (10 minutes each), which your child will experience regularly in their normal day environment. The first situation is a “high attention” in which the researcher will interact with your child while they play with a preferred toy or game. The second condition is a “high demand” condition in which the researcher will ask your child to take part in a less preferred task and will continue to prompt and guide your child throughout the task. The final condition is a “low attention” condition in which your child will again have access to a preferred game or toy but this time the researcher will move their attention away from your child and will talk to the researcher. It is possible that these situations will cause an increase or decrease in particular behaviours. If your child becomes extremely distressed or is at excessive risk of injuring themselves we will immediately stop the session.

During the home visit, we may also conduct some short observations of your child/ person you care for within the home. This will be very similar to the method described above, only this time we will ask *you* to interact with your child/person you care for. The situations will be very natural and will be situations that are part of the regular routine of your child/person you care for. The situations will be similar to the high attention, low attention, and demand conditions conducted during the school assessments (see above) and we will ask you to think of a time in your daily routine where this situation may occur (e.g. high attention may occur when you play a game at a certain point during the day). We will then ask you to run through this situation so that we can observe any changes in behaviour that might occur during that time. In some cases it may be necessary to ask you to think of more specific situations in which self-injurious or aggressive behaviour usually occur and include this situation in the observations (for example it may be the case that you observe higher rates of these behaviours when a preferred activity is terminated. Therefore, we would include this situation in the

observations). There may also be times when we ask you respond to your child/person you care for in a different way to that which you would normally do (e.g. during a low attention condition, we may ask you to turn around and talk to your child/person you care for briefly when behaviour occurs, rather than not paying any attention to the behaviour). Again, this is to observe any changes in behaviour that occur as a result of these situations and responses. It is possible that these situations will cause an increase or decrease in particular behaviours. If your child becomes extremely distressed or is at excessive risk of injuring themselves we will immediately stop the session.

Whilst at the day placement of your child/person you care for, we will also carry out some simple cognitive assessments to give us information about specific problems which may be faced by individuals with Monosomy 1p36 Deletion Syndrome, for example memory, attention and concentration problems.

In order to investigate parents'/guardians' experiences of receiving a diagnosis for their child you will be invited to attend an interview which will last approximately one and a half hours. This interview will be audio taped, and then transcribed and analysed by one of the researchers. Any quotes that are pertinent to the study will be included within the final publication, however all attempts to make such quotes anonymous will be made, and individuals will not be made identifiable. Where possible, these interviews will take place, whilst other members of the research team are conducting cognitive assessments or observations.

What are the benefits/drawbacks of taking part?

Whilst we cannot promise any direct benefits to you for agreeing to take part in the study, the information that we gather will be invaluable in increasing the understanding of Monosomy 1p36 Deletion Syndrome. We hope that a greater understanding of the cognitive impairments and behavioural characteristics of the syndrome would lead to the development of appropriate interventions for these problems.

We do not anticipate any drawbacks of taking part in this study; in fact we hope that you would find it a very worthwhile experience. However, it is possible that parents/guardians may become distressed due to the nature of the questions asked during the interview. Every step will be taken to make sure that any distress is kept to a minimum. It is anticipated that parents/guardians will find the interview to be a positive experience, and will benefit from sharing their experiences, with the knowledge that this research could shape further research relating to their child's condition.

Consent:

It is up to you whether or not you want your child or the person you care for to take part in the study. If your child / the person you care for is under the age of 16 or over the age of 16 but unable to give consent then please fill in consent form A on their behalf. If your child or the person you care for is over the age of 16 and is able to give consent for themselves, please ask them to complete consent form B. If you feel that it is appropriate, you may wish to discuss the project with your child or the person you care for. If you do not wish to take part in all aspects of the research, then there is the opportunity to take part in only the questionnaire study, if you so wish.

Withdrawal:

Should you or the person you care for decide that you no longer wish to be involved in the study, the information that you have provided can be withdrawn at any time without you giving a reason. Even after the questionnaire, interview and observations have been completed, consent can be withdrawn and any data collected will be destroyed. This will not restrict access to other services and will not affect the right to treatment.

Confidentiality:

Contact has been made through Unique, your syndrome support group and we do not know any of your personal details at this stage. All details collected will be kept on a confidential database that is only accessible to those working on the project. Anonymity is ensured by storing the questionnaire data separately from any material that identifies participants. If published, information will be presented without reference to any identifying information.

At the end of the study:

Each parent/ carer will receive a personalised feedback report on their child or the person they care for. A summary of the project's findings will be circulated to anyone involved who wishes to see a copy and a report will be written for the Unique newsletter. Any requests for advice concerning your child/ the person you care for will be referred to Professor Chris Oliver, Clinical Psychologist. It is possible that you may be invited to participate in further research after the study but consenting to participate in this study does not mean that you are obliged to do so.

Review:

If you have any concerns about the conduct of this study please contact Dr Stuart Derbyshire at the School of Psychology Ethics Committee, University of Birmingham, Edgbaston, Birmingham, B15 2TT.

Thank you very much for taking the time to read this information.



CONSENT FORM C: For parents/guardians taking part in interviews

Please read the following statements and place your initials in each box to say that you understand about the content of the information sheet, your involvement, and that you agree to take part in the above named study.

Please initial the boxes

I confirm that I have read and understood the information sheet for the above study or that it has been explained to me and have had the opportunity to ask questions.

I understand that participation in the study is voluntary and that I am free to end my involvement at any time, or request that the data collected in the study be destroyed, without giving a reason.

I agree to take part in the above study

I agree for quotations from my interviews to be used in the above study

I agree to take part in the above study

Please complete the information below

Your name..... Your date of birth.....

Your signature..... Date.....

FOR OFFICE USE ONLY

Signature of researcher.....Date.....

Appendices

APPENDIX 6
INTERVIEW SCHEDULE

Interview Schedule

The following is an example of the types of questions that will be used within the interviews with parents. This schedule is intended to be flexible allowing parents to feel engaged in the interview.

Introduction:

I would like to 'hear about' or 'talk with you about' your experiences of receiving a diagnosis of 1p36 deletion syndrome for your son/daughter [name]. If at any point you feel that you do not wish to continue with this interview; that is ok. No data from this interview will be used without your permission. Should you decide after the interview that you do not wish it to be included in the research, then you can contact me using the contact details on the information sheet, and your data will be removed.

Example questions:

Can I ask you how old [name] is?

I'd like to take you back to when you received the diagnosis of 1p36 syndrome for [name]. Did you have concerns about [name] before you were given the diagnosis? (ask to elaborate)

How long did it take for [name] to be diagnosed, after you had voiced your concerns? *Or* Can you remember how old [name] was when you received the diagnosis?

Can you remember who gave you the confirmed diagnosis? (job title)

Can you think back to what thoughts were going through your head when you were told that [name] had 1p36?

What about feelings, can you tell me what feelings you were experiencing during this time?

What difference did it make to you to have a name for the condition that (name) has? (understood some things differently?)

What did you do after you had received the diagnosis of 1p36 for [name]? (behaviours – telling people/not... Other people have said that they've found it comforting to do.... or may have found it a difficult time and done....(taken from previous interviews).

Can you recall what support you were given at the time of diagnosis? (who was that from...any signposting to other services/support groups...were you given any information to help you understand the condition?)

How did you rate the support that you received (from health care professionals/support groups/services)?

What other support did you use? (friends, family, internet, research)

Would I be right in thinking that you had never heard of 1p36 deletion syndrome until [name] was diagnosed? Have you made any attempts to find out more about this genetic disorder? (When did you do this, what steps did you take to find out more?)

How do you feel about the amount of information available/that you were given about 1p36 deletion syndrome? (Enough? Helpful? Hard to find?)

I was wondering if you could think back to preparing for the birth of your son/daughter, and if you could explain what expectations you had of the birth and the first few months that followed.

I can imagine that this might be different in some ways to the experience that you and your husband/wife actually had during and immediately after [name's] birth, can you explain how the birth and first few months were different to what you expected.

How has your experience of being a parent been compared to your expectations of parenthood? (Do you have other children without difficulties?...how do these experiences compare with each other? Do you find yourself comparing your experiences of parenthood with friends'/family members' experiences)

[Name] is now [age], what feelings/thoughts have you experienced throughout your son's/daughter's life? (if people are struggling, list some feelings/thoughts other people may have mentioned in previous interviews – both positive and negative)

Can you tell me what expectations you have for the future?

Appendices

APPENDIX 7

TRANSCRIPT OF PILOT INTERVIEW

Pilot interview: Sarah, mother of Thomas aged 6 with Angelman's Syndrome

I: Ok then Sarah, errm, the main thing about this interview is to hear about your experiences of receiving a diagnosis for Thomas

S: Right

I: Errm, if you feel at any point that you don't want to carry on...

S: no that's fine...

I: then you can cancel the interview if you think it's going to get too upsetting or ...[that's totally] fine, and no data from this interview is going to be used in any research...

S: [Ok]

S: Okey doke

I: ... without your permission. Should you decide that once the interview is finished, you don't want that to be used then it easy enough to delete the interview.

S: not a problem

I: Can you remind me how old Thomas is? [phone rings, interview stopped] Ok, don't worry if the phone goes, that's fine

S: OK. So you were asking me...

I: Yeah, Thomas just remind me how old he is.

S: He's six and a half now

I: Six and a half, ok. I'd just like to kind of take you back to when you did receive the diagnosis of Angelman's Syndrome

S: Right

I: before you..., actually I'd probably be best to ask, how old was he when he was diagnosed

S: Ooooh dadadada, the actual... umm, I'm gonna guess around about two and a half-ish

I: Right, Ok

S: Yeah around about two and a half, the actual diagnosis of Angelman's

I: right

S: the name

I: that was when you were actually told what was going on? Errm, before you were given that diagnosis did you have any concerns...

S: Well the thing that I found, being a first time mum.....

I: Right,

S: it was "oh little boys are slower than girls, don't worry, he'll walk eventually, he'll do all this, he'll talk eventually, this was obviously the normal health visitors, etc

I: Ah ha

S: Errm, then obviously, it started, it, he went two, which was obviously in the December. Then between that sort of January and the summer, was like right, we'll send him off for a four week assessment blah blah blah, geneticists, urrr. And it was Dr [Name] who did it, she's done one other case of Angelman's

I: Right

S: so she seemed to recognise...

I: what everyone else hadn't?

S: the features and what have you, so that's where the tests were quite quick for the fact that she'd sent it off for Angelman's

I: Right

S: rather than any other syndrome that you can think of

I: Right ok

S: which is errm, you know, we thought it was a very long process, but obviously in the medical world, six months is actually quite quick really

I: Ah ha

S: so err, it all sort of happened about two, two and a half

I: Ok, and did Dr [Name] give you that kind of hint that it could have been Angelman's [from the other child that she'd previously seen]

S: [Yeah I think] it was sort of, we'll send it off for Angelman's, and so of course you wiz home and get on the internet and get it all up and you're like oh my god, you're never gonna do this that and the other

I: [right ok]

S: have a complete panic because obviously it's like with anything you read up on it and you're like "I shouldn't have done that" (laughter)

I: Ok

S: So now it was... she did give us a hint and obviously we waited to see if it was

I: ok

S: and it was

I: OK, so once you had the hint that it could have been Angelman's you went straight to the internet

S: Oh yes (laughter) as you do

I: can you talk me through what that felt like

S: Oh god, well you're reading, because we didn't know the ins and outs of it, you read whatever's put on there, you go to all the websites and it's basically "oh its come from mums genes, oh god it's my fault blah, blah, blah, and then you read a bit into it and it could have come from dad, and it's all very confusing, your chromosomes yours genes and what have ya and all this and I'm like well oh if it's not a deletion, and there's a bit missing, or its upside down inside out, back to front, what are you gonna get told when it all comes back. So of course you're panicking, family life is... you're thinking is it my fault, is it dad's fault, it's nobody's fault but you feel yourself as though you're to blame and then obviously when we were told more details from geneticists and Dr Ibrahim that it was a spontaneous thing for Thomas

I: Right

S: which was a bit of a sigh of relief because obviously it was better chances for a second child, etc rather than coming from my genes and what have you

I: Right ok

S: so when you get to reading what they've actually got as opposed to "they could have all this, you know, madness"

I: So am I right in thinking that when you were first looking, you were getting loads of conflicting [information]

S: [oh god yeah] before obviously she'd given us a bit of a hint oh, it could be Angelman's but wait until the tests, so of course they could have said he's gonna be Prader Willi, what have you. They could have said anything-drome and you would have just come back and looked it up. And that was it. In a way I wish I hadn't but you're just that nosey you think "ahh, whats the matter with him, why isn't he walking, why isn't he talking", so yes, that was the route that we went down.

I: You said, why isn't he walking, talking, were they the main concerns that you had about Thomas

S: Well it was sort of, things that, he's sitting up, his milestones early on sort of one and a half and he's crawling, standing up around the furniture, so the actual, I do remember it was around that summer, two and a half that he actually took a few steps, but, you know I could just plonk him on the grass outside in the summer and he'd be still there. You know he wouldn't toddle off or move or anything, you know he might roll, but that was about it

I: and was he crawling as well or...?

S: he could probably get across a mat or a blanket sort of thing, but he wasn't you know, but it was just like, the health visitor, "oh no no, he's fine, boys are slower, don't worry, everything will be alright" and then it was other friends of mine who've got kids, and cos I wasn't that maternal to be honest, I was 29 when I had Thomas (laughter). But no my friends would be like, I would say, "oh what age did so and so walk and talk, well you know he should be doing it" ... As time ticked by I thought well you know, you can sort of fob off the potty training and all that because you know, some kids are really different aren't they

I: yeah

S: but I thought he should be at least going mamamamam, dadadadad you know, sort of. But as I say if he'd have been a second child, I'd have probably been more wise to it earlier

I: Right, ok. So with it being first child, do you think that made [a difference]

S: [Oh you believe] what the health visitors tell you (laughter) "oh you've gotta have him weighed every week, you've gotta do this every week, rararara, what have you. With your second it's just, you pop over once a month or something (laughter) but it is, you're far more relaxed... As a first time mum you don't like to keep badgering people and keep getting on the phone, "whats this, whats the matter, why isn't he doing dadadada". But I mean, it's like [he] had a bit of a kidney issue (points at other son, Sam) when he was born and it's like rararara, and you get more, because I do it all with Thomas's appointments, you just badger everyone now, one of these nagging mums now (laughter)

I: so it's actually made a difference to how you've parented... Sam is it?

S: Sam yeah hmm. But it does, I think, as you say other parents who've got Angelman's errm children. It's like there's one we know from school, she's got an older sister 18, and the Angelman's is about 11, so of course, mum obviously realised that something was the matter blah blah blah, and you know, I mean the other factor is they've got older siblings to help out as well, you know. I think as a first time mum, not being that maternal, never having many kids in the family around me, I was like, I relied on everybody else's information, and when the health visitor says jump alright then yeah you know, it's like whatever

I: It was Dr [Name] who actually gave you the diagnosis was it?

S: Yeah well, I think I sort of went onto the health visitor a bit as in, surely he should be doing this and they referred us to Wordsley Hospital at the time, for a four week assessment unit, thing, programme where he went off on the bus, kept him there and gave him lunch, Dr [Name] would pop in, different people would pop in, monitor his motorskills etc...And then time ticked by and there was the local Phoenix Centre, I don't know if you've ever heard of that, it's a local from all round, under five special needs

I: Right

S: And he'd pop there a couple of days a week and then gradually got into it, and then it all started you know ticking by, they'd obviously waited from the assessment unit for the results you know, and all the rest of it. It just ticked by from there, oh right, he's going to this under five special needs thing now, and ok, people started talking about statements and god knows what, it's like what's going on, because he was getting older and obviously at the pre-school stage I was like, I don't know what pre-school do, cos obviously not having another child to compare it to. I mean thirty odd years, it was playschool, it wasn't all this you know, it's a completely different education style to what I knew so it was all, it will be new with this one (points at son, Sam) because I've never done it have I.

I: yeah, it's a whole new experience again isn't it

S: Yeah

I: So Dr Ibrahim, is she the geneticist

S: No, it was, she is the urrr, what do you call them, I suppose it's like the Paediatrician. The GP, the consultant at the hospital. She's at Russells Hall now, she was at Wordsley but that's shut. Oh what was her name, Christine Olay, she's the actual geneticist, and there's a girl, a lady called [Name], who works at Netherton Health Centre, who was like the co-ordinator, with the geneticist side, who introduced us to Olay. Blah blah blah

I: right ok

S: (Sighed)

I: So, who actually sat you down and said ok Thomas has got Angel..

S: It was Dr Ibrahim, she'd sort of you know

I: followed you all the way through

S: Yeah, and we still see her every six months or so now, but I mean, because his health isn't as bad with it, she doesn't have to see him as regular, I mean she knows what the outcome is going to be really I suppose

I: And when she sat you down and said "Ok, Thomas has got Angelman's as I suspected", how did that make you feel

S: it was sort of, well in all fairness you saying who sat and told us, I think they were all there Dr Ibrahim and the geneticist

I: the whole team

S: yeah, and it was sort of they were all "oh don't worry, I mean we were you know, I suppose we were still oh my god. They sort of put it to you now you've got to be, like living...I mean I'm quite practical really, Initially oh you know you're all sat there crying oh god what are we gonna do and they sort of approach it, well he's never going to work, errm and you've gotta look logically he's never gonna Sump in a car and pass his driving test when he's seventeen, eighteen you know, you've gotta go, and they bring it back and say whatever, you know, you can't look at the age of eighteen straight away, I mean he was two and a half at the time

I: yeah

S: But here was me thinking so when he's older, I was asking all these questions of when he's like 20 is he gonna live on his own and stuff, not the child, I wasn't that bothered about up to sixteen

I: Ok

S: Cos there's still... Even if he was 'normal' you know what have you, I think up to the age of 16 you think right you've gotta look after them anyway, that's it you know, they're your kids 16 18 what have you, you know they do pass driving tests they do go to

college, they do do this and blah blah blah I'm thinking, I really jumped 20 years down the line I was like....

I: So what do you think it was that made you jump so far ahead

S: I suppose it's because I could still upset him still being in pull-up nappies what have you at a young age, 6,7,8 you know, you go out in public and he whacks somebody or pulls their hair 9, 10 obviously when they get a bit bigger, stronger you know you can tell by his features a bit there's something wrong with him

I: Right

S: but not like you'd pick up on it like a downs you wouldn't you know like the general public are you know, I think I could cope with it, but when it's sort of older I don't think the general public are sowhats the word I'm after, you know so willing [to accept]

I: [forgiving]

S: Yeah forgiving to be honest, if he's kicked someone in the supermarket you know and he's 20 years old you know, it's all gonna go pear shaped. So I did take that big jump and obviously, my other half Rob he brought me back, and he was like, well hold on you can't think that far ahead, so whether it was the motherly thing I don't know, I was like this will happen well in the future, as opposed to the first 6/7/8 years I'd probably you know, I wouldn't bat an eyelid really.

I: so am I right in thinking that you'd were concerned about where he'd be?

S: In yeah, I was thinking obviously we're going to get all this help, far better now than obviously twenty years ago, you're gonna get help, he can go to this preschool, someone will come out and do this and he can go on a bus here there and everywhere do all these bits and bobs, get all this feedback... you know hospital appointments what have you, no problem, then as time ticks by they thin out a bit, and you can expect it, blah blah blah but I'm thinking they get to that age of 16 and obviously in the last few years going to different places you're thinking like oh god you know, when they leave school but I initially panicked over that to start with I don't know why [phone call interrupted, interviews stopped]

I: errm, you mentioned that you probably thought a bit differently being a mum to how your partner Rob [how Rob]

S: [A dad]

I: Can you think what what feelings he was going through at the time

S: I mean ,we were both very teary and oh my god whats going to happen, is he ever going to walk. They gave us the worst case scenario feedback, he could sit there on the sort of cerebral palsy side and not move at all, as in a scale of 1-10 it could be really bad, or some Angelman's kids are blah blah blah, so they give you like 1/10. You know, so Dr Ibrahim was very, middle of the roadish maybe creeping towards the top, and feedback that sort of ...they sort of... give me the impression that he was sort of... medium to severe,

I: Right

S: Rather than more severe in Thomas's case, and obviously that must have sunk into my head because I went on the education side and sent him to a moderate special needs school

I: Right

S: Not happening. He's now at a severe one (laughter) Obviously as a baby, the milestones, their motorskills, etc, aren't as noticeable as now he's got older

I: yeah

S: They did sort of...I remember her saying one thing, whatever he learns up to the age of twelve-ish, that'll be it, he won't carry on learning, and I was like Ok, and then she was on about his speech, and she said roughly about the age of 7 about roughly 7 blah blah blah, not a cat in hell's chance, hence I'm barking up the makaton and sign language tree (laughter). Errm but no, so the things, there's things like that the odd statement, if he hasn't spoke by 7 sort of stick in my head he's never gonna drive a car do you know what I mean?

I: yeah

S: The raw facts stuck in my head, they could be waffling about epileptic fits and I'm thinking he's never had one as yet, he has the medication to control it, touchwood he has a few tremors and that, but I don't know what I'd do if that...you know that's another part of it, that I've never really, it's never bothered me as much as thinking oh god he's never gonna, he's gonna have to rely on public transport and never go anywhere on his own and all this. So as I say, but it did open your eyes and there was a bit, we sort of got a bit upset and got back and looked back on the computer again (laughter) when it was actually officially told to us, errm cos then they explained about deletions and it coming from mums genes and dads genes which would have been in Prader Willi bluh bluh bluh what have you and it's ok alright. They did actually put it into plain English for us, cos we get baffled, cos she even commented I bet you've gone home and looked on the internet haven't you...yeah...and I said err, they even commented that it is, you know as I say, I wasn't a carrier as such so they did say if we had any more children, obviously to inform them, let them know but the percentage was quite low, still had all the tests but you know the percentage was low, as opposed to 50/50 really.

I: So that 6month period waiting for the diagnosis, already having looked on the internet, up until you actually got the official diagnosis, what kind of things were going through your mind

S: It was literally, I think we were both looking for somebody to blame to be honest... I think...As...I think if you go on to one website and have a read, it comes across as though it's paternal and you go on to something else and it's maternal and I'm like it's like well which is it. And I say I'm not that up on chromosomes and genes and yeah Chromosome 15 you read a bit more, you go on and all this oh god imprinting and god knows, I was like phhhh

I: It doesn't come up in normal conversation does it

S: No... there was just one advantage, my auntie is a biology teacher and I was like oh whats this and she put it into a bit of plain English for me and that was basically it was just sitting there, we'd print loads of stuff of the computer, what is Angelman's, obsessed with water bubbles and blah blah blah, usual inaudible happy clappy whatever they come up with, and we were there with... and we sit there and the pair of us, he does do that, ticking them off, ooh he's not, no he doesn't do that one, so he might not be as bad as others, you know, stupid things you've got a list of 20 things, he likes bubbles tick that off, oh no he doesn't do that or whatever it was or is it a flat head at the back of...I said oh no, he's not too bad, his heads not that flat, things like that, so we had got a list that we'd printed off and we were ticking and crossing them trying to work out before we were told, well is he really that bad has he really got all these, or has he only got some of them.

I: and when you were ticking them, how was that making you feel when it's like oh yeah he does like bubbles and...

S: Yeah, it was obviously the things like the water and the bubbles you can cope with but its errm, the list didn't entail headbanging, pinching, it didn't, it said he would flap and [interrupted by son] it didn't involve, it might say, sort of pulling hair and what have you and all this, but because he was so young he hadn't got the strength, he wasn't at the stage

I: Yeah

S: you might get the odd slap round the face, but compared to now... the digging in, the pinching you know, but err, we didn't really tick off too many of those because he was like two and a half, you'd get the odd slap round the face, but he didn't headbutt or anything then, because he couldn't walk up to a wall or a patio, so there were things that we obviously didn't take his age into account and we were like, you know, as you say, it was a list, it doesn't say an age, it's not like a list for under 5 Angelman's

I: they will be doing x,y and z

S: Yeah a list for over 5 Angelman's so this is where we were still quite oblivious to it all, and no so, it was a bit tearful stressful, what are we gonna do, oh god he's never gonna do this that and the other, literally having a moan, but as time has ticked by, you harden up a bit (laughter). Initially you're like oh my god it's the end of our life, you know what are we gonna do. Cos I thought grandparents got more upset

I: right, what was there kind of reaction

S: They're still more upset now. Errrm, well at the time we don't, we'd got Robs dad, he's passed away since, and he was like oh god you know has it come from the man's side of the family. He was sort of very protective of if it was sort of Rob's...not fault, I keep using that word, but you know what I mean

I: Yeah

S: It's like if it was Robs fault he was sort of, oh god it's not our side of the family he was like woah ya know like that. My mum hadn't got Alzheimer's at the time obviously (laughter)...she was sort of "oh god, is he gonna be alright are you gonna cope, blah blah blah". My dad because he was a little boy he thought "great, sport", my dad, he loves sport...he still plays football cricket what have you now, 65 loves it. But my dad couldn't get it round his head that Thomas wouldn't be able to run around and kick a ball

I: Right

S: compared to this two year old now, my dad just couldn't work out that you might have to sit there and read a book or why is he sitting there watching Mr Tumble for 24 hours you know. My dad couldn't... he's not getting any exercise my dad thought he can still move, he's not disabled as in such, so each grandparent found something different to ...but each one was stressing us out because it was like what are you lot panicking about, we've gotta live with it, you know, so as I say it was sort of, yeah they all had something different. And I think friends as well, friends seemed more scared of the fact to ask us anything. Not bothered about Thomas, they'd still, they don't offer now, because he beats them up but as a baby you could get babysitters (laughter) but err nobody offers now, they're a bit more frightened to ask us what was going on, so they'd all wiz off to their computers, they'd all start looking up Angelman's, and say oh well, and I was like why don't people ask because after we had found out that there were that many different varieties, not varieties, severities of it, etc we thought well you know, we'd just tell people in plain English sometimes it comes from mum blah blah blah but this just

spontaneously happened in Thomas, blah blah, cutting it down to about two sentences and that's it.

I: yeah

S: and I still tell people to this day and they'll be like "what's the matter with him, has he got autism", and I'm like no no no they all think they know what he's got

I: everybody trying to diagnose him

I: Yeah but I don't know as I say, the family situation we got to the point why's everyone else flapping, why's everybody else flapping when its, we've gotta live with it. We want the hard facts from the doctors and any problems we go and ask them I wouldn't go and say oh dad you know. I did realise after the first 6 months after going on the internet and stuff, that really that probably wasn't a good idea, but then, you're that impatient you're not gonna wait four, five six months for the blood to be cultured and god knows what, and you're I wanna know now

I: yeah yeah, so when you actually got that diagnosis, a period of shock for you guys?

S: Well because she'd already pre-warned us about Angelman's, I don't think it was a shock. Obviously we were all very teary, and she was like you know, we were there, in her office and like, you know what happens, as I say I jumped 20years into the future. But it was the matter, of, no don't worry, there's all this, and they were very they approached it very well how they told us. They didn't sort of your son's got Angelman's he's never gonna do this, it wasn't very harsh, but they did sort of come out with some facts and as I say those are the facts that stick in my head really. Whether they are facts, obviously I don't know how they can put an age on something, they all vary that much but she's give us a bit of an insight that really, you know, when he get to 15/16 what have you he's not gonna keep learning forever and a day, it might sort of, his life skills, things like that, but you know, he's never gonna count to ten or whatever he's doing, which we've had to accept, which initially, at such a young age to be diagnosed with it, it was like, he's not gonna count anyway when he's three or whatever, but as times ticked by you just realise now that as I said when I sent him to a moderate school, the other autistic kids could count to ten or go to the toilet or whatever, they were just getting way ahead... The early age diagnosis did help in a way that we knew what to expect...if I'd gotta wait until he was like 4 or 5 and I'd started off, putting his name down for schools, I don't know, I think it was to our advantage that Dr Ibrahim has done one other case and picked up on it...

Yeah

S: but as I say, I think if she hadn't have done that we could have been one of these sitting here, come on we've looking for 3 years now, because I wouldn't have any info of Birmingham University and nothing until you actually got the name Angelman's, I wouldn't have know which website, I mean I could have fobbed it off as one of the other ones or anything you know, I hadn't got a clue

I: So it made quite a difference to you and Rob, actually knowing exactly what it was and having a name for it

S: I think initially it was here or there whether we'd got a name, but over the last few years, it's a lot easier to say he's got Angelman's, and if people are interested they'll go home and look it up themselves as I say. And it easier looking at other mums who haven't got diagnoses for their children where they're all fobbed off on the autism ADHD blah blah blah spectrum there they are. All the workshops, all the help is there for autism, got something a bit different, that's it you're stuck. So it is nice as I've said,

some things crop up, I have met two other mums with Angelman's now, there's two I keep in touch with, but you know it sort of it does help having a name and I do honestly thing if I'd just got to sit here now going well I don't know what's the matter with him it's a bit of autism, you know, they'll be giving him medication for ADHD or whatever, it would be like ...you know, on that big rollercoaster with half of the other kids at special schools and that, so it does make a difference, I think it you know, there's that many different syndromes you can't pick up on them all.

I: Is that something that has kind of been brought to your attention more now [that there's a whole gamut]

S: [oh yeah]

I: of other syndromes

S: I mean, as I say, if a child in my eyes, if a child say 6 7 8 whatever was at school, got no name for what's the matter with them, especially this one woman I know, got no name, they are fobbed off on the autism spectrum, it doesn't matter, you know on the education side that's what they're fobbed off as. Errrm... and you know as I say, the ADHD, they land it all on behavioural problems, global developmental delay blah blah blah, all the usual malarkey, but as I say that has put our minds at rest having a name because we haven't got to sit with the more pressure and stress, like my friend who is going to Great Ormand Street with her... faffing about doing this, thinking what the hell is it, trying to find out yourself, looking on the computer yourself. All this, we've got all that pressure took away from us, so we can carry on living...he's got it and that's it, we can't change anything, whereas we'd be fishing if I hadn't got a name

I: So do you feel that now you've got a diagnosis it's shifted your focus

S: Oh yeah, on other things, yeah, it is, if I hadn't got a name I'd be fishing now, you know baffling everybody, ooh you know it's been three years what have you, but having that name it sort of moved us on to think about, well it's silly things like I haven't done my sign language and makaton, I was took up my not that get much spare time, but I'd have took the time up, I'd have took the morning I go to college, I'd have been tapping on the computer trying to find something out, or badgering other doctors and god knows what else, but as you say, we focussed on other things, like Thomas is never gonna talk.. we have got to, you know he can't carry a picture book around with him for the rest of his life, so it would help if he could sign 20 signs things like that. And err, it's one of these things where I try and be practical like as in, right when he's 18 in the middle of a field or Merry Hill somewhere shopping err, with his carer and he wants a drink, if he signs drink he'll get a drink, whereas if he can't point to a tap or a picture and he's thirsty, and you know, cos I mean he's gonna know when he's hungry and thirsty... but because he can't tell you that's when it all kicks off. So this is where I'm thinking I've got to look practically, it might take me 10 years to teach him to sign drink, but (laughter) you know I start as I mean to actually hope he will pick up a couple of ...the living skills [so to]speak, yeah.

I: [useful]

I: So... it's changed your focus more so to ...adapting to the situation [now}

S: [yeah] The first... I mean he was diagnosed what two and half, so his third birthday was Christmas, yeah so we were still... you know, silly things like you know for his birthday he couldn't blow a candle out and I'm thinking ok, is that normal do other three year olds blow candles out, ain't got a clue. Anyway time ticks by, the fourth birthday I thought, we've been to parties now other kids can blow candles things like that so I

mean here's me thinking Thomas's never gonna blow candles out...so I now by sparklers for his birthday cake. Silly things that I would have you know the normal so called things that you would do in life I just get on and adapt over stupid little things and that's it cos I've got it in my head if he can't do that he's never gonna do it, but if he does do it it's a bonus...

I: yes

S: So I'm on a more negative right he's never gonna talk he says mom mom mom mom mom and that's about it really, m m m m m you know sort of a muh thing but that's about it. So... when he's whatever twelve and he says five words beginning with m so be it, fantastic, but in my head he's never gonna talk

I: yeah

S: cos five words is here nor there really it's you know what I mean you can't get through life with five words but you know. So that is where I've hardened up a bit

I: Right

S: and you know between me and Rob we've got different view on things, it's put loads of stress on the relationship as such I mean kids do anyway but it's like, it's sort of well don't do this..we've different views of how we should do things with Thomas,

I: ok

S: like obviously I do most of the dressing and getting him dressed so I'm like "stand up" you know it's easier if he's up on his bed cos he's at that height, stand up, put your feet in your trousers dur dadurdadurm fantastic if they're like tracksuits or pyjamas but if they're school trousers you get beaten to a pulp, don't want them on

I: Ok, he knows what associates with that

S: but if Rob's trying to dress him it's err a wriggle on the floor and I'll say like Thomas, ohhh you know this is where it all kicks off in our house over stupid things like putting a pair of trousers on Thomas. But as I say we've got different views on things... you know, Rob's more "I'll take him out on his bike" which I would, I would expect his dad to do blah blah, whereas I'm sort of the practical "you *will* sit and learn a sign" (laughter) you know but, I don't know as I say it is weird but I think we focus a bit further... .. we live with it, like, as I say we're going on holiday, told them all you know special needs blah blah blah, Oh do you want extra leg room, do you want to pay the extra?" No, if someone complains it's not my fault I've told you I've got a special needs ch/whereas three or four years ago if you know, or if Thomas hadn't got a diagnosis now and I was , oh well, and what's the matter with him oh, it's a bit behaviour and what-have you and you're sort of ...but now I'm like oh yeah he's got Angelman's syndrome he might kick the seat in front but I'm not paying any extra for the seat it's up to you, if you get complaints, I've warned you. And you harden up to the world don't you

I: yeah

S: (oi noisey – to son Sam). No, you harden up to the real world as I say. I think it does help big time and I must say that 6 month period, it was very tearful and stressful and then when we actually got the name and we had to start telling people so that was going up to when he was approaching three. It was still a bit "oh what's that then, I've never heard of that" and I'm like oh here we go again but then after that you've people for six months and you're like sick of it, and you shorten it, that's it got it, might have a fit, might do this might do that, might never do this might never you know

I: so you'd gone from the whole reel that you'd be given at the hospital to ok he's got it this is what he can do this is what he can't, accept it.

S: Yeah, exactly yeah, I mean some parents are very protective over what, you know, we do a lot of social things we take him to this wood class which is like cubs and that and we go off but obviously I have to stay as his carer, and it's like you know I tell people he's prone to epilepsy but he's never had a fit, it could be that I could if I just pop to the shop for two minutes, but if I haven't told people. I'm very open to telling people whereas some parents are very protective, because I think I'm not gonna be here forever, you've gotta tell the world so to speak, so yeah, it does harden you up as a parent (laughter) more so than wrapping them in cotton wool it's like get out do that go on your play scheme and get sorted (laughter)

I: (laughter)... in terms of the team that sat down and said this is where he is and painted the not so bad picture

S: yeah

I: what support did you receive from them

S: The geneticist obviously we sort of the appointments off her were twelve months come back and see me, we did get a bit of feedback from the geneticists co-ordinator [Name] you know she sort of but obviously the actual geneticist was a twelve month things

I: yeah

S: then Dr Ibrahim was basically we'll stick you down for a 6 month slot..... at that point this is the funny thing, when I went over to the doctors to actually say, obviously, handily enough our doctor's is just there (points) so repeat prescriptions and that I keep going over "Right Thomas has been diagnosed with Angelman's" so I went over to the health visitor the doctor, "oh whats that then we've never heard of that" but call me really really stupid but if I was a doctor or a health visitor I'd say "oh I'll to go and research that" no. They turned round, and I, I ended up printing things off my computer and taking them to the doctor and the health visitor

I: Right

S: which I thought was a bit, ok you're getting paid, it's your job to research, even if you've never come across it if I'm one of their patients, you'd think that the sort of but there is a new doctor there now who has come across it in the past, and he was err adamant that little boys couldn't have Angelman's.....

I: Right, ok

S: and I was right ok, he's obviously gone to uni doing his what have you and got it all mixed up with the chromosomes coming from maternal, as in oh right, girls have got Angelman's, he's learnt a little bit about it but he was adamant that our Thomas couldn't have Angelman's.....So I don't know obviously he's done a bit at uni but not read up on too much on it...

I: yeah just taken a snippet

S: Yeah, he's obviously got the err wires crossed, and I said well believe you me he has got it blah blah blah that was the ends of that one, don't see that doctor anymore. (laughter) No but it was a bit wrong that I had to inform people in the profession, the medical side etc, yeah I could probably understand the health visitor, they do come across children with clicky hips whatever's going on but if there is something there they're not doctors as such are they, so I thought the doctor hasn't got a clue but really he should go and look that up so I was a bit cross with that, but then as I say the help and the support was not from the medical side really,... as he was approaching three (will you stop rattling that.... Go and play over there) we had 6 month appointments, 12

month appointments what have you the most help I got then as Thomas was approaching three etc was the education the pre-school special needs send them out do this all your possible statements, helping you fill out DLA, no one comes knocking on your door telling you what you should shouldn't do oh well under 5's he's not really entitled to this that and the other because most children at the age of 3 4 can't cross the road and I thought if there was a big bus coming they might know but you know...and err I was just like so really it was more on the education side with the help (laughter because of Sam – go and play in the kitchen, go play in the kitchen,

I: it was getting the information about what you're entitled to, the assistance in filling out the nightmare of DLA forms

S: the err they were the ones who did the four week assessment unit err because it was all in the focus of Wordsley hospital was gonna shut and it was all moving to Russells Hall etc but err, no as I say the assessment unit were really good "oh we'll send someone out to help you fill a form in blah blah blah" they were more sort of pushing you on "well we'll send someone out from the special needs education preschool blah blah blah" so someone used to come out for an hour and play with Thomas and see how his milestones were progressing and obviously they'd send a copy to Dr Ibrahim, a copy to my doctor blah blah blah so basically he medical side were getting the input from the education side

I: Right ok

S: which they had very funny clauses everywhere, it's like speech and language isn't it, is it education? Under the, if medical, they all argue over where it belongs so you know, that was it they would send people out to do the odd assessment can he post this can you do that, write it down everybody gets a copy goes in his folder that's it, so next time I go to see Dr Ibrahim she has go the information but we're still in the the six months at a time erm.. and then obviously we'll get the letter of so and so and this that and the other and you can apply for this or whatever, so it was more who do I ask and what do I do, and my next step was the disability team

I: and are they kind of a, a community outreach type of

S: yeah the Dudley disability team is basically if your child is on the register you have like a key case worker so to speak hence we barked up the tree for respite at the age of five cos I did listen to the other mums "oh there's no point trying to start respite when he's 9 " because he's going to be used to it isn't he so when he started school it did, it took us over a year to get him used to it, teatime visits what have you he only stays once a fortnight overnight but that's where I was being practical again, "Rob, we've gotta start it now, if I wrap him in cotton wool, and he's 9,10,11,12 we're never gonna even get a night's sleep"

I: yeah

S: do ya know what I mean it's gotta happen, and err cos, he was due when Thomas started school so it was a very big change for Thomas, new school, new brother, respite...It was all going on, but he managed and you know, we stuck to it and you know but things like that as I say the disability team if I've got any queries now where can I take him swimming what can I do, and I sort of phone them and they've got a team that will fish about for me

I: Right

S: saving me doing it all , which under the age of five I was, I think there's that big milestone that when a special needs of disabled child has gone five you get more help

I: Right

S: Under five there sort of “well, you never know” you know and all this and then all the statements started coming and what have you

I: Right what have you, so it was almost like once he reached that point then the help would start coming in and the advice

S: yeah it was like the age of four, and then it was like four and a half it started at the Easter at school so really it was sort of after his fourth birthday we started getting a bit more... so we did sort of pander about for a year just the odd sort of morning down at err under five special needs and blah blah blah and somebody would come out and play with a few toys and you just plodded along as a normal mum really... .. err... .. but we get far more help now.

I: and has the situation changed with the GP, are they [a bit more aware]

S: yeah I mean I presume so, I mean obviously the health visitor you know, sort of, but then that's only my doctor up here, a mile down the road you've got another one if they haven't transferred the information or generally said in passing one health visitor to another, oh have you ever heard of Angelman's cos I've had a patient blah blah blah, you know, they haven't got to give any names out, but you know it's...

I: Sharing information

S: but sort of things like the incontinence service as well, with these pull ups and things, you hear all these rumours “oh right, when he's four you can get free nappies blah blah”, who do I phone? Ask the health visitor, “ooh don't know, if you phone” and I'm thinking well hold on who do I actually go to here, but luckily, we did find out and it's just word of mouth other mums I speak to, and I say “oh did you know you could get free nappies” oh I didn't know that so them all on the phone now but it is, the most information you get as a parent is from other parents,

I: Right

S: That is the top line

I: you mentioned earlier that you're going to the conference in ... September

S: Yeah

I: and... .. is that a specific Angelman's syndrome or is it Angelman's and

S: No just Angelman's, because err, four years ago obviously I clicked on the Angelman's website blah blah blah, as surf, and it said conference blah blah blah but Thomas had... we'd missed it, Thomas had been diagnosed and it was the middle of the summer September and we'd missed it, and then we read that it ran every two years..... Two years after that we had this one (laughter) so two years ago I was I'd just had him, he was eight days old so we weren't gonna obviously go on that..... erm... I thought right we're really gonna do it this year, we're gonna do it, and as it happens, I got to meet this other Angelman's mum at Thomas's new school now, so I said to her, I says right get on the website we'll all go so at least we'll know someone there. There's gonna be hundreds of Angelman's big kids there and what have you adults, what have you, but at least we'll know somebody you know. And err that's when I was reading through the info about and I saw Chris' name on the thing, but err no, we're doing that err basically because I said I don't know whether we'd go every two years but then there's obviously I'm getting the impression we've never been to one, but I get the impression that it's all Angelman's families the workshops will associate with us, wills trusts special needs what have you the Angelman's what have you. But err, Dad will go one day, I'll go to

them the other, gotta look after you pair for one of the days (laughter) cos it's quite difficult, we haven't got carer as yet

I: right

S: so we're in the process of direct payments trying to get one, but there's nobody friend wise you'd singly pick out to look after our children...nobody would do it to be honest to look after the two, they'd have one, but then that defeats the object of, you wouldn't pay another 250 quid for a couple to go so it just means Rob has to tell me what happens on the one workshop and I've gotta tell him

I: Are you kind of...expecting anything from that particular conference

S: The thing I would say is gonna open my eyes cos the two Angelman's girls I've met obviously are girls, I've never an Angelman's boy and I'm very I saw a picture in the paper once of one who lives over Wolverhampton way and err you know I couldn't really ... see any of the features so much in him and I'm like right ok, and I said to Rob it would be so interesting ... obviously they're all families with Angelman's I'm so interested to actually see I don't know a 15 year old boy who's got Angelman's because it does say there's going to be like an AS err somebody doing like maybe a speech or something, one of the, somebody who's got Angelman's, I thought ok so he can obviously talk, you know things that which will open your eyes cos I've got this very image of angleman's in my head... its girls, and the two girls I know are the spit of each other ones 9 ones 11, same hair cut same teeth same eyes you know, they are the spit. Completely different families, but I'm like you know, it would be interesting to see older Angelman's people, and specifically boys or men or what have you

I So the curiosity

S: yeah

I: of boys in particular

S: yeah,

I: being as you have a boy with Angelman's

S: and being as I've only ever met girls

I: but also kind of the ...prognosis, would I be right in thinking

S: yeah, I mean, as I say you sort of...you do get your eyes opened err not just on the Angelman's thing on the whole special needs thing, wills, trusts, whatever happens in the future, I mean there are going to be things aimed at the over 16s etc when they're going to adulthood, which as I say for some reason in my little head I keep jumping that far ahead, obviously I wouldn't go and attend a conference yet as in life after education because I'm not....I'm still curious

I: uh huh

S: what would happen if an 18 year old said, well he started learning life skills at college and that, and I, IU do think that far ahead...errrm because I think right because I think he's just going to (inaudible), because in my eyes he just goes to school and learns what they...he's never going to learn to brush his teeth at school, silly things like that

I: butter his bread, do beans on toast

S: yeah that's it, you know, so life skills I think, they are good in the special needs school because they do take them out and about because they try and teach them as much as they can

I: errm,so with Thomas being your first, would you be able to think back to when you were pregnant with him, what your expectations were

S: in all fairness... I was quite lucky, I worked up to three days before (laughter) I cut my hours down, I found out I was pregnant err in the sort of July, in the summer I cut my hours down, it was Christmas Day Thomas was born so I worked up to the 22nd December, just part-time, I'm only pregnant, I'm not one of these every two minutes, you know I didn't suffer with any sickness or anything I did have a good pregnancy, and obviously we had him. He was early, he was about 11 days early, he wasn't due till the January, but you know as you do. I hated the birth bit that was awful, urrrgh, absolutely vile, but no, they come out "is everything alright" you know, yeah everything's fine" you know, not even so much as they'd say a little bit of jaundice everything was fine, fingers toes the lot, I mean times have changed now, they do hearing at birth now oh I don't know I lose the plot. They did heel prick tests what have you they were fine, no problem. That's it off you go home" then the midwife comes out does her bit you know two weeks later, go and keep getting him weighed every so often, that's it, he's eating, which he was, everything normally happening the early stages, he was having his milk, and you know he was eating his rice and porridge and all this, mash this up you know, nappies no problem, still in nappies still young what have you. Then he started, as you say you get to one, still fobbing it off and you know, as I say as a first time mum I didn't know what to expect him to be doing should he be holding his spoon and feeding his, should he be dunking his spoon in his porridge or what have you I don't know so I'd still sit there doing this, feeding him.but having him (Sam) now, I just stick a bowl in front of him he's like, he's not two till August, stick and there he is, gets it all down him I: He's having a go

S: But if it had been the other way round, I think I would have probably, it would have probably only saved about six months... maybe a year because I would have probably been nagging more at the age of one and a half. I'd have been like well hold on my first was do in that blah blah blah but you know and well you do listen to them when they say boys are slower than girls and you believe everything they say, if someone said Thomas's got Angelman's he's gonna grow three heads, grow three heads when he's twelve and you'd go oh is he? ok (laughter)

I: ok, so in terms of the actual birth everything was fine, no questions, nobody was worried, and then obviously kind of oh well, boys are slower dad a da, being fobbed off with the usual stuff, so no inkling that anything was going to be at all different in your life's to friends that had already had kids.

S: That's it, and I think, on the medical side, what made it as well there was no health issue with Thomas, there was no hole in the heart, no asthma, there was no, in himself he's healthy, if you get what I mean, but obviously the brain side of it isn't that healthy, but obviously I think ... maybe if there had been a bit of a... I don't know asthma thing breathing, kidney whatever something going on, they might have, picked...I don't know I think because he was healthy as such, putting on weight, nobody was concerned, that was it.

I: ok...errm, how would you say your experience of being a first time parent compared with your expectations of parenthood.

S: mmmm, as I say, the first year or, sort of year and a half it was pretty normal what I would expect, I was doing the baby food the nappies the bathing, the usual, down to the toddler group blah blah blah, that was another thing as well the toddler group . Obviously the kids are roughly the same age...and I sit there and I'd be watching and I'd be like ok, "how olds yours again"

I: Making the comparisons

S: yeah to compare, which as I said I was pretty sociable I wasn't going to be one of these mums who sits in the house and not do anything. So even before he was diagnosed of we'd go and then I'd be like right ok, so he's just you know, he's just picked that crayon up and started colouring, how's he got on that chair, how's he climbed on that chair, things like this. And now we'd be sitting down there, and then eventually when Thomas, we did sort of, not take the Mickey but, I said at toddlers, one of these days our Thomas's gonna walk through that door, cos I always had to carry him or he was in his buggy...and then time would tick by "I am he's gonna walk through this door" you will walk (laughter) and then eventually as he was sort of getting too old to go to toddlers he was, though I say he was walking at two and a half he wasn't like walking you know, kind of like down to the shops he was just walking from here to there sort of thing... So, no, it was a bit err, it was a bit err of a shock to compare which I did compare with other mums, but it was what I expected to be honest, until about one and a half

I: when things weren't quite....

S: mmm, well they keep telling me boys are slow, I wouldn't mind but our lads eighteen months and he's ooh what have you you know, so. You can expect the, you can accept it when they say two or three months difference sometimes and then there's always one who say "oh mine was two before he walked". Then what I found after Thomas was diagnosed, was other mums would be panicking and saying "Sarah, oh god, my little lads not walking yet and he's twenty two months...

I: do you think he might have

S: do you think he's got Angelman's, err no, you know, it'[s sort of put the wind up the other mums to be honest thinking so that if they were first time mums themselves they'd try and compare "oh mine's not walking either"

I: and how did that feel when they were saying "do you think he might

S: yeah and I'd be like well no, because obviously at that point I'd say well now because he probably wouldn't have Angelman's because there's loads of syndromes if there is something the matter with him or he's or I mean it could be something as simple as he's deaf in one ear or something, you know I don't know. If something is a problem, it's not going to be Angelman's, I mean you'd be you know it's gonna be quite rare for somebody up the road I mean I was saying one mum lived right down by Wordsley she's in Dubai now, but you know, it is a bit weird so I said no you're barking up the wrong tree really. You know, you try to put them at ease, but they still flap (laughter)

I: So Thomas's now 6...and a half,

S: yeah seven this Christmas yeah

I: Kind of a mixed barrel of emotions I guess throughout his life

S: Yes it sort of I mean he's constantly trying to sign brother, obviously he keeps doing that instead of that. But he's like where's my brother sort of thing, he loves him to bits but you can't leave them. To sit here, they'd play lovely if I was sat here, no Thomas, don't kick, don't throw blah, go in there, that's it, kicking pulling fighting, smacking, chucking Sam up the window and what have you, you know it all goes pear shaped, so he does need the attention, he does need the one to one, I would say, which is obviously very time consuming. It's like from three o'clock when he's back home from school, that's it ... nothings done (laughter). I'm constantly sat here watching the two of them, which on a normal life, the family life, it does, it's a bit weird when you try and do a like a healthy-ish tea, sit down together what have you it's not gonna happen for the fact of I

can't boil some vegetables up in there, and have this going on, or try to cook an omelette or something while I've got Thomas, you know, so if Dad was to work a bit late and come in so this is where convenience foods, or... I've done a meal it's on the plates by three o'clock and we have to microwave it... This is where, it's not as simple as cos Thomas can reach everything now, when he was little, I mean he's what six and a half and I'm buying him aged 9-10 clothes. It's not, he's getting a bit of a belly on him, but he's not fat that way, he's just big for his age. He's solid, I mean I'm quite broad, but he's solid, whereas the two Angelman's girls I've met are quite skinny, and I'm like is he gonna be the only fat Angelman's kid about, well not fat, but big. You know what I mean, but obviously he can reach everything now, cookers switches

I: so you've had to "Thomas-proof" everywhere now

S: oh we've got the catches on the doors there, they were one of my bargains off the internet I found them. Cos I mean I have to have those on the bedroom door and on the bathroom everything, but obviously I can hear him cos there's a gap which is nice rather than shoving a bolt on the door cos I think that's a bit bad.

I: ok, errm, just one more thing, (interrupted to move Sam who'd fallen asleep on mum) so you mentioned the conference being older kids in general, so you have any specific expectations for Thomas, I know you've said I know he won't be able to do this that the other, do you have any kind of hopes for what he will be able to do

S: I mean I'm sort of aiming up the line that when he's sort of left school, on that front, that he will, hopefully he will, well, this might be aiming a bit high but say like a self contained flat with maybe a warden what have you rather than having a live in carer, but now as time ticks by now I'm thinking the live in carers probably looking more realistic. You know there was a time where I thought he could be in a little annex with other other ones, and you know a warden and I'm thinking no I don't think he'd even get to that stage now, so no my mind has changed. And like Rob will come up with ideas, we can convert the garage we can build this and we could get a carer, and I'm thinking getting a carer to live in, 9 times out of 10 carers have go their own families so I'm like you know, this is where you have to sort of... .. I think I do worry more after the age of eighteen. Whatever happens up to then I shall take with a pinch of salt

I: yeah, it's certainly struck me from talking to you, that its post child

S: Childhood-adulthood

I: that transition, almost as if that when he can make a path for himself

S: Yeah, we would sort of like I mean we obviously are realistic and thinking ok 9 times out of 10 parents die before their kids, basically I said to Rob we're not gonna be here forever and a day... when he's sort of eighteen nineteen twenty twenty-one what have you, we've gotta start getting Thomas into a routine with somebody else, he can't always have mum and dad. I'm not gonna wrap him in cotton wool until I'm seventy-odd because when I suddenly die what's he gonna do, that's it, so I do

I: so again, quite practical

S: yeah quite practical as in I couldn't like just disappear and I'd be like I've left, you know, it would be, I think that would be awful for Thomas, that's why get him involved a lot now, even to walk down the pub, we used to go down the pub, I mean everyone knows Thomas on the estate, you know it's like, if he escaped down the front door, someone would bring him back you know it would be like. We do get him involved in a lot of social things for the fact of when he's older I think he's gotta do it, you know they

go on these residential they do this that and the other but I do feel as though he's got to learn to be around other people etc as opposed to me and Rob.

Interrupted by front door

S: sorry about that

I: yeah so kind of in terms of expectations, looking more for him, carving his own way as an adult and it I guess one of the things that struck me is ... still that sense of hope that even though he's not done it kind of at the age of three...there's still a chance that he might do it and not giving up completely.

S: no, yeah. It sort of I mean you do when you speak to [people you're very practical, like my mum and other and as I say grandparents flap a bit more and I'm quite blatant no he's never going to talk don't worry about it, that's just me, when people start badgering me. But then, if I'm with Rob, and like I'll sit here with Thomas "oh look your brothers signing red, Thomas red look there's red look red" and errm you know I still badger him and still go on but to other people I "no he's never going to talk" and I do because you just get that " oh he said mummy, he said mummy" I said "that's all he can say"

I: To you it's nothing particularly eventful but to everyone else

S: it's like when we go over to the campsite and they'll go, "Sarah, Thomas's asking for you" and I'm like running backwards and forwards, what do you want Thomas, "momomomomomom" he doesn't actually say mummy it's momomomomomom or what is it homhomhomhomhomhomhom which is home and mmm which is his cousin Emily that's it, three mmmm s so of course he's sitting there and all these other five year olds going he said mom mom mom, they think its fab cos they he can't talk and he said mum so you know, I think with the little the other kids they're very sort of but he said mum whereas I'm I tell adults no, terrible.

I: so kind of your expectations to others are played down a bit yet you're still, you've still got...

S: I think I am realistically barking up the tree for him to learn some signs now. I've got to the point where ok speech possibly not, if he does it's not going to be very clear, if he does say anything so people still aren't going to understand him. So I'm thinking, right, as I say signs, he's not going to go it's like they're on about communication aids but I thought if he can't, if his motor skills aren't that great to work them in the first place but as I say, sort of, I do trying an aim at practical signs that not a deaf person or a makaton would know, something that if I went like that to you you'd know I was hungry, something to eat, or what have you or knife and fork and what have you, things that Joe Bloggs down the street is gonna know, or if he was pointing in the pub to coke there and he was going like that, you know, not learning the signs for coca-cola, I'm not gonna go that far (laughter) so you know I do sort of I've put that down as the individual education plan, the IEP at school to try and learn him like say learn him teach him (laughter) three basic signs like as in food drink toilet something like that, that would be it for the whole year, learn those three, that is it, you know, which suits me fine you know as long as he can pick up things like that. But that is one of my issues as well, the toileting, I do compare that with other Angelman's kids not other children now, I find myself asking other mums so how old was he when da da da da. Because Thomas is still one of the younger ones that I know, the others are all older now that's another thing that I when's he going to be driving (laughter)

I: is there anything else that you're surprised I haven't asked

S: No not really no, as you say it was the actual, no as I say generally it all happened within the year of the big shock of being you know and as you can imagine if you said you'd got something growing on your foot you'd go on the internet and have a look, it's just one of those things, but twenty years ago you'd have to go to the library there'd be nothing on it whatsoever you know, you'd struggle to find you know things now. But as I say that's the minefield isn't it, the internet. But everybody, all friends and family have done the same as well, and we've had other friends turn round and say the bit I read so has he got... and I'm thinking grrrr because obviously we'd been told from professionals and they've read so can Thomas do this or can Thomas do that, and I'm like well we don't know until he gets older you know, they as I say we have had other famil/friends sort of read up and think they're suddenly doctors you know it's look they all know because they know somebody with Angelman's you find that some people think it's a bit of a like well I know that little boy who's got Angelman's as though it's the best thing in the world and I'm like you don't have to live with him.

I: ok, that's enough of me, errm how have you found that generally

S: fine that was fine, you err obviously wanted to know how as a family we felt when we were actually diagnosed, but obviously kept pulling me back but that's fine, but as I say it's sort of yeah I mean, I think if you had come round here in like 6 years time, I'd be like I can't remember, but because I do tell people regularly that he was two and a half when he started, and he was da da and it's all still quite fresh in the last three four years or so, but I think as I say in 6 years time if you were asking I'd be like oh I don't know, cos since then he's been to three schools and he's done, and everything else has gone on, and it's not like I could always find the information out, I've got every blinking folder in the world but I do think being closer to what you were trying to ask me when he was born especially was he ok, the birth I think most parents would remember, the diagnosis, you sort of

I: it happens

S: yeah, you remember what age it is, and I can still remember how they approached and told us, we were upset in the room, but they weren't vile about it, oh he's never gonna do it, they have gotta be sensitive in that profession. But I don't know things like that when he's twelve I'd be like well I can't remember, I think me and Rob had a bit of an argument and things, but I don't know.

I: Thank you.

My immediate thoughts:

You covered all the areas you had intended in the schedule.

It flows well and you have a nice balance between letting her decide the direction and bringing her back to the schedule.

Surprising just how many interruptions there were, and also realise you had to do the interview with at least the younger child there – is this an issue in continuity? Could anything be done to try and minimise interruptions.

One theme seems to be of how to know if something is normal. The process of comparing notes with others reminds me of illness perception models where this is exactly what is described (e.g. in relation to aches and pains of old age, or in relation to deciding whether a lump is normal).

This makes me then think of other aspects of the health beliefs models (identity, cause, time-line, cure/control etc.) – may be worth checking if all are potentially covered in the

interview. She certainly went on to timeline pretty quickly, thinking about how he would be as an adult.

The power of professionals comes across quite strongly – the way she defers to their knowledge and expertise (how could she do otherwise); the weight of their words; the disappointment with the GP/Health Visitor's lack of knowledge and curiosity; the usefulness when education and health join up and frustration when they don't.

Stages of a journey? (Reminds me of Keady and Nolan in relation to dementia) Normal child/family – possible concerns (issue of being a first time parent)– definite concerns – seeking help - period of limbo between seeking help and getting diagnosis (would have been much worse if prolonged)– diagnosis (issues of self-blame and other's view of where blame might lie; issues of expectations set up by the medics when the diag was given) – adaptation (of expectations, emotional response, ways of coping, impact on relationship with partner).

Seeking further information and trying to second guess and prepare for the future seem to be big themes.

Sense of her being on her own seems quite a big theme too – with 6 month appointments. More of a sense of support once he is school age.

Between the lines – her fears for her own life and independence that may not be as expected in the future?

No mention at all of her feelings towards Thomas.

There is info about the sibling relationship now but little speculation about how it will develop in the future.

Appendices

APPENDIX 8
EXAMPLE OF ONE INTERVIEW

1 RF: It would be helpful if you could remind me how old Andrea is.
2 S: three
3 RF: she's three, and is she in school
4 S: yeah
5 RF: and is she your first child
6 S: yeah
7 RF: and you've got 2
8 S: Holly, one more
9 RF: And how olds Holly?
10 S: 18months
11 RF: OK. Errm, I'd like to take you back to kind of ... when you first received a diagnosis
12 S: yeah
13 RF: of 1p36. Before you actually got that diagnosis, did you have any concerns that things
14 weren't quite right
15 S: oh yeah, we got the diagnosis quite late... erm she started off, ooh gosh as soon as she was
16 born, she was floppy and blue, and cold. Errm, but they discharged/ they said she was fine
17 and they discharged her and within 48hours she wasn't feeding, she wasn't doing anything
18 particularly, erm so she was in special care, she was there for a week or so and they did every
19 scan and test and said there was nothing wrong with her and she picked up a bit and she was
20 ok, and then she started fitting when she was five weeks old and then was back in again and
21 then she had a general sort of diagnosis of epilepsy.
22 RF: Right
23 S: nothing specific, just epilepsy and that was it, and then she stopped developing...
24 completely
25 RF: was this kind of the first few weeks of
26 S: yeah the first two/three months I think there was three, about her three month check in spite
27 of having epilepsey she was actually doing what she should have been doing and then she just
28 stopped.
29 RF: Right ok
30 S: completely stopped and then she didn't develop, she didn't sit up she didn't do anything,
31 erm and then they said she had West's Syndrome
32 RF: Ok
33 S: err
34 RF: did they explain what that possibly was
35 S: they explained in technical terms what is was they didn't/ they kind of put it forward as a
36 diagnosis which I now know it's not a diagnosis its more of a symptom than a diagnosis as
37 such and then just a name given to a collection of problems and symptoms so erm, we kind
38 of hung our hat on that for a while and then that's, we were seeing neurology then obviously,
39 we were going back up the road to Alder Hey erm, and then it was her neurologist, came
40 over from Alder Hey, had a look, literally looked at her, her features and her face and her
41 history and her EEGs all those kinds of things and said he thought she might have West/erm
42 this 1p36 and said "you know, we've never come across this before" and "it was just a long
43 shot". So they tested her for that and err tuberloscerosis and a number of other things
44 similarish things, and then of course that takes forever, you've got to get bloods done, then
45 the results come through, we were April before, April two thousand and seven, [she'd
46 gone one]
47 RF: [So, she was]
48 S: when we got the diagnosis

49 RF: so when they said it was possibly West Syndrome
50 S: yes
51 RF: and you say you kind of hung everything on that
52 S: yes
53 RF: how did it feel to actually get some idea that there could have been a name for what was
54 S: erm it was horrendous, huh huh, we'd just, we'd kind of come to terms with the fact that
55 she was probably epileptic and obviously there are lots of people wandering round who are
56 epileptic who have perfectly normal lives and it was at the point where... they said West
57 Syndrome you come back and you do all the reading up and you realise that you know, it's
58 kind of a long term problem err so yeah we were devastated, quite upset
59 RF: so really, you've kind of had three... separate moments of kind of receiving a diagnosis,
60 with the [epilepsy]
61 S: [yes]
62 RF: West Syndrome and then saying [its 1p36]
63 S: [and then the 1p36] yeah, it's come in stages, I think they're a little more on the ball now
64 they, I know, a lot of people who've had problems and they, the diagnosis has come very
65 quickly because they immediately take bloods now in special care and send them off
66 whereas things have changed a lot.
67 RF: so kind of thinking through in terms of err ... actually having all of this stuff going on
68 first kind of 48hours, she's not quite right but the hospital sent you home
69 S: mmm
70 RF: off you go. In terms of your expectations of first child, how were those kind of first few
71 weeks different/
72 S: erm I think it was weird because I'd never done it before, I didn't really have anything to
73 compare it to and you just go you know , we were back in special care and she was in the CT
74 scanner, she's NG tube up her nose and all the monitors on her, err and you just... deal with it
75 hour to hour, you don't think I'm missing being at home you just immediately focus just on
76 the child and whats going on and at that point they really didn't have any clue what was
77 wrong with her and you know they canulated her, they did lumbar punctures and things which
78 were quite horrible and very painful so yeah we were you know really distressed, but... we
79 kind of because of coming I'm not a medical background but certainly scientific background
80 and with my brother being a doctor aswell erm it didn't really hold any horrid, I understood
81 the procedures I understood what they were doing and I understood why they were doing it
82 RF: Yeah, so maybe you were in a [slightly better situation]
83 S: [I wasn't totally] frightened and didn't have any clue about what they were doing I knew
84 why they were doing it, what they were looking for, I was quite happy to look at scans and
85 xrays and pictures, you know. I know what a lumbar puncture was and why they were doing
86 it so I could rationalise it a little
87 RF: and were there any moments when you were thinking I should be at home kind of
88 cuddling my baby and now I've got to put up with this
89 S: Errrr I think we were, I remember there's sort of like moments that burn into your brain I
90 was sitting on the settee in the parents room in the special care unit while they did a lumbar
91 puncture and I could hear her screaming and crying and I was with my husband and it was a
92 bit like "wow, whats happened" (laughter) yeah you know this is not supposed to happen and
93 you feel quite helpless but no never sort of... it was just something we just had deal with and
94 get over and then return back to normal
95 RF: would you say that you and your husband took quite a practical [approach to what was
96 going on]

97 S: very, yeah, yeah. It was a case of I know what they're doing and I can see that she's ok
98 you know, they thought it was some kind of infection/ had I had any sort of inkling then of
99 what was to come I would have reacted completely differently
100 RF: in what way
101 S: oh I would have been just in pieces
102 RF: right
103 S: or had I known before she was born or anything like that what was going to happen years
104 down the line it would have been different, I wasn't really that, that, I was really upset but I
105 suppose I wasn't really that frightened I could see she wasn't really really sick and I knew that
106 they were treating her for whatever eventuality they were giving her IV antibiotics in case it
107 was some kind of like meningitis type, they did the scans very quickly, everything happened
108 very quickly, so you could rule things out very quickly it was just the case of thinking well
109 she's just slow to pick up, she'll be fine we'll be back home in a week. And then I can open
110 all the cards and the presents and everyone can come see her and then all that kind of all that
111 can yeah, we weren't too..... there were a lot of parents in there who were a lot in a worse
112 state that us put it that way, yeah we kind of coped with it and dealt with it
113 RF: ok, and I'm just thinking it was quite a long time, just over the age of one when you
114 actually got the official diagnosis all the bloods had been done here you go, your child
115 S: yeah we actually went in, went in to another appointment and that's when they gave us the
116 results
117 RF: and was that the neurologist or was the geneticist etcetera involved
118 S: just the neurologist was there at the time, neurology, epilepsy nurse was there, consultant
119 was there errm, and that's when they said "yes this is what it is" errm, and we just looked
120 totally stunned I think because we then the consultant said look you know, we left quite
121 quickly, and then he rang me that afternoon he's a really, he's a really nice guy he said "look
122 you know I appreciate you're probably thinking of a million and one questions come back
123 tomorrow and we'll, when you've had time to digest the information and we'll talk"
124 RF: and was that quite a helpful way round of doing it
125 S: yeah I wasn't in no fit state for him to tell me anything useful there and then
126 RF: What kind of thoughts were going through your head when they actually said
127 S: errm, that it's permanent that was the biggest thing that I knew having done, knowing a bit
128 about genetics and biochemistry it was it was a final, it is definitely permanent it is this, it's
129 not going to get any better...that was the main thing and I think and I didn't recall/ I can tell
130 you where everybody was sitting, the epilepsy nurse was sitting on the window ledge sort of
131 half sitting on the ledge and I remember her saying to me "you know there are some really
132 good special schools about" and it was that was the whack and that was the "woah" you know
133 we really are coming to (inaudible) that was the biggest thing that actually hit me
134 RF: so what kind of things did that conjure up for you "there are some really good special
135 schools around"
136 S: oh, basically immediately she's not going to have any quality of life she's not gonna do
137 anything and you immediately have a negative picture of special schools because of never
138 having experienced anything like that before, although by that stage Andrea had been to
139 groups and you know mixing with lots of other children with disabilities but you know still
140 holding out a hope that she's going to go to the school that I went to and then (laughter) all
141 this kind of stuff and things are going to get better and then it was like it's really not, and she
142 was quite bad at that point as well, so
143 RF: physically she was very bad

144 S: she was very flat and she regressed considerably, and she went downhill... badly she was
145 on a lot of medication, she was dopey she was floppy she wouldn't look at you she didn't
146 make any noise she didn't smile she didn't...she didn't move, you had to move her head for
147 her to stop her getting sores you know you think if this is, this is what it is this is what it's
148 going to be like
149 RF: just kind of thinking through some of the things as a parent, a newborn, one of the things
150 you read about is your child smiling back at you and kind of [being in tune with your child]
151 S: [YES! She wouldn't have cared if it was me there or anybody else yeah it would have
152 made no difference.
153 RF: and was something that you thought about a lot
154 S: yeah, God yeah, it was heartbreaking it was absolutely horrible, erm yeah, I know that
155 that's quite a thing that other friends have ... you know... said
156 RF: other friends, when you say
157 S: other friends as I've met because they're child has a disability, for them to not even look at
158 you and smile, especially when it's *so* labour intensive you know, you sit with her all night
159 and yes it does get quite demoralising after a while but she was just totally flat she didn't do
160 anything until... Christmas of that year
161 RF: Right
162 S: about three days before Christmas
163 RF: Just thinking, Holly is your youngest, and going through all of that one parent I have
164 spoken to before said that it's almost like ... being a first parent again/
165 S: oh it's weird, it's really quite amusing, she does things like when she's learnt to sit up, it's
166 like wow she can do that on her own (laughter) yeah everything works and you think while
167 they develop, by that stage you know a lot about development and what they're supposed to
168 do, you can watch for the signs you can see the rolling and you can yeah, and it was yeah very
169 easy yeah
170 RF: In terms of the erm her being in tune, the smiling, and the communication stuff, how was
171 that having not had it with Andrea
172 S: oh great, a breeze, and an absolute breeze, I've said many times how can anybody say that
173 having a child is difficult because really it's absolutely easy, it happens, she fed and she slept
174 and she you know she's quite advanced for her age as well, she's you know eighteen months
175 you can have a conversation with her and she's you know, she's totally the opposite end of
176 the scale, totally
177 RF: do you think it's made a difference, in the order they were born so would things, would
178 you have picked up on things had Holly been your first
179 S: no, no, I knew straight away when Andrea was born something was not right even though
180 I'd never had a baby before and had no involvement with babies or children before. I
181 immediately knew something was wrong with her and the geneticist actually offered to do
182 genetic tests on Holly when she was born because I wouldn't have any before I had her, err
183 but within seconds of herbeing born I knew that she was fine, immediately I don't know why
184 RF: was that kind of an instinct kind [of thing]
185 S: [YEP!] as much as I knew Andrea wasn't I knew she was so I never had anything, any test
186 or anything done at all
187 RF: Ok, and what was it, if you don't mind me asking that stopped you from going through
188 the amniocentesis test and other test
189 S: errr... risk. Basically, and also I... Andrea's great and we absolutely love having her, I
190 wouldn't change her or that for anything, and I wouldn't now have her without her problems
191 in a sense because she wouldn't be her, she'd be a different child altogether and so you know

192 you have to well, I couldn't. What if I did have the tests and I did find out... what would I
193 do, apart from get very stressed, I certainly wouldn't terminate because I don't believe in it so
194 errm, my consultant was great they did lots of scans for me, they did 3D scans, they did lots
195 and lots of stuff and lots of scans to try to rule out as much as possible because I said I would
196 like to know to prepare myself you know practically, errm but, I'm not really that bothered
197 about knowing that much and I obviously knew by that point that it wasn't hereditary
198 Andrea's condition because they tested both me and my husband so that was the other thing,
199 so my logical mind knew that the chance of it happening again were very slim
200 RF: ok, so the genetics test for you and you husband, was that something that you requested
201 or because you were going to have another child?
202 S: yes, I didn't want to have another one until I was sure that it wasn't genetic, because if it
203 had have been genetic then we wouldn't, we wouldn't have had that, we wouldn't have had
204 another baby
205 RF: some parents I've spoken to about the idea of it possibly being something hereditary have
206 gone through kind of periods where they've blamed themselves saying oh well maybe it's
207 come from my side of the family and the other one saying well maybe it's me
208 S: we knew quickly because they tested us all at the same time
209 RF: Right, so there wasn't that anxiety around
210 S: no I asked them to, as soon as they tested her, I then asked them to do us aswell... because
211 by that stage they knew that it probably was something genetic and I said well rather than wait
212 because I know how long these things take, you know I went in with Andrea and had bloods
213 done and they sent my husband in...Again I think part of that comes from an understanding of
214 physics
215 RF: that's what I was going to ask, maybe you had the upper hand in terms of knowing one
216 that it takes a while to for these things to happen, so it's best to get it done at the same time
217 but also it's something that you needed an answer to, for both of you
218 S: yes definitely, it was a practical decision and the other thing that hurried it along was my
219 cousin, my first cousin was pregnant and I knew I've got a lot of cousins who are all, we're all
220 a similar age and wanted children so it wasn't just me that because its often maternal not
221 paternal errm you know there were wider implications for other family members so right...
222 you know, we'll find out now and then we can make that decision
223 RF: do you think maybe, you felt kind of an obligation to that cousin to find out
224 S: oh yes definitely, yeah, because when I'd spoken to her I knew she was quite concerned
225 about it and when you are pregnant you know you get a bit irrational about it at the best of
226 times errm, yes, so we sat and we went through and we went through generations, did we
227 know anybody on either side of the family who had any kind of issues or problems, but you
228 know we didn't so again in your rational mind says well chances are probably not just done
229 over mutation that's it it's happened
230 RF: just thinking back to you say you can remember where everyone was sitting
231 S: yeah flashbacks (laughs)
232 RF: one of the things you picked up on was the epilepsy nurse saying she's going to go to a
233 special school
234 S:yeah
235 RF: did that give you any kind of ideas around what other people might think about you
236 having a child with special needs
237 S: no, no thoughts no
238 RF: yeah, so yep we've got a daughter and off we go, quite a practical approach

239 S: yeah, it's not erm we've never thought a lot about what other people thought actually,
240 erm, a lot of that comes from there's big family on both sides erm and everybody's great,
241 really supportive, and everybody absolutely adores her, it's not erm, it's never actually no, its
242 its been a whole new world and it has been interesting and I've met some really great people
243 and I don't regret that. There are times when it's incredibly upsetting very stressful, but I
244 wouldn't now change it
245 RF: uh huh
246 S: I would have her not sick but I wouldn't change her, (inaudible) yeah if she wouldn't be
247 sick or in hospital then I'd be happy, that would be fine
248 RF: because you mentioned earlier when we arrived that kind of being in hospital, physical
249 health problems
250 S: Yes
251 RF: what kind of difficulties in that respect does she have
252 S: oh she's a nightmare, erm she's got chronic (inaudible) respiratory? Disease now, erm
253 she's got a severe scoliosis which doesn't help because that squashes one of her lungs and so
254 she's got a lot of lung scarring, she has very poor muscle tone, so erm, she started when she
255 was very young with reflux, and so she suffered a lot from reflux and vomiting, didn't put any
256 weight on erm lots of diarrhoea to then she got an aspiration pneumonia because she couldn't
257 swallow and so that sets for allsorts of other problems then and ultimately they gave her a erm
258 Mickey Button so now she doesn't eat
259 RF: so she's is that PEG fed
260 S: yeah
261 RF: Mickey Button that's a new one on me
262 S: well the PEGs the first stage, the PEGs the one they put in with (inaudible) attached, it's
263 just, it's just the name of the actual equipment. From the PEG, it's got the actual attached tube
264 on it erm and then they put those in first and then they changed it when they did further surgery
265 to a Mickey button (inaudible). It's neat and you put the tube in and then you take it off.
266 RF: right
267 S: it's like a neat little button, it's quite unobtrusive. So she doesn't eat at all now and she's
268 had to have (inaudible) so she's had to have a fairly major surgery on her tummy and she's
269 had her (inaudible) because she was suffering a lot from abdominal cramps and lots of
270 problems, she was just losing weight constantly
271 RF: as a result,
272 S: Yeah she was like a rake, so she was having lots of periods on IV fluids and stuff which is
273 not very good, so she's had a lot of (huh huh) problems, bless her
274 RF: And you said as well, quite a few periods certainly around the winter time of
275 hospitalisation
276 S: yes erm, October November December last year
277 RF: mmm
278 S: approximately two weeks in two weeks out, this year she had surgery at the end of October
279 so she was in Alder Hey. Then she came out, she got a pneumonia the beginning of
280 November that was a short stay in back out and then she became very ill..... middle of
281 November and came out in January
282 RF: Uh huh So a lot of kind of tooing and froing for you and her
283 S: Yeah, well it means we have periods where by like last year when she went to Alder Hey
284 and my husband and I went with her and never stepped foot out of the place for three weeks.
285 Wwhich in itself can be quite hard
286 RF: [stressful]

287 S: Yeah yeah we had Holly as well, whereas the previous year... although I was pregnant
288 obviously there was only me and Andrea really (inaudible) and going to work and
289 stuff.(inaudible) going in Alder Hey, Holly got left with the neighbour for half an hour and
290 then we, it all went horribly wrong and you know I had to ring her up and say look we're in
291 Alder Hey and we're not coming back.[laughter] she had her for about 3 days [laughter] yeah
292 yeah.
293 RF: so it sounds like, not just in terms of family but local, kind of... community, neighbours
294 etcetera there's a lot of support out there in terms
295 S: oh yes they're great I think the Wirral programme (inaudible) aswell, we have a lot of
296 support and care. I think maybe because of the geography, errm, but we have, I know that I
297 get care here that I spoke to a lot of other parents (inaudible) and they don't get anything
298 RF: Thats the local hospice
299 S: yeah, I get twenty-days in care thats a year to be taken whenever I want, that can be
300 anything from a couple of hours, over night, two days, three days, and that is a fantastic place.
301 She absolutely loves going errm we get the continuing care thing, we get carers who come in,
302 I get two full days care a week aswell...
303 RF: Brilliant
304 S:...because I go to work
305 RF: Okay, so is that the biochemical stuff or [something completely different]
306 S: [laughter] [I'm a lawyer][laughter]
307 RF: completely retrained
308 S:completely retrained, now I'm a solicitor I work three days a week and they are here when
309 she gets home from school until half seven at night. Great. We've got them, we've got a
310 great hospital, the home service so we avoid actually going in as much as possible. And then
311 when she's ill, like when she was ill when she came out of hospital in January errm, she was
312 still very, she was on a lot of oxygen and she still had her IVs in because she was on a (?drug
313 withdrawal?)(inaudible) programme and you know she was pretty bad and I had care every
314 night, every day
315 RF: Right
316 S: So they're really, really good
317 RF: so when you say The Wirral, and it's geographical basis kind of helps with that, you
318 mean The Wirral as in terms of kind of local authorities and the hospitals and
319 S: Yes, because we're quite a small area, there's only I think theres 60 parents at the meeting
320 the other day, there are 60children on The Wirral that have got severe multiple learning
321 difficulties and out of those, 30 have got profound so we're quite small in number
322 RF: and so big provision
323 S: so we have (inaudible) the local hospice, they have lots of groups we have hydrotherapy so
324 lots of, yeah you get to know a lot of people and there are lots of services available
325 RF: and I guess kind of knowing that you've got that provision helps with the multitude of
326 professionals that you've got [involved]
327 S: [Yes] yes, we tend to have errm the continuing care co-ordinator does just that you know,
328 liases Dr (name), other people if necessary
329 RF: good communication amongst all of them
330 S: yep so errm, it's err quite a good set up and we're lucky enough to have Alder Hey, on our
331 doorstep. It's a really good hospital, so we don't have to travel you know, they do a lot of
332 clinics in the hospital so we don't have to travel, you know hours, like a lot of people do, just
333 to see the specialist, they're there and then when Andrea needed them, you know for

334 ventilating, you're twenty minutes across in an ambulance and you're there. So we are lucky
335 in that regard.

336 RF: It, it sounds aswell that you kind of hold the Neurologist in quite high regards because of
337 the work that he'd done would that be right?

338 S: err yes, actually yes, he's a strange gu, bedside manner is atrocious errm and he did upset
339 me a few times but errm his knowledge, specific knowledge in relation to genetic conditions,
340 is ... is good and you know to be able to pick up on what she had literally just by looking at
341 her is errm quite, you know he's good, and I don't really care what his bedside manner is like
342 because he knows his stuff.

343 RF: so his professional knowledge is kind of up there and thats the bit you're after

344 S: absolutely,

345 RF: errm and that day when you say you just got out of there really quickly, did you kind of
346 come back and look on the internet, have you looked up for more information about it, or had
347 you already done that from...

348 S: I'd looked up some information before we went because they said it could be this so, the
349 first thing you do is look on the internet you find the one paper that there is on it, which yeah
350 it is incredibly depressing.

351 RF: in what way

352 S: oh you know because they talk about all the other associated problems that they might
353 have, errrm.... so I kind of read a little bit and thought "well there's no point reading all about
354 this, we don't know" so we came home and I phoned my brother on the way home/my parents
355 were there, they were waiting in the cafeteria in the hospital so they came in and they phoned
356 my brother who by the time we had got home had found out a lot more and then when we
357 went back the next day, we went through the papers that were available.

358 RF: sorry with the neurologist

359 S: with the consultant, dug them out, I think downloaded them and handed them over, so we
360 sat and went through them.

361 RF: Yeah, ok . And was that helpful to kind of that/

362 S: Yeah, it was, it was you know we always kind of, I think having a practical plan helped to
363 distract you. You think "Ok, well, this is it, but now what are we going to do" because that's
364 where we sat and made a list of who she needed to see and why and let's get this checked and
365 get that checked out, do this do that, so kind of a kind of a practical, practical plan. Go ahead
366 with something to distract you

367 RF: Yeah

368 S: so when you're in the middle of all these appointments/we were having appointments two,
369 three times a week. You don't obviously think, it hits you afterwards

370 RF: yeah, I would imagine theres not quite a lot of time to actually process what's been said
371 to you

372 S: you just go from day to day, hour to hour, the next one, and I think it really, errm, because I
373 know after the incident in December when she was very bad, it probably hit me in about June,
374 July this year, when she was well, and thats when i, I went to pieces totally.

375 RF: So is that, about the time when you have actually been able to sit back and relax [because
376 she's come out]

377 S: [yes]well she's put some weight on, she's sleeping and all of a sudden I've got time on my
378 hands and you start thinking and you do definitely get tired aswell errm and you know it was
379 very very stressful and you do just tend, I think you just crumble a bit then, but the rest of the
380 time you jsut don't have the time [laughter]. You don't have time stop and get upset and
381 think, you jsut have to deal with it...

382 RF: Erm sitting down, going through the lists of well we need endocrinologist,
383 cardiovascular, all of this kind of stuff, at any point did you, I mean you've mentioned earlier
384 you've thought "oh my god this is permanent", had you thought about kind of a future and
385 erm maybe what that looked like immediately
386 S: erm yeah, you immediately panic and think oh my god, you know she's, and to be fair we
387 initially for some reason and I don't know why, it never clicked with me, that because of this
388 she was going to have a limited life, it just never really entered my head, I don't know why.
389 So immediately you think what's going to happen, certainly think what's happen when I'm
390 not here, that was the first thing that I thought about
391 RF: so were you thinking in terms of what happens in terms of when she hits adulthood
392 S: yes, yes you know who was going to be there for her, whats gonna you know, who's gonna
393 look after her. That was the first thought and then you start thinking about more practical
394 stuff and it was only...it was quite a while after we saw her consultant again and he said it was
395 an aside within conversation "well of course with her having a limited lifespan" you know
396 and it was that/nobody'd said it, I think they just thought that we realised it err and I should
397 have done, I should have done really I suppose
398 RF: Did that prompt you to enquire what, more about what a limited lifespan meant
399 S: no
400 RF: do you have any idea now or
401 S: they don't know, there's nothing about her condition, obviously her condition is stable in
402 the sense of her genetics, it's not gonna it's not a deteriorating condition or anything. So it's
403 not going to be that that she has any problems with, it's probably you know respiratory
404 problems that you know they said would gosh, not this winter thats just gone, the one before
405 you know, "just prepare yourselves for the fact that she's been in hospital and she's on lots
406 and lots of oxygen you know, we might have to ventilate her at some point, and then if we
407 ventilate her she might not come off the ventilator and then and then and then and then, so
408 that was just, she was only one and a half, two, and then she got better and came out and then
409 it did happen this year but she did obviously recover from that
410 RF: so that must put kind of a different spin on how you're looking at those situations when
411 you've been [told this might happen followed by]
412 S: yeah, they'd never said that to us before, they'd never, nobody, they'd never actually say
413 "now we expect this to happen, we expect that to happen" because we've learnt not to expect
414 anything to happen, that she just, seems to see through what she wants to do and she does
415 things in her own way and she never quite follows the same pattern as anybody else and she
416 just you know, we decided a long time ago there was no point in pre-empting what might we
417 just get on with it.
418 RF: So expectations are kind of out the window
419 S: Yes, Oh yeah, it's you know, whereas at the moment even her consultant said to me the
420 other day "I bet you wish you could fast forward now straight through Christmas and New
421 Year to next summer don't you" – Yah, he said "I know, it's going to be very stressful for
422 you" so but you know
423 RF: it sounds from earlier when we were talking that kind of your way of actually coping with
424 that is to kind of keep away from public areas and go off to warmer climates
425 S: If I could keep her at home and never let her out the door then I would, I would
426 RF: for that period of [time]
427 S: For that period of time, but then again, it's not fair, she's enjoys going to school and she
428 likes going to groups, swimming and so. It's got to be a balance but we are you know
429 incredibly careful and you know if she does have to be hospitalised for any reason at all then

430 err I know it's going to be incredibly stressful and the consultant is aware that if we ever go in
431 now we ring him and he makes sure that all (inaudible) have to be there and they canulate her
432 straight away, and they hit it quite hard, because we do a lot at home, oxygen at home, to a
433 certain level, we have quite strong antibiotics we can give her aty home so we keep her at
434 home until absolutely, til we hit the point where she's got to have IV basically.
435 RF: Right, so in terms of having an oxygen supply at home, have yourself and other members
436 of the family had to be specially trained for that side of things, no, there's no special training,
437 you just get on with it. You know she's got an oxygen concentrator upstairs, we've got
438 portable oxygen all over the place, I've got one in the car, you do learn whilst your're in
439 hospital/we have got an oxygen nurse as well though
440 RF: so you've got the different support for all those things
441 S: oh yeah, we have an oxygen nurse, who's just brilliant, she came to see me in hospital and
442 went through all the things like don't smoke – huh! [laughter]the most (inaudible)of safety,
443 don't smoke, but I'm like “ well why on earth would I smoke”, and she said well believe me
444 people do, so erm there's a lot of equipment that goes with it, but you do learn, we've got a
445 suction machine, I've had to use that, obviously the whole Mickey Button, and the feeding
446 pump and stuff like that soso I had to learn to do that
447 RF: Ok, and erm would you say certainly around this time of year it's made yourself and
448 other members of the family maybe more vigilant about what's going on for Andrea, and have
449 other people maybe come in carrying
450 S: oh yeah, definitely, well I mean the whole family know now so, they won't come...if
451 they're not well they stay away and everyone is a lot more careful, just general hygiene and
452 washing hands and just keeping her out the way. So she goes to no routine appointments
453 now at all, and if, school are aware aswell, if anybody in school has a cold, teacherwise they
454 don't go near her/it's not jsut her there are other children as vulnerable as she is, and sthey
455 send reminder letters you know that you shouldn't any of the children to school when there's
456 diarrhea and vomiting, that kidn of thing, because it's not fair on the other kids, so I take her
457 to school everyday, I take her or my husband takes her and we do a check round and if
458 anybody's ill just she just comes home. And you know it may sound extreme but when she
459 first started school, there was a child who had a cold, and I thought no I'm being ridiculous,
460 and I left her and she oh, she was ill for probably about 6 weeks
461 RF: so having kind of said oh don't be ridiculous and gone for it
462 S: and I shouldn't have done
463 RF: did that give you any kind of feelings of guilt [about]
464 S: [yeah] I felt pretty stupid, I should have just stuck with my own gut reaction because then
465 of course there was nothing, no school, nothing she couldn't go anywhere for ages, it gets
466 very labour intensive and I've still gotta work and stuff, it's err so yes err, now I sitck to my
467 gut reaction and if anybody phones I went and heard what the head teacher said and she said
468 you know this child shouldn't ahve been allowed into school err and that's it, so they're a lot
469 more careful now aswell
470 RF: ok, erm... I'm just wondering whether... actually having that diagnosis of monosomy
471 1p36 deletion syndrome or chromosome 1p36 whichever kind of phrase you prefer to use,
472 whether it actually makes a difference having a title such as that
473 S: it did at first...It does in practical terms because obviously it meant that my husband and I
474 had to be tested, and find out whether it was hereditary, that was the biggest, the biggest
475 thing, and also having the checklist of problems so we could be forewarned to check things
476 out/Now it doesn't make any difference at all, all the papers have been ripped up and thrown

477 away a long time ago [laughter] I don't even go on the discussion forums now, it makes no
478 difference, she's Andrea and that's it

479 RF: Was there a point, where you kind of made that decision "I'm not gonna go these
480 discussion forums anymore" [Is there] a particular reason for it

481 S: [yes] err far too depressing...I wouldn't mind being able to contact other people in this
482 country and just out of pure interest, you know it's nice to talk to people that are having the
483 same issues as you, whether it be 1p36 or anywhere else, but particularly, and I'd be intrigued
484 to see a couple of other children with it, just to look at them to see what they look like and I
485 think that would be interesting, but most of the blog sites we looked at, they're just American
486 and they're so flippin dramatic and they are all, it's just too much

487 RF: err yeah, my experience so far of the Yahoo group has been [quite spiritual]

488 S: [dominated by Americans] Oh yeah! Too right.

489 RF: [please pray for my child...] Is that, has that been something else that you have used or

490 S: [yes] I did, we did go on the Yahoo group quite a while but really you want some factual
491 information and sometimes you just want to email someone to say "how are you getting on
492 today" I think, and I found that all too much, they were, everything was to do with insurance
493 position in relation to the States – they do everything with the child, take their adenoids out,
494 take their tonsils out, they do all sorts of stuff with them and you know they are a bit more
495 reactive, no err its a bit, no I didn't like it so I opted out of that one quite quickly

496 RF: So more kind of medical orientated, less of the support, would that be

497 S: apart from the yeah, apart from the everyday, the err God pray for me type I business
498 which err I err, again it's not really much use to me [laughter]

499 RF: Is that not, kind of one of your views that you kind of erm, get your support from that
500 side of things, from [something quite spiritual]

501 S: [no] I think we do it in our own way, but err we haven't, when she was in Alder Hey the
502 Chaplain used to come up every single day they were great, they were really supportive, they
503 were just you know really, they spent a lot of time there, and you she does, we do take her to
504 church and stuff and but to me thats kind of a personal things erm and I found them they
505 were, it was just too over the top, completely over the top yeah, very intense, and unfortunately
506 a lot of the web pages that there are about either that or the facebook one do seem to be
507 dominated by...the Americans, not, err, one of my best friends is American [laughter] but that
508 doesn't count [laughter]

509 RF:I guess we were talking more in respect of support groups

510 S: yes

511 RF: as maybe the Americans would use the, it's a different culture

512 S: they seem to like to just make a drama you know, so they'll do a blog and it will just be
513 really, dramatic I mean, kind a of a huge great thing and you think A) they must have lots of
514 time on their hands, and B) it's like they're so obsessed by what their child's got that they
515 can't even see that it's just you know it's just a child

516 RF: One of the interesting things I thought last week was a parent who ... didn't see her child
517 as having 1p36...she saw her (him sorry) as having a collection of difficulties that could all be
518 addressed in different ways

519 S: Yes, true

520 RF: I was wondering if that might be a similar way that you actually see it now

521 S: yes I don't see it, I don't see Andrea as having 1p36, she's just Andrea and she's the way
522 that she is erm you know and she can be naughty and she can be good and she can be
523 friendly and she can be horrible and just like any, she has some traits of a normal three year
524 old oh yes, the fact that she can't stand up or whatever, it doesn't matter, she's erm, she still

525 does what, the hell she wants to do and she has everybody running round after her, she has a
526 great lifestyle, she really does, a fantastic lifestyle you know she's got people looking after
527 her and caring for her all of the time, they're, whatever she wants she gets. And, you know,
528 she's got loads of fancy kit, she's got bikes you know, we go everywhere with her, she's been
529 round the world and back again, been away nine times this year, away last year, we just go.
530 RF: You mentioned earlier about going off to Australia, that it was actually probably a good
531 thing for her
532 S: Yeah, well it was A)because of my thinking that you have to get on and get used to doing
533 stuff and b) the weather was probably, in hindsight it probably actually helped her through her
534 first winter, having a good few weeks away. I mean, we only got told the day before we were
535 due to go that the consultant only gave the authority to go and we had to cut it in half as well,
536 and it was only because where we were staying we were in Perth, just outside of Perth in
537 Freemantle and he actually knew someone who was a consultant Paedialogist, Paediatrician in
538 Freemantle Hosptial who had been told that we were coming so it was all kind of
539 RF: And whilst you were out there did you need to call on that support or
540 S: no
541 RF: or was it Yes lets just carry on an go with it
542 S: yeah, I mean, she doesn't fit as much, which is fantastic err we've probably got enough to
543 deal with but no we've not, we've been to Montpellier Hospital once when we were in
544 France, and but apart from that we've not been, we haven't, in the kitchen cupboard there in
545 France there's every range of antibiotics known to man, ready and sitting waiting there so if I
546 think there's something going wrong they'll take a sample from her here and then they'll test
547 and then they'll ring up and say its this that, because she has...three (inaudible) MRSA, she's
548 had various, she's got various strains of things floating around so we have the whole night
549 (inaudible)
550 RF: which is quite understanding, the reasons for keeping her out of hospital as much as
551 possible really
552 S: Yes, you know, because thats probably normal
553 RF: thinking of MRSA, C-diff all of those types of things
554 S: Yes we've avoided C-Diff so far, although we couldn't, we're fairly convinced that she has
555 had it, errm but she has that, and Hepatitis aswell, because she gets, she starts, if she starts
556 getting a coldsore it's a complete breakout, all her face, her throat, her bottom, her hands,
557 because she bites her hands so her hands, it goes/ the infection goes down her hands
558 RF: Right
559 S: and then she bites it and it goes back to her mouth so we have as usual things like that,we
560 have the whole pharmacy, again my brother is with us a fair bit
561 RF: Yeah, so your brother being a doctor, is he a doctor in a hospital or GP,
562 S:he's in SellyOak,
563 RF: so stayed local to Birmingham then [laughter]
564 S: he's a major in the army and err he's been (inaudible) and done all that kind of stuff and
565 now he's back at, he's an anaesthetist at Selly Oak. (Inaudible) Helpful in a sense that he's my
566 brother and he can come up and he... is very calm, and I think that's the most helpful part of
567 because he is very very calm. I don't really care about his knowledge because in relation to
568 me, you become an expert in your own child I mean I could tell you all of everything from her
569 normal heart rate bleeping resting and all the stats, the oxygen levels, her CRT levels,
570 everything I could tell you about her you you know more so than probably any medic because
571 you have to ... do that every day you have to know, and it's helpful to know but he's just you

572 know when it's all going horribly wrong he's calm... you know we can sit and just chat about
573 inane ... rubbish

574 RF: as brothers can do

575 S: yeah [laughter] that's it, he's perfectly capable of doing that and you know, he will, I
576 always avoid asking him what he thinks because I think that's unfair, and erm, now if I could
577 tell you, when she was in ITU he'd come in and he'd take in everything, he'd take in all the
578 monitors and just casually look at things and he'd ask people very hard questions, yeah just
579 watch and the staff knew because he was a medic they'd you know just go and look at the
580 charts it's pretty of course you, you know, you could tell by the reaction on his face whether it
581 was good or whether you should be concerned and also you know he came up obviously from
582 Birmingham and then he went back again and as soon as he went back I knew that he thought
583 it was ok, and then she went downhill and then he came back again and then again, a week
584 later, he went back and forth.

585 RF: So you could possibly use your brother as a bit of a barometer for how things are going

586 S: Yes, definitely, yeah, he's quite handy

587 RF: But at the same time he's there as a brother to bring in a bit of normality is that

588 S: yes, yes, tell stupid jokes, yeah

589 RF: take you off track for a bit

590 S: yeah I (inaudible) family members returned home interview suspended for 30seconds

591 RF: so just kind of thinking to now Holly's at special school, as the epilepsy nurse predicted
592 would that be right

593 S: Andrea

594 RF: sorry Andrea do you find you compare your experiences with parents of the other
595 children there

596 S: I don't really see them

597 RF: Right

598 S: yeah, I don't really see them because most children, most parents send their children to
599 school on the bus so they don't really, I mean we have groups in relation to when they're
600 (?trying to close the school down) and there are some parents who I already know through
601 other (you know, through physio groups and things like that erm who are good friends, but
602 they they've been brilliant they really have been a brilliant support the other people you get to
603 meet it crosses every social boundary, and age group the whole you know, indiscriminate...

604 RF: Is there any support that you don't get that maybe you think you should.. or would like

605 S: Yeah, counselling specifically counselling, I found that very hard, cos I you can imagine
606 maybe particularly this summer when I was all falling apart that erm, my GP referred me to a
607 counsellor but she was a fairly general in the sense of you know counsel anybody about
608 anything, I needed something a bit more specific, who maybe understands more about having
609 a child with problems, so I think that is something that really you should be offered and I've
610 struggled through and it doesn't seem to be even in Alder Hey they only counselling they
611 had was a bereavement counsellor and I know quite a few parents when we were in ITU she'd
612 get to speak to everybody, when actual bereavement counselling even though the children
613 were alive, erm because that was the only form they had available

614 RF: I'm quite surprised at that

615 S: erm, you can struggle to get that and I think that should be offered, it's very obvious and
616 very apparent even like we were talking about it with some other parents about setting
617 something up whereby you know in the hospital you could have a leaflet a point of contact
618 even with any one of us, are you having a trauma, look you know, the parents of a group you
619 know stupid mine full of information sometimes like where do i leave me car you know if

620 I've got to stay there for a week you know how do you work the hospital, it's just real, just
621 stuff like that yeah, that you as a parent, perhaps you need to know
622 RF: and maybe it would be useful as kind of a parent who's been through the whole diagnosis
623 to maybe act as a mentor for parents coming through it
624 S: yeah, now obviously every child's different and even children with err the same deletion
625 which is one of the reason I stopped looking at the groups and stuff because (inaudible)
626 RF: there are some many different variations of
627 S: pictures of them walking and running round and stuff and there are children who are worse
628 than Andrea so you know, she crawls, they said she's at the severe end but
629 RF: but then there's gradients of severity there aswell
630 S: yeah yeah
631 RF: Is there anything that you're surprised that I've not asked you about in terms of receiving
632 a diagnosis
633 S:no not really
634 RF: Or anything else that you think would be important for us to know
635 S: Just that it feels like the last three years has taken us thirty years, but I'm really glad I
636 didn't know when I was pregnant I'm really really glad, because now I can see her and I know
637 her and she's great, she's great fun, and you know we have a great time. If somebody had
638 said to me when I was pregnant, by the way, when your child's three she's gonna be about the
639 age of 6months old she's not gonna be able to speak, she's not gonna walk, she's not gonna
640 do this, she's not gonna do that it would have been terrifying, and I think that... people get
641 told they've got children with Downs and stuff and you know I think well ok, because often
642 people with Down's they have a fantastic lifestyle they're great, they are really sociable and
643 great but I think that to be told that, if I got to stage where I could do one of two things/with
644 so common, allegedly if it got to the stage where it was routinely tested for, I think that would
645 be very scary for people definitely. I think unfortunately you would get to the situation where
646 people may think about terminating and really the child could have a fantastic and really
647 rewarding life and that would frighten me a bit, because it does sound so damning, so
648 negative and
649 RF: yeah, quite an internal battle to deal with I guess
650 S: Oh I wouldn't like to be in a position where I would, well I know what I would decide
651 because you know, that's different but I think for people who may be on their own or
652 whatever to know, but I think with advanced knowledge of any genetic disease can be very
653 scary so yeah, thats all really.
654 RF: Thank you very much
655 S: No problem

Appendices

APPENDIX 9
EXAMPLES OF CODING AND ANALYSIS

Example of Open Coding

Interview 8	Open Codes
<p>It was, because at least we knew, at least we knew what it was, and ... it was only when I looked into it a bit deeper erm with the doctor and I said “why, why was this information withheld, did it matter to Dr Hahmeed whether it was genetics or did it matter, why was it so important to withhold information that he knew from the parents who have a right to know” and she said oh Dr, what Dr Hahmeed had wrote in the letter was that he was worried about upsetting me, by saying it was a genetics in case I blamed myself. This was all written in a letter and I was so disgusted and shocked by it, that anybody could judge somebody like that and say “oh well she looks a bit upset, better not tell her it’s genetic in case she has a wobbler or something”. That’s not fair, that’s my, that’s information about my child I have every right to know it regardless of my state of mind, regardless of whether it was on my side of the family or ... how I felt about it, I would just have to come to terms with it wouldn’t I, so I should have been told and he felt that he had the right to do that, and oh my god (sighs)</p>	<p>Knowing something</p> <p>Searching for information Being kept in the dark</p> <p>Professionals having power Keeping things from mother Its a parent’s right to know</p> <p>Protecting</p> <p>Reacting to being kept in the dark Feeling judged</p> <p>Feeling angry</p> <p>Its a parent’s right to know</p> <p>blame Dealing with things</p>

Examples of Open Codes with similar meaning

Being kept in the dark (Int 8)
 Keeping things from mother (Int 8)
 Reacting to being kept in the dark Int (8)
 It’s a parent’s right to know (Int 8)
 Professionals knowing but not sharing (Int 5 and 6)

Focussed Code

Being kept in the dark

Groups of Focused Codes with Common Theme

Being Kept in the dark
 Professionals being negative
 Experience never leaving you

Theme

Being Powerless

Appendices

APPENDIX 10
REFLEXIVITY EXAMPLE

Examples of reflexivity

In one of the early interviews, a mother said that “1p36” didn’t matter, as every difficulty that her son had, could be dealt with by the relevant professional. This was quite a humbling thought, as it brought to mind the exact difficulties that parents are facing on a daily basis. Despite all of these difficulties, parents have taken a practical approach and manage things on a day by day basis. This prompted me to ask in future interviews about how parents deal with the individual difficulties, for example whether all parents take a practical approach to parenting.

Another parent spoke of being offered an amniocentesis but turning this down, because of their beliefs. She told of not wanting to terminate regardless of what the outcome was thus turning it down. This prompted me to discuss with the other researchers on the journey back from this participant’s home, what we would do in this situation. Given the fact that 1p36 is a very common genetic condition, , and the three of us are all in relationships with the intention of having children, it caused us to ask ourselves whether we could parent a child with a disability. These thoughts remained throughout the interviews, and on meeting some of the children, it was quite upsetting to see the degree of their disabilities.