VOLUME ONE

QUALITY OF LIFE OF PEOPLE WITH RARE GENETIC DISORDERS

By

SAMANTHA ELIZABETH CLARKE

A thesis submitted to the University of Birmingham for the degree of

DOCTOR OF CLINICAL PSYCHOLOGY

Clinical Psychology

School of Psychology

College of Life and Environmental Sciences

University of Birmingham

May 2015

Thesis overview

This thesis consists of two volumes. Volume One contains the research components (systematic review and empirical paper) and Volume Two contains the clinical practice reports (models essay, case study, single case experiment, service evaluation and case presentation). Both were completed as part of the Birmingham University Doctorate in Clinical Psychology.

Within Volume One, quality of life (Qol) in those with an intellectual disability is considered. The importance of having pragmatic measures of Qol that services can use to evaluate individuals' outcomes is highlighted. In light of this, a systematic review of the literature is conducted in order to consider the use of eleven measures of Qol within those with intellectual disabilities. These measures were selected as they had been identified by previous reviews as reporting their psychometric properties. The resulting papers are discussed in terms of their utilisation of Qol measures and which factors they explored.

Following on from this, the narrower concept of health related quality of life (HrQol) is considered within the empirical paper. The health and HrQol of individuals with Angelmans, Cornelia de Lange and Cri du Chat is described. In addition to this, an individual's level of challenging behaviour and its impact on health problems and its relationship with HrQol is considered.

Within Volume Two, five clinical practice reports (CPR) are presented. The first CPR presents the case of David*, a nine year old boy, who was referred to the Child and Adolescent Mental Health Service suffering from an increase in Obsessive Compulsive Disorder (OCD) symptoms. Details are provided on the method and results of assessment and then David's case is formulated from both a Cognitive Behavioural and

Psychodynamic approach. The second CPR describes a children's diabetes service and evaluates the wellbeing assessment provided by staff. The completion rate of the wellbeing assessment form was low. Recommendations for service development are made. The third CPR shares the case of Aamira*, a 28 year old Muslim lady, who was referred to the Home Treatment Team suffering from OCD; which incorporated intrusive thoughts about her religion and her relationship with her husband. Her assessment and Cognitive Behavioural formulation are then discussed in depth. This concludes with a discussion on a possible approach to treatment. The forth CPR details the case of Hannah* and her family. Hannah, a 19 year old woman with a longstanding history of OCD combined with High Functioning Autism and Diabetes, was referred to the local Learning Disability team. After presenting the assessment and Systemic formulation, the effectiveness of the communication skills based intervention was measured using an AB experimental design. The final CPR provides an abstract of a case presentation given, detailing the treatment utilising Cognitive Analytical Therapy of Mona*, a 17 year old woman referred to the Youth Support team due to depression.

** All names have been changed to maintain confidentiality*

Dedication

To my family and friends past and present who have each in their own way supported me on my journey to becoming a Clinical Psychologist.

Acknowledgements

I'd like to thank my supervisors Chris Oliver and Dawn Adams for their continued support and guidance throughout this thesis. I'd like to thank my husband for the many hours spent proofreading and support he's given me over these last three years.

I also wish to share my gratitude to all the participants and their families who gave up their time to complete the questionnaires. Without their dedication to research this thesis would not have been possible.

Finally to my fellow trainees I couldn't have asked for a more supportive and friendly cohort. I'm so glad that we've been able to share this journey together.

Thesis Contents

Volume 1

Systematic Review

Empirical Paper

Volume 2

Case Study - CPR3

Single case experimental design - CPR4

Case Presentation – CPR5

Appendices

List of Tables and Figures

Tables

Volume 1

Systematic Review

Empirical Paper

Figures

Volume I

Volume 2

Psychological models - CPR1

Service evaluation - CPR2

Case Study - CPR3

Single case experimental design - CPR4

Systematic Review

Assessing Quality of Life in the Intellectual Disability Population: A Systematic Review of the use of Established Measures.

Abstract

Introduction: There has been a growing focus placed on the importance of ensuring a good quality of life (Qol) for individuals with intellectual disabilities (ID). This importance is seen both in research and health and social policies. In order that services are able to accurately measure and document changes in an individual's Qol, measures which are both robust and pragmatic are required. Combining previous reviews, eleven measures were identified which reported their psychometric properties.

Method: A systematic review of the literature was performed using PsycInfo and Medline databases to identify any paper that utilised one of the eleven measures of interest. Data was extracted in terms of how the measure was used and the factors explored. The quality of the resulting papers was assessed using Downs and Black's (1998) Quality checklist.

Results: 31 papers were identified. Variations were seen in how the papers utilised the measures of Qol. A range of topics were covered in the papers with environmental factors being explored more frequently than personal characteristics.

Discussion: Relatively few papers were identified, suggesting that there has been less of a focus on the utilisation of measures than on their development. There is a call for researchers to provide greater detail on the utilisation of Qol measures, including the use of proxies and individuals in order to allow better comparison between papers. In addition, consideration should be given to the multitude of factors that could be influencing an individual's Qol and in light of this, consider a wider range of confounding factors.

2 Introduction

2.1 Quality of Life (Qol)

Quality of life (Qol) is a social construct typically used to describe a person's overall standard of and satisfaction with life. Whilst there is considerable debate about both its meaning and conceptualisation, it remains an important construct in health and social care. Despite the ongoing theoretical debates, Brown, Hatton and Emerson (2013) adopt the pragmatic position that the use of Qol within the provision of services for those with intellectual disability (ID) has developed from being an implicit organisational goal to an explicit service goal or standard and as such should be operationalised and measured.

Qol is considered to be multidimensional with multiple factors often referred to as Qol domains. It is often hypothesised that these key components of Qol are the same for all people regardless of ability or culture (Bramston, Chipuer, Pretty, 2005; Schalock et al., 2005). There are numerous descriptions of what comprises these domains. For example, Schalock (2004) reviewed 16 published studies and concluded that the most common domains, and therefore what might be considered to be the core Qol domains, are: interpersonal relations, social inclusion, personal development, physical well-being, self-determination, material wellbeing, emotional wellbeing and rights. Although broadly similar, other authors suggest differing domains. Cummins (1997a) suggests seven life domains; material, health, productivity, intimacy, safety, community and emotion. Finally, Felce and Perry (1995) suggest the life domains of physical, material, social and emotional wellbeing in addition to development and activity. It is widely accepted however, and pragmatic, that the importance lies not in seeking to measure every possible domain but in measuring enough to infer an overall level of Qol.

Each domain has a number of different indicators by which it can be assessed. Three of the most common indicators for each of the eight core domains were identified by Schalock and Verdugo (2002) (Table 1.1).

Table 1.1: The indictors and descriptors of the core quality of life domains identified by Schalock and Verdugo (2002)

Core Domain	Indicators (descriptors)
Emotional well-being	Contentment (satisfaction, moods, enjoyment) Self-concept (identify, self-worth, self-esteem) Lack of stress (predictability, control)
Interpersonal relations	Interactions (social networks, social contacts) Relationships (family, friends, peers) Supports (emotional, physical, financial, feedback)
Material well-being	Financial status (income, benefits) Employment (work status, work, environment) Housing (type of residence, ownership)
Personal development	Education (achievements, status) Personal competence (cognitive, social, practical) Performance (success, achievement, productivity)
Physical well-being	Health (functioning, symptoms, fitness, nutrition) Activities of daily living (self-care skills, mobility) Leisure (recreation, hobbies)
Self-determination	Autonomy/personal control(independence) Goals and personal values (desires, expectations) Choices (opportunities, options, preferences)
Social inclusion	Community integration and participation Community roles (contributor, volunteer) Social supports (support network, services)
Rights	Human (respect, dignity, equality) Legal (citizenship, access, due process)

In summary, although there is ongoing debate, it is generally accepted that Qol is a measure of a person's overall 'standard' of life, is multidimensional, and comprises of core

domains with key indicators. The core components of Qol are conceptualised as the same for all people regardless of ability or culture (Keith, Heal & Schalock, 1996).

2.2 The Measurement of Qol

Qol can be measured either subjectively or objectively. Each approach is felt to be equally important, for example the amount of time a person spends in the company of others can be objectively measured and Qol inferred. However, each person will value the company of others differently and therefore a subjective measure is required. Brown, Hatton and Emerson (2013) provide a summary of the current thinking on this aspect of Qol, suggesting that there are limitations to both subjective and objective measurements and that it is currently recommended that both are used together.

2.3 Qol in the Intellectual Disability Population

Qol has become popular in part due to the emergence of the human rights movement within the field of intellectual disabilities (ID) that relates to concepts such as empowerment, inclusion and self-advocacy (Buntix & Schalock, 2010). This changed a society's focus on ID from a 'doing to' to a 'doing with' position. Qol provided a way of considering the objective 'standard' of living and combining it with the person's own values and wellbeing. In this way it is argued that Qol can provide a catalyst for service change and development (Schalock 2004). Examples of this within ID services are given by Keith and Bonham (2005) who suggest that using Qol measures for outcome evaluation allows services to be responsive to individuals' needs and to develop services that keeps the individual at its heart.

In order to measure Qol of individuals with ID, researchers are faced with a number of challenges. Cognitive ability may limit the questions that people are able to understand and

respondents may not be able to answer questions that require abstract thinking, acquiescence may be evident, and multiple options may be demanding of working memory. Each of these compromise reliability and validity. One possible solution is the use of proxy respondents. However, when proxy reports are compared to the reports of those individuals who are able to respond there is often a difference (Cummins, 2002). This can also be seen in the Non-ID population where research into children's Qol has focused on long term health problems. Eiser and Morse (2001) completed a systematic review of health related quality of life in the Non-ID population and concluded that differences between parental and child reports of Qol are common and so recommend that studies measure both.

The alternative to using proxy respondents is to find valid and reliable ways of reporting the subjective experience of individuals, including those with profound disability. Petry and Maes (2008) recognise the difficulty in measuring Qol within ID in general and particularly for those with profound and multiple disabilities but argue that these difficulties should not dissuade researchers from continuing to explore subjective Qol within this population. Petry and Maes (2008) highlight the importance of supporting people to express their views, however, they also recognise that there is likely to remain a group of people who are unable to communicate in this way and so in these situations proxies remain the only option.

In summary, it is particularly important to consider Qol within the ID population as it places the individual at the heart of service provision and development. It also engenders an ethos of maintaining or improving a person's Qol. However, finding ways to measure Qol has proved challenging, particularly for individuals with limited cognitive and communication abilities. In light of the question of the validity of proxy respondents, it

remains important to find ways to include the views of the person with ID. Despite the challenges of measurement pragmatically, a measurement tool is needed. Measures should be relatively simple to conduct and once conducted they should provide results that are valid and reliable and allow for repeated measures to identify change.

2.4 Measures of Qol for Those with ID

Two systematic reviews (Li, Tsoi, Zhang, Chen and Wang, 2013; Townsend-White, Pham and Vassos, 2012*)* have assessed the psychometric properties of Qol measures for those with ID. Townsend-White et al. identified measures which could align with at least one of the eight core domains identified by Schalock, took less than two hours to complete, were in English and reported psychometric properties. The identified measures were then evaluated in terms of reliability, validity, potential for acquiescence and ease of administration.

Li et al. (2013) conducted a similar search of the literature and identified measures that assessed at least one domain which could align with one of Schalock's core domains, included the self-reports of individuals with ID and reported psychometric properties. Li et al. identified nine measures of Qol and evaluated them in terms of reliability, validity, responsiveness, item bias and consideration of floor or ceiling effects.

2.4.1 Measures identified

The search strategies employed by both Townsend-White et al. (2012) and Li et al. (2013) show similarities and identify those Qol measures that report psychometric properties. The results of both reviews are combined and summarised in Table 1.2.

Table 1.2: A description of the measures of quality of life for individuals with intellectual disabilities identified by either Li et al. (2013) or Townsend-White, Pham and Vassos (2012).

Measure		Domains measured	Questions	Scale	Additional information
Choice Questionnaire *Stancliffe & Parmenter (1999)*	Subjective	Domestic activities; Co-residents & staff; Money & spending; Health; Social activities, community access & personal relationships; Work & day activities; Overall choice	26	3 point scale	Measures a person's ability and opportunity to make choices in their daily life. Overall choice reflects the answer of one question not the sum of the other questions
Comprehensive Quality of Life Scale *Cummins (1997b)*	Subjective & Objective	Material wellbeing; Health; Productivity; Intimacy; Safety; Community; Emotional	35	2, 3 or 5 point scale	Pre-test provided
Evaluation of Quality of Life Instrument *Nota et al. (2006)*	Objective	Quality of service received; Satisfaction with social interaction: Satisfaction with Living environment	18	5 point scale	
Health Status Interview Schedule *Ruddick & Oliver (2005)*	Subjective	Physical function; Bodily pain; General health; Vitality; Social functioning; Mental health; Reported health transition; Sensory functioning; Memory functioning	26	Rating scales, forced choice and open questions	
Quality Of Life Interview Schedule *Ouellette-Kuntz (1990)*	Subjective & Objective	Health services; family/guardianship; income maintenance; education/employment; housing& safety; transportation; social/recreational; religious/cultural; case management; advocacy; counselling; aesthetics	NA	7 point scale	Proxies only

Quality of Life Questionnaire *Schalock & Keith (1993)*	Subjective & Objective	Empowerment & independence; competency & productivity; overall satisfaction; Social belonging & community integration	40	3 point scale	Those in paid employment can score up to 30 on competency/productivity compared to the max score of 14 for those not working
Life Satisfaction Scale *Heal & Chadsey-Rush (1985)*	Subjective	Community satisfaction; Friends & free-time satisfaction; Satisfaction with services; General satisfaction; Job satisfaction.	29	yes/no	
Maryland Ask Me Project *Bonham et al. (2004)*	Subjective	Social inclusion; Self-determination; Personal development; Rights; Interpersonal relations; Emotional wellbeing; Physical wellbeing; Material wellbeing	56	3 point graphical scale	
Multifaceted Life Satisfaction Scale *Harner & Heal (1993)*	Subjective	Community; Friends & free-time; Job; Recreation & leisure; client control & self-determination; General satisfaction	58	5 point scale	Measures satisfaction and is a development of the LSS
Personal Wellbeing Index *van Loon et al. (2009)*	Subjective	Standard of living; Personal health; Achievement in life; Personal relationship; Personal safety; Community-connectedness; Future security; Religion/spirituality.	8	2, 3, 5 or 11 point graphical scale	A development of ComQol includes a pre-test
Personal Outcome Scale *Cummins & Lau (2005)*	Subjective & Objective	Social inclusion; Self- determination; Personal development; Rights; Interpersonal relations; Emotional wellbeing; Physical wellbeing; and Material wellbeing	48	3 point scale	

2.4.2 Summary of existing Qol measures for individuals with ID

In summary, together Townsend-White et al. and Li et al. identify 11 measures of Qol that report psychometric properties. Each has a differing approach to what is being measured and how. Some focus on subjective measures only, whereas others comprise both subjective and objective measures. Both authors recognise that further validation of these measures is still required. Although there is no clear 'gold standard' measurement of Qol in individuals with ID, by conducting and publishing tests on their psychometric properties these eleven measures may be considered more useful than those that have not.

2.5 Interim Summary

The importance of Qol is clear both as intrinsically valuable for services and as a reliable outcome measure by which to assess goals and service standards. As an outcome measure, Qol allows a service to recognise and respond to changes within an individual's Qol and in documenting this allows services to openly demonstrate that they value the Qol of the individuals they care for. This becomes increasingly important for those with ID for whom their capacity to act as service consumers and state their needs effectively is compromised. Measuring Qol within this population presents a number of challenges and requires measurements that are both valid and reliable. Recent systematic reviews have identified eleven measures that report psychometric properties.

Services are under increasing pressure to prioritise Qol and require Qol measures that are easy to use, can be used across individuals, provide reliable and valid data and can be used repeatedly. Having identified measures for which these properties are known, it is now possible to explore how the identified measures have been used within published research by conducting a systematic review of the literature to identify all papers that have utilised

these measures. This will provide further information on the use of Qol measures and how researchers have addressed the challenges noted above (e.g. the use of proxy reporting).

2.6 Review Questions

There are two main questions:

1. How have the eleven identified measures been utilised in published research on Qol in the ID population?
    a. What research design was used?
    b. Who was the respondent?
2. What factors have been investigated using the eleven measures and what conclusions were drawn?

3 Method

3.1 Search Strategy

A literature search was carried out on November 4, 2014 combining terms for intellectual disability and the measures identified above. The key words used to define intellectual disability are shown in Table 1.3 and were identified by examining current reviews of literature in this area. Where possible these terms where mapped to subject heading.

Table 1.3: Search terms entered to encompass journal articles regarding intellectual disabilities

Search topic	Terms entered	Alternatives covered
Intellectual Disability	Intellect* disab*	Intellectual, Intellectually; disability, disabilities, disabled
Learning Disability	Learning disab*	disability, disabilities, disabled
	LD	
Intellectual Developmental Disorder	Intellectual Developmen* Dis*	development, developmental; disorder, disability, disabilities, disabled
	IDD	
Mental Retardation	Menta* Retard*	mental, mentally; retard, retardation, retarded
Mental Handicap	Mental* Handica*	mental, mentally; handicap, handicapped
Mental Deficiency	Mental* Deficien*	mental, mentally; deficient ,deficiency

3.2 Databases Searched

Two databases were searched; PsycInfo and MEDLINE. PsycInfo covers the professional and academic literature in psychology and related disciplines, including medicine, psychiatry, nursing, sociology, pharmacology, physiology and linguistics. The MEDLINE database is widely recognized as the premier source for bibliographic and abstract coverage of biomedical literature. MEDLINE encompasses journals which cover both medical and allied health topic areas. Papers dating from 1985 to November 2014 were searched as the oldest measure of interest dates back to 1985.

3.3 Review Process

The initial search identified over 100 potential papers. An abstract search followed by a full text search was conducted.

Studies were included if they met all of the following criteria:

- Publication in a peer reviewed journal.
- Contributed original data.
- Utilised one of the selected measures with the implementation of the Qol measure reported and the psychometric properties of the measure retained by either using the full measure or a defined subscale.
- The measure should be used to assess Qol in individuals with an ID.
- It considered the impact or relationship of a psychological, environmental or social factor on/with Qol.

On the basis of these criteria, papers that focused on the validation of measures or utilised a Qol measure purely to describe a sample without analysis of associated characteristics were excluded.

Although systematic reviews were excluded as they did not contribute original data, their references were searched to identify further papers. This resulted in the consideration of four extra articles, two of which met the inclusion criteria. If during the review process it was unclear whether a paper should be included then an opinion was sought from experienced researchers working within the field of ID. Due to time and resource constraints, only journals written in the English language were reviewed. Four papers were excluded due to language. The details of these papers can be found in Appendix A.

As noted above, these measures were selected due to their psychometric properties. In order to maintain this, if a measure was named in a paper but a differing year or combination of authors was reported, then this new reference was investigated and the paper only retained if the reference referred to a version of the measure that was a match to the number of questions, rating scale and domains covered as originally reported by Li et al. (2013) and Townsend-White et al. (2012). If it still remained unclear what version of the measure had used then the original author was contacted (Appendix B) for further clarification.

Figure 1 shows the numbers of papers excluded at each stage.

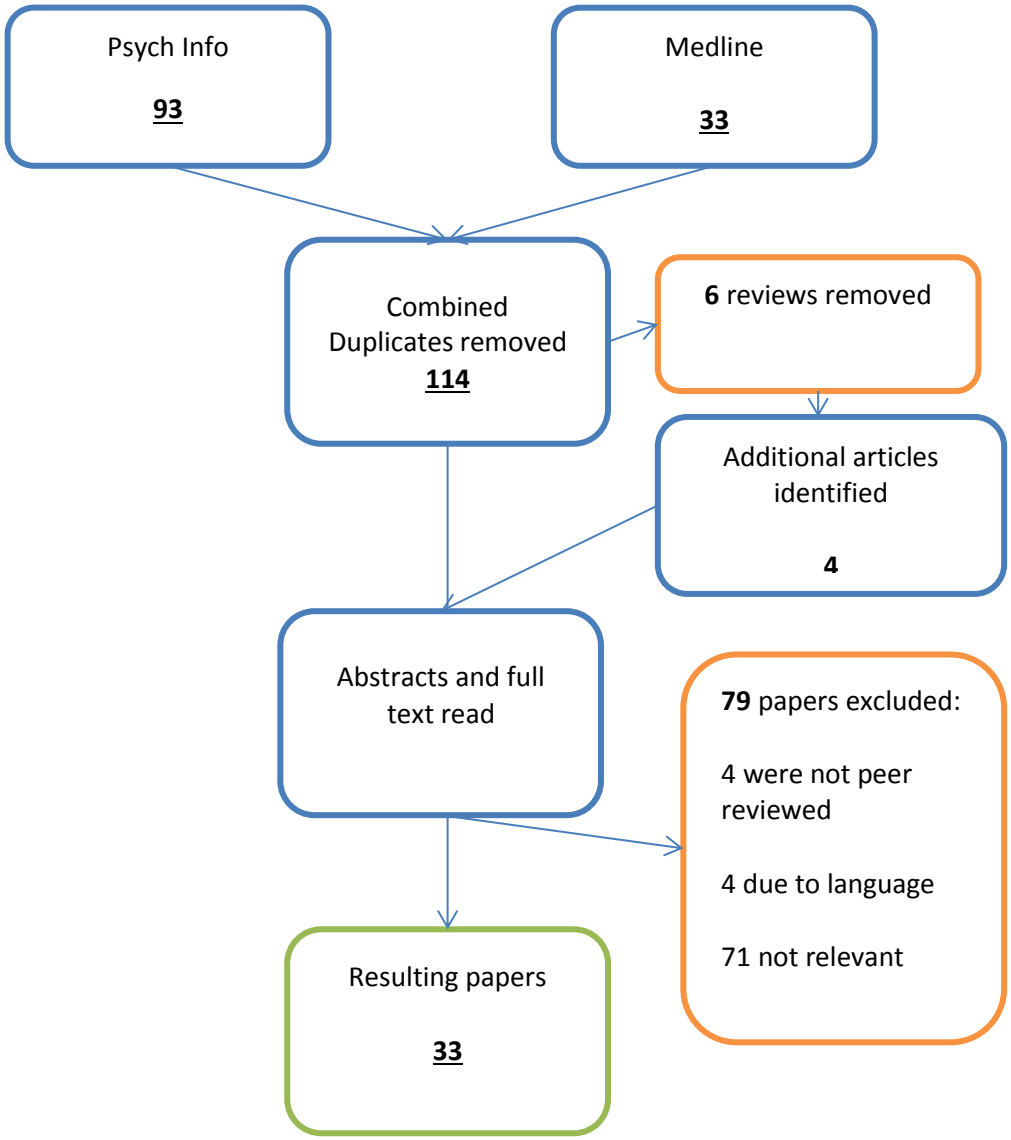


Figure 1: Flow chart showing the identification of included studies.

The initial 114 papers were reduced to a final 33 papers. The reduction included the removal of 71 papers as not relevant. Table 1.4 summarises the reasons for this.

Table 1.4: Summary of the number of paper excluded as deemed not relevant and reasoning for this decision.

Reason why not relevant	Number of papers excluded
Did not relate to QoL or contribute unique data	18
Did not use one of the measures identified	16
Focused solely on psychometric validation	16
Did not involve participants with ID or their proxies	9
Only measured Qol in others (e.g. family, carer and staff)	8
Focused on pharmacological intervention	2
Did not provide enough detail on the method for inclusion	2

Two papers (Appendix C) were excluded due to lack of detail in the method sufficient to apply the inclusion criteria.

The Centre for Reviews and Dissemination (CRD, 2008) highlight the importance of reviews identifying multiple reporting of the same research. From the 33 papers identified, one study had been republished verbatim, four years following original publication. As such the reproduction was removed. Two papers reported different aspects of the same study. In this case only one reported the full Qol result and so this was retained and the other study was removed. This process left 31 papers.

3.4 Quality Assessment

Downs and Black's (1998) checklist was used as this was designed to provide a framework to assess both randomised and non-randomised studies. It has been used in numerous

reviews and is recommended as a suitable measure by Deeks et al. (2003) and the CRD (2008). It has been found to have good inter-rater reliability (r=0.75) (Downs & Black, 1998). In the current review there is particular interest in the Qol result which may not have been the main focus of a paper, therefore a number of adaptations to the framework were made. First, as well as considering the quality of a paper's reporting of main outcome and variability, these two questions were repeated, asking specifically about the reporting of the Qol element of the paper. Questions regarding the adverse impacts of treatment (Question 8), representativeness of treatment (Question 13), data dredging (Question 16) and treatment compliance (Question 19) were inappropriate and removed. Finally, considering the possible challenges of seeking consent within this population, a further question asking if the paper clearly reported how consent was gained was included. A full copy of this checklist and scoring notes can be found in Appendix D

The CRD (2008) does not recommend the use of quality summary scores as a way of comparing the quality of papers and suggest that a checklist is presented instead. A checklist was also preferred as due to the wide variety of methods utilised an overall quality score, as calculated from the Downs and Black framework and would not provide a valid basis on which to compare papers. As such, the full checklist as opposed to a score was compared.

3.5 Data Extraction

Data regarding the research design; overall and Qol specific summary; participant characteristics; and which Qol measure was used and by whom, were extracted from the resulting papers. In addition it was recorded if the paper used additional Qol measures (exclusive of the measures of focus). Finally an overall Qol score was given to the paper ranging from 1-5 to reflect the importance.

Qol scores were awarded as follows:

1 - Uses one measure and reports results briefly.

2 - Uses two or more measures of Qol and reports the results briefly.

3 - Uses one measure of Qol reports results detailing subdomains.

4 - Uses two or more measures of Qol and reports results detailing subdomains.

5 - Considers the factors that contribute to Qol by conducting analysis such as regression or path analysis.

4 Results

The systematic review identified 31 papers of interest. Papers are referred to by first author and date of publication. In order to identify how the eleven Qol measures have been utilised within the ID population, the papers are described in terms of design, participants, quality and treatment of quality of life. Following this, the focus and conclusions of the papers will be discussed.

4.1 Design

The resulting studies evidence a range of designs, with the majority comparing groups (n=13, 41.9%). Nine (29%) explored changes in Qol using repeated measures, five (16.1%) papers combined within and between participant designs and four (12.9%) utilised a correlational design. The number of participants with ID considered within studies ranged from five (McClean et al., 2007) to 248 (Chou et al., 2008) (mean 70.70 (*SD*= 63.12)). Four studies utilised a Non-ID control group (Beyer et al., 2010; Bouras et al., 2004; Bramston et al., 2005; Hematian et al., 2009). Studies were conducted worldwide. The majority of studies focused on Qol within a single country, however four studies measured Qol in two or more countries making up the UK, and one study was conducted across four Western countries (Canada, France, USA and Belgium; Lachapelle et al., 2005).

4.2 Participants

Studies employed participants with a range of levels of ID (mild-moderate, n=3; mild-severe, n=4; mild-profound n=3; moderate-severe, n=2). Those with borderline levels of ID were included in two studies (Wheeler et al., 2013: Horovitz et al., 2014). Six studies only focused on only one level of ID (mild, moderate and severe; two studies each). A number of studies did not specify the level of ID of the participants (n=7) or described them using ability scales without stating a classification level (n=4). The ethnicity of participants was only reported in

three studies (Bouras et al., 2004; Martin et al., 2005; Perry et al., 2013), and within these studies Caucasian formed the majority group followed by Afro-Caribbean and Asian.

The reported mean age of participants ranged from 18.3 (Hematian et al., 2009) to 53.4 (Cooper & Picton, 2000) (overall mean age 35.2 (*SD*=10.4)).

4.3 The use of Qol Measures

The Quality of Life Questionnaire (Qol-Q) was the most frequently used measure (16 studies), followed by the Choice Questionnaire (n=9) and the Comprehensive Quality of Life Scale (ComQol; n=8). The Multifaceted Life Satisfaction Scale (MLSS) was used in three studies and the Life Satisfaction Scale (LSS) and Personal Wellbeing Index (PWI) were used in one study each. None of the other identified measures were reported in the resulting papers. The majority of studies reported that the Qol measure was completed by the individual themselves (n=18), and of these, four studies provided the individual with support by including a main carer or parent during the measure. Proxies were used in nine studies. A further eight studies reported that the Qol measure had been completed by a combination of individuals and staff/caregivers, with only two of these studies specifying the proportions of proxy and self-reports (Beadle-Brown et al. et al., 2009; Horovitz et al., 2014).

The suggested participant (individual, proxy or combination) varies across the differing measurement manuals. With limited information often being provided on the implementation of Qol measures in the resulting papers, it was difficult to assess whether a study was using the selected measure in accordance with the corresponding manual. For example, the ComQol manual (Cummins, 1997b) suggests a differing approach to the completion of the subjective and objective components. None of the papers provide this level of information. Furthermore, the Qol-Q states that if proxies are needed to complete the measure for an

individual then two proxies per person should be used and their score averaged. Only Hartnett et al. (2008) mention using two proxies.

In terms of the Qol score given to these papers in regards to their use and reporting of the Qol measure, over a third of the papers mentioned this only briefly. Eleven papers however reported both the total and subdomain score of the Qol measure used and eight papers considered the factors that contribute to Qol by conducting analysis such as regression or path analysis.

4.4 Quality of Papers

The quality of the 31 papers is presented in Table 1.5 and Table 1.6.

Table 1.5: Quality review of the resulting papers following a systematic review of the use of established quality of life (Qol) measures in the intellectual disability population (including checklist questions 1-10; Downs & Black, 1998).

	Reporting – clearly described										
	Hypothesis/ Aims	Main outcome	Recruitment characteristics	Intervention or grouping	Confounders	Main finding (Qol)	Random variability (Qol)	Individual drop out	Actual p values	Consent	Power considerations
Banks et al. 2010	✔	✔	✔	✔	✖	✔ (✔✖)	✔ (✔)	✔	✖	✖	✖
Barber et al. 1994	✔	✔	✔	✔	NA	✔ (✔)	✔ (✔)	✔	✖	✖	✖
Beadle-Brown et al. 2009	✔	✔	✔	✔	✖	✔ (✔)	✔ (✔)	NA	✖	✔	✖
Beyer et al. 2010	✔	✔	✔✖	✖	✔	✔ (✔)	✔ (✔)	NA	✖	✔	✖
Bouras et al. 2004	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	✔	✔	✔	✖
Bramston et al. 2005	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	NA	✖	✔	✖
Chou et al. 2008	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	NA	✖	✔	✖
Chou et al. 2011	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	✔✖	✖	✔	✖
Chou & Harman et al.2011	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	✔	✖	✔	✖
Cooper & Picton 2000	✔	✔	✔	✖	✔	✔ (✔)	✔ (✔)	✔✖	✖	✔	✖
Duvdevany & Arar 2004	✔	✔	✔✖	✖	✔	✔ (✔✖)	✔ (✔)	NA	✖	✔	✖
Felce et al. 2008	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	NA	✖	✔	✖
García-Villamisar et al. 2013	✔	✔	✔	NA	NA	✔ (✔)	✔ (✔)	NA	✖	✔	✖
Hartnett et al. 2008	✔	✔	✔✖	✖	✔	✔ (✔)	✖ (✖)	NA	NA	✔	NA
Hematian et al. 2009	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	NA	✖	✖	✔
Horovitz et al. 2014	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	NA	✖	✖	✖
Kober & Eggleton 2005	✔	✔	✔✖	✖	✔	✔ (✔)	✖ (✖)	NA	✔	✔	✖
Koritsas et al. 2008	✔	✔	✔	✔	✔	✔ (✔)	✖ (✖)	✔	✖	✔	✔
Kraemer et al. 2003	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	NA	✖	✖	✖
Lachapelle et al. 2005	✔	✔	✖	NA	✔	✔ (✔)	✔ (✔)	NA	✔✖	✖	✖
Martin et al. 2005	✔	✔	✔	✔	✖	✔ (✔✖)	✔✖(✔✖)	✔	✔	✔	✖
McClean et al. 2007	✔	✔	✔	✔	NA	✔ (✔)	NA (NA)	NA	NA	✖	✖
Perry et al. 2011	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔✖)	✔	✔	✔	✖

	Reporting – clearly described										
	Hypothesis/ Aims	Main outcome	Recruitment characteristics	Intervention or grouping	Confounders	Main finding (Qol)	Random variability (Qol)	Individual drop out	Actual p values	Consent	Power considerations
Perry et al. 2013	✔	✔	✔	✖	✔	✔ (✔✖)	✔ (✔✖)	NA	✔	✔	✖
Perry & Felce 2005	✔	✔	✔	NA	✔	✔ (✔)	✖ (✖)	NA	✖	✖	✖
Reiter 2001	✔	✔	NS	✔	NA	✖ (✖)	✔ (✔)	NA	✖	✖	✖
Riches et al. 2011	✔	✔	✔	✔	✖	✔ (✔✖)	✔ (✔)	✔	✔	✖	✖
Stancliffe et al. 2007	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	✔	✔✖	✔	✔
Vine & Hamilton 2005	✔	✔	✔	✔	NA	✔ (✔)	✖ (✖)	NA	✔	✔	✖
Wheeler et al. 2013	✔	✔	✔	✔	✔	✔ (✔)	✔ (✔)	NA	✔	✔	✖
Willner et al. 2013	✔	✔	✔	✔	✔	✔ (✔✖)	✔ (✔)	✔✖	✔	✔	✔

Note: ✔ = Item adequately addressed; ✖ = Item not adequately address; ✔✖ = partially addressed; **NS** = not stated; **NA** = not applicable; Qol = Quality of Life.

Table 1.6: Quality review of the resulting papers following a systematic review on the use of established quality of life (Qol) measures in the intellectual disability population (including checklist questions 11-26; Downs & Black, 1998).

	External Validity		Internal Validity – Bias					Internal validity- Confounding					
	Invited representative of population	Participated representative of population	Subjects blind	Assessor blind	Account for differing lengths of time	Statistical analysis	Full Qol measure used	Case and control from same pop.	Same Recruitment period	Subjects randomised	Randomisation concealed	Confounding variable accounted for in analysis	Drop out accounted for
Banks et al. 2010	✔	NS	✘	✘	✘	✔	✘	✔	✔	NA	NA	✘	✔
Barber et al. 1994	✔	NS	✘	✘	✔	✔	✔	NA	NA	NA	NA	✘	NS
Beadle-Brown et al. et al. 2009	✔	✔✘	NA	NA	NA	✔	✔✘	NA	NA	NA	NA	NA	NA
Beyer et al. 2010	✔	NS	✘	✘	✘	✔	✔	✔	✔	NA	NA	✘	NA
Bouras et al. 2004	✔	NS	✘	✘	✔✘	✔	✔	✔	✔	NA	NA	✔	NA
Bramston et al. 2005	✔	NS	✘	✘	NA	✔	✔	✘	NS	NA	NA	✘	NA
Chou et al. 2008	✔	✔✘	✘	✘	NA	✔	✔	✔	✔	NA	NA	✔	NA
Chou et al. 2011	✔	✔✘	✘	✘	✔	✔	✔	NA	NA	NA	NA	✘	✘
Chou & Harman et al.2011	✔	✔✘	✘	✘	✔	✔	✔	✔	✔	NA	NA	✔	✔
Cooper & Picton 2000	✔	✔	✘	✘	NS	✔	✔	✔	✔	✘	NA	✘	✘
Duvdevany & Arar 2004	NS	NS	✘	✘	✔	✔	✘	✔✘	✔	NA	NA	✔✘	NS
Felce et al. 2008	NS	✔✘	✘	✘	✘	✔	✔✘	✔	✔	NA	NA	✔	NA
García-Villamisar et al. 2013	NS	NS	✘	✘	NA	✔	✔	NA	NA	NA	NA	NA	NA

	External Validity		Internal Validity – Bias					Internal validity- Confounding					
	Invited representative of population	Participated representative of population	Subjects blind	Assessor blind	Account for differing lengths of time	Statistical analysis	Full QoI measure used	Case and control from same pop.	Same Recruitment period	Subjects randomised	Randomisation concealed	Confounding variable accounted for in analysis	Drop out accounted for
Hartnett et al. 2008	NS	NS	✗	✗	✔	✗	✔	✔	✔	NS	NS	NA	✔
Hematian et al. 2009	✔	✔	NA	✗	NA	✔	✔	✔	✔	NA	NA	✗	NA
Horovitz et al. 2014	NS	NS	✗	NS	NA	✔	✔	✔	✔	NA	NA	✔	NA
Kober & Eggleton 2005	✔	NS	✗	✗	✗	✔	✔	✔	✔	NA	NA	✔	NA
Koritsas et al. 2008	NS	NS	✗	✗	NA	✔	✔	NA	NA	NA	NA	NA	✔
Kraemer et al. 2003	✔	✔✗	✗	✗	NA	✔	✔	✔	✔	NA	NA	✔	NA
Lachapelle et al. 2005	✗	✗	✗	✗	NA	✔	✔	NA	NA	NA	NA	NA	NA
Martin et al. 2005	✔	NS	✗	✔	✔	✔	✔	✔	✔	✔	✗	✔	✔
McClean et al. 2007	✔	✔	NA	NA	NA	✗	✔	NA	NA	NA	NA	NA	NA
Perry et al. 2011	✔	✔	✗	✗	✗	✔	✗	✔	✔	✗	NA	✗	✔
Perry et al. 2013	✔	✔✗	✗	✗	✔✗	✔	✗	✔	✔	NA	NA	✔	NA
Perry & Felce 2005	✔	✔	✗	✗	NA	✔	✔	NA	NA	NA	NA	NA	NA
Reiter 2001	✔	NS	✗	✗	NA	✔	✔	NA	NA	NA	NA	NA	NA
Riches et al. 2011	✔	✔	✗	✗	✔	✔	✔	NA	NA	NA	NA	✔✗	✔
Stancliffe et al. 2007	✔✗	✔	✗	✗	✔	✔	✔	NA	NA	NA	NA	✔	✔

	External Validity		Internal Validity – Bias					Internal validity- Confounding					
	Invited representative of population	Participated representative of population	Subjects blind	Assessor blind	Account for differing lengths of time	Statistical analysis	Full Qol measure used	Case and control from same pop.	Same Recruitment period	Subjects randomised	Randomisation concealed	Confounding variable accounted for in analysis	Drop out accounted for
Vine & Hamilton 2005	✔	✔	✘	✘	✔	✔	✔	NA	NA	NA	NA	NA	NA
Wheeler et al. 2013	✔	✔	✘	✘	NA	✔	✔	✔	✔	NA	NA	✔	NA
Willner et al. 2013	✔	✔	NS	✔	✔	✔	✔	✔	✔	✔	✔	✔	✔✘

Note: ✔ = Item adequately addressed; ✘= Item not adequately address; ✔✘= partially addressed; **NS** = not stated; **NA** = not applicable; Qol = Quality of Life.

Tables 1.5 and 1.6 show a variety in the quality of papers, ranging from strong studies, such as Willner et al. (2013) to those with a number of limitations, such as Reiter (2001). All studies clearly reported their hypothesis or aim and how they intended to measure this. The papers clearly reported their main finding and the majority stated the variability. Over half did not report actual p values and only four papers (Hematian et al., 2009; Koritsas et al., 2008; Stancliffe et al., 2007; Willner et al., 2013) considered power either before or after analysis. In relation to their use of Qol measures, the quality of reporting varies greatly and is reflected in the Qol score discussed previously. A number of papers were considered to describe only partially the independent variable. The majority of papers took a quantitative approach with the exception of Hartnett et al. (2008). Considering the challenges of gaining informed consent in this population, the majority of studies document this process although ten studies did not. In terms of experimental design, only two papers state that the assessor was blind to the condition (Martin et al., 2005; Willner et al., 2013).

4.5 Theme of Papers

For the purpose of this review, papers were divided into two categories; those that looked at environmental changes in relation to Qol and those that considered individual characteristics.

4.5.1 Environmental factors

Environmental factors were considered to be those that were external to the person and included the setting of residence or day service, carer approach and employment. The 21 papers that considered environmental factors are presented in Table 1.7.

Table 1.7: Summaries of those papers which address environmental factors from the 31 resulting papers of a systematic review on the use of quality of life measures with psychometric properties in the ID population

	Summary	Participant characteristics	QoL		Other measures used	QoL score
			Measure	Completed by		
Residential or day setting						
Barber et al. 1994	Relocation from institution to community. Repeated Measures (Pre and 1 month post move). **QoL results:** No difference in QoL domains following move to community.	**N=** 15 (Male 46.6%) **Age range (mean):** 30-57 (42.38) **Level of ID:** Mild - severe. **Ethnicity:** NS **Country:** Australia	QoL-Q	Individuals or staff	No	3
Chou et al. 2008	Cost and outcomes of small residential home, group, or institution. Between group comparison. Matched groups. **QoL results:** Small residential home had greater QoL scores (controlling for adaptive and mal adaptive behaviour). Residents' adaptive behaviour is associated with QoL and inclusion in daily activities. Type and location of residence contributed to QoL.	**Residential, group, institute:** **N=** 103,69,76 (Male 77.7%,60.9%,72.4%) **Age range (mean):** NS (28.6,30.5, 29.5) **Full sample** (N=248) **Level of ID:** Mild - profound **Ethnicity:** NS **Country:** Taiwan	QoL-Q	Individuals with main carer	No	5

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	Completed by		
Chou et al. 2011	Service outcomes of a residential scheme. Repeated measures. **Qol results:** Improved at 1 year & 2 years post move. No difference between those from a family home or institution.	**T1** **N=** 49 (Male 67.3%) **Age range (mean):** 19-57 (27.1) **T5** **N**=29 (Male 82.7%) **Age range (mean):** 21-59 (30.7) **Full sample:** **Level of ID:** Mild – profound **Ethnicity:** NS **Country:** Taiwan	Qol-Q	Individual with main carer	No	1
Cooper & Picton 2000*	*<u>*Continuation of Barber et al. 1994*</u> Relocation from institution. Community units vs institution. Repeated measure (pre, post, follow up). **Qol results:** Increase in Qol in both groups at 6 months, remaining at 3 years follow up.	**Community residential unit** **N**=26 (Male 52%) **Age range (mean):**NS(52) **Training centre** **N**=19 (Male 53%) **Age range (mean):**NS(55.2) **Full sample:** **Level of ID:** Mild - severe. **Ethnicity:** NS **Country:** Australia	Qol-Q	Individual or staff	No	1

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	Completed by		
Duvdevany & Arar 2004	Community residential setting vs foster families. **Qol results:** No difference in Qol. Those employed had higher Qol than those not, which was more common in community setting. Higher involvement in leisure activities and lower feelings of loneliness are predictive of higher Qol scores.	**Community** **N**=45(Male 44.4%) **Foster** **N**=40 (Males 55%) **Full sample:** **Age range (mean):**18-55 (NS) **Level of ID:** Mild – moderate **Ethnicity:** NS **Country:** Israel	Qol-Q	Individual	No	5
Felce et al. 2008	Group vs semi-independent setting . Matched groups. **Qol results:** Higher scores on the Choice Questionnaire for those in semi-independent living. No difference in MLSS.	**Staffed** **N=**35 (Male 62.9%) **Level of ID:** ABS M=90 **Age range (mean):** NS (50) **Semi independent** **N=** 35(Male 48.6%) **Age range (mean):** NS (44) **Full sample** (N=70) **Level of ID:** ABS M=95 **Ethnicity:** Mainly Caucasian **Country:** Wales, England	Choice MLSS	Staff and Individuals	Yes	4

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	**Completed by**		
Hartnett et al. 2008	Campus vs community based day service. repeated measures (pre and post). **Qol results:** Community participants increased Qol at six months. Campus based participant's remained the same.	**N**=8 (4 campus and 4 community) (Male 62.5%) **Age range (mean):** 21-32 (NS) **Level of ID:** Severe **Ethnicity:** NS **Country:** Ireland	Qol-Q	Parent and staff member	Yes	3
Perry et al. 2011	Resettlement of **i**ndividuals with severe challenging behaviour. Staggered resettlement over 2 years. Repeated measures. **Qol results:** No change in choice scores. ComQol appeared to decrease whilst MLSS increased	**N=**19 (Male 68.4%) **Age range (mean):** 36-67(47) **Level of ID:** NS, **Ethnicity:** NS **Country:** Ireland	Choice ComQol MLSS	Staff (N=14) Individuals (N=4) Individuals (N=4)	Yes	4
Perry et al. 2013	In vs out of area placements for people with CB. **Qol results:** No difference in Qol seen.	***In Area:*** **N**= 38 (Male 60.5%) **Age range (mean):** NS(46) **Out of Area** **N**= 38 (Male 65.8%) **Age range (mean):** NS(35) **Full sample** (N=76) **Level of ID**: NS **Ethnicity**: Caucasian **Country**: Wales	Choice ComQol MLSS	Paid carers (N=76) Individuals (N=34) Individuals (N=24)	Yes	3

Carers approach.						
Chou & Harman et al.2011	Evaluating AS training for managers and supervisors. Repeated measures (baseline, post, follow up). Non-equivalent control. **Qol results:** Improvement in choice scores between baseline and post training.	**AS group** **N** =49 (Male 63.3%) **Age range (mean):**19-54 (32.3) **Control group** **N**=19 (Male 57.9%) **Age range (mean):**24-53 (32.6) **Full sample** (N= 69) **Level of ID:** Mild - profound **Ethnicity:** NS **Country:** Taiwan	Choice	Individual with staff support	No	1
Koritsas et al. 2008	Evaluating AS training. Repeated measures (pre, post,6 month follow up). **Qol results:** 'Overall choice' increased over time. Improvements in the choice subdomains between pre and post but not follow up.	**N=** 12 (Male 75%) **Age range (mean):** 27-57 (37) **Level of ID:** Moderate - severe, (3 unknown) **Ethnicity:** NS **Country:** Australia	Choice	Staff	No	3
Riches et al. 2011	Evaluating AS training. Repeated measures: before and 3 months after training. **Qol results:** No difference in choice.	**N=** 13 (Male 69.2%) **Age range (mean):** 30-72 (51.5) **Level of ID:** NS **Ethnicity:** NS **Country:** Australia	Choice	Staff	No	1
Stancliffe et al. 2007	Evaluating Active support. Staggered Staff training. Repeated measures. Multiple baseline across group design. **Qol results:** No impact on Choice.	**N=** 22 (Male 36.4%) **Age range (mean):** 27.2-62.1(41.2) **Level of ID:** Mild - severe **Ethnicity:** NS **Country:** Australia	Choice	Staff	No	1

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	Completed by		
Martin et al. 2005	Assertive vs standard community treatment for Psychiatric disorders (affective, psychotic). RCT. **Qol results:** No difference. Improvement in Qol with standard treatment.	**N**=20 (Male 50%) **Age range (mean):** 20-63 (45) **Level of ID:** Mild - moderate **Ethnicity:** 80% Caucasian 20% Afro Caribbean **Country:** England	Qol-Q	Individual	No	1
McClean et al. 2007	Evaluating positive behavioural support. Case study series. Repeated measures. **Qol results:** 3/5 showed significant improvement in Qol.	**N=** 5 (Male 60%) **Age range (mean):** 21-38 (28) **Level of ID:** Mild - severe **Ethnicity:** NS **Country:** Ireland	Qol-Q (1989)*	Informant	No	1
** Author contacted regarding the reporting of the Qol measure. He confirmed that the wrong date was stated, it should have read Qol-Q Schalock (1993)*						
Employment						
Banks et al. 2010	Impact of job loss.. Repeated measures. **Qol results:** No impact. *Qualitative interview suggests some impact.*	**Breakdown** **N**=9 **Full sample** (N= 49) (Male 61.2%) **Age range (mean):** 16-53 (31.2) **Level of ID:** WASI (2 item) M=69 **Ethnicity:** NS **Country:** Scotland	ComQol	Individuals and supporters	Yes	1

	Summary	Participant characteristics	QoL		Other measures used	QoL score
			Measure	**Completed by**		
Beyer et al. 2010	Compares supported employment, employment enterprise workers and day service attendees. Control group of non-disabled co-workers. **QoL results:** Supported employment had higher objective scores. Supported employment had higher subjective QoL than non-disabled controls.	**Supported** **N=** 17 (Male 76%) **Age range (mean):** NS(34) **Level of ID:** ABS percentile ranks 85.0 **Enterprise** **N=** 10 (Male 40%) **Age range (mean):** NS(39) **Level of ID:** ABS percentile ranks 77.8 **Day service** **N=** 10 (Male 90%) **Age range (mean):** NS(42) **Level of ID:** ABS percentile ranks 74.1 **Full sample** (N= 37; exc. 12 non-disabled) **Ethnicity:** NS **Country:** Wales	ComQol	Individuals	No	3

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	Completed by		
Kober & Eggleton 2005	Open vs sheltered employment. **Qol results:** Open employment have higher Qol. Low work ability group: No difference in Qol across differing employment. High work ability group: Open employment had higher total Qol & empowerment/independence and social belonging/community integration domain scores.	**N(open, sheltered)=** 117(64,53) (Male 61.5%) **Age range (mean):** (31) **Level of ID:** NS **Ethnicity:** NS **Country:** Australia	Qol-Q	Individuals or guardians	No	3
Kraemer et al. 2003	Qol in young adults during transition. **Qol results:** School leavers had higher total Qol than school attenders. Those in open /support employment had the highest scores. This difference disappeared when the comp/productivity variable was removed. Adaptive behaviour predicted Qol. Higher functioning , larger social networks and with parents knowledgeable about adult services and less negatively impacted by the individual, predicted higher total Qol scores.	**N=** 188 **Gender:** Male 49% **Age range (mean):** 18-26 (21) **Level of ID:** Moderate - severe **Ethnicity:** NS **Country:** America	Qol-Q	Parents	No	5

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	Completed by		
Social support						
Bramston et al. 2005	Tests three conceptual principals of Qol. **Qol results:** Social support predicts the Qol domains of satisfaction with; safety, emotional wellbeing and material wellbeing.	**N**=80 (+120 non ID controls) **Gender:** 'evenly distributed across groups' **Age range (mean):** 17-25 (20.8) **Level of ID:** Mild to moderate **Ethnicity:** NS **Country:** Australia	ComQol	Individuals	No	5
Socioeconomic						
Hematian et al. 2009	Personal characteristic and Qol. Between those with ID and those without ID from a low socioeconomic background **Qol results:** No difference in total Qol. Regression: those with salesman fathers with a high school diploma or degree had higher Qol score in individual with ID. Suggesting a role of socio economic status.	**N(Id, Non ID)=**82(41,41) **Gender:** 48m,34f **Age range (mean):** 14-26 (18.3) **Level of ID:** NS **Ethnicity:** NS **Country:** Iran	Qol-Q	Individuals	No	5

Note: ABS= Adaptive Behaviour Scale, AS= Active support, ComQol= Comprehensive Quality of Life Scale, Choice= Choice Questionnaire, ID= Intellectual Disability, LSS= Life Satisfaction Scale, MLSS= Multifaceted Life Satisfaction Scale, NS= Not stated, PWI= Personal Wellbeing Index, Qol= Quality of Life, Qol-Q= Quality of Life Questionnaire.

4.5.1.1 Residential or day setting.

Nine studies considered the impact of different residential or day settings on an individual's Qol. Of these, four studies used a within group design (Barber et al., 1994; Chou et al., 2011; Cooper & Picton, 2000; Perry et al., 2011) to consider the impact of relocation from institution to community. Cooper and Picton (2000) also included a between group comparison. These papers predominantly used the Qol-Q although differed in the comparisons made; either comparing the subscale means (Barber et al., 1994), the mean item score (Copper, 2000) or the mean total score (Chou et al., 2011). There was no improvement in Qol at one month post move (Barber et al. 1994), but an improvement was seen at six months and maintained at three years (Cooper & Picton, 2000) and at one and two year post move (Chou et al., 2011). The results provided by Chou et al. (2011) should however be viewed with some caution as individuals who had relocated from family homes and institutions were grouped together and the Qol scores oscillated meaning no improvement was found at six and eighteen months post move. In addition to the improvement found in those moving from institution to community, Cooper and Picton (2000) also found an improvement in Qol of those moving from institution to institution. Finally, in contrast, Perry et al. (2011) found no difference in the pre and post move scores of the Choice Questionnaire.

Four studies (Chou et al., 2008; Duvdevany & Arar, 2004; Felce et al., 2008; Perry et al., 2013) considered the impact of differing living environments, utilising a between groups design. Felce et al. (2008) used both the Choice Questionnaire and MLSS. In addition Perry et al. (2013) also used the ComQol (satisfaction scale only). The remaining papers used the Qol-Q. Those living semi-independently were compared to those who were living in group homes (Felce et al., 2008) and were found to have a higher score on the Choice

Questionnaire, although no difference was found on the MLSS (only the community and recreation subscales were used). Those living in small residential homes had higher levels of Qol compared to those in group homes or institutions when matched on age, sex and level of ID (Chou et al., 2008). No differences in Qol were found between those living in residential settings and those in a foster family, as measured by Qol-Q. However, secondary analysis showed those in foster homes were more likely to work and those that worked were found to have higher Qol. Finally, in vs out of area placements for those with challenging behaviour were compared (Perry et al., 2013) and no difference in Qol was found.

Only one paper considered the location of the residence (Chou et al., 2008), showing that rural residences were associated with higher Qol scores than urban residences. In secondary analysis Perry and Felce (2005, see Section 4.5.2.4 for further details) note that the lower the number of residents, the higher the Qol (based on the Choice score as rated by the individual).

In a slight departure from considering the locations of residence, Hartnett et al. (2008) considered day service locations and found that those attending the community based day service showed an increase in Qol based on the Qol-Q at 6 months compared to those attended a campus based service, although this was not statistically confirmed due to a low sample size.

In summary, in those papers looking at the Qol of individuals with ID in relation to a move from institution to community residence, some improvements in Qol as measured by the Qol-Q were found. However, these differed both between studies and across different time periods. No difference was seen in Qol as measured by the Choice Questionnaire. When

considering differing residential environments, people living semi independently compared to those in group homes had higher Qol scores based on the Choice Questionnaire but not the MLSS. Those in small residential homes had higher Qol than those in group or institutional settings (Qol-Q). Furthermore, no difference in Qol was seen between those living in residential home compared to those in foster homes or those with CB living in area vs out of area.

4.5.1.2 Carers and staff approach.

Six papers considered the impact of staff approaches to the individuals they care for. Four of these used the Choice Questionnaire which was on the whole completed by staff to evaluate Active Support (Chou & Harman, 2011; Koritsas et al., 2008; Riches et al., 2011; Stancliffe et al., 2007). These papers utilised the Choice Questionnaire to identify changes within the environment and did not consider this in relation to Qol. The Choice Questionnaire was either compared on each subdomain score (Koritsas et al., 2008) or combined and the totals compared (Chou & Harman, 2011, Riches et al., 2011, Stancliffe et al., 2007). Following Active Support training, an improvement in the Choice score at four months and a further improvement at 14 months was found (Chou & Harman et al., 2011). However, when compared to participants whose staff had not received full training, no differences between the groups were found. The authors suggest this this may in part be due to some staff and managers of the control group having also received partial Active Support training. Koritsas et al. (2008) found improvement on the Choice Questionnaire immediately following training, however only the improvement in overall choice remained at 6 months post training. Only two papers of these papers included an intervention adherence check documenting the change in staff approach following training, either via questionnaire based measures (Riches et al., 2011) or observation (Stancliffe et al., 2007).

Although both documented positive changes in staff approach, neither found an improvement on the Choice Questionnaire.

Positive behaviour support for individuals with longstanding challenging behaviour was assessed (McClean et al., 2007) through five case studies, showing significant improvements for three participants on their Qol-Q percentile score as scored by proxies.

Assertive community treatment for individuals with mental health problems and ID was assessed (Martin et al., 2005) using a randomised control trial, comparing it with treatment as usual. This found no significant changes in Qol-Q.

In summary, carers and staff approach was considered in terms of the support they provide. The evidence that active or positive behavioural support improves Qol is mixed. Assertive community treatment for those with mental health difficulties was no more effective than treatment as usual.

4.5.1.3 Employment.

As previously stated in section 4.5.1.1, Duvdevany and Arar (2004) found those individuals who were employed had a higher Qol than those who do not. A further four papers considered employment environments and Qol using both within group (Banks et al., 2010) and between group comparisons (Beyer et al., 2010, Kober & Eggleton, 2005; Kraemer et al., 2003). Using the ComQol as completed by individuals with ID, those in supported employment had significantly higher objective Qol than those in enterprise or day service employment, all of which had lower Qol scores than in the Non-ID control group (Beyer et al., 2010). Those in supported employment scored higher subjective Qol than the Non-ID control group. Those in open employment had higher Qol-Q scores than those in sheltered employment (Kober & Eggleton, 2005; Kraemer et al. 2003). However,

when the competence subscale was excluded this difference was no longer found (Kober & Eggleton, 2005). When individuals with high and low work ability were compared, differences in Qol between employment environments were only found in those with high work ability. Here, those in open employment had higher total Qol and higher domain scores for empowerment/independence and belonging/community (Kober & Eggleton, 2005). Employment which broke down either though job loss or by choice, was seen to have no negative impact on Qol, as measured by the ComQol (satisfaction scale only) (Banks et al., 2010).

In summary, those who are employed have a higher Qol than those who are not. Furthermore, certain types of employment (open, supported) are linked to higher Qol than others. This is particularly seen in those with higher work abilities. Although this improvement may not observed in all subscales. No impact of job loss on the ComQol measure was seen.

4.5.1.4 Social support.

Social support was considered by one paper (Bramston et al. 2005) which used the ComQol completed by individuals. The ComQol satisfaction domains of material well-being, safety and emotional well-being were predicted by the levels of social support.

4.5.1.5 Socioeconomic Status.

Hematian et al. (2009) used regression to consider factors which may be predictive of Qol (as rated on the Qol-Q by individuals), Father's career and education were seen to be predictive. Hermatian (2009) suggests this may be representative of socioeconomic status and imply that those with a lower socioeconomic status have lower Qol scores.

4.5.1.6 Interim summary.

The papers considering environmental factors and Qol have covered the topics of location of residence, location of day service, carer or staff approach, employment, socioeconomic status and social support. The measure and method of utilisation has varied across topics. Even in those papers evaluating Active Support where the Choice Questionnaire was dominant, the way that this measure was compared varied between papers. Some papers found that environmental factors such as relocation to community and participation in employment can have a positive impact on an individual's Qol, however this has not been found consistently.

4.5.2 Individual characteristics

Individual characteristics were considered to be those such as mental health, challenging behaviour and ability. Ten papers considered individual characteristics and are presented in Table 1.8.

Table 1.8: Summaries of those papers which address individual characteristics from the 31 resulting papers of a systematic review on the use of Qol measure with psychometric properties in the ID population

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	Completed by		
Mental Health						
Bouras et al. 2004	Schizophrenia-spectrum psychoses in those with and without ID. **Qol results:** No statistical difference in Qol.	**ID** **N=** 53 (Male 51%) **Age range (mean):** 17-77 (39.49) **Ethnicity:** 33 White, 17 Afro-Caribbean, 2 Asian, 1 Other **Non-ID** **N=**53 (Male 77%) **Ethnicity:** 35 White, 14 Afro-Caribbean, 2 Asian, 1 Other **Full sample** (N=106) **Level of ID:** Mild **Country:** England	Qol-Q	Individual	No	1
Horovitz et al. 2014	Comparing individuals with; no axis I disorder, psychotic disorder or mood/anxiety disorder. **Qol results:** No axis I diagnosis had higher total Qol scores (rater and age controlled). The no axis I diagnosis group were higher on the competency/productivity subdomain, compared to those with mood/anxiety. Self raters provided higher Qol scores than proxies.	**No Axis I** **N=** 34 (Male 61.8%) **Age range (mean):** NS (52.94) **Psychotic Disorder** **N=**36 (Male 54.6%) **Age range (mean):** NS (54.58) **Mood/Anxiety** **N=**68 (Male 60.30%) **Age range (mean):** NS (48.40) **Full sample** (N=138)	Qol-Q	Individual or proxy	No	3

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	Completed by		
		Level of ID: Borderline- moderate **Ethnicity:** NS **Country:** America				
Autism and Challenging Behaviour						
Beadle-Brown et al. 2009	Predictors of Qol**.** Correlational design. **Qol results:** No impact of social impairment on Qol. Autism had lower community satisfaction. Lower CB showed higher Community Satisfaction & overall LSS score. An IQ over 50 had significantly higher LSS score & recreation satisfaction.	**N**=72 **Proxy:** 14% Parent, 61% Keyworker, 25% other **Gender:** NS **Age range (mean):** 27-41.5 (34) **Level of ID:** Non-verbal IQ =45.4 **Ethnicity: NS** **Country:** England	LSS	Individual N=9 Proxy N=72	Yes	3
García-Villamisar et al. 2013	Explores the mediatory relationship between CB, ASD and QoL. **Qol results:** A model where Qol acts to mediate the relationship between ASD symptoms and CB is suggested. Qol subdomains (excluding the community subdomain) negatively correlated with ASD traits & CB.	**N=** 70 **Gender:** NS **Age range (mean):** 18-43 (26.6) **Level of ID:** NS **Ethnicity:** NS **Country:** Spain	Qol-Q	Individual or caregiver	No	5

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	Completed by		
Social Abilities						
Reiter 2001	Evaluate a social skills class. Repeated measures (beginning, middle, end). **Qol results:** Significant improvement in the area of social integration.	**N=** 11(Male 45%) **Age range (mean):** 17-18 **Level of ID:** Moderate **Ethnicity:** NS **Country:** Israel	Qol-Q	Individuals	No	3
Ability						
Perry & Felce 2005	Qol of individuals living in fully staffed community residences. Correlational. **Qol results:** Adaptive and challenging behaviour predict staff choice scores. Residents scores were predicted by number of residence per setting and type of residence. No resident characteristics were associated with ComQol or LSS.	**N=** 154(Male 62%) **Age range (mean):** NS(46) **Level of ID:** ABS rank M=60 **Ethnicity:** NS **Country:** England and Wales	Choice ComQol LSS	Individuals and staff	Yes	5
Vine & Hamilton 2005	Characteristics associated with community integration in those previously institutionalised. **Qol results:** No correlation between Qol and adaptive & maladaptive behaviour, age or life circumstance.	**N=** 37(Male 100%) **Age range (mean):** 32-71 (44.60) **Level of ID:** Severe **Ethnicity:** NS **Country:** Australia	ComQol	Staff	No	1

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	**Completed by**		
Self-determination						
Lachapelle et al. 2005	Self-determination and Qol. **Qol results:** Functional analyses, showed a relationship between self-determination and higher levels of Qol.	**N=**182 (Male 50%) **Age range (mean):** NS **Level of ID:** Mild **Ethnicity:** NS **Country:** Canada, France, USA, Belgium	Qol-Q	Individual	No	5
Offending and Anger						
Wheeler et al. 2013	Offender vs non-offenders. Matched on age, gender and level of ID. **Qol results:** No difference on Choice or Personal index. Non-offending participants scored higher rating life as a whole.	**Offenders** **N=**27 (Male 67%) **Age range (mean):** 20-61 (36.48) **Non Offenders** **N=**19 (Male 52.6%) **Age range (mean):** 22-58 (37.79) **Full sample** (N=46) **Level of ID:** Borderline - moderate **Ethnicity:** NS **Country:** UK	Choice PWI	Individuals N=27,16 Individuals N=27,18	Yes	4
Willner et al. 2013	Evaluated CBT anger management. Cluster Randomised control trial. Repeated measures: pre, post, follow up. **Qol results:** No difference found.	**Intervention** **N=**91(Male 70%) **Age:** Median =37 **Level of ID:** IQ Median=59 **Control** **N=**90(Male71.4%) **Age:** Median =38.5 **Level of ID:** IQ Median=55	ComQol	Individual N=129 at 16 weeks N=140 at 10 months	No	1

	Summary	Participant characteristics	Qol		Other measures used	Qol score
			Measure	Completed by		
		Full sample (N=179) **Ethnicity:** NS **Country:** England, Scotland , Wales				

Note: ABS= Adaptive Behaviour Scale, AS= Active support, ASD= Autistic spectrum disorder, CB= challenging behaviours, ComQol= Comprehensive Quality of Life Scale, Choice= Choice Questionnaire, ID= Intellectual Disability, LSS= Life Satisfaction Scale, MLSS= Multifaceted Life Satisfaction Scale, NS= Not stated, PWI= Personal Wellbeing Index, Qol = Quality of Life, Qol-Q Quality of Life Questionnaire

4.5.2.1 Mental health.

Mental health was considered by two papers, using a between groups design and the Qol-Q (Bouras et al., 2004; Horovitz et al., 2014). Horovitz et al. collected the Qol-Q from either the individuals themselves or a proxy and controlled for the different rater in analysis. Those with ID and either a mood or a psychotic disorder had significantly lower total Qol that those with ID without an axis one disorder (Horovitz et al., 2014). Individuals with schizophrenia-spectrum disorders with and without ID, were compared. No difference in Qol as measured by the Qol-Q was found (Bouras et al., 2004)

4.5.2.2 Autism and CB.

Autism Spectrum Disorder (ASD) and challenging behaviour were considered in two papers (Beadle-Brown et al., 2009; García-Villamisar et al., 2013). The LSS was used by Beadle-Brown et al. (2009), and although not considered here due to the psychometric implications, the LSS was combined with some adapted questions from Qol-Q to create a score for overall Qol. Based on proxy rating, those with and without ASD differed only on their community satisfaction subdomain, with those with autism having lower levels of satisfaction. A correlation between higher levels of CB and lower total LSS score was found (Beadle-Brown et al. et al., 2009). The relationship between Qol (as measured by the Qol-Q) and CB and traits of Autistic Spectrum Disorder (ASD) was considered in the García-Villamisar et al. (2013) paper, with higher levels of CB and ASD traits associated with lower Qol. The mediating role of QOL in the relationship between ASD and CB is suggested (García-Villamisar et al., 2013).

In summary, ASD and or CB are associated with lower Qol.

4.5.2.3 Social ability.

Social ability is the primary focus of one paper (Reiter, 2001) which used individuals' repeated reports on the Qol-Q to evaluate a social skills class. There is some suggestion that this resulted in an improvement of Qol, however the prose did not match the figures reported in the tables or graphs. In secondary analysis, Beadle-Brown et al. (2009; discussed in section 3.5.2.2) found no difference between those with and without social impairment on the LSS.

4.5.2.4 Adaptive behaviour.

Vine and Hamilton (2005) considered the correlation between adaptive behaviour and Qol (as measured by proxy reports on the ComQol) however found no association. Perry and Felce (2005) found that over half of the variance in staff rated total Choice scores was associated with participants' characteristics (primarily adaptive behaviour). Only a third of participants in the Perry and Felce study were able to complete the Choice Questionnaire themselves. Considering these scores in isolation to those completed by proxies, the association with adaptive behaviour was not seen.

Secondary analysis in two papers discussed in Section 3.5.1 showed that levels of adaptive behaviour was significantly associated with Qol (Chou et al., 2008; Kramer, 2003). Additionally those with an IQ of below 50 had a lower overall LSS score and lower subdomain score on recreational satisfaction than those with an IQ over 50 (Beadle-Brown et al., 2009).

4.5.2.5 Self-determination.

Lachapelle et al. (2005) considers the role of individuals' self-determination in Qol. Utilising a large sample (n=182) across multiple countries, this suggested that those with higher self-determination were more likely to have high Qol.

4.5.2.7 Offending and Anger

Wheeler et al. 2013 considered the Qol of those who offend in comparison to those who have not. No difference was found on the Choice Questionnaire (completed by the individual). On the PWI non-offenders had a higher score on the 'Life as a whole' question, with no other differences being found. Willner et al. (2013) utilised the ComQol to evaluate a CBT anger management intervention. No difference in anger or Qol was found.

In summary, those who offend only demonstrated a lower Qol in the 'Life as a whole' domain of the PWI.

4.5.2.8 Activities

No paper focused primarily on the impact of activities on Qol, however when comparing locational groups, a relationship between higher involvement in leisure activities and lower feelings of loneliness were predictive of better Qol scores (Duvdevany & Arar, 2004). This was echoed by Chou et al. (2008) finding that those involved in some form of daily activity outside of the residence such as school or day centre predicted higher Qol than those who remained in accommodation all day.

5 Discussion

The recent publication of two systematic reviews (Li et al., 2013; Townsend-White et al., 2012) identified eleven different measures with published psychometric properties for quality of life (Qol) in individuals with intellectual disabilities (ID). The current review gathered information on the use of these measures within published research.

5.1 Utilisation of Measures

With some measures being created as long as thirty years ago (e.g. LSS) there were relatively few resulting papers. Of the eleven measures, half were not utilised within the resulting papers. Perhaps the lack of articles found in some way reflects the challenges of data collection in this population and that the focus has been on the development of measures not their utilisation. There are also circumstances where researchers adopt an individualised approach such as combining elements of differing questionnaires (e.g. Beadle-Brown et al., Murphy & DiTerlizzi, 2009) which in turn makes it difficult to compare the results to the wider field.

The measures were used in a range of differing experimental designs making comparisons both between and within groups. Although covering the range of differing levels of ID, it was more common for those at the milder end to be considered than those at the profound end.

It was rare for studies to blind the assessor and or the participant to the nature of the study. This may have introduced bias and impacted the internal validity of studies, which was particularly a point of concern for the papers evaluating Active Support and is discussed in Section 5.2.2. Studies were conducted across a range of differing countries and although Qol is conceptualised as cross-cultural (Bramston, Chipuer, Pretty, 2005), the differing

cultural values of the individual and their wider system may still have an impact; for example it may play a role in the development of their subjective concept of satisfaction. Oishi, Diener, Lucas, and Suh (1999) found the importance given to predictors of life satisfaction varied between cultures. It is also important therefore to consider the ethnicity of participants. In the resulting papers only 10% (n=3) reported ethnicity and no papers considered possible relationships with Qol.

The Choice Questionnaire was often used as a measure of the change in a person's environment. It is interesting to consider whether from a person's ability and opportunity to make choices, their Qol can be directly inferred. This question is further highlighted in the results of Felce et al. (2008) where two groups significantly differed in their score on the Choice Questionnaire but did not differ on their Multifaceted Life Satisfaction Scale (MLSS) score.

Differences between the results of Qol measures found in some studies (Felce et al., 2008; Perry, Felce, Allen, & Meek, 2011; Perry & Felce, 2005) is contrary to the idea that although measurements may choose to measure differing domains together, they infer the same notion of Qol. This raises some questions about the measurement of Qol which needs further investigation.

A variety of sources were used to complete the Qol measures. Some asked the individual themselves, some used a proxy and others used a combination of the two. Self raters were found to score higher than proxies (Horovitz et al., 2014; Beyer et al., 2010) which raises questions about papers where the reports of individuals and proxies are combined. Furthermore the impact on the reporting of individuals when supported by staff or parent to complete the measures has not been considered.

In light of the continued differences found in the reporting of Qol, it is important that research clearly describes the source. When using both individuals and proxies, the number of both should be clearly stated and, due to the inconsistencies between the raters, analysis should consider these separately. The view of proxies remains a valuable source of information, particularly when the participant is unable to provide the information themselves, however this should be acknowledged as the level of Qol from the perspective of the proxy and not assumed to be equal to that of the individual. Future research should where possible look to gather information for each individual from both themselves and a proxy.

In summary, only half of the identified eleven measures were found to be utilised within the resulting papers. Measures were used to compare differing groups and to measure outcome, with the Quality of Life Questionnaire (Qol-Q) being utilised most. The question of who completes the measure continues to present challenges to researchers. This is not just a question of practicality; different groups of responders appear to provide differing accounts of Qol which requires careful consideration.

5.2 Themes covered

Almost two thirds of the resulting papers considered factors external to the individual. The largest proportion of these was pertaining to residential or day settings.

5.2.1 Residential or day setting

The papers relating to residential or day setting covers both the effect of moving and the difference found in differing settings.

The impact of deinstitutionalisation on individuals found in the papers were mixed and differed both between studies and across different time periods. The Perry et al. (2011)

paper found no difference in Qol as measured by the Choice Questionnaire. In considering this result, the authors reported that the majority of the staff in the new residences had originated from the institution from which the participants came and that staff had commenced training in Positive and Active behavioural support before baseline measurements were taken. Both the consistency of staff and the possible improvement in staff approach before deinstitutionalisation may have minimised the likelihood of improvement in Qol in response to the move.

When comparing different residential environments, differences in the results of the Choice and Multifaceted Life Satisfaction Scale (MLSS) were seen. This may be due to the qualitative difference in the two measures. The Choice Questionnaire measures a combination of a person's ability and opportunity to make choices whereas the MLSS focuses on an individual's satisfaction. It is perhaps unsurprising that those in semi-independent living have more opportunity to make choices.

A common limitation of the location-based papers was that many did not provide detail about the living environment and as such the change or lack of change in Qol may be related to other extraneous differences which were not controlled for, such as access to outdoor space, urban or rural location, relationship with staff etc. Only two papers considered this, finding that rural locations and lower numbers of residents were associated with higher Qol (Chou et al., 2008; Perry & Felce, 2005).

An often overlooked possibility in the deinstitutionalisation research is the novelty effect of moving which could impact Qol regardless of the residence itself. For example an improvement in Qol was seen in individuals moving from one institution to another (Cooper & Picton, 2000). Furthermore, how long after a change would you expect to see a

measurable Qol change? Hartnett et al. (2008) suggests that six months after the start of a new day service may be too soon, however if a change was identified after a year would it be valid to suggest that this is a result of the original change or a response to something more proximal?

5.2.2 Carer approach

Carers and staff approach was only considered in terms of the support they provide. The evidence that Active or Positive Behavioural Support improves Qol is mixed. An increase in Qol was reported in approximately half of the papers. Common limitations of these studies are that the staff who received training and provided intervention also completes the pre and post measures for the participants which may introduce bias in their reporting.

5.2.3 Employment

Individuals in employment, particularly open employment, were found to have higher Qol than those not. Careful consideration needs to be given to the content of the measures used to assess Qol. For example, one domain measured within the Qol-Q is productivity which enables those who work to have the opportunity to score higher on this domain than those who do not. The ability of the individual is likely to impact both the type of employment they are able to participate in and their level of Qol, and as such the difference in Qol for those in open employment may be a reflection of their ability and not the employment alone.

A common limitation for these papers is although they compare differing types of employment, the types of employment are not clearly defined and as such it is difficult to combine the results across papers.

5.2.4 Individual Characteristics

Fewer papers consider factors internal to the person than consider external factors. Those with mental health disorders, increased ASD traits and challenging behaviour had lower Qol scores (Beadle-Brown et al. et al., 2009; García-Villamisar et al.; Horovitz et al., 2014) than those without. Those with higher self-determination, adaptive behaviour or ability achieve higher Qol (Beadle Brown, 2009; Chou et al., 2008, Kramer, 2003; Lachapelle et al.; 2005; Perry & Felce, 2005).

With only one or two papers considering each of the individual characteristics, little can be concluded currently, however it is important that future papers bear the possible impact of differing personal characteristics in mind when comparing groups.

5.3 Themes not Covered

As there were relatively few resulting papers, there are a number of themes yet to be considered. A couple of these will be discussed briefly.

No research was found to consider the personal characteristics of staff/carers/parents and their relation to the individuals Qol. The impact of this may be two fold; it is thought that a person's own personality traits will influence how they subjectively measure Qol and so will impact proxies ratings but also the characteristics of those in our environment may impact how individuals themselves judge Qol.

In regards to individual characteristics there are no papers that primarily focused on an individual's activity. Although secondary analysis suggested that regular leisure activities predicted higher Qol (Chou et al.,2008; Duvdevany & Arar, 2004), which is consistent with existing research which suggests that increased physical activity is associated with increased Qol (Blick, Saad, Goreczny, Roman & Sorensen, 2015).

5.4 Summary of Themes

In summary, a range of topics have been considered within the literature, which has more frequently focused on factors external to the person such as location of residence. The identified research has often taken a broad brush approach to comparisons such as community vs institution. In this example, without providing much greater detail on the differences in facilities and in individual residents it remains difficult to draw valid conclusions. Furthermore, there is a possibility that the Qol measure and independent variable used by a study may overlap, as seen in the employment papers, which raise further concerns about the conclusions they make.

5.5 Possible Limitations of the Present Systematic Review

Guidance produced by NICE (2005) and CRD (2008) for conducting systematic reviews recommends that the inclusion and quality assessment of papers is overseen by two or more researchers for at least a sub-sample of the papers. This is to minimise potential selection bias and in the situation where a disagreement arises it would allow for a tightening of the inclusion criteria. Although a second researcher was consulted during the current review when a paper's inclusion was unclear, involving them at stages throughout the process would improve the replicability of the results.

Only selecting English language articles can also result in a selection bias. Within this review four papers were excluded due to language, however on a later review of their abstracts these excluded papers were also found to meet further exclusion criteria.

Including a wider range of publication types and including grey research was not possible in the current review due to time and cost limitations, however this would improve the breadth of the resulting papers and strengthen this review.

The review considers the importance given to the Qol measures within each paper by awarding them a Qol score between 1-5. This rank was designed to categorise the papers and not to infer overall quality. It should be noted that this score was developed in response to the range of resulting papers and may need further development if it was to be applied to a different range of papers. Furthermore, it would require tests on the validity and reliability of this scale, which should include exploring the inter-rater reliability.

5.6 Future

Future research is needed regarding Qol in those with ID. Primarily this needs to focus on resolving the challenges regarding the measurement of Qol, particularly considering the real life application of measures in services, which requires that measures are reliable and valid but also pragmatic. Future research should also consider the limitations of both the measures and their utilisation noted in this review. Qol is a multidimensional concept and as such requires studies to provide a greater level of detail to allow for the consideration of multiple factors including environmental and individual characteristics which at any one time could be influencing Qol.

5.7 Conclusion

Qol remains an area of active research, which Schalock (2004) states that we are still to fully understand or know how to measure this and use it to consistently impact change. The measures were selected as they had been found to report psychometric properties; however both Townsend-White et al.(2012) and Li et al.(2013) recognise that further validation of each of the measures is required.

Qol is particularly important within the ID population as it values the individual, however due to the challenges in understanding and measuring this, which often stem from

communication and cognitive difficulties, research has generally focused on environmental factors. This could be perhaps seen as moving away from the individuality for which Qol is valued (Hatton, 1998). A compromise needs to be found between theoretic underpinning of Qol and the pragmatic needs of services to be able to quantify, measure and document change. Although some suggest that the measurement of Qol is so flawed that it should be discarded altogether (Hensel, 2001), in doing this we risk throwing the baby out with the bath water; in that the drive for person centred care, a focus on improving individuals' sense of satisfaction/well-being and a move away from the medical model of disease and disorder should be supported and encouraged.

References

Banks, P., Jahoda, A., Dagnan, D., Kemp, J., & Williams, V. (Jul 2010). Supported employment for people with intellectual disability: The effects of job breakdown on psychological well-being. *Journal of Applied Research in Intellectual Disabilities, 23*(4), 344-354. doi:http://dx.doi.org/10.1111/j.1468-3148.2009.00541.x

Barber, J., Cooper, B. & Owen, L. (1994). The short-term effects of relocation on the intellectually disabled. *Research on Social Work Practice,* 4(2), 248-258.

Beadle-Brown, J., Murphy, G., & DiTerlizzi, M. (2009). Quality of life for the Camberwell cohort. *Journal of Applied Research in Intellectual Disabilities*, *22*(4), 380-390.

Beyer, S., Brown, T., Akandi, R., & Rapley, M. (May 2010). A comparison of quality of life outcomes for people with intellectual disabilities in supported employment, day services and employment enterprises. *Journal of Applied Research in Intellectual Disabilities*, *23*(3), 290-295. doi:http://dx.doi.org/10.1111/j.1468-3148.2009.00534.x

Blick, R. N., Saad, A. E., Goreczny, A. J., Roman, K., & Sorensen, C. H. (2015). Effects of declared levels of physical activity on quality of life of individuals with intellectual disabilities. *Research in developmental disabilities*, *37*, 223-229.

Bonham, G. S., Basehart, S., Schalock, R. L., Marchand, C. B., Kirchner, N., & Rumenap, J. M. (2004). Consumer-based quality of life assessment: The Maryland ask me! project. *Journal Information*, *42*(5).

Bouras, N., Martin, G., Leese, M., Vanstraelen, M., Holt, G., Thomas, C., . . . Boardman, J. (Sep 2004). Schizophrenia-spectrum psychoses in people with and without intellectual

disability. *Journal of Intellectual Disability Research, 48*(6), 548-555. doi:http://dx.doi.org/10.1111/j.1365-2788.2004.00623.x

Bramston, P., Chipuer, H., & Pretty, G. (Oct 2005). Conceptual principles of quality of life: An empirical exploration. *Journal of Intellectual Disability Research, 49*(10), 728-733. doi:http://dx.doi.org/10.1111/j.1365-2788.2005.00741.x

Brown, C., Hatton, C., & Emerson, E. (2013). Quality of Life Indicators for Individuals With Intellectual Disabilities: Extending Current Practice. *Intellectual and Developmental Disabilities, 51*(5), 316-332.

Buntinx, W. H., & Schalock, R. L. (2010). Models of disability, quality of life, and individualized supports: implications for professional practice in intellectual disability. *Journal of Policy and Practice in Intellectual Disabilities, 7*(4), 283-294.

Centre for Reviews and Dissemination (2008). Systematic Review: CRD's Guidance in undertaking reviews in health care. York: CRD, University of York.

Chou, Y., Pu, C., Kroger, T., Lee, W., & Chang, S. (Sep 2011). Outcomes of a new residential scheme for adults with intellectual disabilities in taiwan: A 2-year follow-up. *Journal of Intellectual Disability Research, 55*(9), 823-831. doi:http://dx.doi.org/10.1111/j.1365-2788.2011.01394.x

Chou, Y., Harman, A. D., Lin, C., Lee, W., Chang, S., & Lin, M. (May-Jun 2011). Outcome evaluation of active support training in taiwan. *Research in Developmental Disabilities, 32*(3), 1130-1136. doi:http://dx.doi.org/10.1016/j.ridd.2011.01.011

Chou, Y., Lin, L., Pu, C., Lee, W., & Chang, S. (Mar 2008). Outcomes and costs of residential services for adults with intellectual disabilities in taiwan: A comparative evaluation. *Journal of Applied Research in Intellectual Disabilities, 21*(2), 114-125. doi:http://dx.doi.org/10.1111/j.1468-3148.2007.00373.x

Cooper B. & Picton, C. (2000). The long term effects of relocation on people with an intellectual disability: quality of life, behaviour, and environment. Research on social work practice. *10*(2), 195-208.

Cummins, R. A. (1997a). Self-rated quality of life scales for people with an intellectual disability: a review. *Journal of Applied Research in Intellectual Disabilities, 10*(3), 199-216.

Cummins, R.A. (1997b) *Comprehensive Quality of Life Scale (5th edn).* Melbourne: Deakin University.

Cummins, R. A. (2002). Proxy responding for subjective well being : a review. *Int Rev Res Ment Retard, 25*, 183-207.

Cummins R. A. & Lau A. L. D. (2005) Personal Wellbeing Index – Intellectual Disability Manual (3rd edn). Melbourne: School of Psychology, Deakin University.

Deeks, J., Dinnes, J., D'Amico, R., Sowden, A., Sakarovitch, C., Song, F., Petticrew, M., Altman, D.G., (2003). International Stroke Trial Intervention Group and European Carotid Surgery Trial Collaborative Group. *Health Technology Assessment, 7* (27), 1-173.

Downs, S. & Black, N., (1998). The feasibility of creating a checklist for the assessment of the methodological quality of both randomised and non-randomised studies of heath care interventions. *Journal of Epidemiological Community Health, 52*, 377-384.

Duvdevany, I. (2008). Do persons with intellectual disability have a social life? the israeli reality. *Salud Publica De Mexico, 50*(Suppl 2), s222-9.

Duvdevany, I., & Arar, E. (2004). Leisure activities, friendships, and quality of life of persons with intellectual disability: foster home vs community residential settings. *International Journal of Rehabilitation Research.* 27(4), 289-296.

Eiser, C., & Morse, R. (2001). Can parents rate their child's health-related quality of life? Results of a systematic review. *Quality of Life Research, 10*(4), 347-357.

Felce, D., & Perry, J. (1995). Quality of life: Its definition and measurement. *Research in developmental disabilities, 16*(1), 51-74.

Felce, D., Perry, J., Romeo, R., Robertson, J., Meek, A., Emerson, E., & Knapp, M. (2008). Outcomes and costs of community living: Semi-independent living and fully staffed group homes. *American Journal on Mental Retardation, 113*(2), 87-101. doi:http://dx.doi.org/10.1352/0895-8017%282008%29113%5B87:OACOCL%5D2.0.CO;2

García-Villamisar, D., Dattilo, J., & Matson, J. L. (May 2013). Quality of life as a mediator between behavioral challenges and autistic traits for adults with intellectual disabilities. *Research in Autism Spectrum Disorders, 7*(5), 624-629. doi:http://dx.doi.org/10.1016/j.rasd.2012.12.009

Harner, C. J., & Heal, L. W. (1993). The Multifaceted Lifestyle Satisfaction Scale (MLSS): Psychometric properties of an interview schedule for assessing personal satisfaction of adults with limited intelligence. *Research in Developmental Disabilities, 14*(3), 221-236.

Hartnett, E., Gallagher, P., Kiernan, G., Poulsen, C., Gilligan, E., & Reynolds, M. (Jun 2008). Day service programmes for people with a severe intellectual disability and quality of life: Parent and staff perspectives. *Journal of Intellectual Disabilities, 12*(2), 153-172. doi:http://dx.doi.org/10.1177/1744629508091340

Hatton C Who's Quality of life is it anyway? Some problems with emerging quality of life consensus. *Mental retardation, 36*(2), 104-115. (1998) subjective measures should be abandoned

Heal, L. W., & Chadsey-Rusch, J. (1985). The Lifestyle Satisfaction Scale (LSS): Assessing individuals' satisfaction with residence, community setting, and associated services. *Applied research in mental retardation, 6*(4), 475-490.

Hematian, K., Alborzi, S., & Khayyer, M. (Dec 2009). Quality of life of iranian vocational students with and without intellectual disability. *Psychological Reports, 105*(3, Pt 1), 738-746. doi:http://dx.doi.org/10.2466/pr0.105.3.738-746

Hensel, E. (2001). Is Satisfaction a Valid Concept in the assessment of Quality of life of people with Intellectual Disabilities? A review of the Literature. *Journal of Applied Recearch in Intellectual Disabilities, 14*, 311-326.

Horovitz, M., Shear, S., Mancini, L. M., & Pellerito, V. M. (Jan 2014). The relationship between axis I psychopathology and quality of life in adults with mild to moderate intellectual disability. *Research in Developmental Disabilities, 35*(1), 137-143. doi:http://dx.doi.org/10.1016/j.ridd.2013.10.014

Keith, K. D., & Bonham, G. S. (2005). The use of quality of life data at the organization and systems level. *Journal of Intellectual disability Research, 49*(10), 799-805.

Keith, K. D., Heal, L. W., & Schalock, R. L. (1996). Cross-cultural measurement of critical quality of life concepts. *Journal of Intellectual and Developmental Disability*, *21*(4), 273-293.

Kober, R., & Eggleton, I. R. C. (Oct 2005). The effect of different types of employment on quality of life. *Journal of Intellectual Disability Research, 49*(10), 756-760. doi:http://dx.doi.org/10.1111/j.1365-2788.2005.00746.x

Koritsas, S., Iacono, T., Hamilton, D., & Leighton, D. (Sep 2008). The effect of active support training on engagement, opportunities for choice, challenging behaviour and support needs. *Journal of Intellectual and Developmental Disability, 33*(3), 247-256. doi:http://dx.doi.org/10.1080/13668250802282944

Kraemer, B. R., McIntyre, L. L., & Blacher, J. (Aug 2003). Quality of life for young adults with mental retardation during transition. *Mental Retardation, 41*(4), 250-262. doi:http://dx.doi.org/10.1352/0047-6765%282003%2941%3C250:QOLFYA%3E2.0.CO;2

Lachapelle, Y., Wehmeyer, M. L., Haelewyck, M. C., Courbois, Y., Keith, K. D., Schalock, R., . . . Walsh, P. N. (Oct 2005). The relationship between quality of life and

self-determination: An international study. *Journal of Intellectual Disability Research, 49*(10), 740-744. doi:http://dx.doi.org/10.1111/j.1365-2788.2005.00743.x

Li, C., Tsoi, E. W. S., Zhang, A. L., Chen, S. & Wang, C. K. J.(2013) Psychometric Properties of Self-reported Quality of life measures for people with Intellectual Disabilites: A Systematic Review. *Journal of Developmental and Physical Disabilities. 25,* 253-270.

Van Loon, J., Van Hove, G., Schalock, R. L., & Claes, C. (2008). Personal outcomes scale. *Middleburg, Holland: Arduin Steichlich.*

Martin, G., Costello, H., Leese, M., Slade, M., Bouras, N., Higgins, S., & Holt, G. (Jul 2005). An exploratory study of assertive community treatment for people with intellectual disability and psychiatric disorders: Conceptual, clinical, and service issues. *Journal of Intellectual Disability Research, 49*(7), 516-524. doi:http://dx.doi.org/10.1111/j.1365-2788.2005.00709.x

McClean, B., Grey, I. M., & McCracken, M. (Sep 2007). An evaluation of positive behavioural support for people with very severe challenging behaviours in community-based settings. *Journal of Intellectual Disabilities, 11*(3), 281-301. doi:http://dx.doi.org/10.1177/1744629507080791

National Institute for Health and Clinical Excellence (2005) Guideline Development Methods Chapter 7 Reviewing and grading the evidence, 7.1-7.8 London: National Institute for Health and Clinical Excellence.

Nota, L., Soresi, S., & Perry, J. (2006). Quality of life in adults with an intellectual disability: the evaluation of quality of life instrument. *Journal of Intellectual Disability Research, 50*(5), 371-385.

Oishi, S., Diener, E. F., Lucas, R. E., & Suh, E. M. (1999). Cross-cultural variations in predictors of life satisfaction: Perspectives from needs and values. *Personality and social psychology bulletin*, *25*(8), 980-990.

Ouellette-Kuntz, H. (1990). A pilot study in the use of the Quality of Life Interview Schedule. *Social Indicators Research, 23*(3), 283-298.

Perry, J., Allen, D. G., Pimm, C., Meek, A., Lowe, K., Groves, S., . . . Felce, D. (Feb 2013). Adults with intellectual disabilities and challenging behaviour: The costs and outcomes of in- and out-of-area placements. *Journal of Intellectual Disability Research, 57*(2), 139-152. doi:http://dx.doi.org/10.1111/j.1365-2788.2012.01558.x

Perry, J., Felce, D., Allen, D., & Meek, A. (Jan 2011). Resettlement outcomes for people with severe challenging behaviour moving from institutional to community living. *Journal of Applied Research in Intellectual Disabilities, 24*(1), 1-17. doi:http://dx.doi.org/10.1111/j.1468-3148.2010.00567.x

Perry, J., & Felce, D. (Mar 2005). Factors associated with outcome in community group homes. *American Journal on Mental Retardation, 110*(2), 121-135. doi:http://dx.doi.org/10.1352/0895-8017%282005%29110%3C121:FAWOIC%3E2.0.CO;2

Petry, K. & Maes, B. (2008). Quality of Life: People with profound intellectual and multiple disabilities. In Pawlyn, J. & Carnaby, S. *Profound Intellectual and Multiple Disabilities: Nursing Complex Needs (pp.15-36).* West Sussex: Wiley-Blackwell.

Reiter, S. (Jun 2001). Autonomy and social skills: A group-based programme with adolescents with learning difficulties for the enhancement of personal autonomy and social skills. *Journal of Research in Special Educational Needs, 1*(2), Sefe. doi:http://dx.doi.org/10.1111/j.1471-3802.2001.00143.x

Riches, V. C., Harman, A. D., Keen, D., Pennell, D., Harley, J. H., & Walker, M. (Sep 2011). Transforming staff practice through active support. *Journal of Intellectual and Developmental Disability, 36*(3), 156-166. doi:http://dx.doi.org/10.3109/13668250.2011.598499

Ruddick, L., & Oliver, C. (2005). The Development of a Health Status Measure for Self-report by People with Intellectual Disabilities. *Journal of Applied Research in Intellectual Disabilities*, *18*(2), 143-150.

Schalock, R. L. (2004). The concept of quality of life: what we know and do not know. *Journal of intellectual disability research*, *48*(3), 203-216.

Schalock, R. L., & Keith, K. D. (1993). *Quality of life questionnaire*. IDS Publishing Corporation.

Schalock R. L. & Verdugo M. (2002). *Handbook on Quality of Life for Human Service Practitioners.* Washington DC : American association on Mental Retardation.

Schalock, R. L., Verdugo, M. A., Jenaro, C., Wang, M., Wehmeyer, M., Jiancheng, X., & Lachapelle, Y. (2005). Cross-Cultural Study of Quality of Life Indicators. *American Journal on Mental Retardation*, *110*(4), 298-311.

Stancliffe, R. J., Harman, A. D., Toogood, S., & McVilly, K. R. (May 2007). Australian implementation and evaluation of active support. *Journal of Applied Research in Intellectual Disabilities, 20*(3), 211-227. doi:http://dx.doi.org/10.1111/j.1468-3148.2006.00319.x

Stancliffe, R. J., & Parmenter, T. R. (1999). The Choice Questionnaire: A scale to assess choices exercised by adults with intellectual disability. *Journal of Intellectual and Developmental Disability, 24*(2), 107-132.

Townsend-White, C., Pham, A. N. T., & Vassos, M. V. (2012). Review: A systematic review of quality of life measures for people with intellectual disabilities and challenging behaviours. *Journal of Intellectual Disability Research, 56*(3), 270-284.

Vine, X. K. L., & Hamilton, D. I. (Sep 2005). Individual characteristics associated with community integration of adults with intellectual disability. *Journal of Intellectual and Developmental Disability, 30*(3), 171-175. doi:http://dx.doi.org/10.1080/13668250500204125

Wheeler, J. R., Clare, I. C., & Holland, A. J. (2013). Offending by people with intellectual disabilities in community settings: A preliminary examination of contextual factors. *Journal of Applied Research in Intellectual Disabilities, 26*(5), 370-383. doi:http://dx.doi.org/10.1111/jar.12040

Willner, P., Rose, J., Jahoda, A., Kroese, B. S., Felce, D., Cohen, D., . . . Hood, K. (2013). Group-based cognitive-behavioural anger management for people with mild to moderate intellectual disabilities: Cluster randomised controlled trial. *The British Journal of Psychiatry, 203*(4), 288-296. doi:http://dx.doi.org/10.1192/bjp.bp.112.124529

Empirical Paper

Challenging Behaviour, Physical Health and Related Quality of Life in People with Rare Genetic Disorders

1 Abstract

Introduction: Individuals with intellectual disabilities (ID) experience significant health problems. For those with severe or profound ID and complex disabilities it is particularly important that more is known about their potential health difficulties to ensure appropriate care levels are provided. Currently little is known about long term health problems in Angelman (AS), Cornelia de Lange (CdLS) or Cri du Chat (CdC) syndromes; three genetic disorders that are associated with ID; or about the relationship between health and challenging behaviour (CB) or health related quality of life (HrQol).

Method: 43 participants with AS, CdLS or CdC were included based on their contrasting level of CB (either persistently high levels or having no/low levels). Proxies completed questionnaires regarding health, HrQol and CB. Variables were explored across all participants and compared between those with persistent CB and those with low/no levels of CB.

Results: Participants had a high prevalence of health problems and low HrQol. HrQol was negatively associated with the severity of current health problems. When differing levels of CB were compared, although no difference in health problems was found, the *Persistent CB* group had significantly lower HrQol. The level of CB and current health accounted for 20% of the variance in HrQol.

Discussion: This is the first study to explore the relationship between health, HrQol and CB in individuals with ID. The results are consistent with the lower HrQol found in those with chronic health problems in the Non–ID population. However, further research is required comparing the magnitude of the impact of health problems on HrQol for those with and without ID. Following this, attention should be given to the effectiveness of

treatment and in turn its impact on HrQol. It is suggested that further factors may be mediating the relationship between health, HrQol and CB, such as mental health, and therefore further exploration of these is also required.

2 Introduction

Individuals with intellectual disabilities (ID) are at greater risk of both physical and mental health difficulties (Emerson & Hatton, 2007; Gillberg & Soderstrom 2003; Hemmings, Deb, Chaplin, Hardy, & Mukherjee, 2013), with recent reports documenting lower life expectancy (Glover & Ayub, 2010). Despite this, individuals with ID experience poor access to health services (Anderson et al.,2013; Krahn, Hammond, & Turner, 2006; Havercamp, Scandlin, & Roth, 2004). This is particularly evident for those with severe and profound ID (Bittles, Petterson, Sullivan, Hussain, Glasson, & Montgomery, 2002; Cooper, Smiley, Finlayson et al., 2007), as their limited communication skills mean they are reliant on others recognising signs of illness. Consequently, more severe levels of ID are associated with poorer rates of recognition and treatment of health problems (May & Kennedy, 2010). A number of governing bodies have highlighted the need for services to tackle these health inequalities both within the UK (Emerson & Baines 2010) and internationally (Bennett, 2009; Harris, 2005).

Of those with ID, it is estimated that genetic causes are present in 25-50% (McLaren & Bryson, 1987), with this proportion rising significantly in recent years. This prevalence increases proportionally with the severity of ID. Over the last few decades, research within the field of ID has moved from considering those with ID as a single population only to focusing on comparing and contrasting individual syndromes. Exploration of different genetic phenotypes provides information on both typical and atypical development. Furthermore, in identifying common areas of difficulty, greater support and adaptations for individuals can be developed.

Three syndromes that are broadly comparable in terms of levels of ID but differ in social behaviour are Angelman (AS), Cri du Chat (CdC) and Cornelia de Lange (CdLS). These syndromes have been compared in previous research which showed that 50% of individuals with these syndromes will have had at least one health problem within the last six months (Berg, Arron, Burbidge, Moss & Oliver, 2007). In addition to these health problems, parents have commented on the lack of medical awareness of these syndromes (Griffith et al., 2011a).

2.1 Health in Angelman Syndrome

Angelman syndrome (AS) is caused by a lack of expression of the UBE3A gene, often due to a microdeletion of the 15q11.2-q13 region on the maternal inherited chromosome (Dan, 2009). Prevalence is estimated at 1:10 000 – 1:40 000 (Buckley, Dinno, & Weber 1998; Clayton-Smith 1993; Kyllerman, 1995). Individuals with AS show a developmental delay, often considered in the severe range (Clarke & Marston, 2000), impairment in speech with no or limited words, disordered movement or balance, usually ataxia of gait or tremulous movement of limbs (Williams et al., 2006). Heightened sociability and increased frequency of smiling is seen in both social and non-social situations (Oliver et al., 2006).

With regard to health, those with AS often have health problems associated with the nervous system (Berg, Arron, Burbidge, Moss & Oliver, 2007; Clayton-Smith & Laan 2003). Dan (Dan & Plec 2008; Dan, 2009) described a developmental perspective of the health of those with AS, starting with early feeding difficulties, which are often aggravated by gastro-oesophageal reflux and for some individuals this can lead to a failure to thrive. Around 90% of children with AS will experience seizures, often starting between the ages of 18 months and 4 years. These seizures may improve in adolescence but can persist into adulthood. Scoliosis can develop in adolescence and later mobility often decreases into

adulthood. The perspective put forward by Dan suggests continual health difficulties across the lifespan, with specific health problems emerging and dissipating at different ages. Dan (2009) commented that most studies were based on retrospective accounts and that further studies with a longitudinal perspective are required to provide more detail. Furthermore, Dan (2009) states that research should be expanded to consider the impact of aging on individuals and consider the quality of life (Qol) of individuals and carers.

2.2 Health in Cornelia De Lange Syndrome

Cornelia de Lange Syndrome (CdLS) is linked to a number of differing genetic causes. For the majority (60%), a deletion in the NIPBL gene on chromosome 5 (locus 5p13) will be present, whilst others will have mutations on the X linked SMC1 gene or the SMC3 gene on chromosome 10 (Deardorff et al. 2012; Krantz et al., 2004). CdLS is estimated to occur in 1:40 000 live births (Barisic et al., 2008; Ireland, 1996). Individuals with CdLS show a developmental delay, typically severe to profound (Berney, 1999), small stature, limb defects (Barisic et al. 2008), excessive hair growth and often display autistic spectrum disorder-like characteristics (Moss et al., 2008). For those whose CdLS is associated with SMC3 and SMC1A mutations (5% of those with CdLS), a milder phenotype is observed with typically a mild developmental delay and less physical abnormalities (Deardorff et al. 2007). Kline et al. (2007) followed 49 individuals with CdLS over the period of six years and suggest that there is some evidence of accelerated aging.

Individuals with CdLS are described as having more frequent and severe health problems than other genetic syndromes (Hall, Arron, Sloneem, & Oliver, 2008). Gastroesophalgeal reflux is one of the most frequently reported health concerns (Kline et al., 2007, Luzzani, Macchini, Valade, Milani & Selicorni, 2003), with Berg et al. (2007) noting that 15% of individuals with CdLS had health problems associated with digestive diseases.

2.3 Health in Cri Du Chat Syndrome

Cri du Chat Syndrome (CdC) is caused by a deletion on the short arm of chromosome 5 (5p15) (Neibhur, 1978; Overhauser et al., 1994). Prevalence is estimated at 1:15 000 – 1: 50 000 (Higurashi et al., 1990; Niebuhr, 1978). Individuals with CdC show a developmental delay, ranging from moderate to profound (Cornish, Bramble, Munir & Pigram, 1999), a round face with low set ears (Niebuhr, 1978), hypersensitivity to sensory stimuli (Cornish & Pilgram, 1996), and impaired growth which is thought to be related to genotype and indirectly with nutrition and feeding problems (Collins & Eaton-Evans, 2001).

There has been less research documenting the health difficulties in CdC. Frequent eye problems, scoliosis and deformities in the gastrointestinal, respiratory and cardiovascular systems have been noted (Cornish & Bramble, 2002; Mainardi, 2006; Wilkins, Brown, Nance, & Wolf, 1983). There are often problems with asphyxia/cyanosis and feeding during the neonatal years and in the first few years of life, respiratory infections were frequent, as were gastroesophageal reflux, vomiting and constipation (Mainardi et al., 2006). Following on from these years, seizures occurred in 15.7%, and 36.8% of children and adolescents had a dental malocclusion (open bite). The signs of gastroesophageal reflux decrease with age (Tunnicliffe, 2010).

2.4 Impacts of health

It is well documented in the typically developing literature that an individual's health and health difficulties impact upon their life, including their mood and perceived Qol. However, less is known about the impacts of health on those with ID.

2.4.1 Quality of Life (Qol) and Health Related Quality of Life (HrQol)

The exploration of Qol has expanded over the last few decades in both ID and non-ID populations. Although there is still much debate, a consensus was sought by Schalock et al. (2002) by asserting that Qol consists of a number of universal core domains, including emotional wellbeing, personal development and physical wellbeing. In terms of the relationship between an individual's health and their Qol within the ID population, it has been demonstrated that health problems have been linked with decreased Qol. For example, Berg et al. 2007 showed an association between the presence of a health problem and low affect in those with severe and profound ID.

A recent development in the field of Qol measurement is a narrowing of focus to what has been termed health related quality of life (HrQol). HrQol specifically considers the impact of health on an individual's Qol. Mcdougall et al. (2014) suggest that the growing importance of Qol and in turn HrQol has brought about a change in the approach to treatment of long term disorders, moving away from a medical model that is focused on cure to a more holistic view. There are many health, psychological and cognitive disorders that are likely to have a lifetime impact with no cure. However, research within the field of HrQol allows consideration of how people can be supported to live well despite long term conditions. Within the non-ID population, individuals with health problems such as gastrointestinal diseases, chronic pain, hypertension and diabetes or poor oral health have lower HrQol than comparable healthy individuals (Naito et al., 2006; Subramaniam et al., 2013; Varni et al. 2015). Within the ID population there is limited published research on HrQol. One exception is research in Down syndrome showing that the HrQol of those with Down syndrome is lower than the normative population (van Gameren-Oosterom et al., 2011). Two other populations considered were those with High functioning autism (Potvin,

Snider, Prelock, Wood-Dauphinee & Kehayia, 2015) and those with Dravet syndrome, a genetic disorder causing epilepsy and associated ID (Brunklaus, Dorris, & Zuberi, 2011. Here research showed that individuals with either of these conditions had lower HrQol than the normative population. These few papers are currently the extent of the research and reflect exploration of the impact of ID on HrQol and not the impact of health problems on the Hrqol of individuals with ID.

2.4.2 Challenging behaviour (CB)

For individuals with ID it is estimated that 10-15% will display CB (Emerson et al. 2001). CB is persistent with poor treatment rates (Taylor, Oliver & Murphy, 2011). Increased levels of CB are associated with increased cost and requiring higher levels of care (Einfeld, 2010; Emerson et al. 2001).

There is broad research that considers the relationship between specific health difficulties and CB (e.g. Bosch, Van Dyke, Smith & Poulton, 1997; Carr, Smith, Giacin, Whelan & Pancari, 2003; Breau, Camfield, Symons, Bodfish, MacKay, Finley, & McGrath, 2003). In a systematic review, De Winter, Jansen and Evenhuis (2011) found that medical conditions could play a role in CB, particularly in urinary incontinence, chronic sleep difficulties and visual impairment and called for further health problems to be investigated. May and Kennedy (2010) also considered the relationship between health and CB from a behavioural perspective, suggesting that health problems can make CB more frequent due to changing the value of reinforcers and so altering behaviour-consequence relations. May and Kennedy (2010) summarise the current research which links allergies, otitis media (middle ear infection/inflammation), dysmenorrhea, constipation and gastroesophageal reflux disease to increases in CB.

Angelman, Cornelia de Lange and Cri du Chat syndromes evidence a high prevalence of CB (Arron, Oliver, Moss, Berg, & Burbidge, 2011; Collins & Cornish, 2002; Kline et al., 2007). Research looking specifically at the link between health and CB within the three syndromes found health problems associated with self-injury and aggression (Tunnicliffe, 2010) and for those with CdLS, self-injurious behaviour (SIB) was associated with gastroesphageal reflux (Luzzani et al., 2003; Tunnicliffe, 2010). Similarly, Hall et al. (2008) found that health problems in those with CdLS were associated with the severity of SIB but not its presence.

2.5 Interim summary

Health disparities between those with and without ID are clearly evident (Anderson et al., 2013). Few studies consider the long term health of those with genetic syndromes such as AS, CdC or CdLS. A link between health difficulties and CB has been proposed (Hall et al. 2008, Tunnicliffe, 2010) and poor health may be associated with poor Qol (Berg, 2007). Although both these links require further exploration, they strengthen the need to explore the development of health problems within this population.

2.6 Study design

The Cerebra Centre for Neurodevelopmental Disorders at the University of Birmingham conducted a study in 2007-2009 with 60 people (20 with each of Angelman, Cornelia de Lange and Cri du Chat syndromes; including both children and adults), all of whom were displaying high levels of CB (more than once a day). Parents and carers completed questionnaires, including those measuring CB and health difficulties (Tunnicliffe, 2010). This study revisits these families and explores changes or developments in health and appraises HrQol. Furthermore, in order to consider how the presence of CB may impact on

health and HrQoL, a contrast group of participants displaying low or no levels of CB will be selected that are similar in age, genetic syndrome and gender. Postal or online questionnaires based on the reporting of proxies will be used.

2.6.1 Aims

The aims of this study are to:

- describe the current health difficulties and HrQol across the three syndromes
- explore the relationship between health problems, chronic CB and HrQol.

3 Method

3.1 Participants

This study aimed to follow up 60 participants described in Tunnicliffe (2010), Griffiths et al. (2011) and Moss et al. (2013). The original method of recruitment for these participants is described in section 3.1.1. Within this study, these participants will be referred to as the *Persistent Challenging Behaviour* (CB) group. A further 60 participants, comparable on a number of relevant variables but with lower levels of or no CB, were selected to form a contrast group. The method of selection and recruitment for this group is described in section 3.1.2. These participants will be referred to as the *Low/No CB* group.

3.1.1 Original selection of participants.

The 60 participants in the *Persistent CB* group were recruited from a database of families of children with genetic syndromes and intellectual disabilities held at the Cerebra Centre at the University of Birmingham. They were originally selected because they were aged between 2 and 19 years, had either Angelman (AS), Cri du chat (CdC) or Cornelia de Lange (CdLS) syndrome and evidenced clinically significant levels of CB (self-injurious behaviour (SIB) and/or physical aggression during the last month with a frequency of at least daily). Due to ethical approval, only those participants who were residents of England or Wales could be contacted for recruitment to the current study.

3.1.2 Selection of contrast (*Low/No CB*) participants.

The *Low/No CB* group of participants for this study were selected from the same participant database based on their level of CB at the time that the *Persistent CB* group were first assessed, and their comparability to participants in that group. As the CB questionnaire (CBQ) does not collect information on the frequency of aggression towards others, *Low/No CB* participants were selected based only on their SIB.

A staged approach to inviting *Low/No CB* participants was taken. Initially all those now aged between 8 and 30 years, who had either AS, CdC or CdLS, were residents of England or Wales and had previously completed a CBQ[1] which reported SIB occurring weekly or less were invited. Due to the limited numbers of CdC participants on the database, the age range was extended to 8-55 for those with CdC. To ensure a sufficient number of participants for the *Low/No CB* group, the intention was to invite 40 participants per syndrome group, however the selection criteria only resulted in 87 participants (34 AS, 20 CdC, 33 CdLS), all of whom were invited to take part in the study.

In order to maximise recruitment into the *low/no CB* group, the inclusion criterion was adjusted so as to include those who had reported the daily presence of SIB with a severity score of seven or less (this represented the lower 50% of possible severity score). This resulted in eight (2 AS, 3 CdC, 3 CdLS) additional participants being identified.

3.1.3 Recruitment.

From the 60 possible *Persistent CB* participants, four had requested no further contact, four were out of area and three from the CdLS group were deceased. Overall, 49 *Persistent CB* participants (17 AS, 18 CdC and 14 CdLS) and 96 *Low/No CB* participants (37 AS, 23 CdC and 36 CdLS) were invited to take part. Of those contacted, 18 participants (36.7%; 8 AS, 7 CdC and 3 CdLS) of the *Persistent CB* group and 29 (30.2%; 10 AS, 8 CdC and 11 CdLS) of the *Low/No CB* group took part in the study. The parent/carer of two participants

[1] As the original participants completed their measures from January 2007 to February 2009, to identify *Low/No CB* group, data were reviewed from questionnaires that were completed closest to this time (resulting in the review of CBQ questionnaires completed between June 2006 and July 2009). If a participant had completed more than one set of measures during this period, those measures taken closest to the original data collection point were used.

(one CdC *Persistent CB* group member and one CdLS *Low/No CB* group member) were not able to complete the telephone interview and so were excluded.

3.1.4 Participant characteristics.

The age range was 8 to 51 years. The ages of two CdC participants in the *Low/No CB* group were significantly greater than other group members and so they were excluded from analysis. This resulted in a sample of 43 participants with an age range of 8 to 32 years. There were 21 (48.8%) male and 22 (51.2%) females in the whole sample. Over half of the sample was described as non-verbal (knowing less than 30 words or signs) and the majority (76.7%) were able to walk unaided. Using the VABS Daily Living Skills Standard Score as an estimate of ability, 34 (79%) participants had a severe ID, six (14%) had moderate ID and three (7%) had mild ID. Most participants lived at home with carers (*n*= 33, 78.6%). Table 2.1 presents the characteristics of both the *Persistent CB* participants (*n*=17) and *Low/No CB* participants (*n*=26).

Table 2.1: Means, SDs, ranges and statistical analyses of demographic characteristic by group

Characteristic	Persistent CB *n*=17	Low/No CB *n*=26	t/ χ^2 (df)	p
Syndrome				
Angelman (n)	8	10		
Cri du Chat (n)	6	6	2.20	
Cornelia de Lange (n)	3	10	(2)	0.332
Gender (% Male)	52.9(*n*=9)	46.2(*n*=12)	0.19 (1)	0.663
Age in Years				
Time 1				
Mean (*SD*)	8.91 (4.14)	15.25 (5.22)	4.21	
Range	2.92-17.92	2.92-23.83	(41)	<0.001
Time 2				
Mean (*SD*)	15.9 (4.05)	22.67 (6.07)	4.04[a]	
Range	9.67-24.17	8.58-32.08	(41)	0.001
Years between Time 1 and Time 2				
Mean (*SD*)	6.99 (0.37)	7.42 (1.19)	1.72 [ab]	
Range	6.17-7.33	5.67-8.92	(31.82)	0.091
VABS				
Daily living skills standard score				
Mean (*SD*)	34.47 (9.79)	28.96 (11.65)	-1.61[a] (41)	0.114
Communication standard score				
Mean (*SD*)	37.82 (10.56)	29.15 (11.80)	-2.45[a] (41)	0.027
Socialization standard score				
Mean (*SD*)	43.24 (10.52)	35.12 (15.23)	-2.07 [b] (40.83)	0.045
Speech (% verbal)	43.8[c] (*n*=7)	34.6 (*n*=9)	0.35 (1)	0.554
Mobility (% mobile)	88.2 (*n*=15)	69.2 (*n*=18)	Fisher's exact	0.269
Accommodation (% Living at home)	94.1 (*n*=16)	68.0[c] (*n*=17)	Fisher's exact	0.060

Note. CB= Challenging Behaviour, VABS= Vineland Adaptive Behavior Scales 2

[a] Where the assumptions of normality were violated, a bootstrap was preformed utilising 1000 repetitions. [b] As the variance of the two groups for this measure were significantly unequal, a t-test for unequal variances was used. [c] Due to missing data the number of participants is reduced by 1.

Statistical analysis showed that the *Persistent CB* group were significantly younger than those in the *Low/No CB* group. Both groups experienced a similar time gap between Time 1 and Time 2 (6.99 compared to 7.42 years). The *Persistent CB* group had higher VABS standard scores for communication ($t(41)= -2.45$, $p= 0.027$) and socialisation ($t(40.83)= -2.07$, $p= 0.045$) than the *Low/No CB* group. The groups did not differ on VABS daily living standard score, gender, syndrome, number of participants considered to be verbal or mobile. The difference between the groups in CB is presented in Table 2.2.

The data in Table 2.2 show that the *Persistent CB* participants show significantly higher levels of CB than the *Low/No CB* group. This difference is seen at both time points.

Table 2.2: The levels of aggression towards others and self-injurious behaviour (SIB) seen in those with persistent challenging behaviour (CB) and those with Low/No CB

Characteristic	Persistent CB *n*=17	Low/No CB *n*=26	Difference
Presence of either SIB or Aggression Frequency (%)			
Time 1	17(100)	17(65.4)	Fisher Exact p=0.007
Time 2	17(100)	15(57.7)	Fisher Exact p=0.001
SIB and Aggression Severity Mean (*SD*)			
Time 2	25.71(14.26)	9.46(9.73)	$t^a= -4.45$ p>0.001

[a] As the variance of the two groups for this measure were significantly unequal, a t-test for unequal variances was used.

3.2 Consent

Where the participants were over 16, consent was sought from both the participants themselves and their parent/carer. If the participant was under 16 then their assent by proxy was sought. Due to the possible severity of the participant's ID, parents/carers were asked

to consider the participant's capacity to understand and consent/assent to the study. They were provided with the contact details of the research team who could provide advice and support with this decision, if needed. If the participant was over 16 and assessed to be unable to consent for themselves then a personal or a nominated consultee was approached to gain consent for the individual's participation in this research.

3.3 Ethical approval.

This study was approved by Birmingham University and NHS England via the Coventry and Warwick NRES committee (REC Reference 14/WM/0068, Protocol RG_14-009, IRAS Project ID 125728) (Appendix E).

3.4 Procedure

Time one data were collected in one of two ways, depending upon the participant group. For the *Persistent CB* group, parents or carers of each participant had completed the Challenging Behaviour Interview (CBI) and Health Questionnaire as part of Tunnicliffe's study (2010). For the *Low/No CB* group, parents or carers of each participant had completed the CBQ and Health Questionnaire or as part of large-scale, longitudinal data collection (e.g. Arron, Oliver, Berg, Moss & Burbidge, 2011; Burbidge et al., 2010).

In the current study, the parent/carer of the participant completed a questionnaire via post or online (see section 3.5), estimated to take between 20-30 minutes to complete. Following completion of the questionnaire, parents/carers completed a telephone interview containing the Vineland Adaptive Behavior Scales 2 (VABS) interview and the Challenging Behaviour Interview.

3.5 Measures

3.5.1 General measures.

Demographic Questionnaire (Questionnaire pack, Appendix F).

This collected basic information regarding age, gender, abilities (sensory, motor and verbal) and diagnostic status.

Vineland Adaptive Behavior Scales 2 (VABS; Sparrow, Balla & Ciccetti, 2005, Appendix G).

This semi-structured interview assessed the participant's personal and social adaptive behaviour levels and level of ID. The VABS has been used in both individuals with or without ID, and comprises 261 items divided into four domains (communication, daily living skills, socialisation and motor skills). Each domain is then divided into three sub-domains. Internal consistency ranges from 0.83-0.94 for each of the domains and 0.69-0.89 for the sub-domains. Test-retest reliability is also reported as high, and correlations between raters ranges from 0.80 to 0.95.

Challenging Behaviour Interview (CBI; Oliver, McClintock, Smith, Hall, Dagnan & Stenfert Kroese, 2003, Appendix G).

This was assessed the presentation and severity of CB. Part one identified the possible different types of CB present in the last month. Part two gathered further information regarding characteristics of each behaviour identified, such as frequency, duration, response necessary to manage the behaviour and the effects of the behaviour on others. Items in part two were combined to obtain a total severity score for SIB and aggression towards others. The CBI reports a test-retest agreement of 0.91 and inter–rater agreement of 0.86 for SIB. For aggression, 0.86 and 0.62 is reported respectively. If more than one

example of a certain type of behaviour was reported, the highest severity score was recorded. To gain a CB estimate for each participant, the severity score for SIB and aggression were then combined. To allow comparison between Time 1 and Time 2, equivalent CBQ scores for SIB severity and presence of aggression towards others were calculated. The equivalent SIB severity score combined scores for SIB frequency (question 1), duration (question 2) and use of physical restraint (question 13). If more than one example of SIB was recorded, the behaviour with the highest frequency plus duration was selected. If any aggression towards others was reported, this was noted as present.

3.5.2 Health measures.

Health Questionnaire (HQ, Hall, Arron, Sloneem and Oliver, 2008, Appendix F). This measures the presence and severity of 15 health problems and asks about current and lifetime health problems. Only information pertaining to current health problems was utilised here. The parents/carers scored the health problems that the participant has experienced in the last month on a 0 (never occurred) to 3 (is a severe problem) scale. By combining the severity scores of the 15 health problems, the questionnaire produces an overall severity score for current health. A total number of current health problems is also produced. To provide an estimate of the level of chronic health problems, a tally of the current health problems identified as present at both Time 1 and Time 2 was made. This provided information on the type of chronic problem as well as a total number of chronic health problems per participant. Inter-rater reliability for the occurrence of health problems over the last month is reported as a mean Kappa coefficient of 0.76. Intra-class correlations were found for the current severity score (0.65) and current number of health problems (0.73). This measure has been used with the parents/carers of individuals with an ID in studies by Hall, Arron, Sloneem and Oliver, (2008) and Cianfaglione et al. (2015).

3.5.3 Health related Qol measure.

Pediatric Quality of Life Inventory 4.0 (PedsQl, Varni, Seid, & Rode, 1999; Questionnaire pack, Appendix F).

This questionnaire has versions for ages 5-7, 8-12, 13-18, 18-25 and adult; and parent (proxy) reports for each of these age groups. Each questionnaire, although worded differently to aid understanding or to match life stage (e.g. attend school or work), measures the same sets of constructs (physical, emotional, social and school/work) and is comparable across ages. It consists of 23 items designed to cover the core health dimensions identified by the World Health Organisation (WHO) and is estimated to take under five minutes to complete. The PedsQL consists of four subscales (physical, emotional, social and school/work) which when scored provides a total scale score that can be split into two summary scores (physical and psychosocial). All scores are out of 100; where 100 represents a higher standard of health-related quality of life.

The validity and reliability within the general population is reported by Varni, Seid and Kurtin (2001), which reports good internal consistency based on parental reports for the total scale ($\alpha = 0.90$), physical summary score ($\alpha = 0.88$) and psychosocial summary score ($\alpha = 0.86$). Varni, Seid and Kurtin (2001) recommend that the subscale scores only be used for descriptive or exploratory analyses, as the total and summary scores were found to have stronger psychometric properties (i.e. higher reliability and validity). Its use within an ID population has been reported by Golubović and Škrbić (2013) with adolescents experiencing mild to moderate ID. They reported an acceptable level ($\kappa = 0.43$) of agreement in the reports of parents and adolescents with ID.

Due to the level of ID of the participants, only proxy reports were collected.

3.6 Design

This study utilised a longitudinal, group contrast design. Measures of health and CB were taken at Time 1 and were repeated with the addition of a measure of HrQol at Time 2. The study's design allowed for analysis both within and between groups as well cross-sectional explorations. Variables were explored within the total sample and contrasted for the *Persistent CB* and *Low/No CB* groups.

3.7 Data Analysis

A number of variables were found to be significantly skewed, showed kurtosis and/or were non-normally distributed (Appendix H). When one or more variables violated the assumptions of normality, either a bootstrapping method (with 100 repetitions) was used to allow continued parametric analysis, or a suitable non-parametric alternative was used.

T-tests were used to compare health at Time 1 and health at Time 2. A series of comparisons and correlations assessed whether health and HrQol variables had a relationship to or differed with age, gender, syndrome or ability. Finally, Z scores were used to compare HrQol with population norms.

To explore the relationship between current health difficulties and HrQol, Spearmans Rho correlations were used. Health and HrQol between the groups was compared using t-tests. Correlations seen in those with *Persistent CB* and those with *Low/No CB* between Health and HrQol were compared.

Finally, in order to identify the extent to which variables measured can predict HrQol, a regression model was used The criterion variable of PedsQL total score and the predictor variables were those found to be associated with HrQol.

The CBI total SIB and aggression severity score was chosen to represent level of CB as this provided a more detailed continuous variable which was better suited to a multiple regression than a binary variable stating the group membership (either *Persistent CB* or *Low/No CB*). Checks were performed which showed no cause for concern with correlated errors (Durbin-Watson), multcollinearity or heteroscedasity. Furthermore, Cooks D was used to explore the standardised residuals and identify any large residual which was having a significant impact on the regression. This found no residuals of this nature. As such the assumptions on which a multiple regression is based were met and so analysis was conducted.

4. Results

4.1 Health

In order to address the first aim of describing carer reported health problems in this population, four elements of health problems are described; the number, severity, type and chronicity. Two participants (one low/no CB participant with CdC and one persistent CB experimental participant with AS) did not complete this questionnaire at Time 1, so reducing the number of participants to 41 at this time point.

At Time 1, the number of current health problems per participant ranged from 0 to 10 (mean= 2.34, *SD*= 2.00) with 85.4% (*n*=35) of participants having at least one current health problem (see Appendix I, Table I.1). The severity score of those reporting a current health difficulty (*n*=35) ranged from 1-18 with a mean of 4.34 (*SD*= 3.98) (see Appendix I, Table I.2). The most common health problems at Time 1 were dental and skin problems, with each affecting 39% (*n*=16) of participants.

A similar pattern was seen at Time 2 where the number of current health problems reported ranged from 0-6 (mean= 2.16), with 83.7% (*n*=36) of participants reporting at least one current health problem and the severity score ranged from 1-8 with a mean of 3.78 (*SD*= 2.20). The most common health problems at Time 2 were dental (46.3%, *n*=19) and skin problems (37.2%, *n*=16). No significant difference was found for health problems between Time 1 and Time 2 (number: $t(40)=0.34$, $p=0.739$; severity: $t(40)= 0.72$, $p=0.478$).

A total of 53 chronic health problems were reported. The number of chronic health problems reported per individual ranged from 0 – 5, with 63.4% (*n*= 26) of the group reporting at least one, and 38.9% (*n*=16) reporting two or more chronic problems.

A series of comparisons and correlations were conducted to consider whether the number, severity or chronicity of health problems had a relationship with or differed with age, gender, syndrome or ability. No difference or relationship was found (see Appendix I, Tables I.3 - I.12).

In summary, the majority of the total sample has at least one current health problem. Of the current health problems reported, at least one could be considered chronic in the majority of participants. Dental and skin problems were the most frequently reported health problems.

4.2 Health Related Quality of Life (HrQol)

In order to address the second aim of describing the level of health related quality of life of the total sample, the results of the PedsQL were explored. Table 2.3 presents the total score, summary scores and subscales for all participants and then split by gender and syndrome.

Table 2.3: The means (SD) of the total, summary and subscales of the Paediatric Quality of Life Inventory (PedsQL) for the whole sample and split by gender and syndrome.

	All Participants		Demographic				
			Gender		Syndrome		
PedsQL	*n*=43	Range	Male	Female	AS	CdC	CdLS
	Mean *(SD)*		*n*=21	*n*=22	*n*=18	*n*=12	*n*=13
Total Score	51.77 (16.54)	22.83-100	48.88 (11.68)	54.54 (20.01)	47.89 (16.39)	54.98 (19.93)	54.19 (13.16)
Psychosocial health Summary	59.55 (14.86)	31.67-100	55.74 (12.60)	63.18 (16.19)	57.13 (15.14)	63.20 (16.60)	59.53 (13.17)
Physical health Summary [a]	37.35 (28.61)	20-100	36.31 (24.69)	38.35 (32.46)	30.56 (24.12)	39.58 (30.80)	44.71 (32.21)
Social	57.67 (26.22)	0-100	51.67 (28.52)	63.41 (23.01)	57.22 (27.13)	61.25 (28.37)	55.00 (24.58)
Emotional	59.42 (18.26)	20-100	55.95 (15.30)	62.73 (20.51)	60.28 (16.93)	63.33 (19.35)	54.62 (19.41)
School/Work	61.32 (24.82)	0-100	59.13 (22.55)	63.41 (27.18)	53.89 (25.64)	65.00 (22.46)	68.21 (24.81)

Note. AS= Angelman, CdC= Cri du Chat, CdLS= Cornelia de Lange.
[a] The Physical score is both a summary score and a subscale score

The data in Table 2.3 show that the mean participant score was 37.35 on the physical health summary score and 59.55 on the psychosocial health summary score. Participants showed a range of scores from the floor to the ceiling of the assessment (0 to 100). No significant differences between gender or syndrome was found, nor was a relationship between PedsQL total score or summary scores and age or ability found when correlations were conducted (see Appendix I, Table I.13).

In order to consider how the sample's HrQol compared to known populations, a number of Z-scores were calculated (Table 2.4). These scores were based on the population norms for

the PedsQL completed by proxies for healthy, acutely ill and chronically ill children between 2-18 years old produced by Varni, Seid and Kurtin (2001) (Appendix J). There are no norms available for those with ID.

Table 2.4: The Z-score created when comparing the whole sample's total and summary score on the Paediatric Quality of Life Inventory (PedsQL) to those reported by Varni, Seid and Kurtin (2001)

PedsQL	Full group compared to		
	Healthy	Acutely ill	Chronically ill
Total Score	-19.06	-12.31	-8.00
Psychosocial health summary score	-13.86	-8.46	-5.51
Physical health summary score	-20.84	-14.25	-8.72

The data in Table 2.4 show that all the Z-scores are substantially lower than - 1.96, suggesting that total sample is significantly different from the normative and clinical populations on both domains.

In summary, participants had a mean PedsQL total score of 51.77 which is significantly below that reported for chronically ill children. The PedsQL scores in the current sample were not related to gender, age, syndrome or ability.

4.3 Health and HrQol

In order to address the third aim and examine the relationship between Health and HrQol, a series of correlations were conducted (Table 2.5; Time 2 data only).

Table 2.5: Non-parametric correlational coefficients between results from the Health Questionnaire and the Paediatric Quality of Life Inventory (PedsQL) at Time 2

Health Questionnaire	PedsQL					
	Total		Physical		Psychosocial	
	r_s	*p*	r_s	*p*	r_s	*p*
No of health problems	-0.32	.035*	-0.32	.026*	-0.21	.172
Total Severity Score	-0.33	.029*	-0.34	.026*	-0.21	.181
No of chronic health problems[a]	-0.39	.012*	-0.43	.005**	-0.19	.234

[a] *For No of chronic health problems n is reduced to 41*

*p<0.05. **p<0.01.

The data in Table 2.5 show that the PedsQL total and physical summary scores were significantly negatively correlated with number, severity and chronicity of health problems. An higher number or severity of health problems is associated with lower total and physical quality of life scores on the PedsQL. This relationship was strongest between the physical summary score and the chronicity score (r_s= -0.43 *df*=39 p=0.005) suggesting that those with a greater number of chronic health problems had lower physical functioning scores. This negative directional relationship was also seen between the psychosocial summary score and the health variables (number, severity and chronicity), however these did not reach significance.

In summary, individuals with a greater number of current health problems, greater total severity score and a higher number of chronic health problems are likely to have lower HrQol as reflected by a lower PedsQL total score and physical functioning score. The psychosocial summary score was not significantly related to the health of the participants.

4.4 Challenging Behaviour (CB) and Health

In order to evaluate differences in health seen between those with *Persistent Challenging Behaviour* (CB) and those with *Low/No CB*, groups were compared on the health variables of the number, severity and chronicity of health problems (Table 2.6).

Table 2.6: The number, severity and chronicity of health problems reported on the Health Questionnaire compared between those with Persistent and Low/No levels of challenging behaviour (CB)

		Persistent CB	Low/No CB	Difference	
Variable	N (Persistent, Low)	Mean (*SD*)	Mean (*SD*)	*t*	*p*
No of health problems					
T1	41 (25, 16)	2.50 (1.59)	2.24 (2.24)	-0.40[a]	.677
T2	43 (26, 17)	2.41 (1.70)	2.00 (1.50)	-0.84	.408
Severity					
T1	41 (25, 16)	3.63 (2.58)	3.76 (4.73)	0.10[a]	.894
T2	43 (26, 17)	3.53 (2.48)	2.92 (2.45)	-0.79	.434
Chronicity	41 (25, 16)	1.68 (1.58)	1.04 (1.06)	-1.44[b]	.162

[a] Where the assumptions of normality were violated, a bootstrap was performed utilising 1000 repetitions [b] As the variance of the two groups were significantly unequal a t-test for unequal variances was used.

The data in Table 2.6 show that no significant difference was found between those with *Persistent CB* and those with *Low/No CB* for current health problems, severity of current health problems or chronicity of health problems.

The most common type of health problem reported in both the *Persistent CB* and the *Low/No CB* group was dental problems (see Appendix I, Table I.14 for further details). The possible differences between the two groups and the specific health problems could

not be explored further statistically due to the low frequency of each individual health problem.

In summary, there was no significant difference in the number, severity, chronicity or type of health problems between those with *Persistent CB* and those with *Low/No CB.*

4.5 Challenging Behaviour and HrQol

In order to address the aim of describing differences in HrQol between those with *Persistent CB* and those with *Low/No CB,* groups were compared on the total and summary scores of the PedsQL (Table 2.7).

Table 2.7
The difference in the total and summary scores of the Paediatric Quality of Life Inventory (PedsQL) between those with Persistent and Low/No challenging behaviour (CB)

PedsQL	Persistent CB n=17 Mean (*SD*)	Low/No CB n=26 Mean (*SD*)	Difference
Total score	44.02 (12.19)	56.84 (17.23)	*t*= 2.66[a] *p*=.017*
Summary scores			
Psychosocial	54.53 (14.02)	62.82 (14.73)	*t*=1.84 *p*=.073
Physical	24.63 (16.61)	45.67 (31.88)	*t*=2.83[b] *p*=.007**

[a] Where the assumptions of normality were violated, a bootstrap was performed utilising 1000 repetitions [b] As the variance of the two groups were significantly unequal a t-test for unequal variances was used.
*p<0.05. **p<0.01.

The data in Table 2.7 show that there was a significant difference between the PedsQL total for those with *Persistent CB* and those with *Low/No CB*, with those in the *Persistent CB* group having a significantly lower total score. The *Persistent CB* group also showed

significantly lower scores on the physical functioning summary score only (*t* (39.44)= 2.50 *p*=0.007).

4.6 Challenging Behaviour, Health and HrQol

In order to address the final aim of exploring the relationship between health, HrQol and CB, two approaches were taken. Firstly, the relationship between health and HrQol was explored within those with *Persistent CB* and then for those with *Low/No CB*. The variables of health and CB were then explored as possible predictors of HrQoL.

Correlations between health (number, severity and chronicity) and the HrQol (PedsQL total and summary scores) were conducted for those with *Persistent CB* and then for those with *Low/No CB* separately (Table 2.8).

Table 2.8
The relationship between health at Time 2 and health related quality of life as seen in participants with Persistent and Low/No levels of challenging behaviour (CB)

Health	PedsQL					
	Total		Physical		Psychosocial	
	r_s	P	*r*	p	*r*	p
Low/No CB n=26						
No of health problems	-0.37[a]	.0.62	-0.40	.041*	-0.27	.189
Total Severity Score	-0.40 [a]	.044*	-0.43	.030*	-0.21	.295
No of chronic health problems[a]	-0.32 [a]	.118	-0.46	.019*	-0.10	.618
Persistent CB n=17						
No of health problems	-0.13	.626	-0.13	.630	-0.12	.635
Total Severity Score	-0.10	.708	0.01	.985	-0.07	.790
No of chronic health problems[a]	-0.30	.263	-0.26	.316	-0.14	.598

[a] *For No of chronic health problems n was reduced to 41(Low/No, n=25; Persistent, n=16).*
*p<0.05.

With regard to the whole sample (section 4.3), those with greater numbers of health problems, greater total severity score and a higher number of chronic health problems had reportedly lower HrQol, as reflected by a lower PedsQL total score and physical functioning summary score.

The data presented in Table 2.8 show a different pattern of relationships between the *Persistent CB* and Low/No CB groups. In the *Low/No CB* group the pattern is similar to that seen in the overall sample; a greater severity of current health problems correlated with a lower total PedsQL score (r= -0.40 df=24 p= 0.044). Also, the higher the number

and severity of current health problems and the higher the number of chronic health problems reported, the lower the physical functioning summary score (*r*= -0.40 *df*=24 p= 0.041; *r*= -0.43 *df*=24 p= 0.030; *r*= -0.46 *df*=24 p= 0.019; respectively). There was no correlation between the health variables and the psychosocial summary score in either *Low/No* or *Persistent CB* groups.

The second approach to explore the relationship between health, HrQol and CB was to conduct a regression analysis to identify what proportion of HrQol could be predicted based on participants' levels of health and CB. A multiple regression (enter method) was conducted with the variables of CB (represented by total SIB and Aggression score on the CBI), level of current health problems (represented the total current severity score of the Health Questionnaire) and number of chronic health problems (as measured by a health problem being reported as currently present on the Health Questionnaire at both Time 1 and Time 2). A bootstrap was performed utilising 1000 repetitions to increase the power of the multiple regression due to small participant numbers. Two participants were excluded from this analysis as they had not completed the Health Questionnaire at Time 1 and so could not contribute a chronicity score, meaning the multiple regression is based on the scores of 41 participants.

Together these variables explained 20.8% of the variance in total PedsQL ($F(3,37) = 3.25$, p=0.033). The data presented in Table 2.9 show the variable of CB significantly contributed to the regression equation (p=0.045) as did the number of chronic health problems (p=0.038). The remaining variable of current level of health problems made non-significant contributions to the equation (p=0.873).

Table 2.9
Summary of multiple regression analysis of variables predicting Total PedsQL score (N=41)

	Beta	t	*p*
Challenging Behaviour Interview			
Total self-injurious behaviour and aggression	-0.31	-2.07	0.045*
Health Questionnaire			
Total current severity score	-0.03	0.15	0.873
No of chronic health problems	-0.31	-1.43	0.038*

*p<0.05.

In summary, the relationship between HrQol and health differed between participants with *Persistent CB* and those with *Low/No CB*. Within the Low/No CB group only, a greater number and severity of health problems as well as a greater number of chronic health problems were associated with lower physical summary scores on the PedsQL. Also, the higher the total severity scores amongst the low/no CB individuals, the lower their total PedsQL score. The level of health problems experienced by those with *Persistent CB* did not correlate with their HrQol as measured by the PedsQL. Furthermore, following a multiple regression in a model which combines the variables of CB, level of current health problems and number of chronic health problems together, could account for one fifth of the variance in HrQol. The level of CB and number of chronic health problems make a significant contribution to this model.

5. Discussion

5.1 Overview

This study explored the health, health related quality of life (HrQol) and the relationship between these variables in participants with Angelman (AS), Cri du Chat (CdC) and Cornelia de Lange (CdLS) syndromes. Furthermore, the study contrasted individuals with persistent clinical levels of challenging behaviour (CB) with individuals with either low levels or no CB. AS, CdC and CdLS are associated with a higher prevalence of health difficulties (Berg, Arron, Burbidge, Moss & Oliver, 2007) however this is the first study to consider HrQol within these syndromes. By utilising a longitudinal design, the study was able to make inferences on the longevity of an individual's health problems and CB. This study found a high prevalence of health problems and low HrQol. HrQol was associated with the severity and number of current health problems. When those with persistent levels of CB and those with low or no levels of CB were compared, although no difference in health problems were found, the *Persistent CB* group reported significantly lower HrQol. Finally, in considering the relationship between health, CB and HrQol, the level of CB and current health accounted for 20% of the variance in HrQol.

5.2 Health

Health problems were frequent in this population, with the majority (over 80%) having at least one current health problem as reported by carers. This is broadly consistent with the current research which states that health problems are more frequent in those with with severe and profound ID (Gillberg & Soderstrom, 2003; Jansen, Kroi, Groothoff & Post, 2004).

Previous studies documenting health difficulties in AS, CdC and CdLS specifically, noted that 50% will have experienced one or more health problems in the last six months (Berg, Arron, Burbidge, Moss, & Oliver, 2007), which is below the level of 80% as reported by the current sample. Although both studies relied on carers' reporting of health difficulties, Berg et al. used ICD10 definitions of health problems and acknowledged that this did not account for ongoing problems such as epilepsy. As epilepsy was one of the top five health problems in the current sample and is known to be prevalent in those with ID in general (Gillberg & Soderstrom, 2003) and those with AS specifically (Pelc, Boyd, Cheron & Dan 2007), this may account for the difference found.

In the current study, dental and skin problems were the most frequently reported health problems. This is consistent with the increased prevalence of skin problems (McDermott, Platt & Krishnaswami, 1997; Whitfield, Langan & Russell, 1996) and dental problems (Traci, Seekins, Szali-Petree, & Ravesloot, 2002) in those with ID. When looking at the health problems across AS, CdC and CdLS, Berg et al. found the top three health problems were related to the digestive, respiratory and nervous systems. Although not found to be the most common health problems, digestive and nervous system problems were in the top five problems seen within the group but respiratory problems were infrequent. As previously discussed, Berg et al. took a different approach to classifying health problems which may account for some of these differences. Additionally, Berg et al. considered health problems occurring over the last six months, whereas this study focused on only health problems occurring in the last month.

Many syndromes have elevated levels of specific health problems. For example, Hall et al. (2008) found the most common current health difficulties for those with CdLS were dental

and gastrointestinal problems. This seems consistent with the current sample of CdLS participants. Unfortunately however, due to the low participant numbers, it was not possible to explore statistically the differences in the health problems seen in each syndrome. With differing syndromes possibly showing different health problems it may be that specific health problems are more frequent in each of the three syndromes recruited, but as the frequencies were only explored within the total sample, any syndrome-specific prevalence is not identifiable.

Over half of all participants (63.4%) reported at least one chronic health problem. In this study a health problem was considered chronic if it was reported both as currently present and present previously (approximately seven years ago). This allowed for a wider concept of chronic health problems than just considering problems such as diabetes that are chronic by definition. Presence of a chronic health problem under this definition may reflect either a continuous health problem or frequently recurring problems.

One limitation of the health variables of this study is that they were collected via parent/carer reports and as such, rely on their recall of health problems. This method could have been strengthened by combining parent/carer reports with medical records (e.g. Kapell, Nightingale, Rodriguez, Lee, Zigman, & Schupf, 1998), although it may be that medical records do not contain data on certain conditions that do not require specific medical treatment, such as dental problems.

One limitation to both of these data collection methods is that they rely on the carer awareness, recognition and their accurate reporting, and in turn the recognition of medical staff. This is particularly challenging in this population where even if individuals are considered verbal, there may still be difficulties with communicating and reporting health

difficulties. An example of this is among the pain literature which states that pain is under recognised and under treated particularly for individuals who are not able to communicate their pain to others (McGuire, Daly & Smyth, 2010). These limitations are common to studies focused on health in this population.

A further limitation is the construct validity of the severity score on the Health Questionnaire. Here carers are asked to rate severity of each health problem as mild (score of 1), moderate (score of 2) or severe (score of 3). This method of measurement represents an interval measurement and as such assumes these are interchangeable (e.g. three mild problems would have the same combined severity as one severe problem). Also, although it is reported to have a strong inter-rater reliability (Kappa coefficient = 0.76), severity is a subjective measurement and carers' interpretation of what constitutes each severity score may vary. In the current study, this measure is used to provide an estimate of the combined impact of health problems and despite the limitations discussed, when combined with the objective measure of the number of health problems it provides valuable information.

5.3 Health Related Quality of Life (HrQol)

HrQoL was measured by the PedsQL. This showed that participants had a mean PedsQL total score of 51.77. Although the measurement of Qol has attracted a considerable amount of interest within ID research, HrQol has not been considered to the same extent. One paper that utilised the PedsQL for people with ID (Golubović & Škrbić, 2013) reported from a group of 67 adolescents with ID a mean total score of 66.99. This is higher than the mean found in the current sample. One reason for this difference could be the difference in the level of ID of the groups, with Golubović et al. mainly describing participants as having a mild ID of mixed aetiology compared to the severe ID of the majority of the

participants in the current sample. As well as focusing on a smaller age range of participants (13-18), Golubović et al. did not report the current health of the adolescents. It may be that the current sample not only has a higher level of ID but also has greater number and/or severity of health problem than Golubović et al.'s sample, both of which could contribute to a lower total HrQol score. Varni, Seid and Kurtin (2001) published population norms based on parental reporting on the PedsQL for children without ID aged between 2-18 who were classed as either healthy, acutely ill or chronically ill. Varni et al. stated that those who were chronically ill had the lowest total score (mean= 74.22) in the non-ID population. This was shown to be significantly higher than the mean found in the current sample of individuals with ID, which is consistent with the lower HrQol scores found in those with Down syndrome compared to those without ID (van Gameren-Oosterom et al., 2011).

In this study HrQol was based on carers' reports. This is a common approach in researching the Qol of individuals with ID however it does raise questions about validity. There is often a lack of correlation between Qol measured by parents and that measured by individuals (Cummins, 2002), which is also seen within the measurement of HrQol (Potvin, Snider, Prelock, Wood-Dauphinee and Kehayia, 2015). However, Golubović and Škrbić (2013) reported an acceptable level of agreement between adolescences with mild to moderate ID and their parents on the PedsQL total score.

Although the evidence of correlation between proxy and individuals' reporting of HrQol is varied, due to the level of ID in the current sample, HrQol measurement was only possible via proxies. As such, it can be thought of as providing an estimation of the individual's HrQol bearing in mind the limitations discussed. It also provides a method of directly

comparing the HrQOL of this sample to that of normative data for individuals without ID, as these were also collected by proxy methods.

The scores on the PedsQoL provide a quantitative measurement of each carer's perspective of the individual's HrQol. Understanding the carer's perspective on the individual's HrQol, is particularly important with individuals with greater severity of ID, as research shows that carers and medical staff's judgments on an individual's Qol may influence the choices they make regarding medical treatment decisions (Bekkema, de Veer, Wagemans, Hertogh, & Francke, 2014; Wagemans et al. 2013).

A possible limitation of the HrQol variables is that although the PedsQL is a well-regarded measurement of HrQol in children under 18, the young adult 18-25 and adult versions (>25) are new additions to the scale and have limited published reliability and validity. There is also a lack of published norms for the ID population. Due to this, the HrQol could only be compared against the population norms of those aged 2-18 and it is possible that differing levels of HrQol are found in the adult population. For example, Varni & Limbers (2009) found that self-reported HrQol on the PedsQL for young adult were lower than for those younger adolescents, however no proxy norms were produced. Interestingly, no relationship between age and HrQol was found in the current study.

An element that was not considered in the current study was family Qol and wellbeing. Qol and in turn HrQoL is not independent of the environment and so should be thought of holistically. The Qol of the individual may be influenced by the Qol of the family (McIntyre, Kraemer, Blacher, & Simmerman, 2004; Seltzer & Krauss, 2000). Within AS, CdC and CdLS there is currently no research which has explored this relationship, however

parents of children with these syndromes have been found to high levels of anxiety and low mood (Griffith et al., 2011b).

5.4 Health and Health Related Quality of Life

Individuals with poorer health (i.e. a greater number of health problems, greater total severity score and/or a higher number of chronic health problems) are likely to have lower HrQol. This is consistent with research in the non-ID population which suggests that the presence of a health problem such as gastrointestinal conditions, epilepsy and diabetes leads to lower HrQol (Subramaniam et al., 2013; Varni et al. 2015; Varni, Limbers & Burwinkle, 2007). This is also seen in those with skin and dental problems (Hong, Koo & Koo, 2008; Jankovic et al., 2010; Sischo, & Broder, 2011). The psychosocial summary score of the PedsQL in isolation did not show any significant relationship with the level of health problems of the participants. This result contrasts with Berg et al. (2007) who found those with a health problem had significantly lower affect than those who did not have a health problem. However the questionnaire used by Berg et al. (the mood, interest and pleasure, short form, MIPQ-S) and the PedsQL psychosocial summary score differ in a number of ways. The PedsQL psychosocial summary score combines affect with social and school/work functioning, whereas the MIPQ-S consists of 12 questions focused on affect as estimated by parents/carers reports of observable associated behaviour such as the frequency of smiling. It may be that the wider concept of the psychosocial summary score is not sensitive enough identify changes in behavioural expressions of affect that may be associated with health problems over those associated with ID.

5.5 Health and CB

No difference was found in the level of health problems (in number, severity, chronicity or type of problem) of individuals when comparing those with persistent clinical levels of CB and those with low levels or no CB. This is contrary to previous studies which have linked the onset or frequency of CB with health problems such as pain (Breau et al., 2003), skin problems (Hall et al. 2008), dental problems (Sischo & Broder., 2011), epilepsy (Sabaz, Cairns, Lawson, Bleasel & Bye, 2001) gastroesophageal reflux and constipation (Bosch, Van Dyke, Smith, & Poulton, 1997; May & Kennedy 2010).

This is not the first study to find a lack of relationship between health problems and challenging behaviour. The results are consistent with Emerson et al (2001) who found no difference in the levels of epilepsy between those classed as having more or less demanding levels of CB and Hall et al. (2008) who found that the presence of a health problem did not make it either more or less likely that individuals with CdLS displayed self-injurious behaviour (SIB). It may be that just considering "health problems" as a broad concept is not sensitive enough to pick up differences within this population. Future studies with larger samples may wish to consider factors that may be mediators or moderators to this relationship, such as acute and chronic, painful and non-painful, controlled and uncontrolled health conditions.

Due to the study design and limited sample, CB was treated as a grouping variable. This is an acceptable approach to comparing levels of CB however it disregards the topography and intensity of CB seen within the groups. If there had been greater numbers of participants, a wider range of groups could have been identified or the consideration of CB as a continuous variable could have been appropriate.

5.6 HrQol and CB

Individuals with persistent CB had significantly lower levels of HrQol as measured by the PedsQL than those with low/no CB. There is little published research on CB and HrQol however it is consistent with the wider concept of Qol where CB is linked to low levels of Qol (Schalock 2004). The limitations of both the measurement of HrQol and the CB grouping variable were discussed previously.

5.7 HrQol, Health and CB

The relationship between HrQol and health differed when participants were split into those with persistent CB and those with low or no levels of CB. As such, only those with low or no CB showed a correlation between their health and their HrQol. The level of health problems experienced by those with persistent CB did not correlate to their HrQol as measured by the PedsQL. This suggest an additional factor which mediates or moderates the relationship between health and HrQol. The data collected in this study cannot identify such factors. It may be that mental health is an influencing variable in those with CB. In undergraduates with learning difficulties (such as dyslexia), affect/mood were found to have a mediating role in HrQol (Davis, Nida, Zlomke & Nebel-Schwalm, 2009). Low mood has also been shown to be associated with increased CB (Hayes, McGuire, O'Neill, Oliver & Morrison, 2011). A second possible factor is poverty, which has been found to have an impact on Health and HrQol (Elliott, Charyton, Lu, & Moore, 2009) however is an often 'overlooked' risk factor for health problems in the ID population (Anderson, Humphries, Mcdermott, Marks, Sisirak & Larson, 2013). Factors contributing to health, CB and HrQol are likely to be multifaceted and unlikely to form a simple linear

relationship and therefore further research is required to explore the possible relationship between these variables in both the ID and non-ID population.

The multiple regression model which considered the variables of CB, level of current health problems and number of chronic health problems, accounted for one fifth of the variance in HrQol. The level of CB and the number of chronic health problems were noted as making a significant contribution to this model. For this analysis, CB was converted from a grouping variable to a severity score. CB was considered as the combination of both aggression and self-injurious behaviour as in previous studies and allowed for comparisons between Time 1 and Time 2. However, the CBI collects information on a further eleven types of CB. Although these other types of behaviours are less common, their presence and impact if considered could have provided a fuller description of an individual's CB. Additionally, Emerson et al. (2001) found that individuals often displayed two or more types of CB and it may be that by only scoring the most severe behaviour for each type of CB present, the total severity score may have also underestimated the level of CB for some individuals.

In only accounting for a fifth of the variance this also suggests that there are other variables not measured within this study that may be contributing to the individual's HrQol. Although bootstrapped to increase the power of the analysis, due to low participant numbers, caution should still be paid to this result.

5.8 General Limitations

The main limitation of this study was the limited number of participants which constrained the analysis that could be conducted. As AS, CDC and CdLS are rare syndromes, low participant numbers are common and often result in the publication of descriptive or case

studies (e.g. Virbalas, Palma & Tan, 2012). The current study compensates for its limited number of participants by employing a strong design, utilising longitudinal data and comparing within and between groups. As a result of low number of participants, the three syndromes were combined and although similar in communication and levels of ID, there may be other between-syndrome differences that this grouping is disguising. There are also suggestions that the individual syndromes should be further split by the differing genetic causes as these may themselves be associated with different phenotypes (Dan, 2009).

The two groups representing those with persistent clinical levels of CB and those with low levels or no CB differed significantly in age. Although analysis found that neither the variables related to health nor the variables related to HrQol showed a significant association with age, this is important when considering the comparisons between the groups.

It should also be noted that due to the number of significance tests used, there is an increased chance of a making a Type I error.

5.9 Future Development

The need for longitudinal research on the health of people with genetic syndromes has been highlighted and further follow ups are recommended to allow for exploration of the HrQol. Petry and Maes (2008) call for the inclusion of self-reported Qol/HrQol from individuals wherever possible. Future research should consider whether there are ways in which this could be reliably collected from the current sample bearing in mind their level of ID. Even if this becomes possible it should be considered in addition to the proxy reporting and not as a replacement of proxy reporting. Further work needs to be done to consider the possible differences between the three syndromes as well as their similarities.

The current study addressed the number and severity of health problems and it would be useful to next consider whether they had received treatment and the impact of this treatment. Furthermore, there are a number of health problems that have increased frequency in either the three syndrome or in the ID population, such as scoliosis (Kline et al 2007), allergies and menses problems (May & Kennedy 2010) which are not enquired about on the Health Questionnaire and should be considered in the future.

Whilst this study documents physical health difficulties, it does not consider possible difficulties within mental health. Cooper, Smiley, Morrison, Williamson, & Allan (2007) suggest 22% of those with ID have a mental health problem. The relationship between mental health, CB and HrQol should be further explored. Finally research which took a prospective approach and included a non-ID comparison group would enable consideration of the possible differing impact of health problems on those with and without ID.

5.10 Conclusion

This is the first study to explore the relationship between health, HrQol and CB in individuals with ID. It provides information on areas which as of yet have been relatively overlooked in the research and identifies the need for replication with larger samples. This population of participants present a number of challenges to typical research methods which have been discussed. This study set out to provide further information on the health of those with ID and its impact. This remains an important area for research as those with ID, especially those with higher levels of ID, are unable to effectively express their health concerns and if parents and professionals are not aware of the different health problems and how they may be displayed then it is likely that this population will continue to have

untreated health problems and unresolved pain with no way of being able to make others aware.

6 References

Arron, K., Oliver, C., Moss, J., Berg, K., & Burbidge, C. (2011). The prevalence and phenomenology of self-injurious and aggressive behaviour in genetic syndromes. *Journal of Intellectual Disability Research*, *55*(2), 109-120.

Anderson, L. L., Humphries, K., Mcdermott, S., Marks, B., Sisirak, J., & Larson, S. (2013). The State of the science of Health and Wellness for Adults with Intellectual and Developmental Disabilities. *Intellectual and developmental disabilities*, *51*(5), 385-398.

Barisic, I., Tokic, V., Loane, M., Bianchi, F., Calzolari, E., Garne, E., ... & Dolk, H. (2008). Descriptive epidemiology of Cornelia de Lange syndrome in Europe. *American Journal of Medical Genetics Part A*, *146*(1), 51-59.

Bekkema, N., de Veer, A. J., Wagemans, A. M., Hertogh, C. M., & Francke, A. L. (2014). Decision making about medical interventions in the end-of-life care of people with intellectual disabilities: A national survey of the considerations and beliefs of GPs, ID physicians and care staff. *Patient education and counselling*, *96*(2), 204-209.

Bennett, C. C. (2009). A healthier future for all Australians: an overview of the final report of the National Health and Hospitals Reform Commission. *Medical Journal of Australia*, *191*(7), 383.

Berg, K., Arron, K., Burbidge, C., Moss, J., & Oliver, C. (2007). Carer-Reported Contemporary Health Problems in People With Severe and Profound Intellectual Disability and Genetic Syndromes. *Journal of Policy and Practice in Intellectual Disabilities*, *4*(2), 120-128.

Berney, T. P., Ireland, M., & Burn, J. (1999). Behavioural phenotype of Cornelia de Lange syndrome. *Archives of Disease in Childhood*, *81*(4), 333-336.

Bittles, A.H., Petterson, B.A., Sullivan, S.G., Hussain, R., Glasson, & Montgomery, P.D. (2002). The influence of Intellectual Disability on Life Expectancy. *Journal of Gerontology, 57* (7), M470-M472. doi: 10.1093/gerona/57.7.M470

Bosch, J., Van Dyke, D. C., Smith, S. M., & Poulton, S. (1997). Role of medical conditions in the exacerbation of self-injurious behavior: an exploratory study. *Mental Retardation*, *35*(2), 124-130.

Breau, L. M., Camfield, C. S., Symons, F. J., Bodfish, J. W., MacKay, A., Finley, G. A., & McGrath, P. J. (2003). Relation between pain and self-injurious behavior in nonverbal children with severe cognitive impairments. *The Journal of pediatrics*, *142*(5), 498-503.

Brunklaus, A., Dorris, L., & Zuberi, S. M. (2011). Comorbidities and predictors of health-related quality of life in Dravet syndrome. *Epilepsia*, *52*(8), 1476-1482.

Buckley, R. H., Dinno, N., & Weber, P. (1998). Angelman syndrome: Are the estimates too low?. *American journal of medical genetics*, *80*(4), 385-390.

Burbidge, C., Oliver, C., Moss, J., Arron, K., Berg, K., Hill, L., Trusler, K., Furniss, F. and Woodcock, K. A. (2010). The association between repetitive behaviours, impulsivity and hyperactivity in people with intellectual disability. *Journal of Intellectual Disability Research, 54*, 1078-1092.

Carr, E. G., Smith, C. E., Giacin, T. A., Whelan, B. M., & Pancari, J. (2003). Menstrual discomfort as a biological setting event for severe problem behavior: Assessment and intervention. *Journal Information*, *108*(2).

Cianfaglione R, Clarke A, Kerr M, Hastings RP, Oliver C, Felce D. *(*2015). A national survey of Rett syndrome: Age, clinical characteristics, current abilities, and health. *Am J Med Genet* Part A 9999A:1–8.

Clarke, D. J., & Marston, G. (2000). Problem behaviors associated with 15q-Angelman syndrome. *American Journal on Mental Retardation*, *105*(1), 25-31.

Clayton-Smith, J. (1993). Clinical research on Angelman syndrome in the United Kingdom: observations on 82 affected individuals. *American journal of medical genetics*, *46*(1), 12-15.

Clayton-Smith, J., & Laan, L. A. E. M. (2003). Angelman syndrome: a review of the clinical and genetic aspects. *Journal of Medical Genetics*, *40*(2), 87-95.

Cooper, S. A., Smiley, E., Finlayson, J., Jackson, A., Allan, L., Williamson, A., ... & Morrison, J. (2007). The Prevalence, Incidence, and Factors Predictive of Mental Ill-Health in Adults with Profound Intellectual Disabilities. *Journal of Applied Research in Intellectual Disabilities*, *20*(6), 493-501.

Cooper, S., Smiley, E., Morrison, J., Williamson, A., & Allan, L. (2007). Mental ill-health in adults with intellectual disabilities: prevalence and associated factors. *The British Journal of Psychiatry*, *190*(1), 27-35. doi:10.1192/bjp.bp.106.022483

Cornish, K., & Bramble, D. (2002). Cri du chat syndrome: genotype-phenotype correlations and recommendations for clinical management. *Developmental Medicine & Child Neurology*, *44*(7), 494-497.

Cornish, K. M., Bramble, D., Munir, F., & Pigram, J. (1999). Cognitive functioning in children with typical cri du chat (5p−) syndrome. *Developmental Medicine & Child Neurology*, *41*(04), 263-266.

Cornish, K. M., & Pigram, J. (1996). Developmental and behavioural characteristics of cri du chat syndrome. *Archives of disease in childhood*, *75*(5), 448-450.

Collins, M. S., & Cornish, K. (2002). A survey of the prevalence of stereotypy, self-injury and aggression in children and young adults with Cri du Chat syndrome. *Journal of intellectual disability research*, *46*(2), 133-140.

Collins, M. S. R., & Eaton-Evans, J. (2001). Growth study of cri du chat syndrome. *Arch Dis Child*, *85*, 337-338.

Cummins, R. A. (2002). Proxy responding for subjective well being : a review. *Int Rev Res Ment Retard*, *25*, 183-207.

Dan, B. (2009). Angelman syndrome: current understanding and research prospects. *Epilepsia*, *50*(11), 2331-2339.

Dan, B. & Pelc, K. (2008). Natural history of Angelman syndrome. *Developmental Medicine & Child Neurology*, *50*, 392-395.

Davis, T. E., Nida, R. E., Zlomke, K. R., & Nebel-Schwalm, M.S. (2009). Health-Related Quality of Life in College Undergraduates with Learning Disabilities: The mediational

Roles of Anxiety and Sadness. *Journal of Psychopathology and Behavioural Assessment,* 31(3)*, 228-234.*

Deardorff, M. A., Bando, M., Nakato, R., Watrin, E., Itoh, T., Minamino, M., ... & Shirahige, K. (2012). HDAC8 mutations in Cornelia de Lange syndrome affect the cohesin acetylation cycle. *Nature*, *489*(7415), 313-317.

Deardorff, M. A., Kaur, M., Yaeger, D., Rampuria, A., Korolev, S., Pie, J., ... & Krantz, I. D. (2007). Mutations in cohesin complex members SMC3 and SMC1A cause a mild variant of Cornelia de Lange syndrome with predominant mental retardation. *The American Journal of Human Genetics*, *80*(3), 485-494.

De Winter, C. F., Jansen, A. A. C., & Evenhuis, H. M. (2011). Physical conditions and challenging behaviour in people with intellectual disability: a systematic review. *Journal of Intellectual Disability Research*, *55*(7), 675-698.

Einfeld, S. L., Ellis, L. A., Doran, C. M., Emerson, E., Horstead, S. K., Rosamond H. Madden, R. H. & Tonge, B. J. (2010). Behavior Problems Increase Costs of Care of Children With Intellectual Disabilities, *Journal of Mental Health Research in Intellectual Disabilities*, *3*, 202-209.

Elliott, J. O., Charyton, C., Lu, B., & Moore, J. L. (2009). Serious psychological distress and health outcomes for persons with epilepsy in poverty. *Seizure*, *18*(5), 332-338.

Emerson, E., & Baines, S. (2010). *Health inequalities and people with learning disabilities in the UK: 2010.* Durham: Improving Health & Lives: Learning Disabilities Observatory.

Emerson, E., & Hatton, C. (2007). Mental health of children and adolescents with intellectual disabilities in Britain. *The British Journal of Psychiatry*, *191*(6), 493-499.

Emerson, E., Kiernan, C., Alborz, A., Reeves, D., Mason, H., Swarbrick, R., … Hatton, C. (2001). The prevalence of challenging behaviors: a total population study. *Research in Developmental Disabilities, 22*, 77-93.

van Gameren-Oosterom, H. B., Fekkes, M., Buitendijk, S. E., Mohangoo, A. D., Bruil, J., & Van Wouwe, J. P. (2011). Development, problem behavior, and quality of life in a population based sample of eight-year-old children with Down syndrome. *PloS one*, *6*(7), e21879. doi:10.1371/journal.pone.0021879

Gillberg, C. & Soderstrom, H. (2003). Learning disability. *The Lancet, 32*, 811-821.

Glover, G. & Ayub, M. (2010). How people with learning disabilities die. *Durham: Improving Health & Lives: Learning Disabilities Observatory*.

Golubović, Š., & Škrbić, R., (2013). Agreement in quality of life assessment between adolescent with intellectual disability and their parents. *Research in Developmental Disabilities, 34,* 1863-1869.

Griffith, G. M., Hastings, R. P., Nash, S., Petalas, M., Oliver, C., Howlin, P., Moss, J., Petty, J., & Tunnicliffe, P. (2011a). "You Have to Sit and Explain it All, and Explain Yourself." Mothers' Experiences of Support Services for Their Offspring with a Rare Genetic Intellectual Disability Syndrome. *Journal of Genetic Counseling*, 20(2), 165 – 177.

Griffith, G. M., Hastings, R. P., Oliver, C., Howlin, P., Moss, J., Petty, J., & Tunnicliffe, P. (2011b). Psychological well-being in parents of children with Angelman, Cornelia de Lange and Cri du Chat syndromes. *Journal of Intellectual Disability Research*, *55*(4), 397-410.

Hall, S. S., Arron, K., Sloneem, J. & Oliver, C. (2008). Health and sleep problems in Cornelia de Lange Syndrome: a case control study. *Journal of Intellectual Disability Research, 52,* 458-468.

Harris, J. C. (2005). Intellectual Disability : Understanding Its Development, Causes, Classification, Evaluation, and Treatment. Cary, NC, USA: Oxford University Press, USA.

Havercamp, S. M., Scandlin, D., & Roth, M. (2004). Health disparities among adults with developmental disabilities, adults with other disabilities, and adults not reporting disability in North Carolina. *Public Health Reports*, *119*(4), 418.

Hayes, S., McGuire, B., O'Neill, M., Oliver, C., & Morrison, T. (2011). Low mood and challenging behaviour in people with severe and profound intellectual disabilities. *Journal of Intellectual Disability Research, 55*(2), 182-189.

Hemmings, C., Deb, S., Chaplin, E., Hardy, S., & Mukherjee, R. (2013). Review of research for people With ID and mental health problems: A view from the United Kingdom. *Journal of Mental Health Research in Intellectual Disabilities*, *6*(2), 127-158.

Higurashi, M., Oda, M., Iijima, K., Iijima, S., Takeshita, T., Watanabe, N., & Yoneyama, K. (1990). Livebirth prevalence and follow-up of malformation syndromes in 27,472 newborns. *Brain and Development*, *12*(6), 770-773.

Hong, J., Koo, B., & Koo, J. (2008). The psychosocial and occupational impact of chronic skin disease. *Dermatologic therapy*, *21*(1), 54-59.

Hyman, P., Oliver, C., & Hall, S. (2002). Self-injurious behaviour, self-restraint, and compulsive behaviors in Cornelia de Lange syndrome. *American Journal on Mental Retardation, 107.* 146-154.

Ireland, M. (1996). Cornelia de Lange syndrome: Clinical features, common complications and long-term prognosis. *Current Paediatrics*, *6*(2), 69-73.

Jankovic, S., Raznatovic, M., Marinkovic, J., Jankovic, J., Kocev, N., Tomic-Spiric, V., & Vasiljevic, N. (2010). Health-related quality of life in patients with psoriasis. *Journal of cutaneous medicine and surgery*, *15*(1), 29-36.

Jansen, D. E. M. C., Kroi, B., Groothoff, J. W., & Post, D. (2004). People with intellectual disability and their health problems: a review of comparative studies. *Journal of Intellectual Disability Research, 48*(2), 93-102.

Kapell, D., Nightingale, B., Rodriguez, A., Lee, J. H., Zigman, W.B., & Schupf, N. (1998). Prevalence of Chronic Medical Conditions in Adults With Mental Retardation: Comparison With the General Population. *Mental Retardation, 36(*4), 269-279.

Kline, A. D., Grados, M., Sponseller, P., Levy, H. P., Blagowidow, N., Schoedel, C., ... & Tuchman, D. (2007). Natural history of aging in Cornelia de Lange syndrome. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*, *145(*3), 248-260.

Krahn, G. L., Hammond, L., & Turner, A. (2006). A cascade of disparities: health and health care access for people with intellectual disabilities. *Mental retardation and developmental disabilities research reviews*, *12*(1), 70-82.

Krantz, I. D., McCallum, J., DeScipio, C., Kaur, M., Gillis, L. A., Yaeger, D., ... & Jackson, L. G. (2004). Cornelia de Lange syndrome is caused by mutations in NIPBL, the human homolog of Drosophila melanogaster Nipped-B. *Nature genetics*, *36*(6), 631-635.

Kyllerman, M. (1995). On the prevalence of Angelman syndrome. *American journal of medical genetics*, *59*(3), 405-405.

Luzzani, S., Macchini, F., Valade, A., Milani, D., & Selicorni, A. (2003). Gastroesophageal reflux and Cornelia de Lange syndrome: typical and atypical symptoms. *American Journal of Medical Genetics Part A*, *119*(3), 283-287.

Mainardi, P. C. (2006). Cri du Chat syndrome. *Orphanet J Rare Dis*, *1*, 33.

Mainardi, P. C., Pastore, G., Castronovo, C., Godi, M., Guala, A., Tamiazzo, S., ... & Bricarelli, F. D. (2006). The natural history of Cri du Chat Syndrome. A report from the Italian Register. *European journal of medical genetics*, *49*(5), 363-383.

May, M. E., & Kennedy, C. H. (2010). Health and Problem Behavior Among People With Intellectual Disabilities. *Behavior Analysis in Practice*, *3*(2), 4-12.

McDermott, S., Platt, T., & Krishnaswami, S. (1997). Are individuals with mental retardation at high risk for chronic disease? *Family Medicine, 29*, 429-434.

McDougall, J., Baldwin, P., Evans, J., Nichols, M., Etherington, N. & Wright, V. (2015). Quality of Life and Self-Determination: Youth with Chronic Health Conditions Make the Connection. *Applied Research in Quality of Life.* Advance online publication. doi:10.1007/s11482-014-9382-7.

Mcguire, B. E., Daly, P., & Smyth, F. (2010). Chronic pain in people with an intellectual disability: under-recognised and under treated? *Journal of Intellectual Disability Research, 54*(3), 240-245.

McIntyre, L., Kraemer, B. R., Blacher, J., & Simmerman, S. (2004). Quality of life for young adults with severe intellectual disability: Mothers' thoughts and reflections. *Journal of Intellectual and Developmental Disability*, 29(2), 131-146.

McLaren, J., & Bryson, S. E. (1987). Review of recent epidemiological studies of mental retardation: Prevalence, associated disorders, and etiology. *American Journal on Mental Retardation*.

Moss, J., Howlin, P., Hastings, R., Beaumont, S., Griffith, G., Petty, J., Tunnicliffe, P., Yates, R., Villa, D. and Oliver, C. (2013). Social behavior and characteristics of Autism Spectrum Disorder in Angelman, Cornelia de Lange and Cri du Chat syndromes. *American Journal of Intellectual and Developmental Disabilities.* **118,** 262-283.

Moss, J., Oliver, C., Arron, K., Burbidge, C. & Berg, K. (2009). The Prevalence and Phenomenology of Repetitive Behavior in Genetic Syndromes. *Journal of Autism and Developmental Disorders, 39,* 572-588.

Moss, J. F., Oliver, C., Berg, K., Kaur, G., Jephcott, L., & Cornish, K. (2008). Prevalence of autism spectrum phenomenology in Cornelia de Lange and Cri du Chat syndromes. *Journal Information*, *113*(4).

Naito, M., Yuasa, H., Nomura, Y., Nakayama, T., Hamajima, N. and Hanada, N. (2006). Oral health status and health-related quality of life: a systematic review. *Journal of Oral science*, *48*(1), 1-7.

Nelson, L., Moss, J., & Oliver, C. (2014). A longitudinal follow-up study of affect in children and adults with Cornelia de Lange syndrome. *American Journal of Intellectual and Developmental Disabilities, 119,* 235-252.

Niebuhr, E. (1978). The cri du chat syndrome. *Human Genetics, 44*(3), 227-275.

Oliver, C., Horsler, K., Berg, K., Bellamy, G., Dick, K., & Griffiths, E. (2007). Genomic imprinting and the expression of affect in Angelman syndrome: what's in the smile?. *Journal of Child Psychology and Psychiatry, 48*(6), 571-579.

Oliver, C., McClintock, K., Hall, S., Smith, M., Dagnan, D. and Stenfert-Kroese, B. (2003) Assessing the Severity of Challenging Behaviour: Psychometric Properties of the Challenging Behaviour Interview. *Journal of applied research in intellectual disabilities*. 16 (1), 53-61.

Oliver, C., & Woodcock, K.A. (2008). Integrating levels of explanation in behavioural phenotype research. *Journal of Intellectual Disability Research, 52,* 807-809.

Overhauser, J., Huang, X., Gersh, M., Wilson, W., McMahon, J., Bengtsson, U., ... & Wasmuth, J. J. (1994). Molecular and phenotypic mapping of the short arm of chromosome 5: sublocalization of the critical region for the cri-du-chat syndrome. *Human molecular genetics, 3*(2), 247-252.

Pelc, K., Boyd, S. G., Cheron, G., & Dan, B. (2007). Epilepsy in Angelman syndrome. *Seizure, 17*(3), 211-217.

Peters, S. U., Horowitz, L., Barbieri-Welge, R., Taylor, J. L. & Hundley, R.J. (2012). Longitudinal follow-up of autism spectrum features and sensory behaviors in Angelman

syndrome by deletion class. *Journal of Child Psychology and Psychiatry and Allied disciplines, 53,* 152-159.

Petry, K. & Maes, B. (2008). Quality of Life: People with profound intellectual and multiple disabilities. In Pawlyn, J. & Carnaby, S. *Profound Intellectual and Multiple Disabilities: Nursing Complex Needs (pp.15 36).* West Sussex: Wiley-Blackwell.

Poppes, P., Van der Putten, A. J. J., & Vlaskamp, C. (2010). Frequency and severity of challenging behaviour in people with profound intellectual and multiple disabilities. *Research in Developmental Disabilities*, *31*(6), 1269-1275.

Potvin, M. C., Snider, L., Prelock, P. A., Wood-Dauphinee, S., & Kehayia, E. (2015). Health-related quality of life in children with high-functioning autism. *Autism*, *19*(1), 14-19.

Sabaz, M., Cairns, D. R., Lawson, J. A., Bleasel, A. F., & Bye, A. M. (2001). The Health-Related Quality of Life of Children with Refractory Epilepsy: A Comparison of Those With and Without Intellectual Disability. *Epilepsia*, *42*(5), 621-628.

Schalock, R. L. (2004). The concept of quality of life: what we know and do not know. *Journal of Intellectual Disability Research, 48,* 203 216.

Seltzer, M. M., & Krauss, M. W. (2000). Quality of life of adults with mental retardation/developmental disabilities who live with family. *Mental retardation and developmental disabilities research reviews, 7*(2), 105-114.

Sischo, L., & Broder, H. L. (2011). Oral Health-related Quality of Life What, Why, How, and Future Implications. *Journal of dental research*, *90*(11), 1264-1270.

Sparrow, S. S., Cicchetti, D. V., & Balla, D. A. (2005). *Vineland adaptive behavior scales: Second edition (Vineland II), survey interview form/caregiver rating form*. Livonia, MN: Pearson Assessments.

Subramaniam, M., Abdin, E., Vaingankar, J. A., Nan, L., Heng, D., McCrone, P., & Chong, S. A. (2013). Impact of psychiatric disorders and chronic physical conditions on health-related quality of life: Singapore Mental Health Study. *Journal of affective disorders*, *147*(1), 325-330.

Taylor, L., Oliver, C., & Murphy, G. (2011). The Chronicity of Self-Injurious Behaviour: A Long-Term Follow-Up of a Total Population Study. *Journal of Applied Research in Intellectual Disabilities*, *24*(2), 105-117.

Traci, M. A. Seekins, T., Szali-Petree, A., & Ravesloot, C. (2002). Assessing secondary conditions among adult with developmental disabilities. *Journal of Policy and Practice in Intellectual Disabilities, 7*, 204-210.

Tunnicliffe, P. L. (2010). *Self-injurious and aggressive behaviour in Angelman, Cri du Chat and Cornelia de Lange syndromes*. PhD. The University of Birmingham: U.K.

Wagemans, A. M., Lantman-de, H. M. V. S., Proot, I. M., Metsemakers, J., Tuffrey-Wijne, I., & Curfs, L. M. (2013). End-of-life decisions for people with intellectual disabilities, an interview study with patient representatives. *Palliative medicine*, *27*(8), 765-771.

Whitfield, M., Langan, J. & Russell, O. (1996). Assessing general practitioners' care of adult patient with learning disability: case-control study. *Quality in Health Care, 5*, 31-35.

Wilkins, L. E., Brown, J. A., Nance, W. E., & Wolf, B. (1983). Clinical heterogeneity in 80 home-reared children with cri du chat syndrome. *The Journal of pediatrics, 102*(4), 528-533.

Williams, C. A., Beaudet, A. L., Clayton-Smith, J., Knoll, J. H., Kyllerman, M., Lann, L.A., … Wagstaff, J. (2006). Angelman Syndrome 2005: Updated Consensus for Diagnostic Criteria. *American Journal of Medical Genetics, 140A*, 413-418.

Varni, J. W., Bendo, C. B., Nurko, S., Shulman, R. J., Self, M. M., Franciosi, J. P., ... & Pohl, J. F. (2015). Health-Related Quality of Life in Pediatric Patients with Functional and Organic Gastrointestinal Diseases. *The Journal of pediatrics*, *166*(1), 85-90.

Varni, J.W., & Limbers, C.A. (2009). The PedsQL™ 4.0 Generic Core Scales young adult version: Feasibility, reliability and validity in a university student population. *Journal of Health Psychology, 14*, 611-622.

Varni, J. W., Limbers, C. A., & Burwinkle, T. M. (2007). Impaired health-related quality of life in children and adolescents with chronic conditions: a comparative analysis of 10 disease clusters and 33 disease categories/severities utilizing the PedsQL™ 4.0 Generic Core Scales. *Health and quality of life outcomes*, *5*(1), 43.

Varni, J., Seid, M., & Kurtin, P. (2001). PedsQL 4.0: Reliability and Validity of the Pediatric Quality of life Inventory Version 4.0 Generic Core Scales in Healthy and Patient Populations. *Medical care, 39*(8), 800-812.

Varni, J. W., Seid, M., & Rode, C. A. (1999). The PedsQL™: measurement model for the pediatric quality of life inventory. *Medical care*, *37*(2), 126-139.

Virbalas, J. M., Palma, G., & Tan, M. (2012). Obstacles to Communication in Children With Cri du Chat Syndrome. *Journal of Voice*, *26*(6), 821-822.

Executive Summary

The term quality of life (Qol) is used in research and health and social care policies as a concept that provides a way of considering a person's life as a whole. Quality of life is made up of a number of factors such as a person's health, wellbeing, financial circumstance and relationships with others. It can also include wider factors such as a person's rights and their inclusion in society. Qol is a complex concept and there is much theoretical discussion about the exact factors which should be included. What is agreed is that to measure Qol, researchers should include reports on things that they can see/measure such as income and time spent with others. These are called objective measures. It should however also include subjective measures, where the researcher asks the individual in question how happy/satisfied they are with certain things in their life. Together, the subjective and objective measures are thought to provide the best estimate of a person's Qol.

For people with intellectual disabilities (ID), the measurement of Qol has become particularly important as it has encouraged services to consider the individual and their likes and wishes. As the importance of Qol has grown, so has the need to find ways to measure and record it. In order for a measure to be of use to a service, it needs to be easy to use and should provide results which can help document change in an individual. There are a number of Qol measures, however only eleven have been found to provide information on their validity and reliability. The systematic review within this thesis identified any paper that had used one of these eleven measures. 31 papers were found and they covered a range of topics including both environmental factors such as where a person lived and individual factors such as a person's ability and mental health. Different researchers used

the same Qol measures differently making it difficult to compare studies. Overall, it was found that more studies had focused on environmental factors then individual factors. Measuring Qol in people with ID, although important, can be difficult and further research is needed to consider how these measures can be used in service to help maintain or increase a person's Qol.

A further development in the study of Qol is to focus on the impact of health on Qol. This narrower definition of Qol is known as health related quality of life (HrQol). The impact of long term health problems have been explored in both adults and children without ID. Although people with ID are at increased risk of long term health problems, the impact of these on their HrQol has as of yet not been considered. The empirical paper in this thesis explores the health of people with rare genetic syndromes (Angelmans, Cornelia de Lange or Cri du Chat) and also measures their HrQol. This found that health problems were common in these people and that many of them had reported the same type of health problem almost seven year ago. People with more health problems and more severely impacted by health had lower HrQol scores. There are a range of behaviours that can be classed as challenging behaviours which includes behaviour that hurts the individual such as head banging or behaviours that hurt someone else such as hitting. People in this paper were split into those who showed continuing high levels of challenging behaviour and those that showed low levels or no challenging behaviour. When compared, although there was no difference in the level of health problems found those with challenging behaviour had lower HrQol. It is important that we understand what health problems people with ID may develop and how these may impact them as many of them are unable to communicate their discomfort and rely on others noticing difficulties and seeking support on their behalf.

Appendices

Contents

Appendix A- Papers removed due to language

Four papers were excluded from the results as they were not available in English. The details of these are below.

1. Del Pilar Poblete, Y. and Jimenez Figueroa, A. E. (2013). Psychosocial intervention proposal in self-determination, social skills and entrepreneurial capacity: Contribution from psychology to the integration of entrepreneurs women with intellectual disabilities. *Psychologia: Avances de la Disciplina*. 7(2), 55-67.

Language: Spanish

Abstract

It constructs an intervention on subjective well-being, self-determination, social skills and entrepreneurship in women micro-entrepreneurs with intellectual disabilities from a diagnosis realized in 20 women with intellectual disabilities. It administered a battery of five instruments: Satisfaction Scale life Diener et ál (1984), subjective happiness scale Lyubomirsky and Lepper (1999), Self-Concept Scale (La Rosa y Díaz, 1999), Questionnaire social skills Goldstein (1999) and Entrepreurship Questionnaire (Spencer & Spencer, 1993). The descriptive analysis indicates that women in the sample expressed high levels of development of their self-concept (self-determination) ($x = 382.6$, $\sigma = 53.32$), social skills ($x = 91.6$, $\sigma = 6, 65$) and subjective well-being, both in subjective happiness ($x = 21.45$, $\sigma = 3.96$) and life satisfaction ($x = 27.85$, $\sigma = 5.79$) and moderate levels of entrepreneurial ability ($x = 65.85$, $\sigma = 5,66$). An adequate level for all variables in women in the sample, the proposed intervention focusing on enhancing their skills and competencies, developing parallel knowledge to support existing ones.

Comment: *If this paper were to be available in English it is unlikely that it would meet the inclusion criteria as it does not seem to be using one of the Qol measure of interest, however this would have to be confirmed by a full text search.*

2. Moraes, A.M., Magna, L.A. and Marques-de-Faria, A. P. (2006). Prevention of mental retardation: knowledge and perception by health professionals. *Cadernos de Saude Publica*. 22(3):685-90.

Language: Portuguese

Abstract

This article aimed to evaluate the prevention of mental disability in primary healthcare services in Maringá, Paraná, Brazil. The sample consisted of 90 male and female physicians from different fields, namely gynecology and obstetrics, pediatrics, general practice, and family health, as well as 66 male and female nurses. A multiple-choice questionnaire was filled out by the subjects themselves from August to December 2003. Qualitative variables were compared using the qui-square test at 5% significance level. Partial data relating to both the perception and knowledge of health professionals

concerning mental disability were as follows: 75% were unable to choose the correct alternative on prevalence; 25% did not know how the genome contributes to etiology; 37% were unaware of prevention for mental disability; 28% were not confident in providing orientation on the teratogenic effect of ethanol; 35% demonstrated insecurity in orienting patients on amniocentesis. The data showed that participants had an unsatisfactory perception of the relevance of mental disability within the overall population disease profile, and that they need more information on the respective genetic and environmental issues.

Comment: *From the abstract this paper does not seem relevant it is evaluating the knowledge of help professionals not assessing Qol in individuals with an ID.*

3. Maalouf, D., El Hachem, H., Kesrouani, A., Hleis, S., Rohayem, J., Chammai, R., Haddad, G., Haddad, R. and Richa, S. (2011). Awareness and knowledge about risks of drinking during pregnancy in pregnant Lebanese women. *Encephale*. 37(2):94-100.

Language: French

Abstract:

OBJECTIVE: To assess the awareness and knowledge of pregnant Lebanese women about the risks of drinking during pregnancy and the factors that influence their drinking patterns. **MATERIALS AND METHODS:** A prospective study was conducted on a sample of 107 women consulting the gynecology outpatient department of Hôtel-Dieu de France in Beirut, Lebanon, who completed the T-ACE screening test included in a 21 multiple choice questionnaire which examine knowledge and beliefs about alcohol use during pregnancy, drinking patterns and awareness of fetal alcohol exposure. **RESULTS:** The 107 women of our sample were all married, between 20 and 41 years old and had mostly a high educational level (86%). Most of the women (47%) were at their first pregnancy. Of the 20 women who self-reported drinking during pregnancy, 60% obtained a positive score on the T-ACE questionnaire, which indicates that more than 11% of the women engaged with potentially high risk drinking for the baby. There is not a significant difference between the different age categories or educational levels. This proportion is lower than that found in international publications. However, the rate of excessive drinking (4 drinks or more on any one occasion in females) was higher and one woman in five reported excessive drinking in the previous year. There is a high level of knowledge that alcohol use during pregnancy is harmful to the child, and the more consumption the more harmful and likely the effects, but there is confusion about the safety of small amounts of alcohol. Women (37%) think that there is a safe level of drinking during pregnancy; 29% tolerate up to one drink a month, 9% tolerate up to one drink a week and one woman thinks having one drink a day is safe. Women who actually drink during pregnancy are more likely to think that alcohol consumption to a certain level is safe. Women (31%) think that beer and/or wine are safe alcohols to a certain level during pregnancy. When asked about the source of this belief, 22% mention a gynecologist but the majority (61%) says it is a personal belief. Women (65%) in our sample are aware that alcohol use during pregnancy can lead to life-long disabilities in a child, such as delayed development (36%), birth defects/deformities (35%) and mental retardation (32%). However, up to 33% of the

respondents report having no information about the effects of alcohol on the fetus and two women believe alcohol is not harmful at all. Women with lower levels of education are somewhat less knowledgeable about the risks of alcohol use during pregnancy than those with higher levels of education. There is no association between the drinking patterns of the women with their age, their professional habits and the alcohol consumption of their husbands. The women in our sample seem to be more aware of the necessity to stop smoking rather than stop drinking during pregnancy. **CONCLUSION:** Lebanese women are not fully aware of the recommendations and risks related to drinking during pregnancy. This is the reason why action must be taken to ensure better diffusion of these recommendations and better assessment of alcohol intake during prenatal visits.

Comment: *From the abstract this paper does not seem relevant it is evaluating the knowledge of pregnant women regarding the risk of alcohol and not assessing Qol in individuals with an ID.*

Mannfeld, S., Strauss, A. and Schulze, A.(2012). The cognitive development of triplets in school age and its impact on the quality of family life--a follow-up study from a perinatal centre. *Z Geburtshilfe Neonatol*. 216(6):269-76.

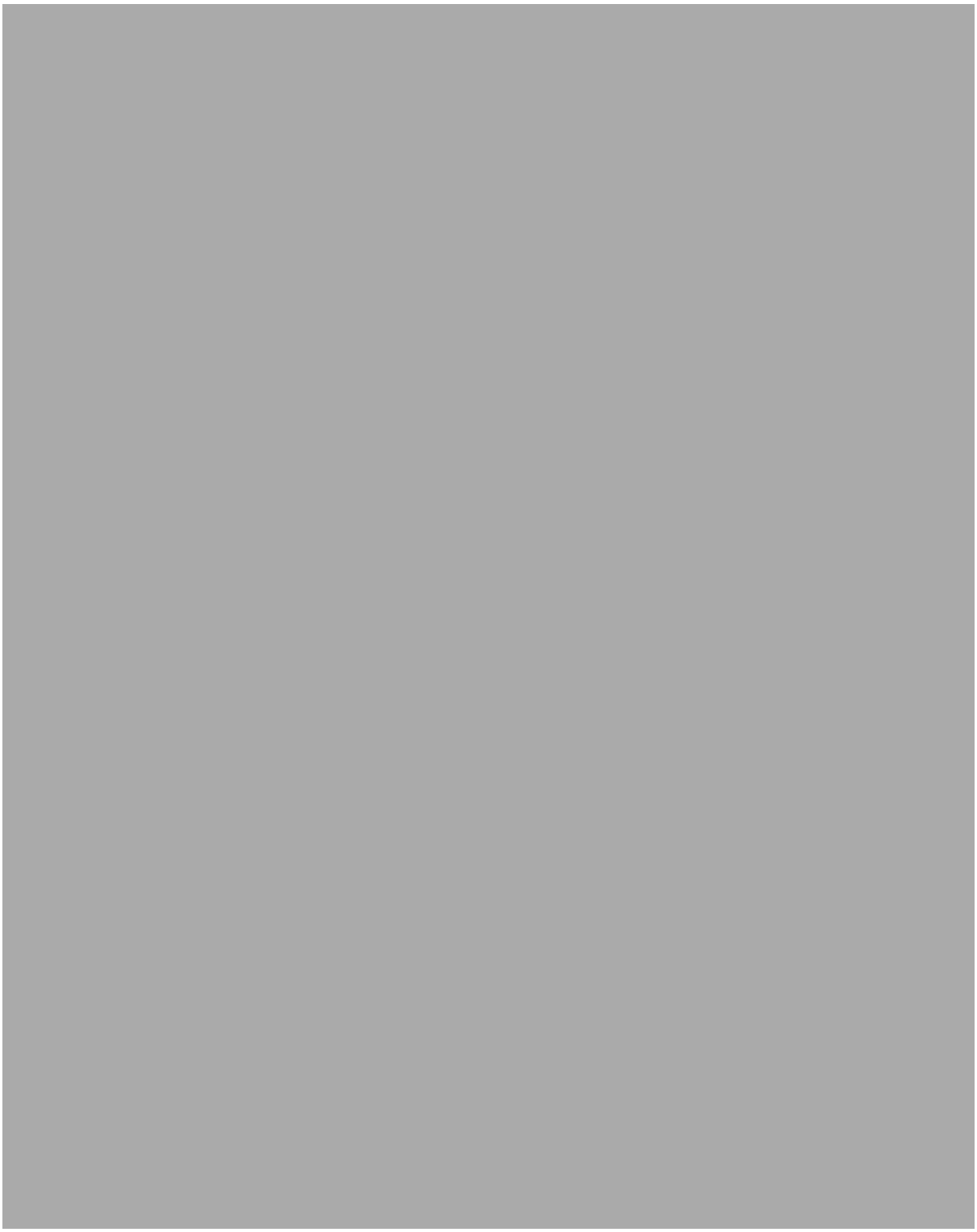
Language: German

Abstract:

OBJECTIVE: The aim of this study was to assess the developmental outcome of 7- to 9-year-old triplets and to determine parenting stress and quality of family life. **METHODS**: Cognitive development (Wechsler intelligence scales for children III, WISC III) and quality of family life (Kansas family life satisfaction scale, KFLSS; parenting stress index, PSI) of 48 triplets born between 1996 and 1998 at a perinatal centre were compared with controls born at a gestational age ≥37 weeks. Index and control children/mothers were matched by age (birthday: ±14 days/maternal age±2 years). **RESULTS:** Triplets and their families showed a mean IQ score and parenting satisfaction and stress within the normal range. Full IQ score (97 SD 16) as well as verbal and performance IQs (102 SD 16; 92 SD 19) were significantly lower than the controls' corresponding data (full IQ 111 SD 15, $p<0.001$; verbal IQ 116 SD 21, $p<0.001$; performance IQ 102 SD 17, $p=0.001$). Parents of triplets tended to show a higher total PSI score (222 SD 39) than the parents of the controls (205 SD 47). The differences reached statistical significance only in the parent domain (triplets 125 SD 24; controls 111 SD 27; $p<0.01$). Satisfaction with family life was good in triplet families and did not differ significantly in any of the subscales from control parents' satisfaction. **CONCLUSIONS:** This study demonstrates a favourable outcome of triplets with respect to their cognitive development and the quality of their families' lives at school age. The difference in children's IQ was statistically, but perhaps not clinically, significant, because the triplets' data were within the normal range.

Comment: *This paper would be excluding as it is measuring Family Qol not an individuals. In addition the individuals did not have a ID.*

<u>Appendix B -</u>A copy of message sent to Author regarding the version of the questionnaire used in his paper.



Appendix C- Papers removed due to not containing enough detail

Two papers were excluded from the results as they did not contain enough information to full assess the inclusion criteria. The details of these are below.

1. Keith, K. D. & Bonham, G.S (2005). The use of quality of life data at the organization and systems level. *Journal of Intellectual Disability Research.* 49(10). 799-805.

Abstract

Background: To date researchers have given little attention to the use of quality of life (QOL) data for organization and systems-level change. This article presents two state-level examples of how QOL data are currently used in the USA. **Method:** Individuals with intellectual disability (ID) were assessed on an ongoing basis using two multidimensional QOL instruments. Data were analysed at the individual and organizational level. **Results:** Examples of state-wide data utilization include: (1) determining significant predictors of quality outcomes; (2) developing provider profiles; (3) comparing individuals with ID with those without ID; (4) developing state-level performance standards; and (5) implementing continuous programme improvement. **Conclusions:** The availability of this type of data allows service delivery systems to: (1) significantly alter the relationship between individual consumers and service providers; (2) open the system to scrutiny by citizens with and without ID; (3) improve responsiveness and quality outcomes; and (4) shape future directions of the service delivery system for people with ID.

Comment: *This paper describes two different unpublished studies one of which utilised a Qol measure of interest however it did not provide sufficient enough information for it to be included in the review.*

2. McIntyre, L. L., Kraemer, B. R., Blacher, J. & Simmerman, S. (2004). Quality of life for young adults with severe intellectual disability: mothers' thoughts and reflections. *Journal of Intellectual and Developmental Disability*, 29(2), 131-146.

Abstract

Thirty mothers of transition-aged young adults (18-24 years) with severe intellectual disability were interviewed regarding their son or daughter's quality of life. All mothers completed the standardised Quality of Life Questionnaire and responded to several open-ended questions to further delineate quality of life for their child. Mothers were asked to describe quality of life for their young adult child and to evaluate their child's quality of life. Most mothers (73%) mentioned recreation, activities, and hobbies as important components of their young adult child's quality of life. Other common responses included having their son or daughter's basic needs met (53%), having their son or daughter belong to a social network (40%), and having their son or daughter be happy or content (37%). Less common responses included work (7%) for their son or daughter, communication capabilities (10%), health (13%), and consistency (17%) in their son or daughter's life. Mothers' visions for their sons and daughters, environmental and social supports for family members, and family quality of life issues are explored. Discussion focuses on contributions of this study to the burgeoning quality of life literature.

Comment: *This paper uses the Qol-Q (Cummins 1997b) as a descriptive of the group The substantial part of the paper is giving a qualitative account of mothers' narrative about their child's Qol.*

Appendix D - Checklist for the assessment of methodological quality based to Downs and Black (1998)

Below is the quality checklist used in this systematic review it is based on Downs and Black (1998). Scoring guidance is in italics. Adaptions to the original checklist and scoring notes are denoted by bold typeface.

Reporting

1 Is the hypothesis/aim/objective of the study clearly described?

2 Are the main outcomes to be measured clearly described in the Introduction or Methods section? *If the main outcomes are first mentioned in the Results section, the question should be answered no.*

3 Are the characteristics of the patients included in the study clearly described? *In cohort studies and trials, inclusion and/or exclusion criteria should be given. In case-control studies, a case-definition and the source for controls should be given.*

4 Are the interventions of interest clearly described? *Treatments and placebo (where relevant) that are to be compared should be clearly described*. ***In relocation studies the location of interest must be clearly described.***

5 Are the distributions of principal confounders in each group of subjects to be compared clearly described? *E.g. age, sex,* ***ID***

A not applicable rating was given to either single case or within group designs. A rating of no was given to paper which only described the principal confounders in the full sample not the groups.

6 Are the main findings of the study clearly described? *Simple outcome data (including denominators and numerators) should be reported for all major findings so that the reader can check the major analyses and conclusions.*

Addition 6a Are the Quality of Life (Qol) findings of the study clearly described? ***Partial rating given if analysis is preformed but not discussed in text***

7 Does the study provide estimates of the random variability in the data for the main outcomes? *In non normally distributed data the inter-quartile range of results should be reported. In normally distributed data the standard error, standard deviation or confidence intervals should be reported*

Addition 7a, Does the study provide estimates of the random variability in the data for Qol?

8 **(REMOVED)** ~~Have all important adverse events that may be a consequence of the intervention been reported?~~

9 Have the characteristics of patients lost to follow-up been described?

This should be answered yes where ther are no losses to follow up or where losses to follow-up were so small that findings would be unaffected by their inclusion. This should be answered no where a study does not report the number of patients lost to follow-up.

10 Have actual probability values been reported (e.g. 0.035 rather than <0.05) for the main outcomes except where the probability value is less than 0.001?

Addition, Is the method of consent clearly described?

External validity

11 Were the subjects asked to participate in the study representative of the entire population from which they were recruited? *The study must identify the source population for patients and describe how the patients were selected. Patients would be representative if they comprised the entire source population, an unselected sample of consecutive patients, or a random sample. Random sampling is only feasible where a list of all members of the relevant population exists. Where a study does not report the population from which the patients are derived the question should be answered* ***NS***

12 Were those subjects who were prepared to participate representative of the entire population from which they were recruited? *The proportion of those asked who agreed should be stated*. ***A partial rating was given to studies which discussed reason for none participation.***

13 **(REMOVED)** ~~Were the staff, places, and facilities where the patients were treated, representative of the treatment the majority of patients receive?~~

Internal validity- Bias

14 Was an attempt made to blind study subjects to the intervention they have received? *For studies where the patients would have no way of knowing which intervention they received, this should be answered yes.*

15 Was an attempt made to blind those measuring the main outcomes of the intervention?

16 **(REMOVED)** ~~If any of the results of the study were based on "data dredging", was this made clear?~~

17 In trials and cohort studies, do the analyses adjust for different lengths of follow-up of patients, or in case-control studies, is the time period between the intervention and outcome the same for cases and controls? *Where follow-up was the same for all study patients the answer should yes. Studies where differences in follow-up are ignored should be answered no.*

18 Were the statistical tests used to assess the main outcomes appropriate?

19 Was compliance with the intervention/s reliable? *Where there was non compliance with the allocated treatment or where there was contamination of one group, the question should be answered no.* …….

20 Were the main outcome measures used accurate (valid and reliable)? *Where outcome measures are clearly described, which refer to other work or that demonstrates the outcome measures are accurate, should be answered yes.*

Internal validity- confounding (selection bias)

21 Were the patients in different intervention groups (trials and cohort studies) or were the cases and controls (case-control studies) recruited from the same population? *For example Patients for all comparison groups should be selected from the same hospital. The question should be answered **NS** for cohort and case control studies where there is no information concerning the source of patients.*

22 Were study subjects in different intervention groups (trials and cohort studies) or were the cases and controls (case-control studies) recruited over the same time?

23 Were study subjects randomised to intervention groups? *Studies which state that subjects were randomised should be answered yes except where method of randomisation would not ensure random allocation.*

24 Was the randomised intervention assignment concealed from both patients and health care staff until recruitment was complete and irrevocable? *All non-randomised studies should be answered no. If assignment was concealed from patients but not from staff, it should be answered no.*

25 Was there adequate adjustment for confounding in the analyses from which the main findings were drawn? *In nonrandomised studies if the effect of the main confounders was not investigated or no adjustment was made in the final analyses the question should be answered as no.*

26 Were losses of patients to follow-up taken into account? *If the numbers of patients lost to follow-up are not reported the question should be answered **NS.** If loss to follow up was too small to affect the main findings, the question should be answered yes.*

Power

27 **(Adapted)** ~~Did the study have sufficient power to detect a clinically important effect where the probability value for a difference being due to chance <5% Sample sizes have been calculated to detect a difference of x% and y%.~~

Did the study report consideration of power either in method or in discussion of the results?

Appendix E – Ethical Approval

Ethical approval letter for full project of which this study is a part of.

REMOVED FROM E VERSION

Appendix B - Questionnaire pack

As this was part of a larger study entitled 'Describing and Understanding Challenging Behaviour and Parent/Carer Well-being', the participants were asked to complete a number of questionnaires either online or by post. A full list of the questionnaires is below. Those specific to this study are underlined and reproduced in the following pages.

DEMOGRAPHIC QUESTIONNAIRE

WESSEX QUESTIONNAIRE

HOSPITAL ANXIETY AND DEPRESSION SCALE

POSITIVE AFFECT SCALE

RAISING A CHILD WITH AN INTELLECTUAL DISABILITY (PGS)

THE PARENT AND FAMILY SUBSCALE OF THE QUESTIONNAIRE ON RESOURCES AND STRESS-SHORT FORM.

GENETIC SYNDROME STRESSORS SCALE

HEALTH QUESTIONNAIRE

PAEDIATRIC QUALITY OF LIFE INVENTORY (PEDSQL)

The Cerebra Centre for Neurodevelopmental Disorders

Cerebra
Positively Different

Describing and Understanding Challenging Behaviour and Parent/Carer Well-being

Questionnaire Pack

UNIVERSITY OF BIRMINGHAM

DEMOGRAPHIC QUESTIONNAIRE

Please tick or write your response to these questions concerning background details:

Today's date: _____

Your name: _____

*The following questions regard information about the **person you care** for:*

1. Gender: Male ☐ Female ☐

2. Date of Birth: ___/___/____ **Age:_____**

3. Is the person you care for verbal? (i.e. more than 30 signs/ words in their vocabulary)

Yes/ No (delete as appropriate)

4. Is the person you care for able to walk unaided?

Yes/ No (delete as appropriate)

5. Has the person you care for been diagnosed with a syndrome?

Yes/ No (delete as appropriate)

If yes, please indicate which syndrome in 5a. and answer questions 6 to 8. If no, please move on to question 9.

5a. Cornelia de Lange syndrome ☐ Cri du Chat syndrome ☐
Angelman syndrome ☐ Other
_____

6. What is the genetic mechanism causing the syndrome in the person you care for?

Uni-parental disomy ☐ Sequence repetition ☐
Deletion ☐ Translocation ☐
Unknown Other ☐
_____

7. When was the person you care for diagnosed?

_____

8. Who diagnosed the person you care for?

Paediatrician ☐ Clinical Geneticist ☐
GP Other _____ ☐

9. Has the person had any medical / health difficulties in the last six months? If yes, please give details:

_____

_____

The following questions ask for background information about you, your child with Angelman, Cornelia de Lange or Cri du Chat syndrome, and your family. Please tick the appropriate boxes or write in the spaces provided.

1. Are you male or female? Male ☐ Female ☐

2. What was your age in years on your last birthday? _____ years

3. What is your current marital status?

Married, and living with spouse..... ☐

Living with partner..... ☐

Divorced/Separated/Widowed/Single and NOT living with a partner.... ☐

4. In total how many people currently live in your home? _____ Adults _____ Children

5. Please tick the boxes next to all of the educational qualifications that you hold

No formal educational qualifications..... ☐

GCSE, GCE, O Levels or equivalent..... ☐

A Levels, GNVQ, or equivalent..... ☐

Polytechnic/University degree..... ☐

Masters or Doctoral degree..... ☐

6. What is your relationship to your child (e.g., mother, father, stepmother, grandmother, adoptive parent)? _____

*7. **Does your child normally live with you?***

Yes ☐ ***No*** ☐

If no, then where do they live? _____

8. Recent data from research with families of children with special needs has shown that a family's financial resources are important in understanding family member's views and

experiences. With this in mind, we would be very grateful if you could answer the additional question below. We would like to be able to look at whether those with high versus lower levels of financial resources have different experiences.

What is your current total annual family income? Please include a rough estimate of total salaries and other income (including benefits) before tax and national insurance/pensions.

Total family income

Less than £15,000..... ☐

£15,001 to £25,000..... ☐

£25,001 to £35,000..... ☐

£35,001 to £45,000..... ☐

£45,001 to £55,000..... ☐

£55,001 to £65,000..... ☐

£65,001 or more..... ☐

Once this questionnaire is complete, we have a few questions that are easier to ask over the telephone. Can you please confirm the best telephone number to reach you on?

_____

And what time of day is it best to try and reach you? (please tick all that apply)
Check any that apply

Weekday mornings (9am-12.30pm) ..... ☐

Weekday afternoons (12.30-3.30pm) ..... ☐

Weekday early evening (4.30-7pm) ..... ☐

Weekday evening (7pm onwards) ..... ☐

Weekends ..... ☐

* And what is the best email address to email you at?

_____

Health Questionnaire

PART A

Instructions:

- Have these problems **EVER** affected your child or person you care for?
- Please rate as **0** – if the problem has never affected the person you care for, **1** – if it has been a mild problem, **2** - if the problem has been moderately serious, or **3** – if the problem has been severe.
- If the person you care for has had these problems please state whether any treatment has been implemented by circling **yes** or **no**.

	Never	Mild	Moderate	Severe
1a. Eye Problems (e.g. glaucoma / blocked tear duct/s).....	**0**	**1**	**2**	**3**
1b. Corrective surgery / medication / treatment: **yes / no**				
2a. Ear Problems (e.g. infections, glue ear)	**0**	**1**	**2**	**3**
2b. Corrective surgery / medication / treatment (e.g. grommets): **yes / no**				
3a. Dental Problems (e.g. toothache / gum problems / mouth ulcers / delayed eruption of teeth).....	**0**	**1**	**2**	**3**
3b.Dental surgery / treatment (e.g. teeth removal): **yes / no**				
4a. Cleft Palate.....	**0**	**1**	**2**	**3**
4b. Repaired: **yes / no**				
5a. Gastrointestinal Difficulties (e.g. reflux / stomach problems).....	**0**	**1**	**2**	**3**
5b. Corrective surgery / medication / treatment (e.g. nissen fundoplication): **yes / no**				
6a. Bowel Problems (e.g. obstruction).....	**0**	**1**	**2**	**3**
6b. Corrective surgery / treatment: **yes / no**				
7a. Heart Abnormalities or Circulatory Problems (e.g. congenital heart lesions or murmur).....	**0**	**1**	**2**	**3**
7b. Corrective surgery / medication / treatment: **yes / no**				
8a. Problems with Genitalia (e.g. prostate/ testicular problems i.e. undescended testes) ……..	**0**	**1**	**2**	**3**
8b. Corrective surgery / treatment: **yes / no**				
9a. Hernia (e.g. inguinal or hiatal).....	**0**	**1**	**2**	**3**
9b. Repair / treatment: **yes / no**				
10. Limb Abnormalities (e.g. malformed arm).....	**0**	**1**	**2**	**3**
11a. Epilepsy / Seizures / Neurological Referrals.....	**0**	**1**	**2**	**3**
11b. Medication: **yes / no**				
12a. Lung or Respiratory Problems (asthma/bronchitis).....	**0**	**1**	**2**	**3**
12b. Corrective surgery / medication / treatment: **yes / no**				
13a. Liver or Kidney Problems.....	**0**	**1**	**2**	**3**
13b. Corrective surgery / medication / treatment: **yes / no**				
14a. Diabetes or Thyroid Function Problems.....	**0**	**1**	**2**	**3**
14b. Corrective surgery / medication / treatment: **yes / no**				
15a. Skin Problems (e.g. tinea, eczema, psoriasis, dry skin)…….....	**0**	**1**	**2**	**3**
15b. Medication / treatment: **yes / no**				
16a. Other (please specify problem, severity from 0-3).....	**0**	**1**	**2**	**3**
16b. Corrective surgery / medication / treatment: **yes / no**				

PART B

Instructions:

- Have these medical problems affected the person you care for in the past **MONTH**

- Please rate as **0** – if your child has not been affected by this problem in the past month, **1** - if they have been mildly affected, **2** – if the problem has moderately affected your child and **3** - if your child has been severely affected by the problem.

	No	Mild	Moderate	Severe
17. Eye Problems (e.g. glaucoma / blocked tear duct/s).....	**0**	**1**	**2**	**3**
18. Ear Problems (e.g. infections, glue ear).....	**0**	**1**	**2**	**3**
19. Dental Problems (e.g. toothache / gum problems / mouth ulcers / delayed eruption of teeth).....	**0**	**1**	**2**	**3**
20. Cleft Palate.....	**0**	**1**	**2**	**3**
21. Gastrointestinal Difficulties (e.g. reflux / stomach problems).....	**0**	**1**	**2**	**3**
22. Bowel Problems (e.g. obstruction).....	**0**	**1**	**2**	**3**
23. Heart Abnormalities or Circulatory Problems (e.g. congenital heart lesions or murmur)……..	**0**	**1**	**2**	**3**
24. Problems with Genitalia (e.g. prostate / testicular problems i.e. undescended testes)....	**0**	**1**	**2**	**3**
25. Hernia (e.g. inguinal or hiatal).....	**0**	**1**	**2**	**3**
26. Limb Abnormalities (e.g. malformed arm).....	**0**	**1**	**2**	**3**
27. Epilepsy / Seizures / Neurological Referrals.....	**0**	**1**	**2**	**3**
28. Lung or Respiratory Problems (asthma / bronchitis).....	**0**	**1**	**2**	**3**
29. Liver or Kidney Problems.....	**0**	**1**	**2**	**3**
30. Diabetes or Thyroid Function Problems.....	**0**	**1**	**2**	**3**
31. Skin Problems (e.g. tinea, eczema, psoriasis, dry skin).....	**0**	**1**	**2**	**3**
32. Other (please specify problem and severity from 0-3) ……..	**0**	**1**	**2**	**3**

Paediatric Quality of Life Inventory (PedsQL)

REMOVED FROM E VERSION

Appendix C- Telephone interview

As this was part of a larger study entitled 'Describing and Understanding Challenging Behaviour and Parent/Carer Well-being', during the telephone interview a number of questionnaire were used. A full list of the questionnaires used in the phone interview is below. Those specific to this study are underlined and reproduced in the following pages.

CHALLENGING BEHAVIOUR INTERVIEW

VINELAND ADAPTIVE BEHAVIOUR SCALES 2 (VABS)

CLIENT SERVICE RECEIPT INVENTORY :CHILDREN WITH INTELLECTUAL DISABILITIES

Challenging Behaviour Interview

Name: _____ Date of interview: ____/ ____/ 19____ Male ☐ Female ☐ Date of Birth: _____/ ____/ 19____

Current Address: _____ Name of Respondent: _____

_____ Profession/job: _____

Administration

1. Identify a respondent who has known the person well for at least 3 months.

2. Let the participant see a copy of the interview to help administration.

3. For part I, ask the respondent if each category of challenging behaviour has occurred in the last month by naming the category and then giving some examples from the appendix. Check the response by ensuring the month criterion and that the behaviour fulfils the operational definition. Tick the appropriate box.

4. In part II, enter the behaviour categories in the boxes above question 1. This will help you enter the ratings later on. For challenging behaviours which are included, read each question whilst the respondent looks at the question and then ask for a rating for each of the behaviour categories which have been chosen. Check the rating by asking for an example.

CHALLENGING BEHAVIOUR INTERVIEW (PART I)

1. Which of the following behaviours have occurred in the last month? (See appendix for definitions and examples)

Challenging Behaviour Category	List behaviours from examples in appendix
❑ Self –Injury (SIB)	
❑ Physical aggression (PAG)	
❑ Verbal aggression (VAG)	
❑ Disruption and destruction of property or the environment (DST)	
❑ Anal poking (AP)	
❑ Stereotyped behaviours (STB)	
❑ Inappropriate vocalisations (IV)	
❑ Inappropriate removal of clothing (IRC)	
❑ Pica (PIC)	
❑ Inappropriate or unacceptable sexual behaviour (ISB)	
❑ Smearing (SMR)	
❑ Stealing (STL)	
❑ Self-induced vomiting and regurgitation (SIV)	

CHALLENGING BEHAVIOUR INTERVIEW (PART II)

In each box, enter the category of challenging behaviour that is being considered

1. Think about how often this behaviour occurred in the last month. If there was no change and you watched this person now, then would you *definitely* see the behaviour:

⑤	④	③	②	①
In the next 15 minutes	In the next hour	By this time tomorrow	By this time next week	By this time next month

2. In the last month, for how long did the *longest* episode or burst of this behaviour last?

①	②	③	④	⑤
Less than a minute	Less than 5 minutes	Less than 15 minutes	Less than an hour	More than an hour

3. In the last month, for how long have episodes or bursts of this behaviour *typically* lasted or lasted on average?

①	②	③	④	⑤
Less than a minute	Less than 5 minutes	Less than 15 minutes	Less than an hour	More than an hour

4. For the worst episode of behaviour in the last month, what response was necessary[2]?

⓪	①	②	③	④
Nothing	Verbal discouragement or reminder	Informal physical intervention by one member of staff e.g. blocking, holding an arm briefly, taking objects from an individual Removal to a safe environment Removal of staff or others from immediate environments	Informal physical intervention by more than one member of staff Formal restraint procedure Protective or restrictive devices employed	Seclusion PRN medication Legal involvement or legal advice has been sought Section of MHA invoked

[2] To score, identify any items which have occurred and take highest scoring item.

5. In the last month, what has been the worst effect of this behaviour on the individual's *physical health*?

⓪	①	②	③
No effect at all	**Minor, temporary injury,** such as reddening of the skin, but *no* bruising or tissue damage	**Moderate injury,** such as bruising , cuts or abrasions **or illness lasting less than a day,** e.g. brief stomach upset, a single episode of vomiting	**Significant injury** e.g. fractured bones, sutures required, minor or major operation required **or illness lasting more than a day**

6. In the last month, what has been the worst *direct* effect of this behaviour on the *physical health* of staff or carers?

⓪	①	②	③
No effect at all	**Minor, temporary injury,** such as reddening of the skin, but *no* bruising or tissue damage	**Moderate injury,** such as bruising , cuts or abrasions **or illness lasting less than a day,** e.g. brief stomach upset, a single episode of vomiting	**Significant injury** e.g. fractured bones, sutures required, minor or major operation required **or illness lasting more than a day**

7. In the last month, what has been the worst ***direct* effect of this behaviour on the ***physical health*** of other service users?**

⓪	①	②	③
No effect at all	**Minor, temporary injury,** such as reddening of the skin, but *no* bruising or tissue damage	**Moderate injury,** such as bruising , cuts or abrasions **or illness lasting less than a day,** e.g. brief stomach upset, a single episode of vomiting	**Significant injury** e.g. fractured bones, sutures required, minor or major operation required **or illness lasting more than a day**

8. Throughout the whole of the last month, has the behaviour had any negative effects on the well-being of other service users e.g. disruption to planned activities, service users are frightened or upset, belongings or clothing are damaged or lost, meals are spoiled etc.?

⓪	①	②	③	④
No effect at all on the well-being of other service users	Effect on the well-being of other service users **about once in the last month**	Effect on the well-being of other service users **about once a week**	Effect on the well-being of other service users **about once every 3 days**	Effect on the well-being of other service users **nearly every day**

9. In the last month, what has been the *direct* effect of this behaviour on the environment in which the individual lives?

⓪	①	②	③	④					
No damage or loss at all	**Disruption or mild damage** to property or the living areas e.g., objects thrown, furniture tipped, doors slammed, meals spoiled, paint scratched. **Item does not require repair or replacement.**	**Moderate damage** to property or living areas e.g. curtains torn, furniture partly broken. **Item requires repair but can be used.**	**Significant damage** to property and living areas. **Item requires repair and cannot be used.**	**Extreme damage** to property or living areas. **Item requires replacement and cannot be used or repaired** e.g. windows broken, furniture unusable.					

10. In the last month, as a result of this behaviour, have restrictive or protective devices (e.g. arm splints, helmet) or specially designed clothing (e.g. all-in-one suit) been worn by the individual?

⓪	①	②	③	④					
Never	**Some of the time**	**About half the time**	**Most of the time**	**Almost continuously**					

(If so was it: Arm splint(s) ❑, Helmet or headgear ❑, Gloves/mittens/other items on hands ❑, Specially designed clothing ❑, Other ❑, (please specify) _____)

11. Has the environment in which the individual currently lives been modified because of this behaviour (examples of modification are given in the box below)?

⓪	①	②	③
No modifications	**Modifications to the person's possessions but not elsewhere** e.g. padding on a wheel chair, clothing which is strengthened	**Modifications have been made to the environment but are not noticeable unless pointed out e.g. curtains on Velcro, window locks**	**Modifications have been made to the environment and are noticeable**

Examples of modification to the environment: windows are not made of glass, TV is in a protective cabinet or out of reach, furniture is secured, a cupboard door is secured, a door is secured, curtains are absent (because they will be torn down), pictures are out of reach, hard or sharp surfaces are padded, service users are always visible, a room is out of bounds, cutlery is plastic, furniture is deliberately heavy, door closers are fitted to prevent slamming, wallpaper is washable in rooms apart from kitchen and bathroom, fridge is secured, ornaments are out of reach, furniture or fittings have been removed, furniture is chosen because it has particular qualities e.g. no sharp edges etc.

12. In the last month, as a result of this behaviour, has a verbal response by staff or carers been necessary e.g. discouraging the behaviour, distraction to another activity, reminder, reprimand?

⓪	①	②	③	④
Never	**At least once a month**	**At least once a week**	**At least once a day**	**At least once an hour**

13. In the last month, as a result of this behaviour, has physical contact or prevention or restraint by staff or carers been necessary e.g. blocking, taking objects from an individual, temporary restraint of an arm?

⓪	①	②	③	④					
Never	**At least once a month**	**At least once a week**	**At least once a day**	**At least once an hour**					

(If so was it a written procedure ❑ or an informal procedure ❑, please tick.)

14. In the last month, for this behaviour, was it necessary for more than one member of staff to respond when the behaviour occurred?

⓪	①	②	③	④					
Never	**At least once a month**	**At least once a week**	**At least once a day**	**At least once an hour**					

15. In the last month, has there been any contact with any of the following regarding this behaviour?

	Name and Contact Number
❑ Clinical Psychologist or Psychology Assistant working with a Clinical Psychologist	
❑ Psychiatrist	
❑ General Practitioner	
❑ Challenging Behaviour specialist or team	
❑ Speech and language therapist	
❑ Legal advisor	
❑ Other	
❑ Other	
❑ Other	

Summary of Scores

	Behaviours				
Qu.					
1					
2					
3					
4					
5					
6					
7					
8					
9					
10					
11					
12					
13					
14					
Total					

NOTES:

Vineland Adaptive Behaviour Scales 2 (VABS)

This Questionnaire is under copyright and so we are currently unable to attach a copy. A copy can be provided on request.

Appendix H- Exploring the Distribution of Variables

Table H.1: Distribution of variables within the Full group

Variable		Full group									
		Mean	SD	Skewness	SE skewness	z score	Kurtosis	SE Kurtosis	z score	Kolmogor	sig
Health											
No. health problems	T1	2.34	1.995	1.566	0.369	4.244	4.12	0.724	5.690	1.138	0.15
	T2	2.16	1.57	0.373	0.361	1.033	-0.574	0.709	-0.810	1.085	0.189
Severity score	T1	3.71	3.99	1.949	0.369	5.282	4.592	0.724	6.343	1.295	0.07
	T2	3.16	2.45	0.353	0.361	0.978	-1.059	0.709	-1.494	0.968	0.306
Average Severity	T1	1.23	0.71	-0.032	0.369	0.088	-0.259	0.724	-0.358	1.468	0.027
	T2	1.21	0.67	-0.497	0.361	1.377	-0.356	0.709	-0.502	1.405	0.038
Chronicity		1.23	1.31							1.46	0.028
HrQol											
Pedsql Total		51.77	16.53	0.961	0.361	2.662	1.013	0.709	1.429	0.756	0.617
Physical		37.35	28.61	0.927	0.361	2.568	-0.336	0.709	-0.474	1.172	0.128
Psychosocial		59.55	14.86	0.52	0.361	1.440	0.15	0.709	0.212	0.62	0.836
Challenging Behaviour											
CBQ SIB	T1	3.79	3.71	0.506	0.361	1.402	-1.033	0.709	-1.457	2.731	0.000
	T2	3.41	3.25	0.225	0.369	0.609	-1.388	0.724	-1.917	1.717	0.006
CBI											
SIB severity		8.48	7.99	0.457	0.365	1.252	-0.754	0.717	-1.052	1.533	0.018
Physical aggression severity		7.36	9.24	1.025	0.365	2.808	-0.018	0.717	-0.025	2.015	0.001
SIB + Aggression		15.83	14.26	0.856	0.365	2.345	0.262	0.717	0.366	0.864	0.444

Note. CBI= Challenging Behaviour Interview, CBQ= Challenging Behaviour Questionnaire, HrQol= Health related Quality of Life, Pedsql= Paediatric quality of life Inventory , SIB= Self injurious behaviour, T1=Time one, T2= Time Two.

Table H.2: Distribution of variables within the Persistent Challenging Behaviour group

Variable		**Persistent Challenging behaviour**									
		Mean	**SD**	**Skewness**	**SE skewness**	**z score**	**Kurtosis**	**SE Kurtosis**	**z score**	**Kolmogor**	**sig**
Health											
No health problems	T1	2.5	1.59	-0.057	0.564	-0.101	-0.883	1.091	-0.809	0.743	0.639
	T2	2.41	1.7	0.475	0.55	0.864	-0.286	1.063	-0.269	0.619	0.838
Severity score	T1	3.63	2.58	-0.031	0.564	-0.055	-1.557	1.091	-1.427	0.633	0.818
	T2	3.53	2.48	0.281	0.55	0.511	-0.902	1.063	-0.849	0.591	0.877
Average Severity	T1	1.22	0.64	-0.257	0.564	-0.456	0.815	1.091	0.747	0.958	0.318
	T2	1.32	0.64	-0.714	0.55	-1.298	0.538	1.063	0.506	0.79	0.56
Chronicity		1.68	1.58	0.591	0.564	1.048	-0.524	1.091	-0.480	0.679	0.746
HrQol											
Pedsql Total		44.02	12.19	0.133	0.55	0.242	-0.718	1.063	-0.675	0.603	0.86
Physical		24.63	16.61	0.343	0.55	0.624	-0.721	1.063	-0.678	0.551	0.755
Psychosocial		54.53	14.02	0.266	0.55	0.484	-0.991	1.063	-0.932	0.921	0.618
Challenging Behaviour											
CBQ SIB	T1	7.18	3.09	-0.881	0.55	-1.602	0.53	1.063	0.499	0.797	0.548
	T2	5.63	2.47	-0.473	0.564	-0.839	0.881	1.091	0.808	0.601	0.863
CBI											
SIB severity		13.35	6.93	0.307	0.55	0.558	0.111	1.063	0.104	0.478	0.976
Physical aggression severity		12.35	11.33	0.204	0.55	0.371	-1.583	1.063	-1.489	0.887	0.411
SIB + Aggression		25.71	14.26	0.608	0.55	1.105	-0.778	1.063	-0.732	0.867	0.44

Note. CBI= Challenging Behaviour Interview, CBQ= Challenging Behaviour Questionnaire, HrQol= Health related Quality of Life, Pedsql= Paediatric quality of life Inventory , SIB= Self injurious behaviour, T1=Time one, T2= Time Two.

Table H.3: Distribution of variables within the Persistent Challenging Behaviour group

Variable		**Low or No Challenging behaviour**									
		Mean	**SD**	**Skewness**	**SE skewness**	**z score**	**Kurtosis**	**SE Kurtosis**	**z score**	**Kolmogor**	**sig**
Health											
No health problems	T1	2.24	2.24	1.991	0.464	4.291	5.114	0.902	5.670	1.31	0.064
	T2	2	1.5	0.233	0.456	0.512	-1.004	0.887	-1.132	0.872	0.432
Severity score	T1	3.76	4.73	1.993	0.464	4.295	3.686	0.902	4.086	1.426	0.034
	T2	2.92	2.45	0.433	0.456	0.950	-1.106	0.887	-1.247	0.859	0.451
Average Severity	T1	1.23	0.76	0.043	0.464	0.093	-0.524	0.902	-0.581	1.285	0.074
	T2	1.14	0.69	-0.387	0.456	-0.849	-0.552	0.887	-0.622	1.166	0.132
Chronicity		1	1.06	0.658	0.456	1.443	-0.801	0.887	-0.903	1.184	0.121
HrQol											
Pedsql Total		56.84	17.23	0.997	0.456	2.186	0.343	0.887	0.387	0.881	0.419
Physical		45.67	31.88	0.536	0.456	1.175	-1.406	0.887	-1.585	1.098	0.179
Psychosocial		62.83	14.73	0.721	0.456	1.581	0.168	0.887	0.189	0.609	0.853
Challenging Behaviour											
CBQ SIB	T1	1.58	2.02	0.789	0.456	1.730	-0.848	0.887	-0.956	1.831	0.002
	T2	2	2.9	0.99	0.464	2.134	-0.678	0.902	-0.752	1.463	0.028
CBI											
SIB severity		5.38	6.94	0.831	0.456	1.822	-0.691	0.887	-0.779	1.825	0.003
Physical aggression severity		4.08	5.44	0.714	0.456	1.566	-1.331	0.887	-1.500	1.981	0.001
Sib + Aggression		9.46	9.73	0.539	0.456	1.182	-0.989	0.887	-1.115	1.313	0.064

Note. CBI= Challenging Behaviour Interview, CBQ= Challenging Behaviour Questionnaire, HrQol= Health related Quality of Life, Pedsql= Paediatric quality of life Inventory , SIB= Self injurious behaviour, T1=Time one, T2= Time Two.

Appendix I- Additional Results Tables

Within this appendix the following results tables can be found

Table I.1
Number of Current Health Problems Reported On the Health Questionnaire

Number of health problems	Time 1 n=41 (%)	Time 2 n=43 (%)
0	6 (14.6)	7 (16.3)
1	10 (24.4)	10 (23.3)
2	9 (22)	8 (18.6)
3	7 (17.1)	9 (20.9)
4	4 (9.8)	6 (14.0)
5	3 (7.3)	2 (4.7)
6	1 (2.4)	1 (2.3)
10	1 (2.4)	0 (0)

Table I.2

The Severity Score of Those Reporting at Least One Current Health Problem Within the Last Month on the Health Questionnaire

Severity score	Time 1 n=35 (%)	Time 2 n=36 (%)
1	9 (25.7)	7 (19.4)
2	7 (20.0)	6 (16.7)
3	2 (5.7)	4 (11.1)
4	3 (8.6)	6 (16.7)
5	4 (11.4)	5 (13.9)
6	3 (8.6)	1 (2.8)
7	3 (8.6)	6 (16.7)
8	0 (0)	1 (2.8)
9	2 (5.7)	0
16	1 (2.9)	0
18	1 (2.9)	0

Table I.3
The Relationship Between the Demographics Variables (Age and Ability) and the Health Questionnaire Variables (Number of Health Problems and Total Severity)

	Demographic Variable			
	Age		Ability	
Demographic	r_s	p	r_s	p
Total Health Problems				
Time 1	-0.27	.092	-0.01	.930
Time 2	-0.14	.376	0.25	.101
Total severity				
Time 1	-0.27	.086	0.02	.909
Time 2	-0.16	.303	0.23	.134

Table I.4
The Number and Severity of Current Health problem as reported by the Health Questionnaire Split by Syndrome and Gender

		Number of Health problems				Severity of Health			
				Difference				Difference	
Demographic	*N*	*mean*	*SD*	$t/\chi^{2\,b}$	p	Mean	*SD*	$t/\chi^{2\,b}$	p
Gender									
Time 1									
Male	20	2.15	2.48			3.10	4.23		
Female	21	2.52	1.44	-0.60[a]	.568	4.29	3.76	-0.95[a]	.366
Time 2									
Male	21	2.14	1.71			2.86	2.41		
Female	22	2.18	1.47	-0.08	.936	3.45	2.50	-0.80	.431
Syndrome									
Time 1									
AS	17	2.12	1.41			2.94	2.41		
CDC	11	2.00	1.41	.232	.890	3.45	3.08	0.24	.887
CDLS	13	2.92	2.90			4.92	5.91		
Time 2									
AS	18	1.94	1.30			2.78	2.21		
CDC	12	1.67	1.67			2.67	2.67		
CDLS	13	2.92	1.66	4.32	.115	4.15	2.44	3.35	.187

[a] Where the assumptions of normality were violated, a bootstrap was performed utilising 1000 repetitions [b] Kruskal Wallis Test

Table I.5

The Frequency and Percentage of Participants Showing Each Type of Health Problem Within the Past Month as Reported by the Health Questionnaire

	Time 1 (n=41)	Time 2 (n=43)
Health problem	Frequency (%)	Frequency (%)
Dental	16 (39.0)	19 (44.2)
Skin	16 (39.0)	16 (37.2)
Bowel	13 (31.7)	12 (27.9)
Epilepsy/ Seizures/ Neurological	11 (25.6)	9 (20.9)
Gastrointestinal	9 (22.0)	13 (30.2)
Eye	9 (22.0)	4 (9.3)
Limb Abnormalities	6 (14.0)	7 (16.3)
Ear	5 (11.6)	7 (16.3)
Heart or Circulatory	3 (7.0)	2 (4.7)
Lung / Respiratory	3 (7.0)	2 (4.7)
Cleft Plate	2 (4.9)	0
Genitalia	1 (2.3)	0
Hernia	1 (2.3)	2 (4.7)
Diabetes or Thyroid	1 (2.3)	0
Liver/ Kidney	0 (0)	0

Note. Percentages reported to 1 d.p.. As a participant can have multiple problems the cumulative percentage will exceed 100.

Table I.6
Parent/Carer Responses to the "Any Other Health Problems?" Question on the Health Questionnaire

Other health problems reported	*Includes*	Frequency Time 1 n=41	Frequency Time 2 n=43
Leg and Feet	*Ankle and knee problems Contracted Achilles tendon and foot turning in Stiff legs, Orthotic shoes*	2	2
Scoliosis	*Curve of spine*	2	1
Allergies	*Hayfever*	2	1
Hypotonia		1	
Postural Hypotension			1
Tonsillitis			1
Hip and back pain			1
Hands	*Slipped tendon in thumb*		1

Table I.7
The Top Five Current Health Problems as Reported on the Health Questionnaire Split by Gender and Syndrome Group

		Dental			Skin			Bowel			Epilepsy…			Gastrointestinal		
		freq	Difference		*freq*	Difference		*freq*	Difference		*freq*	Difference		*freq*	Difference	
Demographic	*N*	*(%)*	χ^2	p	*(%)*	χ^2	p	*(%)*	χ^2	p	*(%)*	χ^2	p	*(%)*	χ^2	p
Gender																
T1																
Male	20	5			7			4			5			4		
		(25.0)			**(35.0)**			(20.0)			(25.0)			(20.0)		
Female	21	11			9			9			6			5		
		(52.4)	3.23	.072	(42.9)	0.27	.606	(42.9)	2.47	.116	(28.6)	0.07	.796	(23.8)	–	–
T2																
Male	21	10			7			5			3			5		
		(47.6)			(33.3)			(23.8)			(14.3)			(23.8)		
Female	22	9			9			7			6			8		
		(40.9)	0.20	.658	**(40.9)**	0.27	.607	(31.8)	0.34	.558	(27.3)	–	–	(36.4)	0.80	.370
Syndrome																
T1																
AS	17	5			7			3			10			3		
		(29.4)			(41.2)			(17.6)			**(58.8)**			(17.6)		
CDC	11	5			4			7			1			2		
		(45.5)			(36.4)			**(63.6)**			(9.1)			(18.2)		
CDLS	13	6			5			3			0			4		
		(46.2)	1.13	.569	(38.5)	0.07	.967	(23.1)	–	–	(0)	–	–	(30.8)	–	–
T2																
AS	18	6			6			6			8			2		
		(33.3)			(33.3)			(33.3)			**(44.4)**			(11.1)		
CDC	12	4			3			5			0			3		
		(33.3)			(25.0)			**(41.7)**			(0)			(25.0)		
CDLS	13	9			7			1			1			8		
		(69.2)	4.74	0.94	(53.8)	–	–	(7.7)	–	–	(7.7)	–	–	(61.5)	–	–

Note: The most common health problem per subgroup is in bold. _ denotes where analysis was not conducted due to low expected frequencies.

Table I.8

The Results of T Tests Between Type of Health Problem as reported by the Health Questionnaire and Demographic Variables of Age and Ability

	Demographic Variable			
	Age		Ability[a]	
Health problem	*t*	p	*t*	p
Dental				
T1	-0.57	.575	0.49	.633
T2[c]	0.64	.526	-0.83	.422
Skin				
T1	-0.31	.760	-0.35	.738
T2	-0.77	.448	0.01	.994
Bowel				
T1	-1.14	.262	-0.31	.771
T2	1.18	.246	-1.43	.156
Epilepsy/ Seizures/ Neurological				
T1	2.19	.035*	1.08	.145
Gastrointestinal				
T2	0.57	.572	-0.84	.444

[a] Where the assumptions of normality were violated, a bootstrap was performed utilising 1000 repetitions _ denotes where chi- squared analysis was not conducted due to low expected frequencies.

**p<0.05*

Table I.9

The Frequency and Percentage of Different Types of Chronic Health Problems as reported by the Health Questionnaire. Presented Across the Whole Sample Then Split by Gender and Syndrome

Chronic Health Problem	Frequency (%)					
	Full group	Gender		Syndrome		
		Male	Female	AS	CDC	CDLS
	n=41	n=20	n=21	n=17	n=11	n=13
Dental	12 (29.3)	6 (30.0)	6 (28.6)	4 (23.5)	2 (18.2)	6 (46.2)
Skin	11 (26.8)	5 (25.0)	6 (28.6)	5 (29.4)	2 (18.2)	4 (30.8)
Gastrointestinal	6 (14.6)	2 (10.0)	4 (19.0)	2 (11.8)	1 (9.1)	3 (23.1)
Bowel	6 (14.6)	2 (10.0)	4 (19.0)	2 (11.8)	4 (36.4)	0 (0)
Epilepsy/ Seizures/ Neurological Referrals	6 (14.6)	1 (5.0)	5 (23.8)	6 (35.3)	0 (0)	0 (0)
Limb Abnormalities	4 (9.8)	2 (10.0)	2 (9.5)	0 (0)	1 (9.1)	3 (23.1)
Ear	3 (7.3)	0 (0)	3 (14.3)	1 (5.9)	1 (9.1)	1 (7.7)
Eye	3 (7.3)	2 (10.0)	1 (4.8)	0 (0)	0 (0)	3 (23.1)
Heart or Circulatory	1 (2.4)	1 (5.0)	0 (0)	0 (0)	0 (0)	1 (7.7)
Hernia	1 (2.4)	0 (0)	1 (4.8)	1 (5.9)	0 (0)	0 (0)

Table I.10
Number of Chronic Health Problems Reported on the Health Questionnaire by All Participants and Split by Gender and Syndrome

No of chronic health problems	Frequency (%)					
	All participants	Gender		Syndrome		
		Male	Female	AS	CDC	CDLS
	n=41	n=20	n=21	n=17	n=11	n=13
0	15 (36.6)	9 (45.0)	6 (28.6)	6 (35.3)	5 (45.5)	4 (30.8)
1	10 (24.4)	6 (30.0)	4 (19.0)	4 (23.5)	2 (18.2)	4 (30.8)
2	8 (19.5)	3 (15.0)	5 (23.8)	4 (23.5)	3 (27.3)	1 (7.7)
3	6 (14.6)	0	6 (28.6)	3 (17.6)	1 (9.1)	2 (7.7)
4	1 (2.4)	1 (5.0)	0 (0)	0 (0)	0 (0)	1 (7.7)
5	1 (2.4)	1 (5.0)	0 (0)	0 (0)	0 (0)	1 (7.7)

Table I.11
The Difference in Number of Chronic Health Problems on the Health Questionnaire Between Gender and Syndrome Groups

Chronic health Problems	Gender			Syndrome			
	Male n=20	Female n=21	Difference t	AS n=17	CDC n=11	CDLS n=13	Difference f
Mean (*SD*)	1.05 (1.39)	1.52 (1.21)	-1.16[a] p=.261	1.24 (1.15)	1.00 (1.10)	1.61 (1.66)	.676 p=0.515

[a] Where the assumptions of normality were violated, a bootstrap was performed utilising 1000 repetitions.

Table I.12
The Correlation Between Number of Chronic Health Problems Reported on the Health Questionnaire and Ability and Age.

Chronic health Problems	Age		Ability	
	r_s	p	r_s	p
	-0.25	.117	0.11	.481

Table I.13

The Difference Between the Total and Summary PedsQL Scores when Compared by Gender and Syndrome and the Relationship Between These PedsQl Scores and Participants Ability and Age.

PedsQL	Gender		Syndrome		Age		Ability	
	t	p	$\chi^{2\,a}$	p	r_s	p	r_s	p
Total Score	-1.14[b]	.261	2.32	.314	0.27	.076	-0.06	.694
Psychosocial health Summary	-1.68	.101	1.04	.594	0.27	.0.76	-0.26	.096
Physical health Summary [a]	-0.23[c]	.831	0.90	.636	0.18	.260	0.07	.664

[a] Kruskal Wallis test [b] Equality of variance not assumed. [c] Where the assumptions of normality were violated, a bootstrap was performed utilising 1000 repetitions

Table I.14

The Frequency of Common Health problems identified by the Health Questionnaire split by syndrome and level of Challenging behaviour

	Persistent CB			Low/No CB		
Health problem	*AS*[a] *n=7,8*	CdC n=6	CdLS n=3	*AS n=10*	CdC[a] n=5,6	CdLS n=10
Dental						
Time 1	7	2	1	5	1	4
Time 2	2	3	3	4	1	6
Skin Time 1	3	1	3	4	3	2
Time 2	2	1	3	4	2	4
Bowel						
Time 1	0	5	0	3	2	3
Time 2	3	4	0	3	1	1
Epilepsy/ Seizures/ Neurological Time 1	4	1	0	6	0	0
Gastrointestinal Time 2	0	2	2	2	1	6

Note. AS= Angelman, CdC= Cri du Chat, CdLS= Cornelia de Lange.

[a] Time 2 had 2 less participants complete the Health Questionnaire so the n is reduced to 41

Appendix J- Population Norms

Table J.1 shows the population norms score as presented by Varni et al. 2001, as reported by proxies for healthy, acutely ill and chronically ill children between 2-18 years old.

Table J.1

The PedsQL (proxy) population norms as reported by Varni, Seid and Kurtin (2001)

Scale	Mean	SD
Total Score		
Chronically ill	74.22	18.40
Acutely ill	80.42	15.26
Healthy	87.61	12.33
Physical Health		
Chronically ill	73.28	27.02
Acutely ill	81.81	20.46
Healthy	89.32	16.35
Psychosocial Health		
Chronically ill	74.80	18.16
Acutely ill	79.56	15.51
Healthy	86.58	12.79

Note. Chronically ill was defined as those who had a chronic health condition (n=638), Acutely ill were those who had attended a specialist clinic (orthopaedics, cardiology, rheumatology and diabetes) but did not have a chronic health condition (n=207). Healthy individuals were those who attended well-child checks and did not report a chronic health problem (n=730).

Appendix K- Author Guidelines

Below are the guidelines for the Journal, Volume one of this thesis was written with these in mind.

REMOVED FROM E VERSION