Quality of Life in Irish Children with Down Syndrome: A Cross-Sectional Study

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CARL Research Project

in collaboration with

DOWN SYNDROME IRELAND

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What is Community-Academic Research Links?

Community Academic Research Links (CARL) is a community engagement initiative provided by University College Cork to support the research needs of community and voluntary groups/ Civil Society Organisations (CSOs). These groups can be grassroots groups, single-issue temporary groups, but also structured community and voluntary organisations. Research for the CSO is carried out free of financial cost by student researchers.

CARL seeks to:

- provide civil society with knowledge and skills through research and education;
- provide their services on an affordable basis;
- promote and support public access to and influence on science and technology;
- create equitable and supportive partnerships with civil society organisations;
- enhance understanding among policymakers and education and research institutions of the research and education needs of civil society, and
- enhance the transferrable skills and knowledge of students, community representatives and researchers (Living Knowledge Network).

What is a CSO?

We define CSOs as groups who are non-governmental, non-profit, not representing commercial interests, and/or pursuing a common purpose in the public interest. These groups include: trade unions, NGOs, professional associations, charities, grass-roots organisations, organisations that involve citizens in local and municipal life, churches and religious committees, and so on.

Why is this report on the UCC website?

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How do I reference this report?

Author (year) *Dissertation/Project Title*, [online], Community-Academic Research Links/University College Cork, Ireland, Available from: https://www.ucc.ie/en/scishop/rr/ [Accessed: date].

How can I find out more about the Community-Academic Research Links and the Living Knowledge Network?

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Abstract

Objectives:

Our main aim was to investigate quality of life (QoL) in Irish children with Down Syndrome (DS). We evaluated the impact of chronic health conditions on QoL, and determined whether frequency of screening for these conditions impact QoL.

Design:

This is a quantitative, cross-sectional study.

Setting:

This research was community-based, involving children registered with Down Syndrome Ireland (DSI) Munster and Dublin branches.

Participants:

Target population was parents of children with DS aged 8-18 years old, living in the Republic of Ireland (ROI). A parental survey, the "Kidscreen-27 Health-Related Quality of Life (HRQOL) Questionnaire for Children and Adolescents aged from 8-18 years" (Parents' version), with additional demographic and health questions was distributed via email by DSI branches to members.

-**Exclusion criteria:** Age >18 years, or residing outside ROI.

Primary & Secondary Outcome Measures:

Overall HRQOL score for each child was calculated, and sub-category scores within the questionnaire evaluated. Impact of demographic variables, chronic health conditions, and screening intensity for these conditions on HRQOL scores was investigated, along with effect of number of chronic conditions on screening intensity. Analysis included a subgroup of children aged 0-8 years with DS, and parental qualitative feedback.

Results:

Mean QoL scores were below that of the normative population, with social QoL scores significantly lower than the reference data. Those with higher frequency of medical screens had lower mean QoL. The subgroup with multiple underlying health conditions had the highest frequency of medical screens, and lowest mean QoL scores.

Conclusion:

Regular screening of children with DS minimises impact of co-morbidities. Unsurprisingly, those with high numbers of co-morbidities have low QoL, but frequent screening and medical appointments also impact QoL. Greater focus on friendship-building and social interaction is hugely important. Strategies for improving screening in the community at General Practitioner (GP) and Area Medical Official clinics should be explored.

Introduction

Down Syndrome

DS is the commonest genetic cause of intellectual disability. It is characterised by an extra copy of chromosome 21 ("trisomy 21"), which significantly affects physical and cognitive development (1). DS is associated with increased risk of chronic health conditions, such as congenital heart and gastro-intestinal anomalies, congenital and acquired impairments of hearing and vision, haematological, respiratory, musculoskeletal, and endocrine problems (2). These conditions can impair the everyday functioning and well-being of the child with DS, from both a health perspective, and a social and developmental perspective (3).

Incidence, Life Expectancy and DSI

Ireland's incidence of DS is approximately 1:500 live births, the highest in Europe (4). Average life expectancy of a baby with DS has increased from 12 years in 1949 to 60 years today, due to medical care improvements (5). Nowadays, children with DS undergo cardiac investigations at birth (ECG and ECHO tests), along with vision, hearing, haematologic and thyroid screens, and have follow-up appointments or surgery if any abnormalities are identified (6). Physiotherapy, speech and language and occupational therapy provided by the Health Service Executive (HSE) in Ireland via regional disability services have improved health outcomes in these children in recent years (7).

DSI, a volunteer-led organisation which advocates for the welfare of people with DS, estimates there are approximately 7,000 people with DS living in Ireland (8). DSI was established to provide a support network for parents of people with DS due to a lack of services available for people with this condition at the time (9).

Quality of Life

The World Health Organisation defines QoL as a person's perception of their position in life, affected by their health, psychological state, and social relationships (10). Health-related QoL (HRQOL) examines the link between the individual's health, and their perceived QoL (11).

Research to date shows QoL in children with DS is generally lower than in children who do not have additional needs (12). The burden of illness associated with chronic medical conditions in children with DS may contribute to poorer QoL in these children (3). Depression and anxiety are common among children with DS, leading to emotional and behavioural problems, negatively affecting their QoL (13). Studies show that social functioning, school support and psychosocial health are lower in children with DS compared with their peers (12), resulting in lower QoL, while strong social relationships are associated with better QoL (14).

Certain demographic variables affect QoL in children. For example, one study found children living in urban areas reported a better QoL than rural dwellers (15), while another found that children with a mother of a high level of education had higher QoL scores (16). An Australian study found that adolescents with DS had lower QoL than children with DS across all dimensions explored, including physical and psychological well-being, social and peer support, and autonomy (17).

Screening for Chronic Conditions

The need for screening protocols in the management of children with DS was identified by a study in the National Children's Hospital, Dublin, demonstrating that treatable medical conditions occur at a high rate in children and adolescents with DS (18)https://pubmed.ncbi.nlm.nih.gov/15835512/. Early intervention positively impacts patient outcomes; regular health checks (well-child exams) are important to make sure underlying conditions are not missed (19). Regular screening is the most effective way of reducing the impact of chronic disease, as earlier detection allows for earlier treatment (20). This is supported by a US study which found, within their population of children with DS diagnosed with coeliac disease, 82% were diagnosed on routine screening as opposed to symptomatic presentations (21).

The Down Syndrome Medical Interest Group (DSMIG) is an organisation consisting of healthcare professionals from Britain and the ROI. They produce regularly updated guidelines to standardise management of people with DS, promoting follow-up and continuity of care from birth to adulthood (see Appendix 2).

A US study from 2021 showed that less than 50% of children with DS had preventative activities performed by their primary care provider over four years, including well-child checks, hearing tests, and vaccinations (22). An audit on thyroid screening practice in University Hospital Limerick revealed only 23% of children with DS aged from 12-17 years who attended the hospital had been screened in accordance with guidelines (23).

Importance of this Research Area

Despite the growing recognition of the importance of QoL across many chronic medical conditions, there is a paucity of existing studies examining QoL in children with DS (15). Even fewer studies examine the effect of chronic conditions on QoL, or indeed the impact of regular health screening on QoL. This is an important area of research, as the goal in management of all chronic conditions should be to maximise QoL (19).

Aims and Objectives

The primary aim of this project is to explore QoL in children with DS living with chronic health conditions in Ireland.

The objectives of this research are:

1. To assess QoL in children with DS living in Ireland, and to determine which groups have the lowest QoL.

2. To investigate whether the presence of chronic health conditions impact QoL.

3. To assess if intensity of screening for complications of DS impact QoL.

Methods

Study Ethics:

Ethical approval was received from the Social Research Ethics Committee (SREC) of the Cork Teaching Hospitals in July 2020 (see Appendix 4).

Study Design:

This is a quantitative, cross-sectional survey assessing QoL in children with DS in Ireland, using a parent completed, validated QoL questionnaire (detailed below).

-Inclusion criteria: Parents of children with DS living in Ireland aged 8-18 years of age.

-Exclusion criteria: Age >18 years, or residing outside ROI.

Study Methods:

Participants:

The target population was parents of children with DS aged 8-18 years old, living in the ROI. Participants were recruited with the help of DSI (Munster and Dublin branches) via Facebook and email.

Data was collected between September 1st - October 31st, 2020.

Demographics questionnaire: Data was collected on each child to include:

-Parental characteristics: Age, gender, area of residence, education.

-Child characteristics: Age, place in the family, number of chronic health conditions, number of visits to hospital/GP in past year.

Screening Intensity: Number of screening tests performed in childhood.

QoL questionnaire: "Kidscreen-27" (Parents' Version), is a validated parental questionnaire, widely used across Europe (24) to study QoL in children. Parents completed this online questionnaire, comprising twenty-seven questions presented as statements, grouped together

in five categories: “Physical Activity and Health,” “General Mood and Feelings,” “Family and Free Time,” “Peers and Social Support,” and “School and Learning” (see Appendix 1).

The two questionnaires were combined and distributed using “Google Forms.” No participant identifiers were collected. Respondents consented to use of their data before completing the questionnaire.

Statistical Analysis

Questionnaires were collected and stored using “Google Forms,” and data was analysed using IBM SPSS-version 28.

All responses were examined for missing data, and for children outside the ages of the study protocol.

Statistical significance was designated at the conventional level of $p<0.05$.

Descriptive statistics used included mean and standard deviation (SD) for parametric distributions.

An overall raw QoL score for each child was calculated by totalling the Likert-scale responses for each participant. Negatively formulated items were recoded using SPSS software to have scorings from 1-5 with higher values indicating higher QoL, i.e. 5 was substituted for 1 on the Likert scale, and vice versa. Raw scores were transformed into z-values and then t-values using specialised syntax provided by “Kidscreen” designers, where the difference between the raw total score and the population mean was divided by (the standard deviation divided by the square root of the sample size), for each participant. This standardised each individual score for comparison with the normative data; t-values represent the difference between population means, in units of standard error (25). Normative data provided by “Kidscreen” comprised the mean HRQOL score, and subcategory mean scores, for children aged 8-18 years from twelve European countries.

Independent t-tests compared QoL scores of the study population with the European mean score (total and sub-categories). The same method was used to examine the effect of screening intensity on QoL, and the effect of different demographic variables, and chronic health conditions, on QoL. One-way Analysis of Variance explored the effect of demographic variables on QoL and screening intensity. Correlation studies explored the relationship

between screening intensity and QoL, and between screening intensity and number of chronic health conditions.

Timeline

The study took place over a period of two years. Literature was reviewed from October-December 2019, ethical approval obtained in July 2020, and data collected, analysed, and presented between September 2020-December 2021.

Results

Number of emails sent: 729

Responses: 105 **Number excluded:** 15

Response rate: 12%

Figure 1

Demographic Variables: Parents and Children

Parents who completed the questionnaire:			
Gender			
Female: 99%	Male: 1%		
Age			
<41 years: 9%	41-55: 82%	>55: 9%	
Employment:			
Full-time: 22%	Part-time: 40%	Within the home: 36%	Unemployed: 2%
Demographics of Children:			
Age:			
Mean Age: 11 years	Range: 4-18 years	SD: 3.5	
Residence:			
Rurally: 43%	Urban area: 57%		
Mean Number of Health Conditions by Age:			
Number of children <13 years old: 35	Number of teenagers: 55		
Mean number of health conditions in children <13 years old: 1.4	Mean no. health conditions in teenagers: 1.4		
SD in children <13 years old: 0.5	SD in teenagers: 0.5		

HRQOL Scores

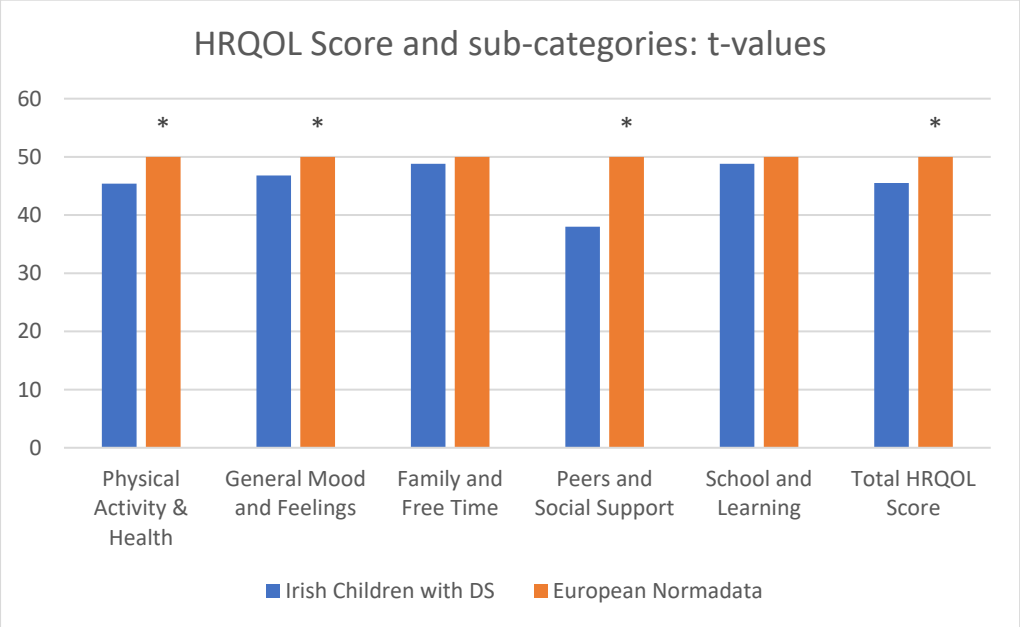
Data was analysed within two groups; an n=90 population of children with DS aged 0-18, and an n=78 subgroup of those aged 8-18. Children under 8 were included, due to small sample size.

The mean total HRQOL score for Irish children with DS aged 0-18 was 45.5 (SD:7.4, range:31-66), significantly lower than the European score of 50 (SD:10, range:45-55), $t(89)= -5.8$, $p<.05$

All sub-category scores in the study population were below the European sub-category means of 50; "Physical Well-being:" 45.4, "Psychological Well-being:" 46.8, "Autonomy and Parents:" 48.8, "Peers and Social Support:" 38, "School and Learning:" 48.8.

Of these sub-categories, "Physical well-being," ($t(89)= -4.7$, $p<.05$)), "Psychological well-being," ($t(89)= -3.4$, $p<.05$), and "Peers and Social Support" ($t(89)= -8.9$, $p<.05$) were significantly below European means (Figure 2 (*)).

Figure 2



Subgroup

There was no significant difference in the HRQOL scores of the subgroup of Irish children with DS aged 8-18 compared to the entire study population; their total HRQOL score was 45.6, with similar sub-category scores, so we included children under 8 years of age in the analysis.

Demographics

Demographic variables explored had no significant effect on QoL or screening intensity of participants in this study.

Area of residence: Of children surveyed, 57% lived in an urban area, and 43% lived in a rural area. Urban-dwellers had slightly lower QoL (M=44.4, SD=6.4) than rural-dwellers (M=47, SD=8.3), but this did not reach statistical significance, $t(88)=1.64$, $p>.05$.

Screening intensity was not significantly different between urban (M=21.4 screens, SD=21.5), and rural settings (M=23.8 screens, SD=22.9), $t(88)=0.51$, $p>.05$.

Parental education: Of those surveyed, 13% of parents completed their education in secondary school, and 87% of parents had a third-level qualification. Those who had finished with secondary school (M=45.1, SD=11.5) did not report a better QoL in their children than those with a third-level degree (M=45.6, SD=6.6), $t(88)= -0.231$, $p>.05$.

Children of parents with secondary school education alone had a lower screening intensity (M=16.1, SD=17.2) compared with children of parents with a university degree, (M=23.4, SD=22.6), but this did not reach statistical significance, $t(88)= -1.1$, $p>.05$.

Place in the family: There was no significant effect for the child with DS being born as the mother's first child (32% of those surveyed), or in a subsequent pregnancy (68% of those surveyed), on their QoL. First-born children (M=45.3, SD=6) did not report a better QoL than those born in subsequent pregnancies (M=45.6, SD=7.9), $t(88)= -0.2$, $p>.05$.

Neither did first-born children have significantly more childhood screens (M=24.4, SD=20.2) than those born in later pregnancies (M=21.5, SD=22.9), $t(88)=0.576$, $p>.05$.

Child's Age: QoL did not correlate with the child's age, $r(88)=0.15$, $p>.05$.

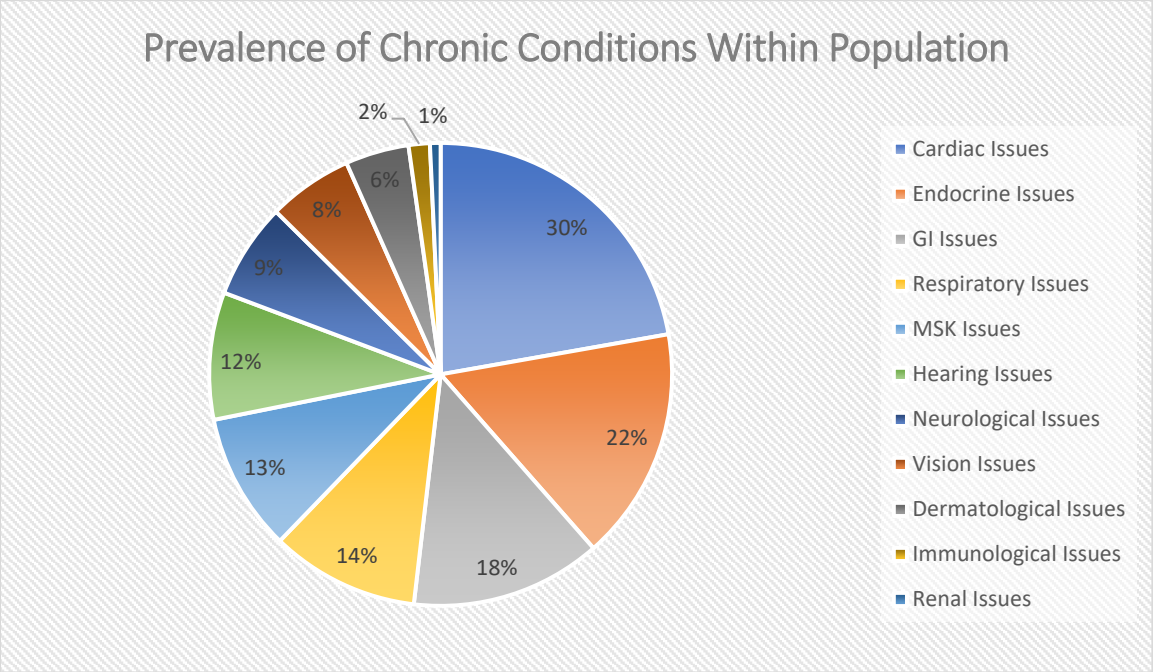
No association was found between the age of the child and the presence of chronic health conditions, $t(88)=0.072$, $p>.05$.

Effect of Health Conditions on QoL

Of the children studied, 57% had at least one underlying chronic health condition (Figure 3).

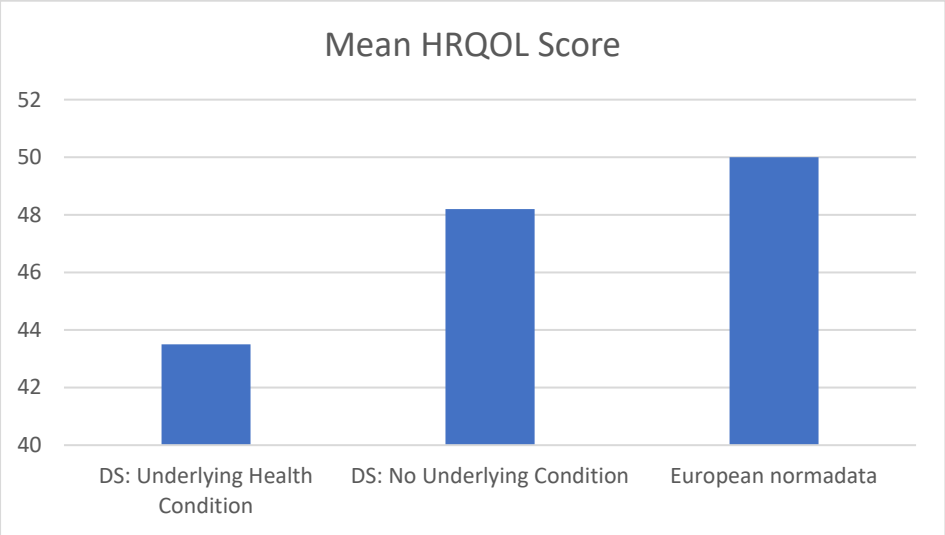
Mean number of conditions per child was 1 (range:0-9 conditions, SD:1.5).

Figure 3



QoL of children with at least one underlying health condition (M=43.5, SD=7.15) was significantly lower than of those without underlying conditions (M=48.2, SD=6.85), t(88)= -3.12, p<.05 (Figure 4).

Figure 4



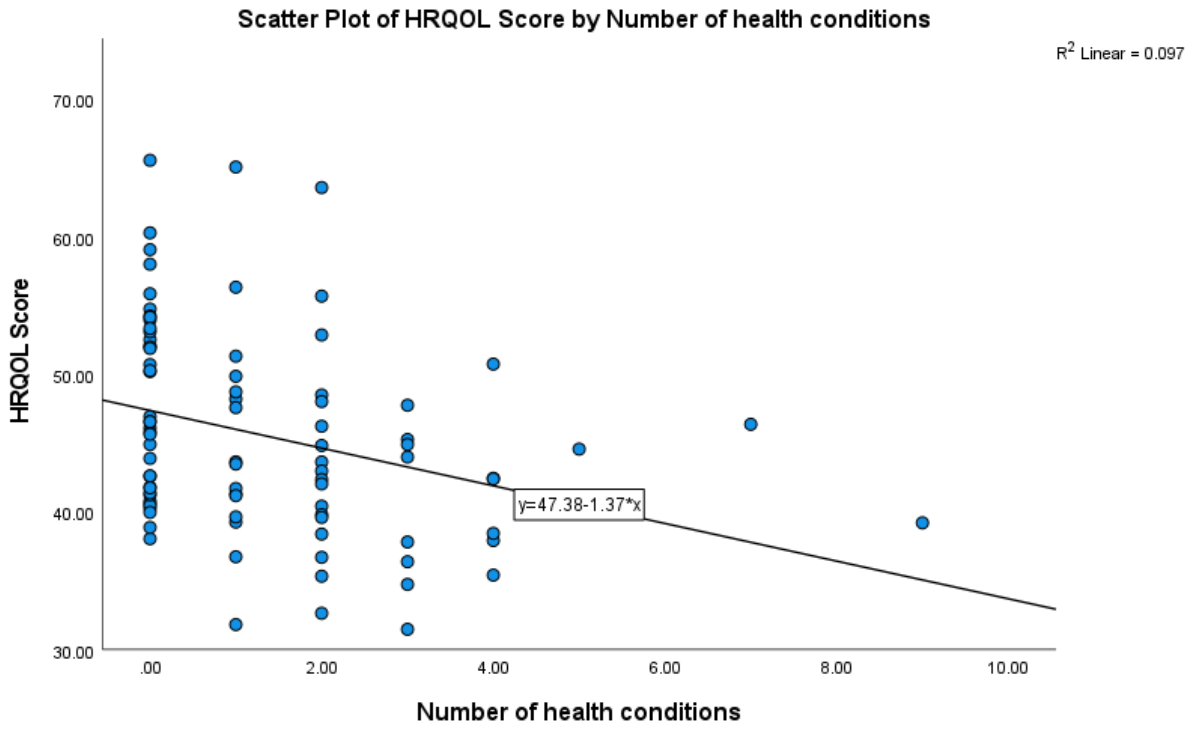
Of those studied, 87% had at least one GP check-up over the past year, while 53% had at least one hospital visit during that time, and 50% had at least one visit to both. Nine children (10%) had no health check in the past year. Only one child not seen by healthcare services in the last year had an underlying condition.

Mean age of those who had a well-check was 11 years, SD: 3.5, while the mean age of those who did not have a well-check was 12 years, SD: 3.3.
There was no correlation between age and having a well-check; age did not influence whether the child was examined by their GP or in hospital, r(88)=0.068, p>.05.

Correlation between number of chronic health conditions and QoL:

Mean number of hospital and GP visits combined within the past year was 5 (SD:4.5, range: 0-18). As number of underlying conditions increased, QoL scores decreased: r(88)= -0.301, p<.05 (Figure 5).

Figure 5



Screening Intensity

Mean number of health screens in childhood was 23 (range:0-92, SD:22).

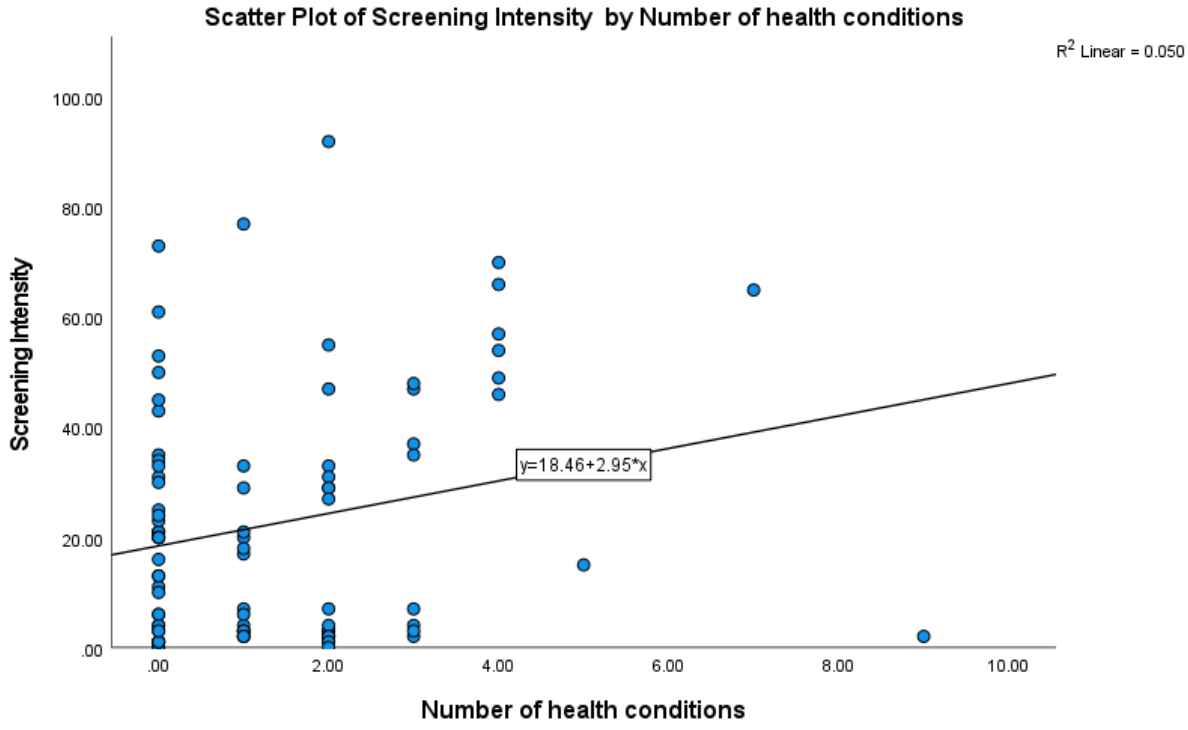
Thyroid function was not tested regularly in 21% of children, 39% had never been tested for coeliac disease, 41% never tested for atlanto-axial subluxation, 48% never tested for obstructive sleep apnoea (OSA), and 81% never tested for juvenile idiopathic arthritis (JIA).

Vision was not tested regularly in 3% of children, and 10% did not have regular hearing tests.

As the number of conditions a child was screened for increased, their QoL decreased: r(88) = -.248, p <.05.

Higher numbers of underlying health conditions in the child correlated with higher numbers of health screens, r(88)=.224, p<.05 (Figure 6).

Figure 6



Parents

Difficulties in securing appointments for screening tests were cited by 14% of parents responding to the study, either due to their concerns being ignored by healthcare professionals, or due to the Covid-19 pandemic. For example, one child who has heart, hearing and vision tests annually had all tests cancelled within the previous year, due to Covid-19.

Many parents reported making pleading phone calls to services to request assessment for their child. One mother felt her child was "let down by the HSE" and that her phone calls requesting audiology and ophthalmic reviews were constantly ignored. Another said her child's hearing and vision was followed up locally, but appointments were "scarce." Children with heart conditions were followed up in Crumlin, and there was greater parental satisfaction

with cardiac services than with other services, e.g. audiology, vision. One mother was frustrated by her GP's refusal to refer her daughter with sore knees for an arthritis review, and also reported taking her daughter for an ophthalmology review in Derry, where visual issues were spotted which had gone undiagnosed in HSE services and which qualified her for a visually impaired teacher in school.

Parental Knowledge

Of respondents, 16% reported their child was not tested for cardiac conditions in their lifetime, while 33% did not know whether their child was screened for at least one of ten common conditions associated with DS.

Only one child had been tested for nine out of the ten conditions specified (all except blood disorders).

Key findings:

1. Our study sample of Irish children with DS aged between 0-18, and between 8-18 years, have a significantly lower QoL than their European counterparts. The subcategories, "Physical Well-being," "Psychological Well-being" and in particular "Peers and Social Support," were significantly lower for both groups.
2. QoL in children with at least one underlying health condition (57% of children studied) is significantly lower than for those without underlying health conditions.
3. No well-check was performed on 10% of our population during the past year.
4. The higher the intensity of screening for complications of DS in childhood, the lower the child's QoL was in this study.
5. Number of chronic conditions was positively correlated with screening intensity.
6. Of parents studied, 33% answered "don't know" as to whether their child was screened for at least one of ten common conditions associated with DS.
7. Communication difficulties and Covid-19 were barriers highlighted by 14% of parents to securing screening tests.
8. Demographic variables had no significant effect on the child's QoL or screening intensity.

Discussion

This research was undertaken to explore QoL in children with DS, and to pinpoint various markers which best determine their QoL. Ireland's incidence of DS is the highest in Europe, yet much of the existing research into QoL in people with DS, and its determinants, has been carried out in other European countries and in Australia. This sample of children with DS aged between 0-18, and between 8-18, had lower QoL than their European counterparts. An overall HRQOL score of 45.5 was recorded for our population (0-18), significantly below that of the reference data score of 50.

This finding is echoed in much of the existing research into QoL in children with DS; QoL scores were lower in the population of children with DS than the reference population across the literature. One study investigated QoL in young people with DS in Australia aged 16-31 using the "Kidscreen" questionnaire, reporting a mean QoL score of 42 for the population, showing low QoL scores persist into adulthood (20).

A number of contributors to low QoL scores were identified in the study above (15). They found that of those surveyed, children with several friends scored higher on their questionnaire, identifying paucity of friendships as a marker of lower QoL. This is one of the main findings of our study: Irish children with DS recorded very low scores in the "Peers" category of our questionnaire, M=37.9, M(reference data)=50. This study similarly found that decreased social outlets negatively impacted QoL. Lack of support outside the family and decreased social contact trumped other factors investigated, such as health conditions and physical wellbeing, as the best marker of QoL in the child. The focus of many interventions in the management of children with DS is on the treatment of their chronic conditions (26), however further research into the domains of friendship-building and social integration within the community is clearly warranted.

Haddad and colleagues above found that children with three or more friends had a better QoL (15). This is corroborated by another Australian study, which found that one-third of children with DS did not have any friends (27). When examining participation of children with DS in play and leisure activities they found most were restricted to solitary pastimes, a likely contributor to low QoL scores in the "Peers" category of our own questionnaire. This is echoed by another study (16), which found that people with DS spent most of their time in the company of their parents and siblings, and little time with friends, negatively affecting

their QoL. Similarly, Pikora and colleagues (3) found the burden of chronic medical conditions associated with DS restricted the child's participation in community activities and employment opportunities, having a detrimental effect on their QoL. Therefore, to improve leisure participation, children with DS require continuous health surveillance. Research demonstrates that certain approaches towards participation are more effective than others; for example, communication tools such as iPads allow children to tell stories and part-take in conversations where speech difficulties are otherwise a barrier (14), and regular dance classes improved physical and mental health, and social functioning, in children with DS in one study (28). During adolescence, children develop significantly both physically and emotionally, and the gap between children with DS and their peers widens; Shields and colleagues attribute lower QoL scores in terms of friendship within their study to a growing disparity in social skills between children with DS and their peers as they get older (17). Therefore, to ameliorate QoL in terms of social support and friendships, interventions should be targeted at adolescence. A qualitative study looking at QoL from the perspective of young adults with DS themselves found community participation was important to them; therefore more projects like the "Field of Dreams" founded by DS Cork (29), which helps young people with DS gradually transition into the workforce by providing them with skills classes and work placements, should be undertaken, and also provided to younger teenagers. Another study found young people with DS desired inclusion and integration with their peers (30); this could be achieved through amalgamation of Special Olympics clubs with community sports clubs, to encourage integration between children with DS and their typically developing peers.

One possible contributor to lower social QoL scores is whether the child attends a "mainstream" or "special educational needs" (SEN) school. A UK study found teenagers with DS educated in mainstream schools had improved communication levels and fewer behavioural problems than those in SEN schools (31), however few studies have examined the effect of the child's schooling on their QoL. Further research should examine differences in social QoL scores based on the child's school environment, determining whether this influences QoL, directing parents as to the best education route for their child.

Previous studies found that family socio-demographics impact QoL of people with DS, however our demographic analysis did not reach statistical significance. Haddad and colleagues found a significant effect for the child's area of residence, where their QoL and health status decreased the further away from a city they lived (15). The discrepancy between

our results could be due to better provision of services for children with DS in Ireland compared to Australia, with access to supports in Ireland more equal between rural and urban dwellers (32). The RESONATE study in Australia identified a disparity in healthcare provisions between regional areas and urban settings, likely contributing to rural dwellers with DS there having lower QoL (33); DSI has twenty-five regional branches, which may contribute to better services nationwide.

In terms of chronic conditions, 57% of children studied had at least one underlying health condition. They had significantly lower QoL scores than those without underlying conditions. Haddad and colleagues (15) reported a similar detrimental effect of the burden of both physical and mental health issues on "Kidscreen" QoL scores within their population. These findings are not surprising, nor are they unique to the population of people with DS; a European study found significantly lower QoL in children and adolescents with chronic health conditions than their healthy peers (34), corroborated by studies on children with demyelinating disease (35), and fibromyalgia (36).

Early diagnosis of medical co-morbidities improves long-term health and QoL, and contributes to the life expectancy of 60 years in people with DS today (19). These children require regular health checks to detect underlying conditions pre-symptomatically. Provision of regular screening visits is the most effective method of decreasing the impact of chronic diseases, and improving health outcomes (20). In our study we identified nine children, i.e. 10% of our population, who were lost to follow-up and did not have a well-child check by a clinician over the year preceding our questionnaire distribution. A US study on health supervision in DS found very low compliance with preventative screening guidelines; adherence to ophthalmology screening was 33%, audiology 43%, and thyroid 61%. Adherence rates were higher in children referred to paediatricians and specialists (37), reflecting our finding that more ill children had more screening tests performed. A UK study in adults with DS had a similar low screening rate (38). Although these studies reflect our findings, the high incidence of DS in Ireland means urgent action is needed to ensure these children are not lost to follow-up.

The DSMIG recommends that children with DS have annual thyroid and hearing tests, vision tests every two years, and other tests at various intervals (39). We found that children screened more frequently for medical complications of DS had lower QoL scores, but also had more underlying medical conditions, suggesting these children were more likely to be

examined if they were symptomatic. A 2014 audit in University Hospital Limerick found their compliance with thyroid screening guidelines was 53%; our research shows that little has changed since then (23). Among our population, many children did not undergo regular screening for various conditions; 39% had never been tested for coeliac disease, which is worrying as this condition is eighteen times more prevalent in people with DS than in the general population (40). Since early intervention prevents severe complications arising in the management of medical problems in children with DS (33), a change in practice is needed going forward; the focus should be on early diagnosis of medical problems, before significant symptoms affecting QoL develop.

A US study identified long-term benefits of routine input from the primary care provider in the care of children with DS, in terms of parent education, and referral to specialists and early intervention services. This is associated with better health outcomes (41), and presents an opportunity to empower GPs in Ireland as a central point of care for children with DS. In our study, communication difficulties impeded access to screening tests, with parents feeling ignored by health services, and frustrated with long waiting lists for appointments. Many reported their child was only tested because the parent persistently requested they were seen for medical review. Parents also referred to Covid-19-related interruptions to services, which is mirrored across all areas of healthcare; one study showed that 40-80% of cancer patients in England were affected by a decrease in provision of health services since the pandemic began, highlighting Covid-associated interruptions are not unique to the care of children with DS (42). This study identified the important role of the GP in mitigating long-term risks associated with such interruptions to healthcare. Therefore, further research should examine awareness amongst Irish GPs of the DSMIG guidelines.

The fact that 16% of parents reported that their child was not tested for cardiac conditions in their lifetime suggests that we are failing as healthcare providers to adequately educate parents of children with DS; all children are screened for cardiac complications at birth (43). Of course, recall bias is also a factor, as some of these ECHOs will have taken place ten or more years ago, depending on the age of the child. Similarly, 33% of parents answered “don’t know” as to whether their child was screened for at least one of ten common conditions associated with DS, suggesting they are either unaware their child is at risk of developing one of these conditions, or have not been kept abreast of medical investigations carried out on their child. Only one parent reported her child had been tested for nine out of ten conditions

specified; the parent is a GP, benefitting from more medical knowledge than a parent of a non-medical background.

An unexpected diagnosis of DS in one's child can be overwhelming, and a study examining parental coping in the early period after their child's diagnosis found that communication styles among clinicians strongly influenced parental adaptation to their child's health requirements. Many parents in that study would have appreciated access to a liaison worker when caring for their child's medical needs (44). With that in mind, improved communication, and access to services as highlighted by respondents to our questionnaire may improve health outcomes for these children. One way of facilitating this is to create a specialised clinic for children/adults with DS within the HSE, where all screening tests (ophthalmology, thyroid, etc.) may be performed in one location, at regular intervals, e.g. annually, during the birthday month. A service of this kind exists in Tallaght University Hospital, a "one stop" health surveillance clinic for children with DS which has demonstrated substantial improvements in adherence to guidelines since its foundation (45https://adc.bmj.com/content/104/Suppl_3/A216.1). Parental satisfaction with the clinic was surveyed to be 100% (46), all the more significant considering the frustrations reported by parents in our study. This could potentially remove some of the barriers cited by parents above, such as difficulty accessing services, lack of awareness of tests, and lack of well-child exams. While some studies suggest adopting medical checklists as preventative health routines (47) and having people with DS treated by an age-focused primary care provider (22), there is little reference to a centralised approach such as this within the literature, and therefore this represents a fresh strategy to a global issue which urgently needs to be addressed.

Strengths and Limitations

The validated questionnaire used allowed for comparison of results against European normative data. Being self-completed and available online meant our project was largely unaffected by Covid-19 restrictions, and facilitated greater anonymity among participants.

As well as quantitative data, we collected qualitative feedback from parents outlining their frustration with lack of access to timely screening visits; therefore this is an important first

step in understanding factors contributing to QoL in children with DS, and it underlies the need for more studies.

Our study's most notable limitation was the small sample size of ninety participants. The response rate was lower than expected, and some responses had to be excluded from analysis as the child in question was over 18. Wide advertising and promotion on social media, along with reminder emails, were used, but our response rate was disappointing. There was a very high level of education amongst our population, reflecting some responder bias which may affect the generalisability of our results.

Another limitation was the use of a proxy questionnaire to evaluate the child's QoL. This assumes the parent's responses accurately reflect the child's feelings and situation, which may not always be the case. A qualitative section for responses from the child would give them a voice in further studies of QoL in DS.

Further research

A similar cross-sectional study aimed at people with DS aged over 18 living in Ireland is an important follow-up area to study, to identify discrepancies in QoL over different age brackets.
Further research should examine other variables, such as how gender differences affect QoL and screening intensity, and whether QoL scores are affected by mainstream or SEN education. Another important area of research is exploration of particular conditions which have the biggest impact on QoL, allowing for more thorough screening for such co-morbidities. GP awareness of the DSMIG screening guidelines should be evaluated as a follow-up to this study.

Conclusion

QoL in people with DS is an important area of continuing research, especially considering Ireland's high incidence of DS. Our population of ninety Irish children with DS had lower QoL scores than the European normative data, while the burden of medical illness in these children decreased their QoL scores even further. A higher intensity of preventative screening tests was associated with lower QoL and greater number of medical co-morbidities, suggesting children are not being screened until they are symptomatic.

Our results highlight the need for more regular screening of children with DS for chronic conditions impacting QoL, such as through a centralised service to which these children are referred annually, and where all investigations may be performed at the same visit. Management of children with DS children tends to focus on chronic conditions, however further research into domains of friendship building and social integration are equally important. Since illness has been identified as a barrier to leisure participation, screening interventions may also have a positive impact on the low QoL scores reported by our population from a social and friendship point of view.

This is the first study to explore QoL in children with DS living in Ireland. Those with the most underlying health conditions have the lowest QoL, and also the highest number of health screens. There remains an unacceptably high number of children not being screened according to guidelines, putting them at risk of future health issues and reduced QoL as they get older. There is a need to improve parental knowledge of screening protocols, and to adopt a more co-ordinated approach to screening.

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Student Declaration

This study was designed by my supervisor, Dr. Louise Gibson, and myself. Data was collected and analysed, and this paper written, by us alone.

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Thanks also to Down Syndrome Ireland's regional branches, for distributing and promoting our questionnaire.

Appendix 1

Questionnaire

Information Leaflet for Parents

Quality of life in Irish children with Down Syndrome: a cross-sectional study.

I would like to invite you to take part in a research study. Before you decide, you need to understand why we are undertaking this research, and what it will involve for you. Please take time to read the following information carefully. Ask questions if any parts are not clear, or if you would like more information. Take time to decide whether to take part.

WHO I AM, AND WHAT THIS STUDY IS ABOUT:

My name is Ella Curtin, and I am a 4th year medical student from UCC. As part of my studies, I am required to complete a research project, and I have chosen to examine quality of life in children with Down Syndrome. I aim to investigate any correlation between quality of life and chronic health conditions in children with Down Syndrome, and whether screening for these conditions enhances quality of life in these children, and also whether demographics, such as where the child lives, affects their quality of life.

WHAT WILL TAKING PART INVOLVE?

You will be asked to take 10-15 minutes to fill out a simple questionnaire, the 'Kidscreen-27 Health-Related Quality of Life Questionnaire for Children and Adolescents aged from 8 to 18 years,' available through your email, or on the Down Syndrome Cork Facebook page. This will include questions on your child's activity levels, school life, general happiness, and physical well-being. There will be 5 options for each question, e.g. from 'never' to 'always.' There will also be some preliminary questions on your family's demographics, such as your child's age and area of residence, along with a few questions on your child's medical history.

WHY HAVE YOU BEEN INVITED TO TAKE PART?

As a parent of a child with Down Syndrome, and a member of your local Down Syndrome Ireland branch, you are welcome to participate.

DO YOU HAVE TO TAKE PART?

Participation is completely voluntary; you have the right to refuse participation, refuse any question, and withdraw within two weeks of participation. Your data will then be destroyed.

WHAT ARE THE POSSIBLE RISKS AND BENEFITS OF TAKING PART? WHAT IF THERE IS A PROBLEM?

No harm is anticipated, however some of the questions included are of a personal nature. If you have concerns about your child following your participation in the study, it is advisable to see your GP, and share your concerns with them. If sharing about your experiences in this way causes you any distress, you can talk to a fellow parent of a child with Down Syndrome by emailing info@downsyndrome.ie.

WILL TAKING PART BE CONFIDENTIAL?

Data will be kept confidential for the duration of the study. No patient identifiers will be used in data analysis, and access will be limited to my supervisor, Dr. Louise Gibson, and I.

WHAT WILL HAPPEN TO THE RESULTS OF THE STUDY?

Results of the study will be presented in my thesis. They will be seen by my supervisor, a second marker, and an external examiner. The thesis may be read by future students on the course, and may also be published in a research journal.

WHO HAS REVIEWED THIS STUDY?

Approval must be given by the Social Research Ethics Committee of UCC before studies like this can take place.

WHO SHOULD YOU CONTACT FOR FURTHER INFORMATION?

Dr. Louise Gibson, Senior Lecturer/Consultant Community Paediatrician, CUH
L.Gibson@ucc.ie

Ella Curtin, Medical Student, 4th Year
117345663@umail.ucc.ie

If you agree to take part in the study, please complete and submit the following questionnaire.

THANK YOU,

Ella Curtin

Questionnaire

Preliminary questions relating to parent of child with Down Syndrome/ child themselves

In questions related to the parent, we are looking for age, gender etc. of just the parent completing the questionnaire

Please tick the answer which applies

- **Parent's: Age:** Under 40…….. 41-55…….. 55+……..
- **Parent's Gender:** M…….. F……
- **Residence:** Urban……. Suburban…… Rural …….
- **Employment:**

  Full-time……. Part-time……. Within the home……. Unemployed…….
- **Your level of education?**

  Secondary School Completed……3rd level completed ……
  Postgraduate……
- **Were you aware that your baby had Down Syndrome during your pregnancy?**

  Yes……. No…….
- **What age is your child with Down Syndrome?** …….
- **Was this child your first-born, or from a subsequent pregnancy?**

  ……

- **Does your child have any underlying health conditions? If so, give details:**

_____

_____

_____

_____

_____

_____

_____

- **How many hospital visits has he/she had during the past year?**

…….

- **How many GP visits has he/she had during the past year?**

…….

<u>Screening</u>: (answer yes (y) or no (n))

Was your child ever tested for:

1) **Coeliac disease: _____**
2) **Cervical Spine subluxation: _____**
3) **Hypothyroidism or hyperthyroidism: _____**
4) **Vision problems: _____**
5) **Hearing loss: _____**
6) **Epilepsy: _____**
7) **Blood disorders: (please specify which) _____**

8) **Sleep apnoea:**_____

9) **Arthritis:**_____

10) **Cardiac condition:**_____

11) **Other:**_____

If you answered yes to any of the above, please include details below of when this was tested for; if it was tested more than once, state how regularly the test has been carried out. Please also include the result of the test(s).

KIDSCREEN-27

Health Questionnaire for Children and Young People

Parent Version

Dear Parents,

How is your child? How does she/he feel? This is what we would like to know from you.

Please answer the following questions to the best of your knowledge, ensuring that the answers you give reflect the perspective of your child. Please try to remember your child's experiences over the last week...

1. Physical Activities and Health

1. **In general, how would your child rate her/his health?**

- ○ excellent
- ○ very good
- ○ good
- ○
- ○ fair

Thinking about the last week ...

	not at all	slightly	moderately	very	extremely
2. Has your child felt fit and well?	○	○	○	○	○
3. Has your child been physically active (e.g. running, climbing, biking)?	○	○	○	○	○
4. Has your child been able to run well?	○	○	○	○	○

Thinking about the last week ...

	never	seldom	quite often	very often	always
5. Has your child felt full of energy?	○	○	○	○	○

2. General Mood and Your Child's Feelings

Thinking about the last week…

	not at all	slightly	moderately	very	extremely
1. Has your child felt that life was enjoyable?	○	○	○	○	○

Thinking about the last week...	never	seldom	quite often	very often	always
2. Has your child been in a good mood?	○	○	○	○	○
3. Has your child had fun?	○	○	○	○	○

Thinking about the last week...	never	seldom	quite often	very often	always
4. Has your child felt sad?	○	○	○	○	○
5. Has your child felt so bad that he/she didn't want to do anything?	○	○	○	○	○
6. Has your child felt lonely?	○	○	○	○	○
7. Has your child been happy with the way he/she is?	○	○	○	○	○

3. Family and Your Child's Free Time

Thinking about the last week...	never	seldom	quite often	very often	always
1. Has your child had enough time for him/herself?	○	○	○	○	○
2. Has your child been able to do the things that he/she wants to do in his/her free time?	○	○	○	○	○
3. Has your child felt that his/her parent(s) had enough time for him/her?	○	○	○	○	○

		never	seldom	quite often	very often	always
4.	Has your child felt that his/her parent(s) treated him/her fairly?	○	○	○	○	○
5.	Has your child been able to talk to his/her parent(s) when he/she wanted to?	○	○	○	○	○
6.	Has your child had enough money to do the same things as his/her friends?	○	○	○	○	○
7.	Has your child felt that he/she had enough money for his/her expenses?	○	○	○	○	○

4. Friends

Thinking about the last week...

	never	seldom	quite often	very often	always
1. Has your child spent time with his/her friends?	○	○	○	○	○
2. Has your child had fun with his/her friends?	○	○	○	○	○
3. Have your child and his/her friends helped each other?	○	○	○	○	○
4. Has your child been able to rely on his/her friends?	○	○	○	○	○

5. School and Learning

Thinking about the last week...

	not at all	slightly	moderately	very	extremely
1. Has your child been happy at school?	○	○	○	○	○
2. Has your child got on well at school?	○	○	○	○	○

Thinking about the last week...

	never	seldom	quite often	very often	always
3. Has your child been able to pay attention?	never ○	seldom ○	quite often ○	very often ○	always ○
4. Has your child got along well with his/her teachers?	never ○	seldom ○	quite often ○	very often ○	always ○

Appendix 2

DOWN SYNDROME MEDICAL MANAGEMENT GUIDELINES

Suggested schedule of health checks taken from Guidelines Growth

	Growth	Heart	Thyroid	Sight	Hearing
Birth - 6 wks	Length / Weight / Head circumference Plot on 2011 revised Down syndrome specific charts. (Use NICAM charts for preterm babies)	Clinical Examination ECG and Echocardiogram 0-6 weeks	Routine Guthrie test	Eye Examination; Check for congenital cataract, congenital glaucoma + any other eye abnormality	National Neonatal Hearing screening
6-10 months	Growth assessment - As above at each routine visit*			Visual behaviour, check for squint	Full audiological review (Otoscopy, Impedance, Hearing thresholds)
12 months	Growth assessment - As above at each routine visit*	Dental Advice, Infective endocarditis advice/information if necessary	Full Thyroid function tests or TSH (finger prick)** yearly where available	Visual behaviour, check for squint	
18-24 months	Growth assessment - As above* Chart those ≥2years of age on BMI conversion charts if concerns about overweight.	Dental Advice and Examination of teeth Infective endocarditis advice/information if necessary	Full Thyroid function tests or TSH (finger prick)** yearly when available	Ophthalmological examination including Orthoptic screening, refraction and fundal examination and focusing ability	Full audiological review as above
3 – 3 ½ years	Growth (Height/Weight) assessment and advice*. Chart on BMI conversion charts if concerns about overweight.	Dental Advice and Examination of teeth Infective endocarditis advice/information if necessary	Full Thyroid function tests or TSH (finger prick)** yearly when available		Full audiological review as above

4 – 4 ½ years	Growth (Height/Weight) assessment and advice as above*	Dental Advice and Examination of teeth Infective endocarditis advice/information if necessary	Full Thyroid function tests or TSH (finger prick)** yearly when available	Ophthalmological examination as above	Full audiological review as above

**Encourage a healthy lifestyle (healthy eating and regular exercise) at all times*

***TSH (finger prick)- capillary whole blood thyroid stimulating hormone (TSH) sample –using one circle on National Newborn Screening Programme card)*

From age 5years to 19 years

Paediatric Medical Review Annually

Cardiology Echo in early adult life to rule out mitral valve prolapse.

Infective endocarditis information to be given later in life for those with cardiac history.

Hearing 2 yearly audiological review as above

Vision 2 yearly ophthalmological exam incl. refraction & fundal exam, & focusing ability

Thyroid 2 yearly from 5 years (venous) or TSH (fingerprick)** annually, when appropriate structures, personnel and funding are in place

A comprehensive history and careful clinical examination should be undertaken to detect other emergent health issues such as, respiratory and rheumatological complications

Updated December 2015. (SIGHT updated July 2009)

Appendix 3

STROBE Statement

	Item No	Recommendation	Page No
Title and abstract	1	(*a*) Indicate the study's design with a commonly used term in the title or the abstract	1
		(*b*) Provide in the abstract an informative and balanced summary of what was done and what was found	2, 3
Introduction			
Background/rationale	2	Explain the scientific background and rationale for the investigation being reported	4, 5, 6
Objectives	3	State specific objectives, including any prespecified hypotheses	6
Methods			
Study design	4	Present key elements of study design early in the paper	7
Setting	5	Describe the setting, locations, and relevant dates, including periods of recruitment, exposure, follow-up, and data collection	7
Participants	6	(*a*) Give the eligibility criteria, and the sources and methods of selection of participants	7
Variables	7	Clearly define all outcomes, exposures, predictors, potential confounders, and effect modifiers. Give diagnostic criteria, if applicable	7
Data sources/ measurement	8*	For each variable of interest, give sources of data and details of methods of assessment (measurement). Describe comparability of assessment methods if there is more than one group	7, 8
Bias	9	Describe any efforts to address potential sources of bias	8
Study size	10	Explain how the study size was arrived at	7
Quantitative variables	11	Explain how quantitative variables were handled in the analyses. If applicable, describe which groupings were chosen and why	7
Statistical methods	12	(*a*) Describe all statistical methods, including those used to control for confounding	8, 9
		(*b*) Describe any methods used to examine subgroups and interactions	8, 9
		(*c*) Explain how missing data were addressed	8
		(*d*) If applicable, describe analytical methods taking account of sampling strategy	N/A
		(*e*) Describe any sensitivity analyses	N/A

Results			
Participants	13*	(a) Report numbers of individuals at each stage of study—eg numbers potentially eligible, examined for eligibility, confirmed eligible, included in the study, completing follow-up, and analysed	10, 12
		(b) Give reasons for non-participation at each stage	10, 11
		(c) Consider use of a flow diagram	N/A
Descriptive data	14*	(a) Give characteristics of study participants (eg demographic, clinical, social) and information on exposures and potential confounders	10, 11, 12
		(b) Indicate number of participants with missing data for each variable of interest	N/A
Outcome data	15*	Report numbers of outcome events or summary measures	10 - 17
Main results	16	(*a*) Give unadjusted estimates and, if applicable, confounder-adjusted estimates and their precision (eg, 95% confidence interval). Make clear which confounders were adjusted for and why they were included	10 - 17
		(*b*) Report category boundaries when continuous variables were categorized	N/A
		(*c*) If relevant, consider translating estimates of relative risk into absolute risk for a meaningful time period	N/A
Other analyses	17	Report other analyses done—eg analyses of subgroups and interactions, and sensitivity analyses	12, 16, 17
Discussion			
Key results	18	Summarise key results with reference to study objectives	17
Limitations	19	Discuss limitations of the study, taking into account sources of potential bias or imprecision. Discuss both direction and magnitude of any potential bias	23
Interpretation	20	Give a cautious overall interpretation of results considering objectives, limitations, multiplicity of analyses, results from similar studies, and other relevant evidence	18, 19, 20, 21, 22
Generalisability	21	Discuss the generalisability (external validity) of the study results	19, 20, 23
Other information			
Funding	22	Give the source of funding and the role of the funders for the present study and, if applicable, for the original study on which the present article is based	N/A

*Give information separately for exposed and unexposed groups.

Appendix 4

UCC University College Cork, Ireland Coláiste na hOllscoile Corcaigh	**ETHICS APPROVAL FORM** **Social Research Ethics Committee (SREC)** **School of Medicine Sub-Committee Form**

APPLICANT(S) DETAILS

Name of UCC applicant(s)	Ella Curtin	**Date**	19/12/2019
Department / School / Research Institute / Centre / Unit / College	UCC School of Medicine	**Contact No.**	087 3618198
Correspondence Address	Coolnegera, Coachford, Co. Cork	**Email Address**	117345663@umail.ucc.ie
Name and year of course (students only)	3rd year, Medicine	**Name of supervisor(s)** (students only)	Dr. Louise Gibson, Senior Lecturer/Consultant Community Paediatrician, CUH
Is this a resubmission?	No	SREC Log No. (if known):	
What type of SREC approval are you seeking?[i]	Full approval X Outline approval ☐ Funding approval ☐		

Obtaining ethical approval from SREC does not free you from securing permissions and approvals from other institutional decision-makers and agency ethical review bodies. These bodies may accept the SREC approval, but researchers are responsible for ensuring they are compliant in advance of collecting data.

Project working title	Quality of Life in Irish Children with Down Syndrome: a Cross-Sectional Study

If this is a collaborative project / community-based participatory research project / *joint* application with another agency, please complete this additional section:

Names of research partners / civil society organisations collaborating on this project (this section must be completed for participatory / community-based participatory research studies)	CARL, 'Community Academic Research Links', UCC
Agency contact person and position	Dr. Anna Kingston, CARL co-ordinator
Agency address	School of Applied Social Studies, University College Cork, Ireland
Details of the partnership (roles, type of partnership, etc.)	Dr. Kingston from CARL is helping to facilitate my project; she sourced a supervisor for me and helped me to contact Down Syndrome Ireland.

DESCRIPTION OF THE PROJECT

*Ethical review requires that you **reflect** and seek to **anticipate** ethical issues that may arise,*

rather than reproduce copious text from existing research proposals into these boxes.

*Entries should be **concise** and relevant to the point / question.*

24. Very brief description of your study (15-25 words max.)
[i.e. This is a qualitative study of primary school teachers' attitudes towards religious teaching using focus groups to collect original data]

This is a quantitative study of quality of life (QoL) in Irish children with Down Syndrome (DS), using a self-completion questionnaire to collect original data.

25. What is your study about? (100-200 words max.)

For my project, I will investigate QoL in Irish children (8-18 years old) with DS, and the effect of chronic health conditions and the need for multiple appointments and tests on their overall QoL.

DS is the most common chromosomal abnormality in humans, caused by an additional chromosome 21. Children with DS have an increased risk of health complications. They are more susceptible to hearing and vision problems, under-active thyroid, sleep apnoea, coeliac disease, gastro-intestinal issues, cervical spine subluxation, epilepsy, blood disorders, arthritis, and recurrent respiratory infections. Chronic conditions such as these can impair their everyday functioning, such as attendance at school and participation in extra-curricular activities. In addition chronic health conditions can reduce job prospects in adult life or putting them at risk of developing 'depressive symptomology' approaching adulthood.
Regular screening for these conditions is largely at the discretion of parents. They must take the initiative and request such tests from their healthcare provider; this can result in late and severe presentation to health services, where they were not aware of pertinent tests. Few studies have examined QoL in DS, and the impact of regular health screening and chronic health conditions on QoL.

26. What are your research questions?[ii]

1. What is quality of life like in Irish children with Down Syndrome?
2. Does the presence of chronic health conditions impact quality of life?
3. Does screening for complications of Down Syndrome impact quality of life?
4. Are there are any differences in screening and quality of life in different populations of children with Down Syndrome?

27. Brief description and justification of methods and measures to be used (attach questionnaire / interview protocol / discussion guide / etc. for **full** SREC approval. **Not** required for SREC outline approval)

A validated, widely used, self-completed cross-sectional questionnaire called the 'Kidscreen-27 Health-Related Quality of Life Questionnaire for Children and Adolescents aged from 8 to 18 years,' (Parents' version) will be used gather our data. Parents will complete this online questionnaire, which consists of 27 headings, e.g. 'General Mood and Feelings,' and tick one of 5 possible answers to each question, e.g. from 'excellent' to 'poor.'

This survey is clear and simple, and only takes 10-15 minutes to complete.
In addition the parents will complete some questions related to demographics, underlying medical conditions, date of most recent health screen.
1) Demographics: Parents' age, gender, area of residence, profession, level of education, knowledge of DS during pregnancy, number of children with DS, age of child with DS, child with DS born in first/subsequent pregnancy, child's underlying health conditions, child's number of visits to hospital and GP/year.
2) Screening: Whether or not their Down Syndrome child was tested for complications of DS, and if so, when, and what was the result.
Initial statistical analysis will compare mean QoL scores with the normative data, and further analysis will identify variables associated with lower QoL.

28. Participants (recruitment methods, number, age, gender, exclusion/inclusion criteria, detail permissions to be sought / secured already)

Participants: My target population is parents of children with Down Syndrome aged 8-18 years old, living in the Republic of Ireland. My estimated response rate is 15% (based on the response rate of a similar research project being undertaken by a student in Fourth year, on the same target population), so I am expecting a sample size of 225 families. Participants will be recruited using the DS Ireland database of families of children with DS. DS Ireland currently has 1500 member families across Ireland.

Inclusion Criteria: Child must be between 8 and 18 years of age for parents to part-take.

Data Collection: The 'Kidscreen-27 Health-Related QoL Questionnaire for Children and Adolescents (aged from 8 to 18 years,') will be emailed by regional branches of Down Syndrome Ireland to their members, and posted on the Down Syndrome Cork Facebook page, from 01/09/20 - 31/10/20. The questionnaire will be promoted using Facebook posts for wider participation.

The anonymous data will be stored on a password protected computer for analysis.

The participants will complete an informed consent form giving permission to use the data elicited from the questionnaire prior to completing the questionnaire and supplementary questions.

29. Concise statement of *anticipated* ethical issues raised by your project. How do you intend to deal with them? Please address *all* items where your answers fell into a shaded box in the self-evaluation above. (350 words max.)

1) Consent- A consent form will be provided with the online questionnaire and information sheet; prospective candidates will have to tick a box online before accessing the questionnaire to indicate they consent to the use of their data.
2) Anonymity- Responses to the questionnaire will be anonymised through coding, and data will be stored in a password protected file, in a password protected computer, for confidentiality.
3) I do not envisage any risks to participants, as there is no intervention involved.

30. Data: **(a) What type of data will you be storing?** **(b) How and where will you store your data?**[iii] (provide details for both physical *and* electronic documents). [iv] **(c) For how long will you store the data?** (A minimum storage period of 10 years is required) **(d) Who will have access to the dataset?** (*Sample* prompts: If you plan to make your raw research dataset available publicly as part of the open data movement, please address your protocol here. For collaborative/community-based participatory research, please address issues such as shared ownership of data, publication of findings, etc. If your funder contractually requires you to give them access to the 'raw' dataset, examine relevant implications, including appropriate anonymisation, protocols for secure access to the dataset, etc.). **(e) If you are planning to analyse an existing dataset, please outline how the original consent process allows for your analysis.**
(a) I will be storing quantitative data. **(b)** Data will be stored in a password-protected file on a password-protected computer. **(c)** Data will be stored for 10 years, and then destroyed. **(d)** Access to the data will be limited to my supervisor and myself. No no patient identifiers, such as names, will be collected.

31. Arrangements for informing participants about the nature of the study (cf. Question 3)
An information sheet will be included with the online questionnaire, which potential candidates may read before undertaking to participate in the study. (See enclosed).

32. How you will obtain Informed Consent? (cf. Question 4 - attach relevant form(s))
A consent form will be provided with the online questionnaire and information sheet; prospective candidates will have to tick a box online before accessing the questionnaire to indicate they consent to the use of their data.

33. Outline of debriefing process (cf. Question 9). If you answered YES to Questions 19a or 19b, give details here. State what you will advise participants to do if they should experience problems (e.g. who to contact for help).
Upon completion of the questionnaire, a message will appear thanking the person for their participation, and reminding them that all data will be treated with full anonymity, with no patient identifiers being published.

34. Estimated start date and duration of project
Project to commence (i.e. data collection) in September 2020, and should take roughly one year to complete.
35. Additional information of relevance to your application

This project is taking place in collaboration with Down Syndrome Ireland, who fully support the research and its goals.

36. Declarations	
I/we agree that should there be unexpected ethical issues arising during the course of this study, that I/we will utilise my/our professional/disciplinary code of ethics, and/or notify UCC SREC, where appropriate	Yes / No
I/we have consulted the UCC *Code of Research Conduct* (2016) and believe my/our proposal is in line with its requirements	Yes / No
I/we have consulted the UCC *Child Protection Policy* and believe my/our proposal is in line with its requirements	Yes / No / NA

37. Signatures	
UCC Applicant(s)	**Academic Supervisor / Tutor / Principal Investigator** (where applicable)
	
Date: 21/04/20	Date: 21/04/20

[i] *Full approval* is required for study design, data collection *and* data analysis. *Outline approval* is for activities such as early-stage research design and participatory processes where there is *no* data collection at this time. For *outline* approvals, a further application will be necessary should there be a subsequent data collection phase. *Funding approval* should be ticked if your funding grant requires approval within a short time frame (e.g. 2 months).

[ii] If your study approach does not normally require that research questions are set in advance, please comment in this box.

[iii] Data management should follow the FAIR guiding principles (Findability, Accessibility, Interoperability & Reusability). See, for example, Wilkinson, M. D. *and collegaues* (2016) *The FAIR Guiding Principles for Scientific Data Management and Stewardship*. Full text: http://www.nature.com/articles/sdata201618.

[iv] It is required that all staff and student researchers store those data which are required to replicate research findings, and the information required to enable re-use of data. Details of the UCC policy on research data storage can be found in section 8 of the *Code of Research Conduct* (2016): https://www.ucc.ie/en/media/research/researchatucc/documents/UCCCodeofResearchConduct.pdf. UCC's staff IT service can assist with encrypting staff laptop/desktop computers (see http://www.ucc.ie/en/it/services/encryptionlaptop/) and with providing storage space on a secure Network Attached Server for your data (UCC staff only - see http://www.ucc.ie/en/it/services/networkfilestorenas/). SREC advises against storing research data on cloud-based storage services.