Prevalence and Impact of X-ray Screening for Atlantoaxial Instability in Children with Down Syndrome

Daniel Shenoda

CARL Research Project
in collaboration with
Down Syndrome Cork

Name of student	Daniel Shenoda
Academic supervisor(s):	Dr. Louise Gibson
Department	Paediatrics
Name of community group:	Down Syndrome Cork
Name of community group liaison person:	Karen O’Sullivan
Name and year of course:	Graduate Entry Medicine 4
Date submitted:	10/02/2021

TABLE OF CONTENTS:

ABSTRACT

Introduction:

Atlantoaxial instability (AAI) is defined as excessive movement between the first and second cervical vertebra. This can lead to spinal cord compression, resulting in myelopathic and radicular symptoms. These symptoms occur in 1-2% of the down syndrome (DS) population. DS athletes are often required to undergo pre-participation X-ray screening for AAI to help identify those at risk. However, the evidence for plain cervical spine X-ray as a form of screening is poor.

Aim:

This project aims to explore the use, sensitivity and specificity of X-ray screening for AAI in the Irish DS population, to investigate the prevalence of symptomatic AAI, to identify the rate of sport exclusion based on an abnormal X-ray and to investigate whether neuromuscular conditions, type of schooling or activity level are significantly correlated to an abnormal X-ray.

Method:

This is a nation-wide cross-sectional online survey. It was rolled out via email by Down Syndrome Ireland to 1511 registered families. The survey asked parents if their child has had to undergo x-ray screening, the result of the x-ray, the impact of the result and if they developed symptoms. The survey also assessed parental knowledge of symptoms of AAI.

Results:

Out of 240 responders, 7 responders had symptomatic AAI and 5 of these had normal X-rays (29% sensitivity). Chi-Squared testing showed no variables significantly correlated with having an abnormal X-ray. Of the total group of 146 who underwent X-ray pre-participation screening, 20 had abnormal results and were excluded from playing their desired sports (specificity 86.7 %).

Conclusion:

X-ray screening for AAI in Ireland is very common and can result in the exclusion of many from participating in sports. Plain X-ray has low sensitivity. Therefore, it is not an optimal

screening tool for AAI in asymptomatic children. More should be done to improve parental knowledge of this condition.

INTRODUCTION

Background:

AAI is defined as laxity between the first and second vertebrae [1]. It is reported to occur in up to 27% of the down syndrome population [1]. Most cases are asymptomatic but rarely (1-2%) it can present with myelopathic symptoms due to spinal cord impingement [1, 2]. Special Olympics require all athletes with DS to undergo a pre participation screening before being allowed to take part in what they call high risk sports ray to rule out asymptomatic AAI [3]. Some of these sports are gymnastics, diving, butterfly stroke and diving start in swimming, pentathlon, soccer, rugby and high jump [2]. The pre-participation screening involves plain cervical radiographs taken in the neutral, flexed and extended positions [3]. From these radiographs the Anterior Atlanto Odontoid Distance (AAOD), also known as the Atlantodens Interval (ADI) is measured [3]. However, the methodology for this measurement is prone to error and there is inconsistency in the literature on what measurement is diagnostic of AAI [1, 4]. For example, 3 studies report that an ADI measurement of 3mm or more is diagnostic of AAI [5, 6, 7]. 4 studies report that an ADI measurement of 4mm or more is diagnostic [8, 9, 10, 11]. 1 study reports a measurement of 4.5mm or more is diagnostic [12]. 2 studies report a measurement of 5mm, or more is diagnostic [13, 14].

This inconsistency has resulted in the exclusion of many asymptomatic children with down syndrome from playing their desired sports [4]. In addition to this it has also resulted in the exposure of children to unnecessary radiation. It is well known that the thyroid gland is one of the most radiosensitive organs in the human body [15]. It has also been shown that radiation exposure during childhood is a recognized environmental risk factor for thyroid cancer [16}. One study proposed that it would be better if children with down syndrome were regularly assessed via history and physical examination for symptoms of AAI along with parental education about early signs [17]. The British Gymnastics program has adopted a pre-participation screening for AAI in children with down syndrome that does not involve an X-ray [18]. It involves a physical exam to check for normal neck flexion, good muscular control of the head, and the absence of symptoms of progressive myopathy [18]. In fact, the American academy of pediatrics also recommends that only symptomatic children with down syndrome should be X-rayed [19]. This would mean that physicians and parents have to be vigilant in identifying symptomatic AAI.

Rationale:

In Ireland, this issue has caused some confusion among parents of children with down syndrome. They do not know if they should be exposing their children to X-rays whenever Special Olympics or other sport organizations require such. There has never been a study done that looked at the prevalence of the use of X-ray as a form of pre-participation screening among children with down syndrome or the efficacy of such in Ireland. For this reason, this project aims to explore the use of X-ray and its sensitivity and specificity in screening for symptomatic AAI in Ireland and hopefully provide some answers to parents of children with down syndrome.

Aims and objectives:

Primary objectives:

This project aims to explore the use of neck X-rays as a form of pre-participation screening for AAI in children with down syndrome. This project seeks to determine the prevalence of this practice in Ireland and its impact on sport exclusion. This projects also aims to answer questions like: at what age are children with down syndrome asked to get this X-ray? and are they asked to get this once only or periodically in order to continue playing sports? This project also aims to determine the prevalence of symptomatic AAI in Ireland and explore what factors they have in common.

Secondary objectives:

This project also aims to look for associations with having an abnormal (AAI) X-ray. For example, is level of exercise, type of schooling (special school, mainstream), and/or underlying neuromuscular comorbidities associated with having AAI on X-ray? In addition, this project also seeks to assess parental knowledge of symptoms of AAI and what is their primary source of information when it comes to this. This project also seeks to determine if parental level of education and/or source of information is associated with their knowledge of symptoms of AAI.

METHODOLOGY

Study Design:

This was a nation-wide cross-sectional online survey study. It was done in collaboration with Community Academic Research Links (CARL) and Down Syndrome Cork. CARL is a community engagement program at University College Cork (UCC) that connects Civil Society Organizations (CSO) to students who are interested in doing a research project that would directly benefit the community organization. See Appendix A for more information on CARL. The inclusion criteria for this study were:

i) Confirmed diagnosis of DS
ii) Age 17 years or younger
iii) Registered with Down Syndrome Ireland

Participants:

The population of study was children with DS. The online survey was sent out via email by a representative of Down Syndrome Cork to 1511 Irish families (parents/guardians) registered with Down Syndrome Ireland.

Study Measures:

Due to the novelty of this study, a newly developed standardized questionnaire survey was created. See Appendix B for questionnaire. The questionnaire measured the prevalence of X-ray screening for AAI among children with DS in Ireland, the prevalence of symptomatic AAI, the rate of sports exclusion based on an abnormal X-ray, the sensitivity and specificity of X-ray screening for AAI. The questionnaire was also used to look for correlation between activity level or comorbidities with having an abnormal X-ray. It also assessed parental knowledge of symptoms of AAI.

Procedures:

The online questionnaire was created on Survey Hero. Survey Hero is a free online survey building website. It captures responses as they come in and tabulates results. For a small fee it allows the user to download the results onto excel sheets for statistical analysis. This was done in this study. The questionnaire was comprised of 19 items on 1 screen. Items were not randomized. Adaptive questioning was not used as this would have costed additional fees. Not all questions were compulsory as not all questions were relevant to every participant. However, certain items were compulsory for completion of the survey. For example, on the last item participants needed to click “I agree” to a statement which said, “by submitting this survey I consent for my responses to be used in this study.” Participants were able to review and change their answers but not after they have submitted the survey. The questionnaire was not a closed survey. Although the survey was not password protected, it was accessible only to parents who received a unique link through their emails. The link was not posted on social media or any websites. Prior to wide distribution of the online questionnaire, the survey was piloted for usability and technical functionality among a select few parents and staff at Down Syndrome Cork.

After the questionnaire was finalized on Survey Hero it was emailed to a Down Syndrome Cork representative who on July 1st, 2018 emailed it along with a participation information document (Appendix C) to the 1511 families who met the inclusion criteria. This was the initial and only contact with the potential participants as there were no advertisements or incentives used to increase response rate. The participation information document explained the purpose of the study, who the primary and co-investigators are, the length of time of the survey, what data is stored and where and for how long, and that completion of the survey is voluntary. On September 1st, 2018, after 240 parents anonymously submitted their responses, the data was downloaded from Survey Hero onto a password protected lap top accessible only to the primary and co-investigators.

Out of the 1511 participants emailed, the survey was viewed 309 times and completed by 240 people. Although cookies and IP addresses were not used, limiting multiple entrees from the same individual was done by only allowing participants one submission per browser. As mentioned above not all questions were relevant to every participant; however, some participants did not answer questions that did apply to them. In these instances, those participants were not included in the corresponding statistical analysis.

Study Ethics:

This project was formally approved by the Clinical Research Ethics Committee (CREC) of the Cork teaching hospitals. See Appendix D for ethical approval and Appendix E for the Checklist for Reporting Results of Internet E-Surveys (CHERRIES).

Data Analysis:

The data was first saved on excel sheets, where preliminary analysis was done. This initial analysis consisted of finding the prevalence of X-ray screening, age at first X-ray, rate of sport exclusion, prevalence of symptomatic AAI, sensitivity and specificity of X-ray screening. After that, the data was inputted into SPSS for Chi squared testing. Chi squared testing was used to look for correlation between activity level or comorbidities with having an abnormal X-ray. It was also used to look for correlations between knowledge of symptoms of AAI and their source of information and their educational level.

RESULTS

Prevalence of X-ray Pre-participation Screening for AAI:

Out of the 1511 families that were sent the online questionnaire, 240 (16%) responded. Form the 240 responders, 146 (61%) were required to undergo X-ray screening before being able to participate in sports, 88 were not required to undergo any form of screening, and 5 were screened via history and/or physical examination (Figure 1).

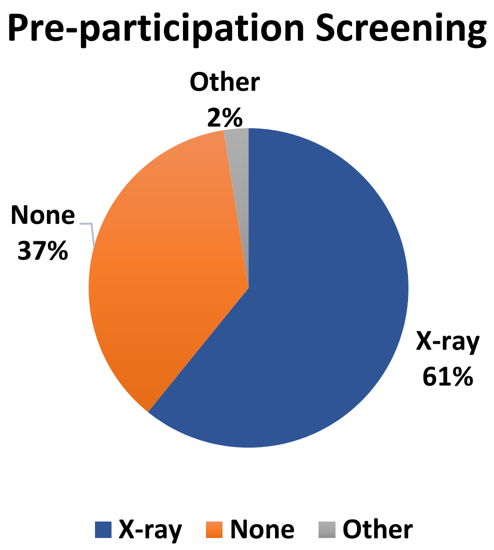


Figure 1. Pie chart showing type of pre-participation screening children were required to undergo. Note: Other refers to children whose pre-participation screening only consisted of history and/or physical examination

97 (66%) of children required to get an X-ray as part of their pre-participation screening, were only required to undergo 1 X-ray, 27 (19%) were required to get 2 X-rays, 12 (8%) were required to undergo 3 or more X-rays, and 6 (4%) did not respond (Figure 2).

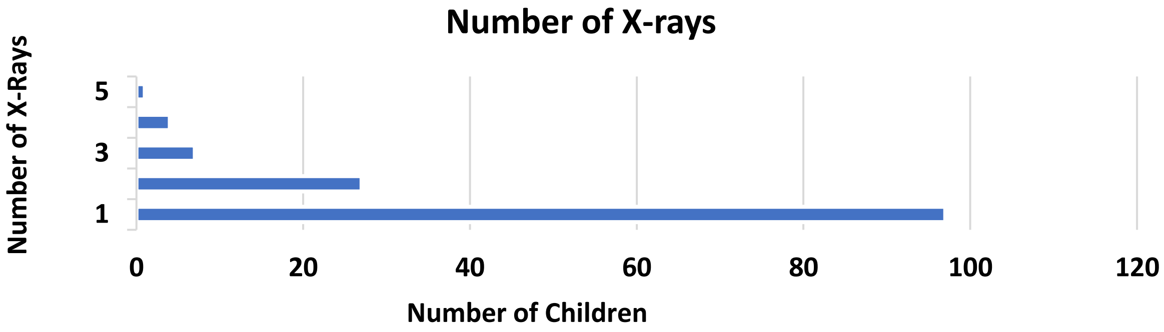


Figure 2. Bar graph showing the number of X-rays children with down syndrome were required to undergo as part of their pre-participation screening

Among the 146 children required to get a pre-participation X-ray, the average age at time of the first X-ray was 6.5 years old, with some children having their first X-ray as early as 1, 2 and 3 years old (Figure 3).

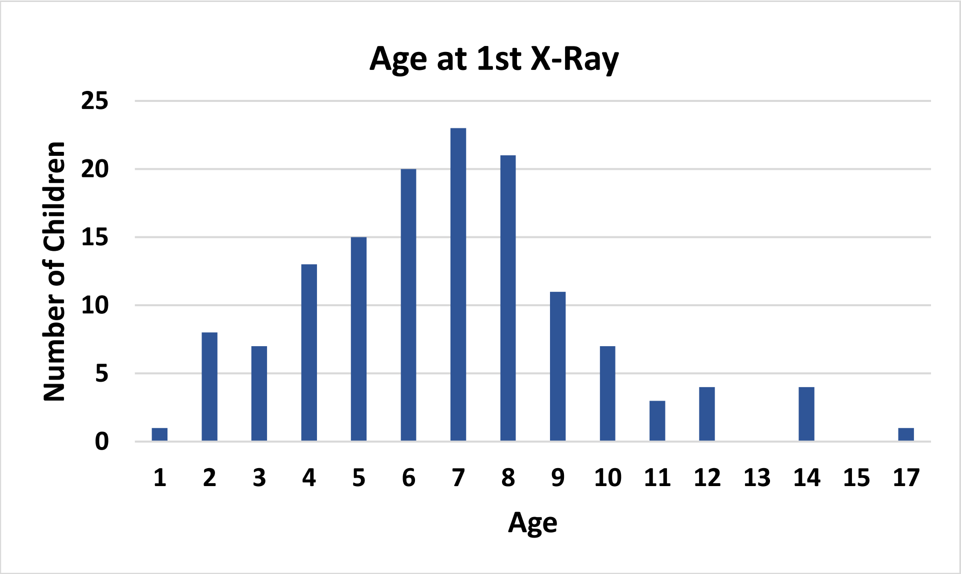


Figure 3. Bar chart showing the ages of children at the time of their first X-ray

Impact of X-ray:

Out of the 146 children who were required to get an X-ray, 20 (14%) had abnormal (asymptomatic AAI) X-rays and were restricted from playing their desired sports. 122 (84%) had normal X-rays and 4 (3%) did not respond.

Prevalence of Symptomatic AAI:

Out of the 240 total responders, 7 (3%) were diagnosed with symptomatic AAI. Comparing their activity levels and medical comorbidities, there is nothing that they all had in common. See Table 1 for a visual summery of these 7 children.

Table 1. Chart showing the X-ray results, activity level and medical comorbidities of the 7 children with symptomatic AAI.

	X- Ray Result	**Participation**	**Activity Level**	**Medical Issues**
1	Normal	Participated	>4 days/week	Cardiac, Hearing
2	Normal	Participated	3 days/week	Thyroid, vision, Hearing
3	Normal	Participated	2 days/week	Cardiac
4	Normal	Participated	2 days/week	Hearing
5	Normal	Participated	2 days/week	GI, MSK
6	AAI	Restricted	3 days/week	GI, Derm, Thyroid, Vision, hearing
7	AAI	Restricted	<1 day/week	Cardiac, Vision, Hearing

Sensitivity and Specificity:

Given the information presented in table 1, sensitivity of X-ray as a screening tool for developing symptomatic AAI was calculated to be 29%. Specificity was calculated to be 87%. This is because out of the 122 children with normal X-rays, 5 had symptomatic AAI, and from the 20 children with abnormal X-rays, only 2 had symptoms of AAI.

Associations with Abnormal X-rays:

Chi-squared testing for associations with having an abnormal X-ray revealed no significant findings. Children with a lower level of activity were more likely to have an abnormal X-ray; however, this was not statistically significant (Table 2).

Table 2. Chart showing Chi-squared test for 3 different variables and their association with having an abnormal X-ray

Variables	Categories	Chi- Squared	P value
Neuromuscular Conditions	• Yes • No	0.053	0.818
Type of School	• Special School • Mainstream School	0.020	0.888
Level of Activity	• < 1 day per week • 2 days per week • 3 days per week • >4 days per week	6.363	0.095

Parental Knowledge of Symptoms of AAI:

58% of parents were only able to identify 2 or less symptoms of AAI out of 6. 200 parents responded to a question regarding their source of information. 38% of parents used the internet as their source of information, 22% got their information from a doctor, 18% guessed, and 14% from another parent, and 8% from various other sources like teachers, physios, leaflets and Special Olympics.

Chi-squared testing found no significant associations between parental level of education or source of information with ability to identify symptoms of AAI (Table 3). On average, parents with postgraduate education did better than those with post-secondary education, and parents with post-secondary education did better than those with secondary education; however, this was not statistically significant. When it came to source of information, parents who used the internet did best, then those who got the information from another parent, then those who guessed, then those who got it from a doctor; however, this was also not statistically significant.

Table 3. Chart showing Chi-squared test for 2 different variables and their association with ability to identify 3 or more symptoms of AAI.

Variables	Categories	Chi- Squared	P value
Level of Education	1. Postgraduate 2. Post-Secondary 3. Secondary School	3.077	0.215
Source of Information	1. Internet 2. Other Parent 3. Guess 4. Doctor	7.753	0.051

DISCUSSION

Main Findings:

The use of X-ray as a form of pre-participation screening for AAI in children with down syndrome is very common (61% of responders). This practice has led to the exclusion of asymptomatic children from participating in their desired sports. This accounts for 14% of children who have an X-ray. From 240 responders, 7 (3%) had symptomatic AAI. The sensitivity of a neck X-ray as a screening tool for developing symptomatic AAI is 29% while its specificity is 87%. Chi squared analysis found no statistically significant association between neuromuscular conditions, level of activity and type of schooling with having an abnormal X-ray. Parental knowledge of symptoms of AAI is quite poor with 58% of parents only capable of identifying 2 or less symptoms out of 6. Chi squared analysis found no statistically significant associations between parents' source of information and/or level of education with their ability to identify symptoms of AAI.

Exploring the Use of X-ray for Pre-participation Screening in Ireland:

As was stated above, the use of X-ray as a form of pre-participation screening for AAI in Ireland is quite common. Most (66%) of the children who had to undergo an x-ray did so only once. This indicates that children in Ireland are not often required to undergo periodic neck X-rays in order to continue playing sports. This is consistent with the recommendations from the American Academy of pediatrics who recommend that children should not be routinely X-rayed unless symptoms develop [19]. From the 146 children who were X-rayed the average age at first X-ray was 6.5 years old; however, there was children as young as 1 and 2 years old who were X-rayed. This is quite concerning, not only because children should not be X-rayed unless they are symptomatic but also because before the age of 3 vertebral bone mineralization is not sufficiently adequate for an accurate radiological evaluation [20]. Therefore, these children are being exposed to harmful radiation with no benefit.

Symptomatic AAI:

From 240 responders, 7 (3%) had symptomatic AAI. According to the literature the prevalence of symptomatic AAI in children with down syndrome is 1-2% [1]. This could mean that the prevalence of symptomatic AAI is slightly higher in Ireland. However, it is more likely that this study is overestimating the true prevalence due to response bias.

As was mentioned above, the sensitivity of neck X-ray as a form of screening for the development of symptomatic AAI is 29% while its specificity is 87%. This is simply not good enough to be a screening tool. This is precisely why asymptomatic children should not be X-rayed. In fact, 1 of the 7 children who developed symptomatic AAI was X-rayed after she developed symptoms and her X-ray was not diagnostic of AAI. It was not until she had an MRI that she received her diagnosis.

Parental Knowledge of Symptoms of AAI:

Parental knowledge of symptoms of AAI was poor, with more than half (58%) unable to identify more than 2 symptoms. This lack of knowledge is a serious issue. It is serious because children who are symptomatic are the ones who need to be X-rayed. For this reason, parents should be vigilant and capable of identifying symptoms such as change in gait, neck pain/stiffness, bowel/bladder dysfunction, weakness, hypotonia, hyperreflexia, radicular and myelopathic symptoms [21, 22].

When the parents were asked about their source of information regarding their knowledge of the symptoms of AAI, on average parents who said internet did best, then those who put other parents, then those who guessed and then those who put doctor. Not only did parents who used the internet did best, but it was also the most common source of information among parents. Although this was not statistically significant it shows that if the correct information was online and parents knew where to find it, their knowledge of AAI symptoms would be much better. Websites like downsyndromecork.ie and downsyndrome.ie would be great places for such information. Seminars and workshops hosted by Down Syndrome Cork would also be a great way for parents to learn more about the topic from experts and from other parents.

Strengths and Limitations:

Strengths:

One of the strengths of this project was the large number of responses. The response rate was 16%, 240 responders out of 1511. This is quite good for an online questionnaire, especially considering the absence of advertisements or incentives. Future research among children with down syndrome through Down Syndrome Ireland can be extremely successful if the appropriate advertisements, education and incentives are used to increase the overall engagement of parents with research.

This project was also the first of its kind. Not only was this project the first to explore the use of X-ray as a form of pre-participation screening in Ireland, but it was also the first time Down Syndrome Ireland used their data base of registered members to send out a nation-wide online survey for research purposes.

Finally, the impact this project has on the community is a huge strength. This is a CARL project done in collaboration with Down Syndrome Cork. The purpose of the project is to answer a research question posed to Down Syndrome Cork by parents of children with down syndrome. Down Syndrome Cork are a non-profit community organization caring for children with down syndrome and supporting their families. They do not have the human resources to conduct research. For this reason, this research project really benefits the community.

Limitations:

One of the major limitations to this study is responder bias. Parents whose children were affected by symptomatic AAI were more likely to respond. This is evident in that 3% of responders had symptomatic AAI which is more than what is in the literature. Also, parents with a higher level of education were more likely to participate in the study. 56% of responders had post graduate education, 32% had post-secondary education and 12% had secondary education. Another limitation to this study was that not every question was completed. Not every question applied to everyone so there were some questions that did not need to be completed by everyone; however, some responders who should have responded to such questions did not. The final limitation to this study is that not every child with down syndrome in Ireland is registered with Down Syndrome Ireland.

Future Research:

Future research on AAI should explore the use of MRI as a diagnostic tool for symptomatic AAI in order to limit radiation. Future research should also look to see if children with down syndrome are more likely to suffer harm from radiation than their counterparts. Finally, there is a huge potential for research through Down Syndrome Ireland among families impacted by down syndrome. This would be extremely beneficial for the community.

CONCLUSION

In conclusion, the use of X-ray as a form of pre-participation screening for AAI in children with down syndrome is very common. It has led to the exclusion of many asymptomatic children from participating in their desired sports. It is a poor screening method and should only be used for diagnostic purposes in symptomatic children. More research should be done on the use of MRI as a diagnostic tool in symptomatic patients. Parents of children with down syndrome lack crucial knowledge of the symptoms of AAI and for that reason more should be done to educate them on the topic.

REFERENCES

1. Tomlinson C, Campbell A, Hurley A, Fenton E, Heron N. Sport Preparticipation Screening for Asymptomatic Atlantoaxial Instability in Patients With Down Syndrome. Clinical Journal of Sport Medicine. 2018;.
2. Cohen W. Atlantoaxial Instability. Archives of Pediatrics & Adolescent Medicine. 1998;152(2).
3. Brockmeyer D. Down Syndrome and Craniovertebral Instability. Pediatric Neurosurgery. 1999;31(2):71-77.
4. Myśliwiec A, Posłuszny A, Saulicz E, Doroniewicz I, Linek P, Wolny T et al. Atlanto-Axial Instability in People with Down's Syndrome and its Impact on the Ability to Perform Sports Activities – A Review. Journal of Human Kinetics. 2015;48(1):17-24.
5. El-Khouri M, Mourão M, Tobo A, Battistella L, Herrero C, Riberto M. Prevalence of Atlanto-Occipital and Atlantoaxial Instability in Adults with Down Syndrome. World Neurosurgery. 2014;82(1-2):215-218.
6. Ali F, Al-Bustan M, Al-Busairi W, Al-Mulla F, Esbaita E. Cervical spine abnormalities associated with Down syndrome. International Orthopaedics. 2006;30(4):284-289.
7. Roy M, Baxter M, Roy A. Atlantoaxial Instability in down Syndrome-Guidelines for Screening and Detection. Journal of the Royal Society of Medicine. 1990;83(7):433-435.
8. Morton R, Khan M, Murray-Leslie C, Elliott S. Atlantoaxial instability in Down's syndrome: a five year follow up study. Archives of Disease in Childhood. 1995;72(2):115-119.
9. Cremers M, Ramos L, Bol E, van Gijn J. Radiological assessment of the atlantoaxial distance in Down's syndrome. Archives of Disease in Childhood. 1993;69(3):347-350.
10. Cremers M, Bol E, de Roos F, van Gijn J. Risk of sports activities in children with Down's syndrome and atlantoaxial instability. The Lancet. 1993;342(8870):511-514.
11. Cremers M, Beijer H. No Relation Between General Laxity and Atlantoaxial Instability in Children with Down Syndrome. Journal of Pediatric Orthopaedics. 1993;13(3):318-321.
12. Selby K, Newton R, Gupta S, Hunt L. Clinical predictors and radiological reliability in atlantoaxial subluxation in Down's syndrome. Archives of Disease in Childhood. 1991;66(7):876-878.

13. Pueschel S, Scola F, Pezzullo J. A Longitudinal Study of Atlanto-Dens Relationships in Asymptomatic Individuals With Down Syndrome. Journal of Pediatric Orthopaedics. 1992;12(6):831.
14. Pueschel S, Scola F. Atlantoaxial instability in individuals with Down syndrome: epidemiologic, radiographic, and clinical studies. Pediatrics: American Academy of Pediatrics. 1987;80(4):555-60.
15. Zhang Y, Chen Y, Huang H, Sandler J, Dai M, Ma S et al. Diagnostic radiography exposure increases the risk for thyroid microcarcinoma. European Journal of Cancer Prevention. 2015;24(5):439-446
16. Sinnott B, Ron E, Schneider A. Exposing the Thyroid to Radiation: A Review of Its Current Extent, Risks, and Implications. Endocrine Reviews. 2010;31(5):756-773.
17. Dedlow E, Siddiqi S, Fillipps D, Kelly M, Nackashi J, Tuli S. Symptomatic Atlantoaxial Instability in an Adolescent with Trisomy 21 (Down's syndrome). Clinical Pediatrics. 2013;52(7):633-638
18. Atlanta-Axial Instability Information Pack 2018 [Internet]. British-gymnastics.org. 2018 [cited 1 January 2021]. Available from: https://www.british-gymnastics.org/technical-information/discipline-updates/disabilities/9316-atlanto-axial-information-pack-1/file
19. Bull M. Health Supervision for Children With Down Syndrome. PEDIATRICS. 2011;128(2):393-406
20. Locke G, Gradner J, Van Epps E. Atlas-dens interval (ADI) in children: a survey based on 200 normal cervical spines. Am J Roent- genol Radium Ther Nucl Med. 1966;97(1): 135–140
21. Davidson R. Atlantoaxial instability in individuals with Down syndrome: a fresh look at the evidence. Pediatrics. 1988;81(6): 857– 865
22. Pizzutillo P, Herman M. Cervical spine issues in Down syndrome. J Pediatr Orthop. 2005; 25(2):253–259

STUDENT DECLARATION

Dr. Louise Gibson, the research supervisor and I, the co-investigator designed the research study. Karen O’Sullivan (Down Syndrome Cork coordinator) sent out the online questionnaire. I collected and analyzed the data and wrote the report. The authors involved have not and will not receive financial gain from any company or institution related to this manuscript.

ACKNOWLEDGEMENTS

Special thanks to Karen O’Sullivan, Down Syndrome Cork coordinator who sent out the online survey along with the participation information leaflet to 1511 families registered with Down Syndrome Ireland. Special thanks also to Anna Kingston, CARL coordinator who facilitated the partnership between Down Syndrome Cork and University College Cork School of Medicine.

APPENDICES

Appendix A:

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 grass roots groups, single issue temporary groups, but also structured community 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 (www.livingknowledge.org).

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?

The research agreement between the CSO, student and CARL/University states that the results of the study must be made public through the publication of the final research report on the CARL (UCC) website. CARL is committed to open access, and the free and public dissemination of research results.

How do I reference this report?

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

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

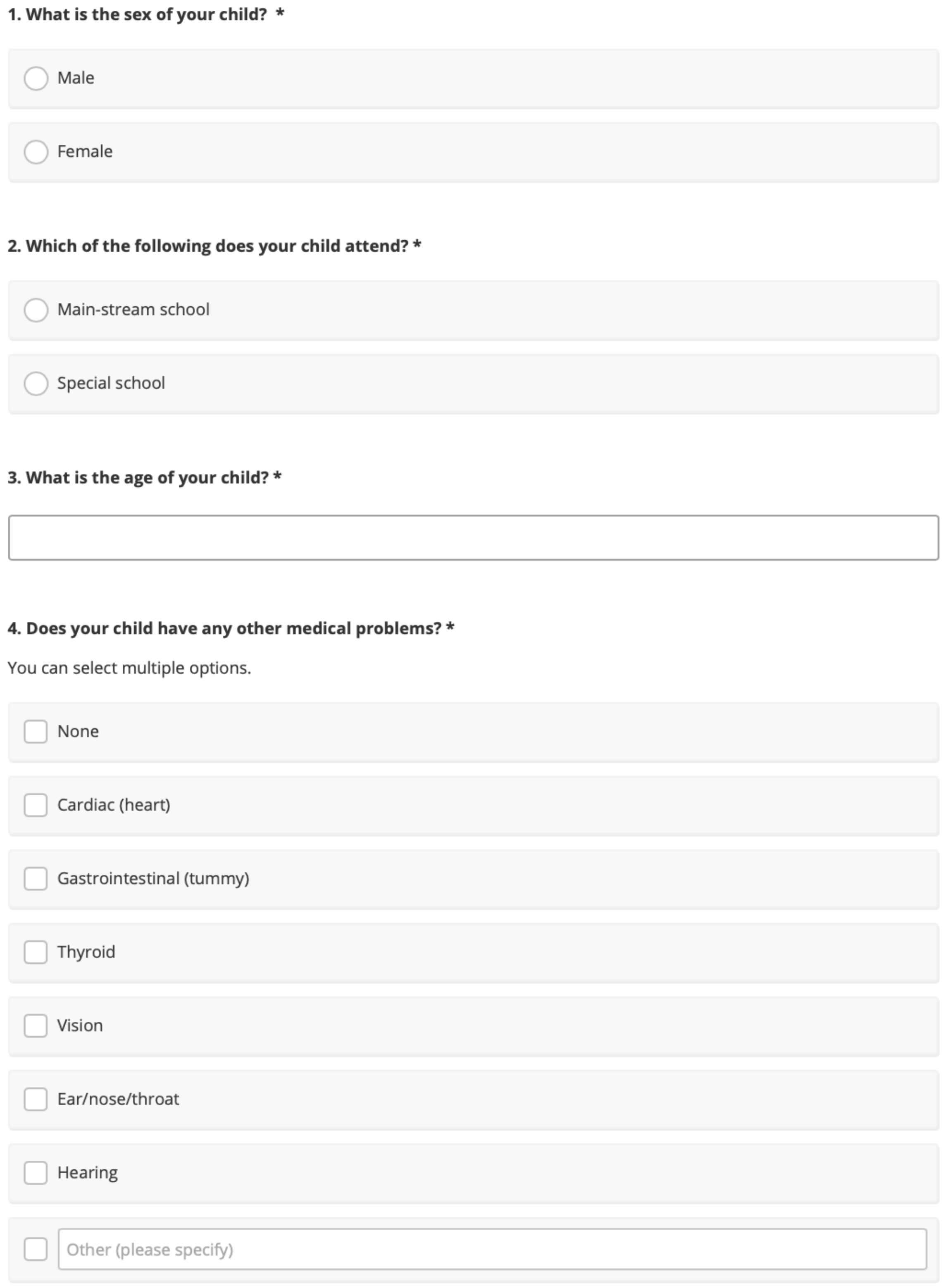
The UCC CARL website has further information on the background and operation of Community-Academic Research Links at University College Cork, Ireland. http://carl.ucc.ie. You can follow CARL on Twitter at @UCC_CARL. All of our research reports are accessible free online here: http://www.ucc.ie/en/scishop/rr/.

CARL is part of an international network of Science Shops called the Living Knowledge Network. You can read more about this vibrant community and its activities on this website: http://www.scienceshops.org and on Twitter @ScienceShops. CARL is also a contributor to Campus Engage, which is the Irish Universities Association engagement initiative to promote community-based research, community-based learning and volunteering amongst Higher Education students and staff.

Are you a member of a community project and have an idea for a research project?
We would love to hear from you! Read the background information here http://www.ucc.ie/en/scishop/ap/c&vo/ and contact us by email at carl@ucc.ie.

Disclaimer
Notwithstanding the contributions by the University and its staff, the University gives no warranty as to the accuracy of the project report or the suitability of any material contained in it for either general or specific purposes. It will be for the Client Group, or users, to ensure that any outcome from the project meets safety and other requirements. The Client Group agrees not to hold the University responsible in respect of any use of the project results. Notwithstanding this disclaimer, it is a matter of record that many student projects have been completed to a very high standard and to the satisfaction of the Client Group.

Appendix B:



1. What is the sex of your child? *

○ Male

○ Female

2. Which of the following does your child attend? *

○ Main-stream school

○ Special school

3. What is the age of your child? *

4. Does your child have any other medical problems? *

You can select multiple options.

☐ None

☐ Cardiac (heart)

☐ Gastrointestinal (tummy)

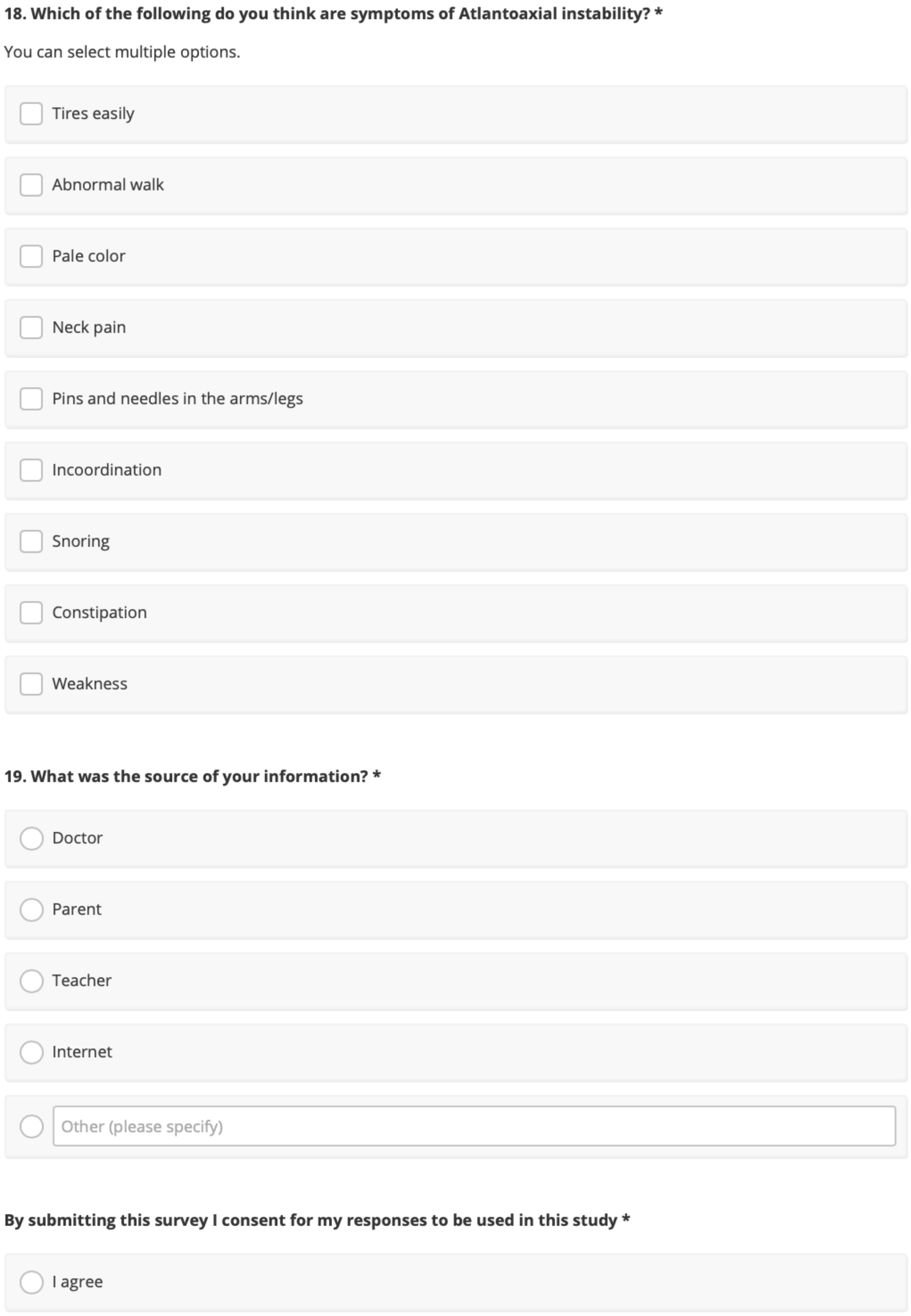
☐ Thyroid

☐ Vision

☐ Ear/nose/throat

☐ Hearing

☐ Other (please specify)



18. Which of the following do you think are symptoms of Atlantoaxial instability? *

You can select multiple options.

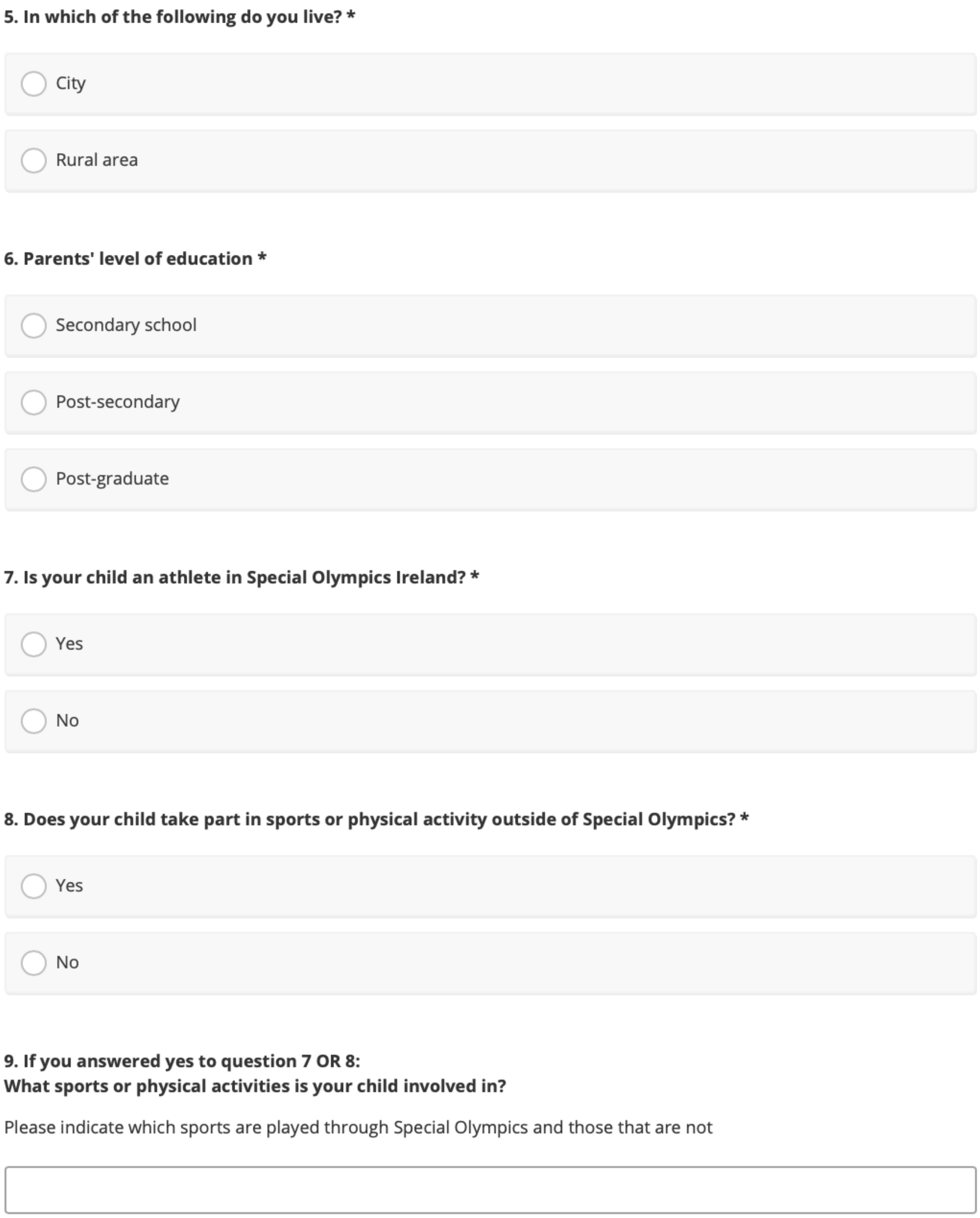
- ☐ Tires easily
- ☐ Abnormal walk
- ☐ Pale color
- ☐ Neck pain
- ☐ Pins and needles in the arms/legs
- ☐ Incoordination
- ☐ Snoring
- ☐ Constipation
- ☐ Weakness

19. What was the source of your information? *

- ○ Doctor
- ○ Parent
- ○ Teacher
- ○ Internet
- ○ Other (please specify)

By submitting this survey I consent for my responses to be used in this study *

- ○ I agree



5. In which of the following do you live? *

○ City

○ Rural area

6. Parents' level of education *

○ Secondary school

○ Post-secondary

○ Post-graduate

7. Is your child an athlete in Special Olympics Ireland? *

○ Yes

○ No

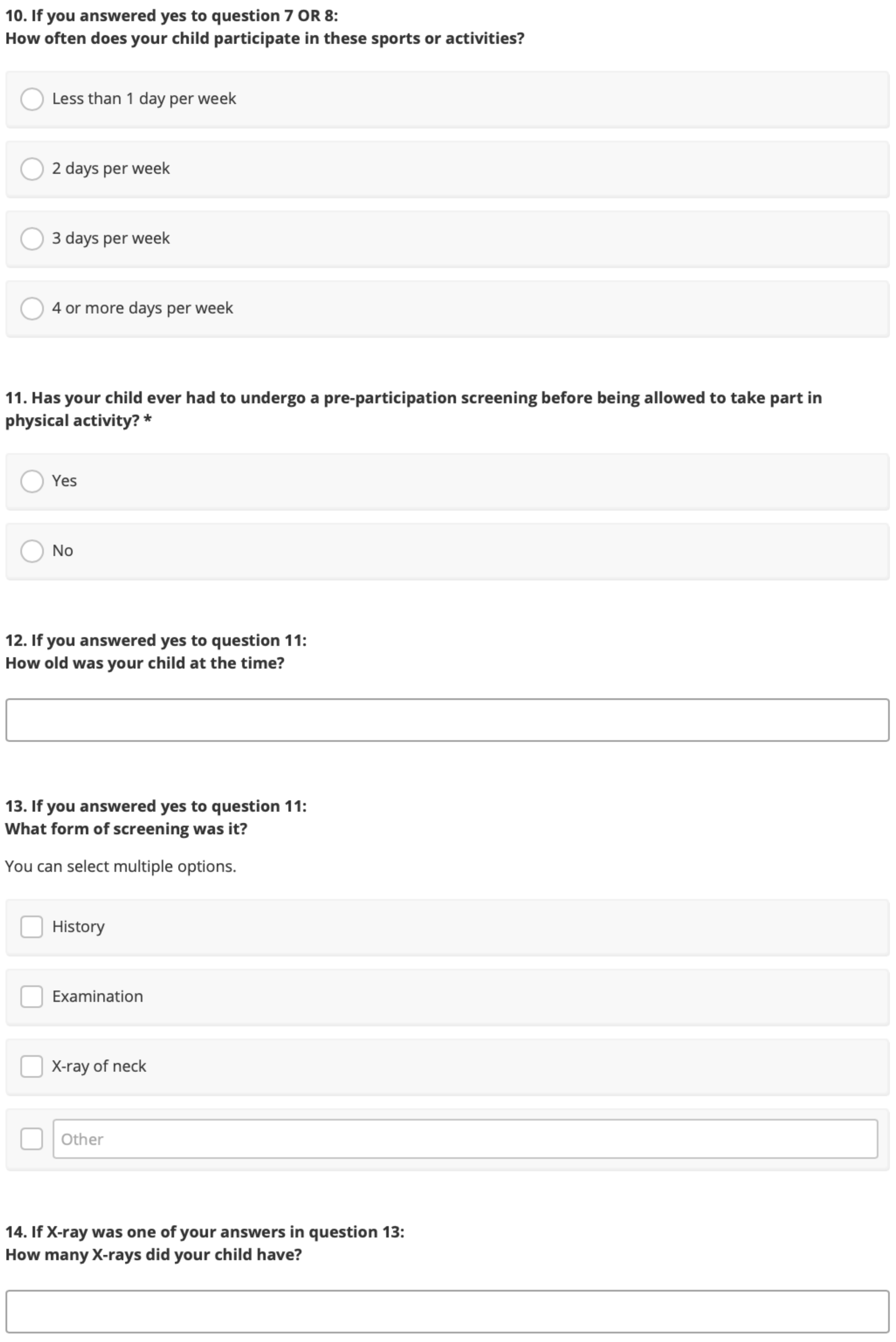
8. Does your child take part in sports or physical activity outside of Special Olympics? *

○ Yes

○ No

9. If you answered yes to question 7 OR 8:
What sports or physical activities is your child involved in?

Please indicate which sports are played through Special Olympics and those that are not



10. If you answered yes to question 7 OR 8:
How often does your child participate in these sports or activities?

○ Less than 1 day per week

○ 2 days per week

○ 3 days per week

○ 4 or more days per week

11. Has your child ever had to undergo a pre-participation screening before being allowed to take part in physical activity? *

○ Yes

○ No

12. If you answered yes to question 11:
How old was your child at the time?

13. If you answered yes to question 11:
What form of screening was it?

You can select multiple options.

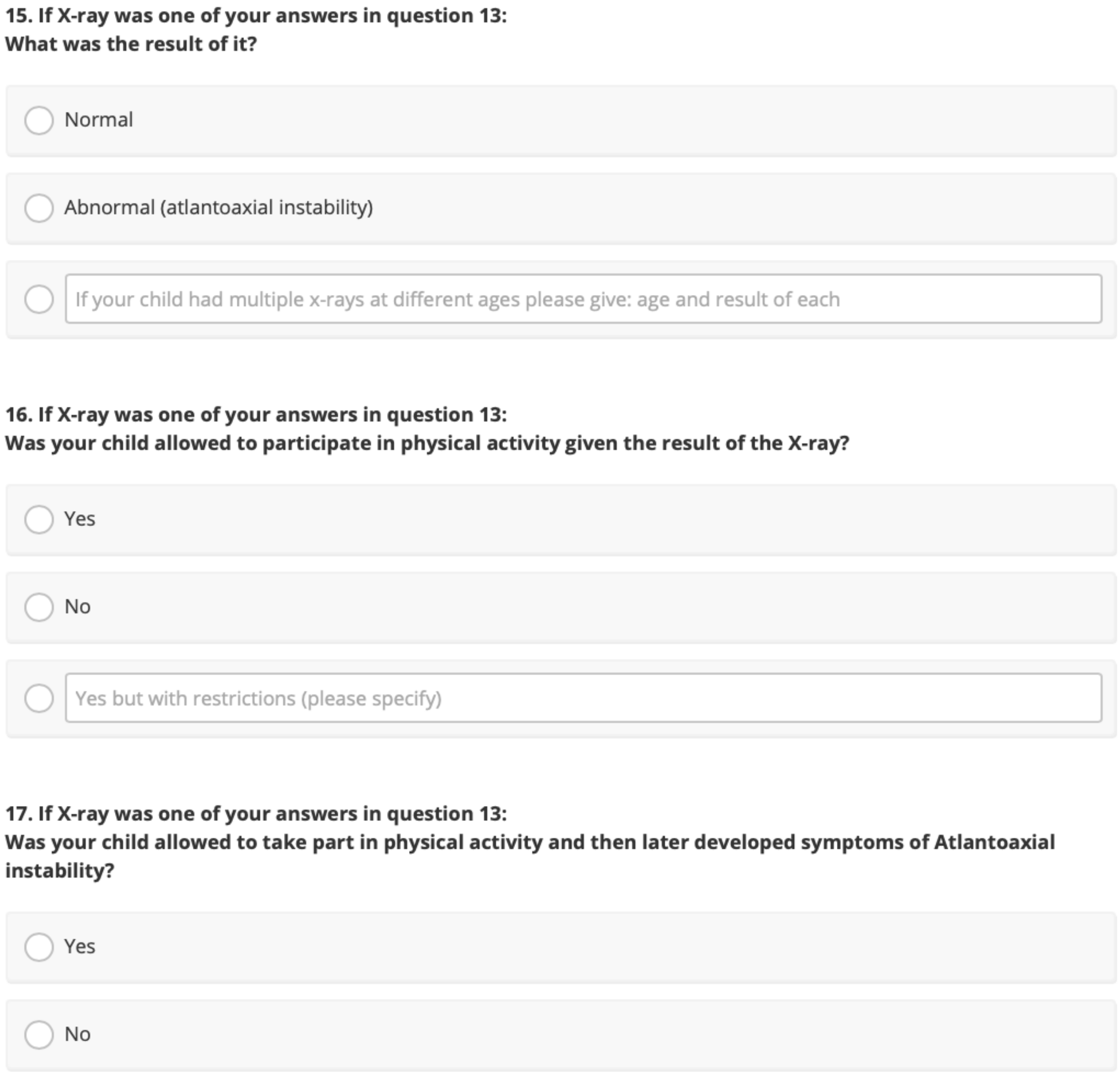
☐ History

☐ Examination

☐ X-ray of neck

☐ Other

14. If X-ray was one of your answers in question 13:
How many X-rays did your child have?



15. If X-ray was one of your answers in question 13:
What was the result of it?

○ Normal

○ Abnormal (atlantoaxial instability)

○ If your child had multiple x-rays at different ages please give: age and result of each

16. If X-ray was one of your answers in question 13:
Was your child allowed to participate in physical activity given the result of the X-ray?

○ Yes

○ No

○ Yes but with restrictions (please specify)

17. If X-ray was one of your answers in question 13:
Was your child allowed to take part in physical activity and then later developed symptoms of Atlantoaxial instability?

○ Yes

○ No

Appendix C:

Participant Information Leaflet

Study Title: Prevalence and Impact of X-ray Screening for Atlanto-Axial Instability in Children with Down Syndrome

Chief Investigator: Dr. Louise Gibson, Consultant Pediatrician, Cork University Hospital

Contact Number for Chief Investigator: 0876758196

Medical Student: Daniel Shenoda, 2nd Year Medical Student, UCC
Email Address: 117105320@umail.ucc.ie

Project Description

We are trying to determine whether or not children with down syndrome in Ireland are required to undergo a pre-participation screening test in the form of an x-ray prior to being allowed to take part in physical activity and the impact this has on the children.

Why have I been chosen?

You have been asked to take part in this project because you have a child (or children) with Down Syndrome, and I would like to know if they have ever had to undergo pre-participation screening before being allowed to play sports.

What will it involve?

You will be asked to complete an online questionnaire which will be emailed to you. You can do it from the comfort of your own home on your computer or any smart device. It will take approximately 5 minutes. Once you have completed the questionnaire all you have to do is click submit and you are done. This questionnaire will ask if your child has ever had to undergo a pre-participation screening test in the form of an x-ray and what impact this may have had.

What will happen to the information collected?

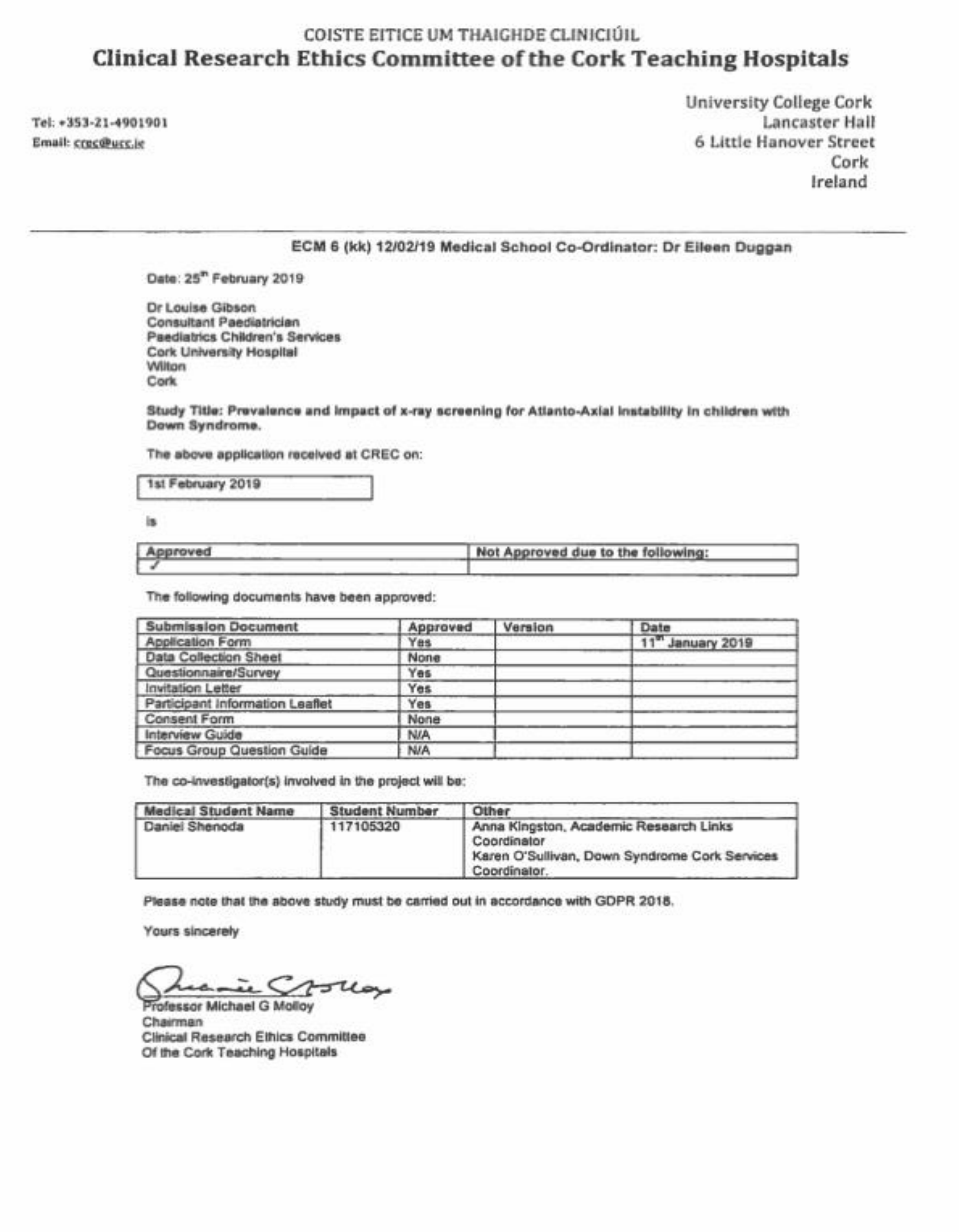
Your data will be stored in a password protected computer accessible only to the Dr. Louise Gibson, the medical student, and Down Syndrome Cork. Your data will be securely stored by the Chief Investigator for 10 years in a secure storage facility and will be destroyed after 10 years. Your information will be used in this study only.
The data will be analyzed and presented in the medical student’s final year research project report, however no personally identifiable information to you, will be analyzed or included in the findings. If an opportunity arose to publish this data in a research journal, again all personal details will be kept anonymous.

Will the information I give be confidential?

Yes. No personal identifiable information will be collected in this study.

This study has been approved by the Clinical Research Ethics Committee of the Cork Teaching Hospitals, Lancaster Hall, 6 Little Hanover Street, Cork, 021 4901901.

Appendix D:



COISTE EITICE UM THAIGHDE CLINICIÚIL
Clinical Research Ethics Committee of the Cork Teaching Hospitals

Tel: +353-21-4901901
Email: crec@ucc.ie

University College Cork
Lancaster Hall
6 Little Hanover Street
Cork
Ireland

ECM 6 (kk) 12/02/19 Medical School Co-Ordinator: Dr Eileen Duggan

Date: 25th February 2019

Dr Louise Gibson
Consultant Paediatrician
Paediatrics Children's Services
Cork University Hospital
Wilton
Cork

Study Title: Prevalence and impact of x-ray screening for Atlanto-Axial instability in children with Down Syndrome.

The above application received at CREC on:

1st February 2019

is

Approved	Not Approved due to the following:
✓	

The following documents have been approved:

Submission Document	Approved	Version	Date
Application Form	Yes		11th January 2019
Data Collection Sheet	None		
Questionnaire/Survey	Yes		
Invitation Letter	Yes		
Participant Information Leaflet	Yes		
Consent Form	None		
Interview Guide	N/A		
Focus Group Question Guide	N/A		

The co-investigator(s) involved in the project will be:

Medical Student Name	Student Number	Other
Daniel Shenoda	117105320	Anna Kingston, Academic Research Links Coordinator Karen O'Sullivan, Down Syndrome Cork Services Coordinator.

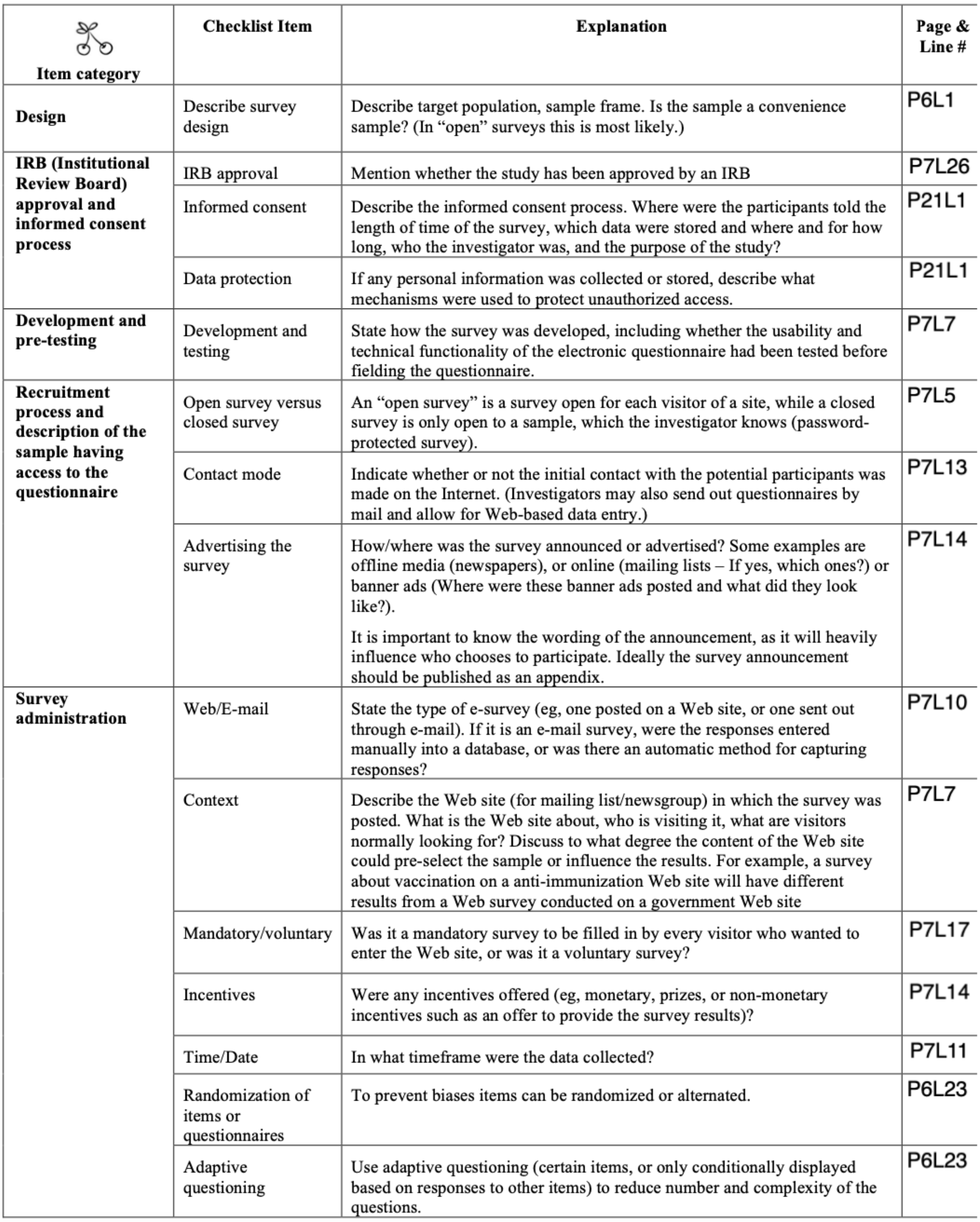
Please note that the above study must be carried out in accordance with GDPR 2018.

Yours sincerely

Professor Michael G Molloy
Chairman
Clinical Research Ethics Committee
Of the Cork Teaching Hospitals

Appendix E:

Checklist for Reporting Results of Internet E-Surveys (CHERRIES)



Item category	Checklist Item	Explanation	Page & Line #
Design	Describe survey design	Describe target population, sample frame. Is the sample a convenience sample? (In "open" surveys this is most likely.)	P6L1
IRB (Institutional Review Board) approval and informed consent process	IRB approval	Mention whether the study has been approved by an IRB	P7L26
	Informed consent	Describe the informed consent process. Where were the participants told the length of time of the survey, which data were stored and where and for how long, who the investigator was, and the purpose of the study?	P21L1
	Data protection	If any personal information was collected or stored, describe what mechanisms were used to protect unauthorized access.	P21L1
Development and pre-testing	Development and testing	State how the survey was developed, including whether the usability and technical functionality of the electronic questionnaire had been tested before fielding the questionnaire.	P7L7
Recruitment process and description of the sample having access to the questionnaire	Open survey versus closed survey	An "open survey" is a survey open for each visitor of a site, while a closed survey is only open to a sample, which the investigator knows (password-protected survey).	P7L5
	Contact mode	Indicate whether or not the initial contact with the potential participants was made on the Internet. (Investigators may also send out questionnaires by mail and allow for Web-based data entry.)	P7L13
	Advertising the survey	How/where was the survey announced or advertised? Some examples are offline media (newspapers), or online (mailing lists – If yes, which ones?) or banner ads (Where were these banner ads posted and what did they look like?). It is important to know the wording of the announcement, as it will heavily influence who chooses to participate. Ideally the survey announcement should be published as an appendix.	P7L14
Survey administration	Web/E-mail	State the type of e-survey (eg, one posted on a Web site, or one sent out through e-mail). If it is an e-mail survey, were the responses entered manually into a database, or was there an automatic method for capturing responses?	P7L10
	Context	Describe the Web site (for mailing list/newsgroup) in which the survey was posted. What is the Web site about, who is visiting it, what are visitors normally looking for? Discuss to what degree the content of the Web site could pre-select the sample or influence the results. For example, a survey about vaccination on a anti-immunization Web site will have different results from a Web survey conducted on a government Web site	P7L7
	Mandatory/voluntary	Was it a mandatory survey to be filled in by every visitor who wanted to enter the Web site, or was it a voluntary survey?	P7L17
	Incentives	Were any incentives offered (eg, monetary, prizes, or non-monetary incentives such as an offer to provide the survey results)?	P7L14
	Time/Date	In what timeframe were the data collected?	P7L11
	Randomization of items or questionnaires	To prevent biases items can be randomized or alternated.	P6L23
	Adaptive questioning	Use adaptive questioning (certain items, or only conditionally displayed based on responses to other items) to reduce number and complexity of the questions.	P6L23

	Number of Items	What was the number of questionnaire items per page? The number of items is an important factor for the completion rate.	P6L22
	Number of screens (pages)	Over how many pages was the questionnaire distributed? The number of items is an important factor for the completion rate.	P6L22
	Completeness check	It is technically possible to do consistency or completeness checks before the questionnaire is submitted. Was this done, and if "yes", how (usually JAVAScript)? An alternative is to check for completeness after the questionnaire has been submitted (and highlight mandatory items). If this has been done, it should be reported. All items should provide a non-response option such as "not applicable" or "rather not say", and selection of one response option should be enforced.	P7L1
	Review step	State whether respondents were able to review and change their answers (eg, through a Back button or a Review step which displays a summary of the responses and asks the respondents if they are correct).	P7L4
Response rates	Unique site visitor	If you provide view rates or participation rates, you need to define how you determined a unique visitor. There are different techniques available, based on IP addresses or cookies or both.	N/A
	View rate (Ratio of unique survey visitors/unique site visitors)	Requires counting unique visitors to the first page of the survey, divided by the number of unique site visitors (not page views!). It is not unusual to have view rates of less than 0.1 % if the survey is voluntary.	N/A
	Participation rate (Ratio of unique visitors who agreed to participate/unique first survey page visitors)	Count the unique number of people who filled in the first survey page (or agreed to participate, for example by checking a checkbox), divided by visitors who visit the first page of the survey (or the informed consents page, if present). This can also be called "recruitment" rate.	N/A
	Completion rate (Ratio of users who finished the survey/users who agreed to participate)	The number of people submitting the last questionnaire page, divided by the number of people who agreed to participate (or submitted the first survey page). This is only relevant if there is a separate "informed consent" page or if the survey goes over several pages. This is a measure for attrition. Note that "com- pletion" can involve leaving questionnaire items blank. This is not a measure for how completely questionnaires were filled in. (If you need a measure for this, use the word "completeness rate".)	P7L24
Preventing multiple entries from the same individual	**Cookies used**	Indicate whether cookies were used to assign a unique user identifier to each client computer. If so, mention the page on which the cookie was set and read, and how long the cookie was valid. Were duplicate entries avoided by preventing users access to the survey twice; or were duplicate database entries having the same user ID eliminated before analysis? In the latter case, which entries were kept for analysis (eg, the first entry or the most recent)?	P7L25
	IP Check	Indicate whether the IP address of the client computer was used to identify po- tential duplicate entries from the same user. If so, mention the period of time for which no two entries from the same IP address were allowed (eg, 24 hours). Were duplicate entries avoided by preventing users with the same IP address access to the survey twice; or were duplicate database entries having the same IP address within a given period of time eliminated before analysis? If the latter, which entries were kept for analysis (eg, the first entry or the most recent)?	P7L25
	Log file analysis	Indicate whether other techniques to analyze the log file for identification of multiple entries were used. If so, please describe.	P7L25
	Registration	In "closed" (non-open) surveys, users need to login first and it is easier to prevent duplicate entries from the same user. Describe how this was done. For example, was the survey never displayed a second time once the user had filled it in, or was the username stored together with the survey results and later eliminated? If the latter, which entries were kept for analysis (eg,	P7L5

		the first entry or the most recent)?	P7L5
Analysis	Handling of incomplete questionnaires	Were only completed questionnaires analyzed? Were questionnaires which ter- minated early (where, for example, users did not go through all questionnaire pages) also analyzed?	P7L23
	Questionnaires submitted with an atypical timestamp	Some investigators may measure the time people needed to fill in a questionnaire and exclude questionnaires that were submitted too soon. Specify the timeframe that was used as a cut-off point, and describe how this point was determined.	P7L17
	Statistical correction	Indicate whether any methods such as weighting of items or propensity scores have been used to adjust for the non-representative sample; if so, please describe the methods.	N/A

Eysenbach G. Improving the quality of Web surveys: the **Checklist** for Reporting Results of Internet E-Surveys (**CHERRIES**). J Med Internet Res. 2004 Sep 29;6(3):e34.