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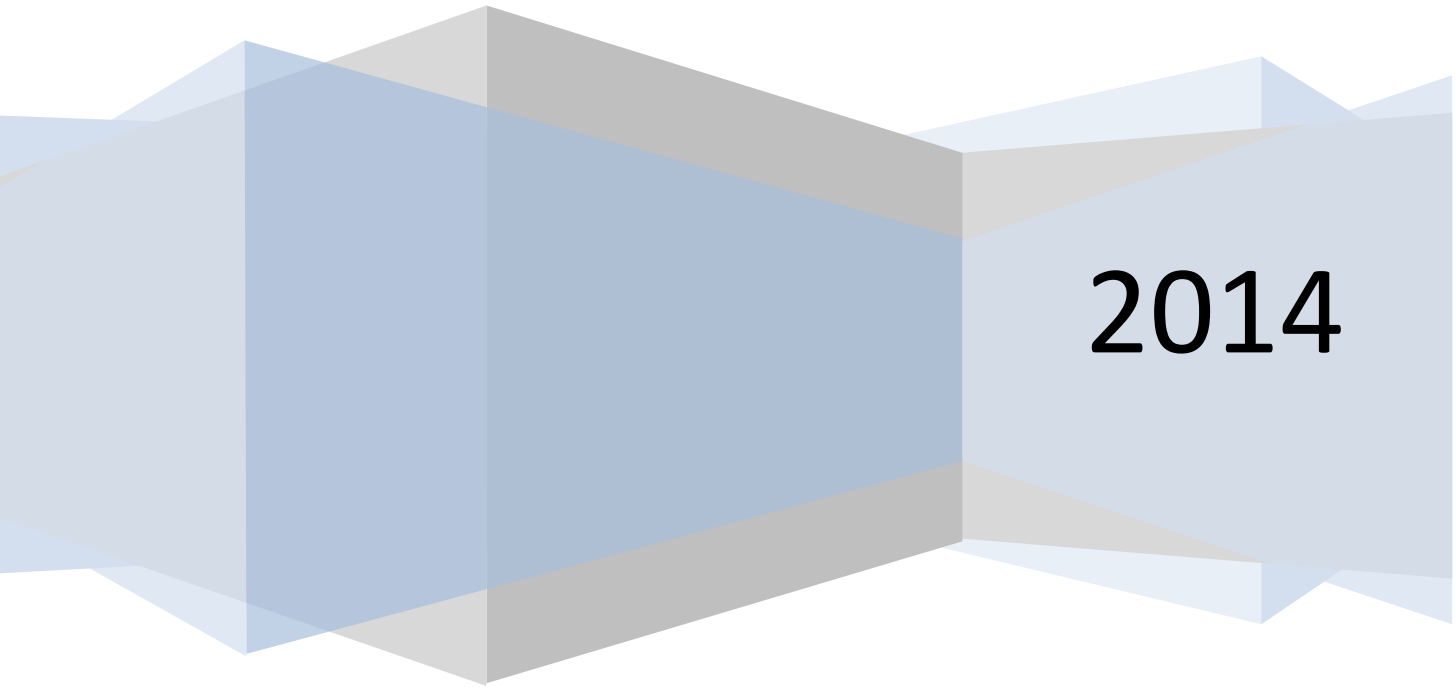
Report of the Consultation for a National Registry and Biobank for Autism and Neurodevelopmental Disorders

National Stakeholder Consultation 2013-2014



2014

Report of the Consultation for a National Registry and Biobank for Autism and Neurodevelopmental Disorders

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2013-2014

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Executive Summary

One in 68 children has been identified as having autism spectrum disorder (CDC, 2014). Autism spectrum disorders (ASD) and neurodevelopmental disorders (NDD) are lifelong heterogeneous conditions with implications for the family, state services and society at large presenting a significant public health challenge. Currently in Ireland there is no effective health information system or biobank gathering essential data for ASD/ NDD. This report presents compelling evidence of the timely need to create a comprehensive registry and biobank and is informed by a national stakeholder consultation process which took place from October 2013 to April 2014. Achieving national collaboration is an important part of the process to lay sound foundations for building a sustainable registry and biobank. The scope and aims of this registry and biobank is to act as a transformational innovation to accelerate world-class research in Ireland, deliver best practice service provision, support and education for individuals with ASD/NDD and enable best quality of life.

Methods: A national stakeholder consultation process was implemented from October 2013 through April 2014 and comprised of four town hall meetings in Dublin, Galway, Sligo and Cork and a national online survey, completed by 425 of parents, self-advocates and services providers.

Results: Ninety three percent of survey respondents agreed that a registry and biobank are needed. The most important focus for the registry, according to survey results, is to inform health, education and social care aspects of service development. The primary concern emerging from both the consultation process and survey was around data privacy and protection.

Conclusions: The registry would be a tremendous tool for mapping real needs for individuals, while the biobank would enrich Ireland's capacity to make major advances in healthcare - a transformative innovation for improved health. It would facilitate a major investment in Ireland's knowledge economy; expedite world-class research and development resulting in both clinical and economic benefits.

Landscape Analysis

2.1 Background

In March 2013, a private members bill, The Autism Bill, was brought forward to government to require the development of a national strategy for autism, including the creation of a national database to provide an accurate estimate of the number of people affected with ASD in Ireland. Further, the National Review of Autism Services in 2012 expressly called on the HSE to support a database for autism and other neurodevelopmental disabilities. In response, NUI Galway and Trinity College Dublin, in collaboration with Autism Speaks, launched a national consultation to understand the views of self-advocates, families, service providers and researchers to creating an autism and neurodevelopmental disabilities registry and biobank. Over the past year, a national survey was launched, and a series of town hall meetings hosted throughout Ireland informed the development of a comprehensive consultation report. Progress to date has also been informed by meetings with Irish government stakeholders as well as the consultation Parent and Self-advocate Advisory Group.

Health registries gather large amounts of data including diagnostic histories, pathways to care and service use with the ultimate goal of improving systems of care. Registries have been effectively used in other health areas such as cancer and stroke in Ireland. In Scandinavia, national health registries have helped not only to uncover important risk factors for ASD but also to inform improvements to systems of care for affected families. Unique opportunities have been created for researchers through internet-mediated research to study medical and healthcare data and genetics. One excellent working model is the Interactive Autism Network (IAN). The IAN has spearheaded a movement in autism research with more than 38,000 participants. The dataset that IAN has collected has enabled IAN to provide much-needed subject recruitment assistance to over 300 autism research projects. Furthermore, it has enabled IAN investigators, collaborators and independent researchers to publish on a variety of topics ranging from population genetics to twin studies and the stability of diagnosis.

2.2 Objectives

The main objective of this initiative is to create a national registry and biobank that will inform the development of clinical practice, future research and services for people with autism and neurodevelopmental disorders. The scope of a fully operational registry and biobank will provide critical data useful for the development of services and to identify critical research questions to be investigated further. In addition health policy decisions can be made at the highest level of government with reliable evidence of the social and economic cost of these conditions. A comprehensive health information system for ASD has never before existed in Ireland and will facilitate opportunities for researchers, clinicians and patients to connect, empowering and informing research, health services and education.

The aim of the consultation process was to engage with stakeholders affected by, living with or working with people with autism and other related neurodevelopmental disorders and to learn as much about their multifarious needs as possible. Feedback on the development of both a clinical registry and a biobank to support biomedical research is vital for envisioning, planning and implementing a sustainable model of excellence to advance research and improve services and supports.

2.3 What are Neurodevelopmental and Autism Spectrum Disorders?

The World Health Organisation (WHO, 2011) defines neurodevelopmental disorders as "disabilities in the functioning of the brain that affect a child's behaviour, memory or ability to learn e.g. mental retardation, dyslexia, attention deficit hyperactivity disorder (ADHD), learning deficits and autism" (WHO, 2003).

More recently, the American Psychiatric Association's *Diagnostic and Statistical Manual of Mental Disorders 5th edition* (DSM-5) uses the term autism spectrum to describe a range of neurodevelopmental conditions (DSM-5, 2013). The DSM-5, published in 2013, redefined the autism spectrum to encompass the previous (DSM-4-TR) diagnoses of autism, Asperger syndrome, pervasive developmental disorder not otherwise specified (PDD-NOS), childhood disintegrative disorder, and Rett syndrome. These disorders are characterized by social deficits and communication difficulties, stereotyped or repetitive behaviours and interests, and in some cases, cognitive delays. The term "spectrum" refers to the wide range of symptoms, skills, and levels of impairment, or disability, that individuals with ASD can possess. Some are mildly impaired by their symptoms, but others are severely disabled (National Institute for Mental Health, 2013). ASD are a lifelong disorder and has profound effects on an individual's social, emotional and cognitive development. Widely varying degrees of difficulty have significant mental, emotional, physical, and economic consequences not only for individuals, but their families and society in general.

There is no cure for ASD and the specific causes have yet to be found. In the research literature, risk factors contributing to the development of ASD include genetics, prenatal and perinatal factors, as well as neuroanatomical abnormalities (Glasson et. al, 2004; Kolevzon et al. 2007; Larsson et al. 2004). Environmental factors have also been identified. Fragile X syndrome is the most common known cause of autism or autism spectrum disorders and the most common identified cause of inherited intellectual disability (Hagerman, R. 2011). Fragile X syndrome is associated with the expansion of the CGG trinucleotide repeat affecting the *Fragile X mental retardation 1* (*FMR1*) gene on the X chromosome, resulting in a failure to express the fragile X mental retardation protein (FMRP), which is required for normal neural development.

Research has proven that early intervention, therapies and behavioural interventions can target specific symptoms and bring about substantial improvement. Therefore early diagnosis and intervention is critical, yet many children cannot avail of this due to misdiagnosis or no diagnosis at all. Furthermore there is a huge lack of understanding and investment regarding services and interventions for adults. In Ireland, screening only takes place if the child is referred for an assessment whereas the American Academy of Paediatrics has recommended screening for ASD in all children aged 18 months to two years (Johnson and Myers, 2007). ASD tend to be highly comorbid with other disorders. Distinguishing between ASD and other diagnoses can be challenging because the traits of ASD often overlap with symptoms of other disorders and the characteristics of ASD make traditional diagnostic procedures difficult.

2.4 What is a Clinical Registry?

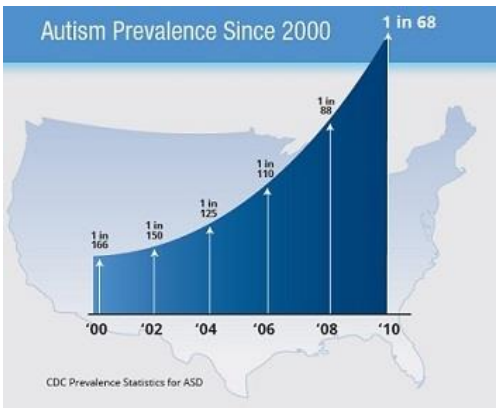
Clinical registries or databases gather clinical information and other data on patients to inform the development of clinical practice, services and future research. Some of the best-known examples of registries are those that exist in Scandinavian countries where there are well-established patient databases for a variety of physical and mental illnesses and disabilities (SFARI, 2009). These well-organised databases provide critical data that is useful for the development of services and to identify critical research questions to be further investigated.

2.5 What is a biobank and why is it so important?

The term biobank is used to describe collections of patient samples and corresponding clinical research and its translation into medical, scientific, economic and societal benefits. Samples can be made available for basic medical research, development of new pharmaceutical treatment, for developing biomarkers for diagnosis or treatment response and to help identify molecular pathways for complex genetics. The reliability of data derived from these collections is dependent on the quality and consistency of the bio-specimens that are collected and how they are managed and stored. Biobanks such as the Autism Genetics Resource Exchange (AGRE) have delivered numerous important research discoveries and informed state and federal health, education and social policies. Biobanks form a critical bridge to enable translational research and are a critical component in twenty-first century research with increasing emphasis placed on predictive and preventative treatments. It is proposed to develop a biobank in Ireland that would collect DNA samples from people with ASD/NDD to advance clinical and economic benefits. This would provide more samples to Irish researchers and to large-scale international studies for genetic research. Such a biobank would also be useful for pharmaceutical companies interested in researching new treatments for these conditions.

2.6 Epidemiology

With a population of 4.58 million (CSO, 2011) Ireland is an ideal sample to study. Despite being a much smaller country physically, the population is genetically relatively homogenous so a registry and biobank would hold much greater validity than countries with more heterogeneous ethnicity. This provides scientific credibility to the uniqueness and attractiveness of this initiative. Prevalence rates for ASD are not yet available in Ireland. Without accurate data, effective planning and decision-making cannot happen. Data from the Irish National Disability Survey (2006) indicates that prevalence is likely to be similar to the international estimates of 1%. The Centre for Disease Control's Autism and Developmental Disabilities Monitoring (ADDM) Network released data in March 2014, revealing that about one in 68 children in the United States has an ASD, five times more common among boys than among girls. Epidemiological studies in many countries around the world have identified increased prevalence of ASD in recent years. This increase is said to be partly attributable to better diagnosis, screening and broadening of diagnostic criteria. New prevalence numbers also accentuate the reality that well over a half-million children with autism will reach adulthood in the United States over the next decade with sweeping effects beyond the individual. Efforts must be urgently made to develop and deliver the supports needed to help these individuals become fulfilled valued members of their communities. With no epidemiological data or specific register in this country, services and requirements cannot be effectively planned leading to insufficient services and misallocation of resources.



2.7 Current activity at a national level

There are two relevant databases in Ireland, the National Intellectual Disability Database and the Physical and Sensory Disability Database but these do not capture all individuals with ASD/NDD. There is a critical need for this type of comprehensive database and biobank to

provide a complete and adequate umbrella framework with long-term benefits for all concerned.

In 2012, a proposed Autism Bill was put forward by TD Michael McCarthy to provide Ireland with a coherent and national framework for addressing the specific needs of people with autism drawing from similar legislation which has been enacted in Northern Ireland (Autism (NI) Act 2011) and England (Autism Act 2009). The development of a national registry and biobank would also align with the National Strategy for Rare Disease which is under development to look at areas of centres of expertise, orphan drugs and technologies, research and information and patient empowerment and support (EUCERD, 2013).

2.8 What are the benefits of a registry and biobank?

Establishing an Irish registry and biobank has the potential to provide new insight into the whole field of ASD/NDD offering pathways to research discoveries in diagnosis and new treatments. The Autism Genome Project is an excellent demonstration of the unprecedented power of statistical and data leveraging (Autism Speaks, 2007). The Autism Genome Project (AGP), which began in 2002, involved an autism genetics consortium from over 50 institutions in 19 countries. Researchers came together and shared samples, data and expertise to facilitate the identification of autism susceptibility genes. The results of this study revealed previously unidentified region of chromosome 11, and neurexin 1, a member of a family of genes believed to be important in neuronal contact and communication, among other regions and genes in the genome. The neurexin finding in particular highlights a special group of neurons, called glutamate neurons, and the genes affecting their development and function, suggesting they play a critical role in autism spectrum disorders.

Is there one best model that could be taken and used in Ireland? Drawing on the analysis of other registries and biobanks in Table 1, the answer is not to take one particular model, but to learn from and adapt the best practices of existing, successful models which deliver a variety of benefits from driving unique research possibilities and breakthroughs to increasing the scope of online learning, public awareness and education. For example, Denmark is at the very front when it comes to epidemiological research, boasting registries which provide detailed information on the entire population. With the establishment of a very large national biobank and linkage between the biological specimens and the information contained in the Danish registers, Danish research is given new and competitively unique possibilities (Statens Serum Institut, 2014).

The Phelan-McDermid Syndrome International Registry is an excellent example of a parent-driven registry, founded, driven and governed by parents. This registry collects demographic, genetic, clinical and developmental data on patients diagnosed with PMS with registrations from over 43 countries.

The ASD-UK is a model of a research-driven family database of children with an ASD. The aim of ASD-UK is to provide accurate data to facilitate research and give families the opportunity to take part in research studies. ASD-UK is funded by a charity called Autistica.

Another example, the Interactive Autism Network is a great model with two spokes: IAN Research and IAN Community. The IAN Research solves a major problem faced by many researchers: recruiting participants for research projects. IAN Community is an online learning environment that includes an interconnected learning and online knowledge forum. An additional strength of IAN that should be incorporated in the Irish registry and biobank is measurement of non-biological phenomena including behaviour, communication and social styles, which is essential for ASD research.

The transformative and powerful leveraging capacity of a registry and biobank for ASD/NDD in Ireland cannot be emphasised enough.

A longitudinal study of ASD/NDD through the registry and biobank would indicate the most effective approaches for professionals and caregivers for both early intervention and treatments for young children as well as adults and the elderly. As reported in the research findings, there are huge gaps in service provision and care for adults and the elderly with ASD. They can be left undiagnosed, receive inappropriate services or struggle with no access to proper enabling environment. Employment and independent living are problems that need to be addressed and the registry and biobank would be of great benefit for this.

Consultation Methodology

In response to a call from the HSE and the National Review of Autism Services in 2012, NUI Galway and Trinity College Dublin, in collaboration with Autism Speaks, launched a national consultation to understand the views of self-advocates, families, service providers and researchers to creating an autism and neurodevelopmental disabilities registry and biobank.

In June 2013 an Expert Group was formed including Professor Louise Gallagher, Trinity College Dublin; Dr. Geraldine Leader, National University of Ireland, Galway; June O'Reilly, Trinity College Dublin, Dr. Amy Daniels, Autism Speaks; and Dr. Andy Shih, Autism Speaks.

The first step in the stakeholder consultation process was to establish a Stakeholder Advisory Committee consisting of individuals with ASD, parents and representatives from relevant ASD and neurodevelopmental organisations to ensure equal representation, expertise and balance. Round table meetings with the Stakeholder Consultation Advisory Committee were held in June 2013 and October 2013 to identify and discuss key issues and concerns. Outside of these meetings the Stakeholder Consultation Advisory Committee worked virtually, communicating regularly by email due to geographical locations.

A pilot survey was launched in June 2013 at the "Autism Spectrum Disorders: From Research to Practice" Conference which took place at NUI Galway. This survey assessed if respondents felt there is a need for a registry and biobank, what type of data should be included, who should access the data and if they had any concerns about the registry. The pilot survey also asked questions about the biobank, if they supported the establishment of a biobank for ASD, who they felt should access the biobank and what it would be useful for.

Following analysis of the pilot survey and subsequent consultation with the Stakeholder Consultation Advisory Committee, the final survey was implemented using Survey Monkey, an online survey system (Appendix A). The web-based questionnaire provided a flexible platform for delivery of a range of questions providing all people with an opportunity to contribute their perspectives. Getting appropriate information from a variety of individuals including families, professionals and service providers involved the effective use of different questioning methods including open, closed, probe, hypothetical and reflective. Participation in the online survey was voluntary and accessed via a website. A website specifically for the Irish Autism and Neurodevelopmental Registry and Biobank was designed and developed using Wordpress in January 2014. The website address is www.iarb.ie.

Targeted and customised communications with multifarious stakeholder groups required a multi-targeted strategy to promote and encourage participation. In parallel with the launch of the website and online survey a particular PR focus was applied across national TV, national and local radio, online and print media. Facebook and Twitter were set up as communication tools to increase awareness about the initiative and encourage engagement in the survey. Links to the survey webpage and communication updates on the process were issued via Facebook, Twitter and listserv emails on a regular basis. On World Autism Day, on 2nd April 2014, a media drive to promote the consultation was pushed on national press, online, and through relevant Facebook pages and Twitter.

As this initiative targeted the entire country, four regional public consultation meetings were scheduled to take place in each of the four provinces of Ireland. The purpose of the public consultation meetings was to reach out directly to the public and to inform them about the registry and biobank ensuring they received accurate information and to attract buy in. It was also to learn and appreciate the diverse perspectives about the initiative. An easily accessible central venue was selected for each meeting. The meetings were advertised in national and local press and through effective use of social media. Refreshments were provided to welcome attendees and allow for informal interaction. Three meetings were held on midweek evenings, however one was held on a Saturday afternoon. The time and day did not affect attendance numbers however weather conditions did affect attendance with severe flooding and red weather alerts around the time of the meetings. The Southern meeting had to be postponed from February to the end of March for this reason. Refreshments and room hire were funded by Autism Speaks.

East (Trinity College Dublin)	14 January 2014
West (National University of Ireland Galway)	28 January 2014
North and Northwest (Sligo Park Hotel)	13 February 2014
South (University College Cork)	22 March 2014

The format of the public consultation meetings was consistent across regions. An overview of the Registry and Biobank was presented. The panel of experts were introduced and three key questions were posed first to the panel and then to the audience:

1. What do you see as the primary purpose of a registry/biobank for ASD/NDD?
2. What are your biggest concerns with having a registry/biobank for ASD/NDD?
3. What has been your own experience with registries and biobank in the past?

Following discussion of the key questions, there was a mediated open floor discussion to assess key issues. Facilitating the diverse emotive and social intelligence of the audience was important when sensitive issues arose. All comments were recorded. All attendees were asked to disseminate the information and to encourage their networks to complete the online survey.

Meetings were conducted with Governmental agencies to ensure they understood the objectives and goals of this registry and biobank and to gain governmental support. These meetings also provided an opportunity to learn what the forthcoming plans and strategies of the relevant bodies are and how this registry and biobank can work in partnership with government strategy. Meetings were held with the Health Research Board, the Department of Health and the Department of Education.

Key Findings

The results presented in this section based on the methodology outlined above include the pilot survey, a nationwide online survey, and an analysis of other database and biobank models.

In summary, the research findings strongly indicate that Ireland needs an ASD/NDD registry to be established with a focus on clinical, biomedical and environmental research. It should focus on informing service development and act as a central reliable information resource. Parents said it was "the first time they had ever been asked what will make my child's life better" and "it is like being handed a microphone to voice our needs and concerns". The compendium of comments presents a diverse insight into the perspectives and viewpoints of the public.

4.1 Key findings of pilot survey

A pilot survey was carried out in June 2013 at the "Autism Spectrum Disorders: From Research to Practice" Conference in NUI Galway. Of the 62 respondents, that included parents, self-advocates and service providers, 84% strongly agreed with establishment of a National Autism Data Repository and the inclusion of data pertaining to clinical presentation, health and education service needs and use, occupational and residential needs and community supports. A significant number were concerned (64%) that the data could be accessed inappropriately and were most concerned about medical insurance companies. There was support for data entry by parents, professionals and adults with ASD.

When asked specifically about the biobank, 67% supported the establishment of a biobank for use by researchers in Ireland (92%) and outside (89%) with less enthusiasm for use by private companies (56%). Again there was concern about inappropriate access by medical insurance companies (66%), the Gardaí (56%) or 'others' (55%). The biobank was thought to be useful for supporting research (93%), providing info on genetic risk (86%), to develop better treatments (86%).

Some common qualitative themes emerged such as the **sense of urgency** -
"Long needed, just do it" and "long overdue".

Access to the information system as a tool to benefit personal health was raised -
"Parents and adults with ASD must have access to their own data & to be able to use it in a way that is beneficial to them".

The importance of **partnership** was stressed - *"Excellent initiative. We need to partner with research community to get answers and to help o ur community".*

Expectations for **improved regional service delivery** also presented -
"I expect regional services would benefit from accurate numbers when planning heal th and educational services."

4.2 Online Survey results

The online survey consisted of four sections. The first section explores the profile of the respondent. The second section asks questions specific to the registry. The third section seeks responses and comments specific to the biobank. The final section of the survey asks respondents about their perspectives on linking the registry and biobank, but also establishing links with other databases. As of January 2015 there were a total of 425

respondents who completed at least 60% of the survey. Below is a summary of findings from each of the four sections of the survey based on these 425 individuals.

Section 1: Respondent profile

The majority of respondents were mothers aged between 35 and 44, with children under the age of 18. Only 13% of respondents were male.

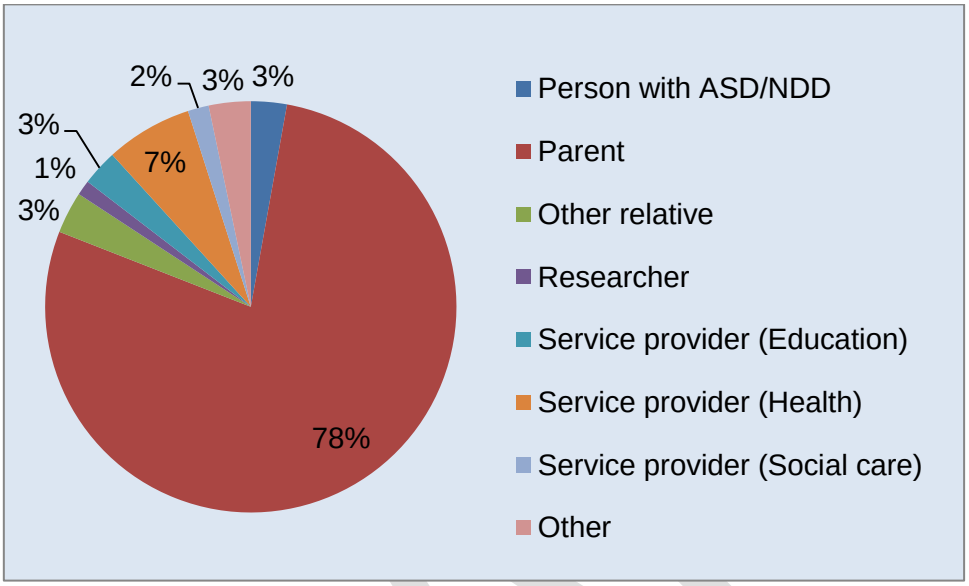


Figure 1: Profile of Respondents **(Question 1)**

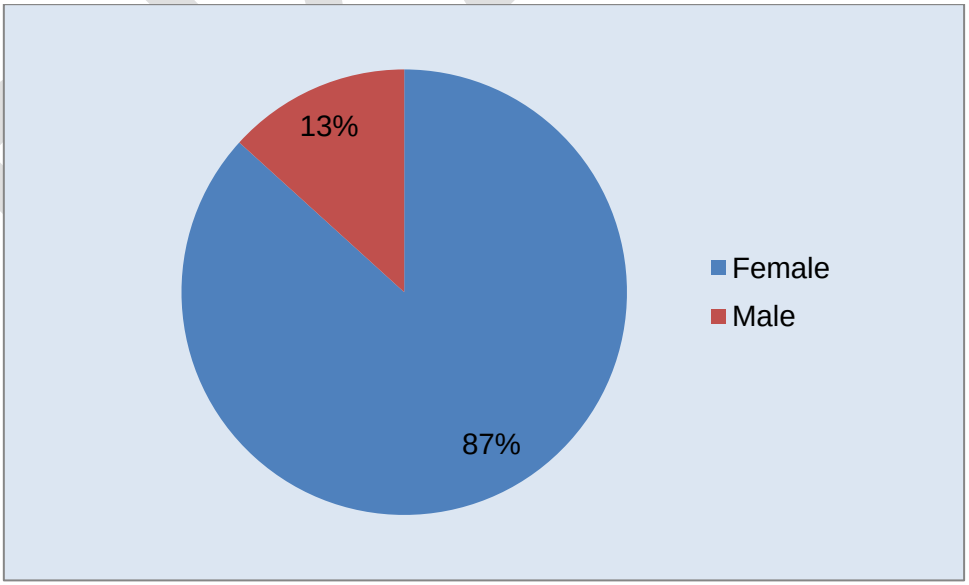


Figure 2: Gender of respondents **(Question 2)**

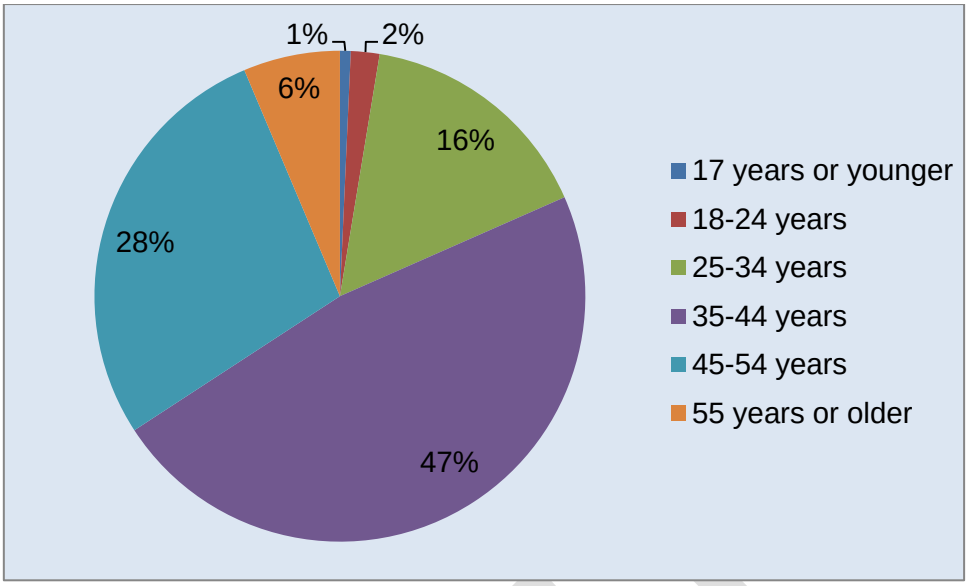


Figure 3: Age of respondents **(Question 3)**

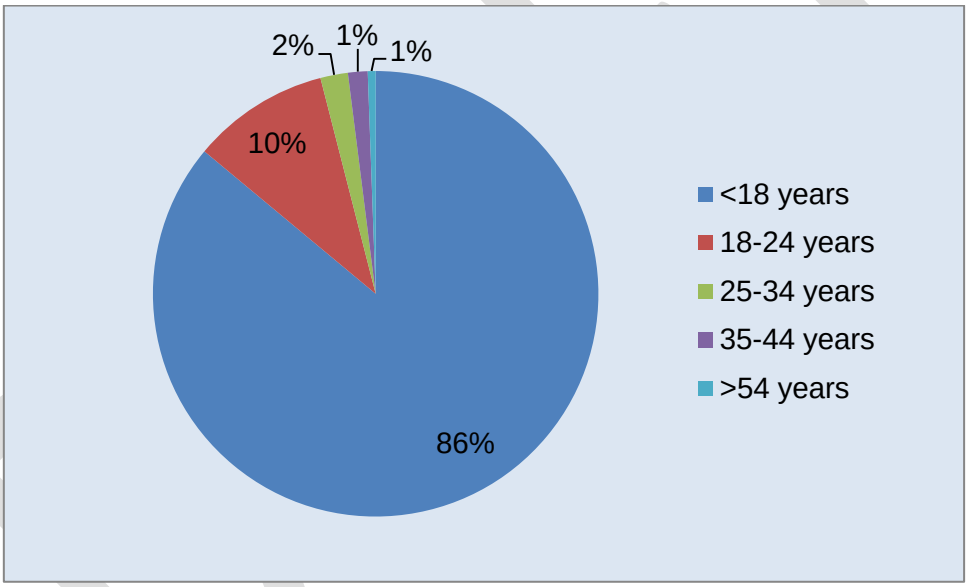


Figure 4: Age of relative with ASD/NDD **(Question 4)**

Section 2: Registry

Section 2 asks about views on establishing an ASD/NDD registry in Ireland. 92% either strongly agreed or agreed that Ireland needed a national ASD/NDD registry. Only 2% did not agree that it is required. Furthermore, 85%% were in favour of their details or their relatives details being included in the registry.

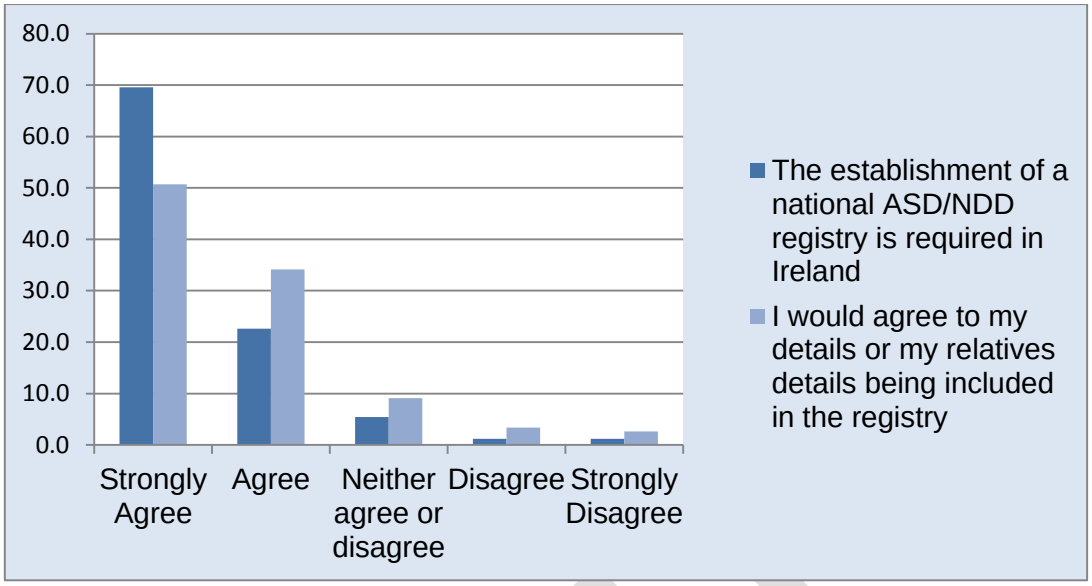


Figure 5: Establishment of a national ASD/NDD registry (%) and including relative's details in the registry (%) **(Question 5)**

Regarding the scope and aims, most respondents agreed that it is a priority for research including clinical, biomedical and environmental research. The most important focus for the registry according to respondents is to inform health, education, social care aspects of service development.

	Strongly Agree	Agree	Neither agree or disagree	Disagree	Strongly Disagree
An important focus for a registry for ASD / NDDs is to inform service development (health, education, social care)	80	17	2	.5	.9
An important focus for a registry for ASD / NDD is to provide information about ASD and related NDDs to affected individuals/ families	74	20	3	1	1
An important focus for a registry for ASD/ NDD is to provide information regarding the extent of ASD/ NDD in Ireland	72	24	3	.7	1
An important focus for a registry for ASD /NDD is to facilitate research (e.g. by providing contact between affected individuals and their families and researchers)	73	21	2	1	1
Biomedical research – i.e. support the identification of genetic or other biological causes of ASD/ NDD	65	26	6	1	2
Environmental research – i.e. supporting identification of environmental factors	61	31	7	.7	.7
Clinical research – i.e. identification of clinical problems that are associated with ASD/ NDD	71	26	3	.2	.7

Table 1: Scope and aims of an ASD/NDD registry in Ireland **(Question 5)**

Over 90% agreed that data regarding clinical information, health service use, education, disability service, occupational activities, residential arrangements and community supports should be included.

	Strongly Agree	Agree	Neither agree or disagree	Disagree	Strongly Disagree
Clinical details regarding autism or NDD	67	28	3	1	1
Clinical details regarding associated medical conditions	65	29	4	0.5	1
Information on educational placement	71	23	4	1	1
Information on educational resources received	72	22	3	1	1
Health Service use (e.g. medical or mental health services)	70	25	3	1	1
Disability service use	69	27	2	1	1
Occupational activities	70	24	4	1	1
Residential arrangements	62	26	8	2	1
Community based supports	72	22	4	1	1

Table 2: Type of data that should be recorded **(Question 6)**

In the same question, respondents were also asked to comment about when they feel is an optimal time to put data into a registry, e.g. point of diagnosis, sometime after the diagnosis or at different time points? The most common comment was at the point of diagnosis and with regular updates.

Question 7 of the online survey asked respondents to give their experience of participating in other registries. Among the 147 who reported participating in other registries, the largest group belonged to those with participating with the NIDD. However, many reported they were unsure of the use or benefits of the NIDD.

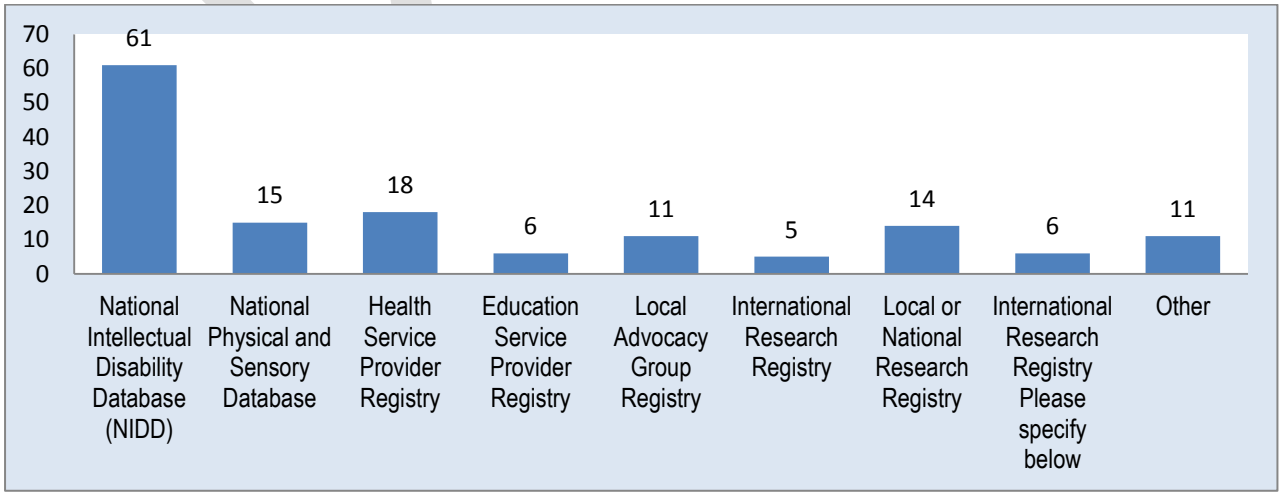


Figure 6: Previous experience with other databases **(Question 7)**

When asked about potential barriers in **Question 8**, insufficient time was selected as the main barrier, followed by consent. Most respondents disagreed that lack of interest would be a barrier.

Question 9 related to data privacy and protection.

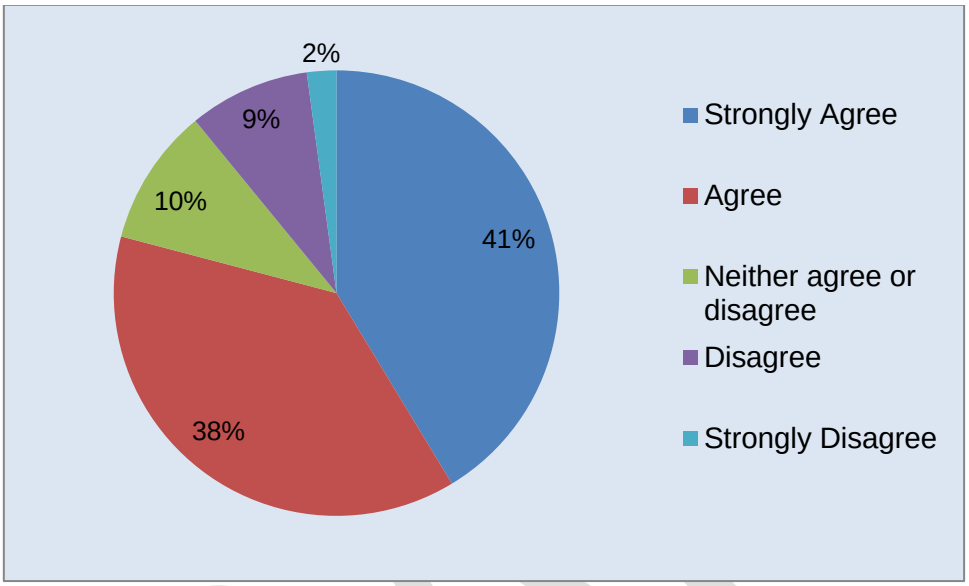


Figure 7: Concern that data could be accessed inappropriately **(Question 9)**

Respondents were most concerned about access to data by medical insurance companies, followed by private companies such as pharmaceutical companies. There was also concern that police and certain government departments could access data.

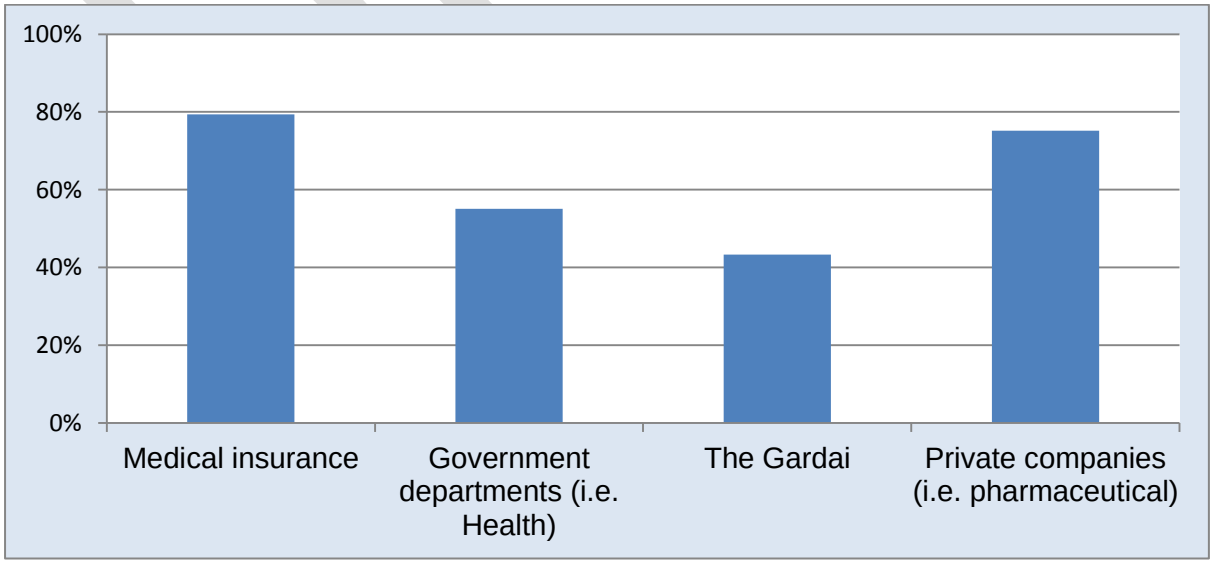


Figure 8: Specific data privacy and protection concerns (strongly agree or agree) **(Question 9)**

Question 10 asked who respondents felt should be permitted to enter data in the registry. As depicted in Figure 10 below, based on a likert scale of 5 (strongly agree) to 1 (strongly disagree) there was strong support for health professionals followed by parents or relatives, educational professionals and also adults with ASD/NDD to information to the registry.

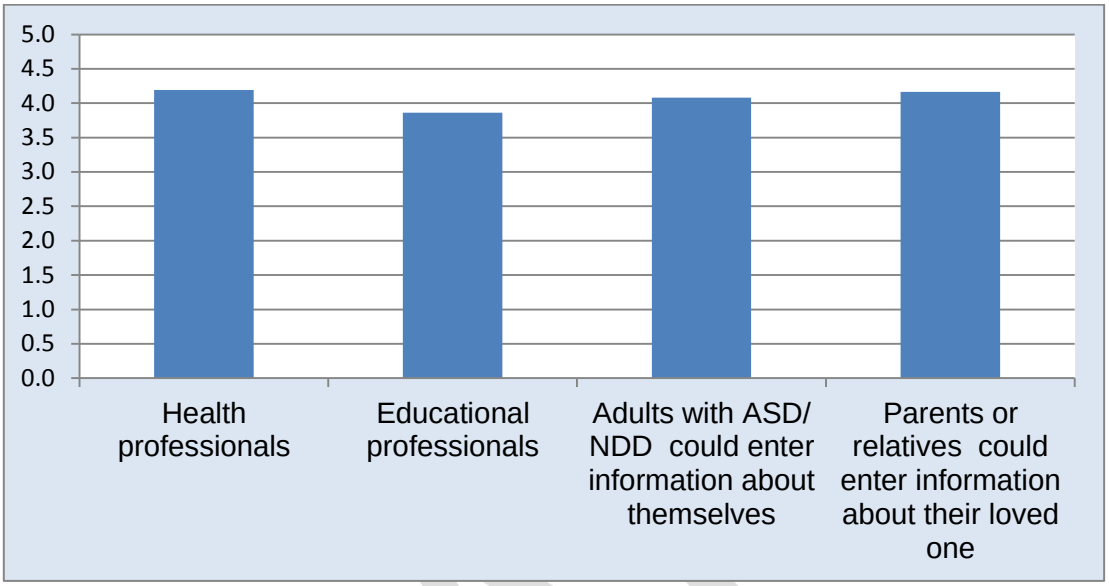


Figure 9: Who should enter data in the registry? **(Question 10)**

Question 11 of the online survey relates to accessing registry data.

	Strongly agree	Agree	Neither agree nor disagree	Disagree	Strongly Disagree
Health professionals working in HSE service provision	53	38	4	2	2
Health professionals working in non-statutory service provision	33	34	20	9	4
Health professionals working in private service provision	32	32	17	12	5
Education professionals	45	37	10	5	3
Professionals working in government or state run agencies (e.g. Dept of Health, HSE, Dept of Education, NCSE)	40	39	12	6	3
Researchers in clinical services	46	34	12	4	2
University/ higher education based researchers	42	33	16	5	3
Advocacy groups supporting individuals with ASD/ NDD	43	35	13	6	3
Individuals with ASD/ NDD or their family members	54	32	8	2	3

Table 3: Who should be able to access the data in the registry? **(Question 11)**

The following is a selection of additional comments from **Question 11**:

"If anonymous I would have no problem with any access to this information"

"I don't think this is something we need to be overly careful of as if it is anonymised data we do not really have anything to be concerned about. I would suggest however that, as a model to ensure long-term funding for the service, data access could be licensed to anyone who is interested but a fee being required for the year. I also think, in the interest of getting maximum participation, we should perhaps consider offering individuals to absent their data from versions not given to Statutory bodies or non-commercial researchers to allay any fears, as unfounded they may be, that they might have; All the above should be approved and monitored by an ethics committee or named board of responsible persons".

"Confidentiality is a huge issue. In my experience nothing is anonymous".

Question 12 asked about perspectives on linking data to other registries

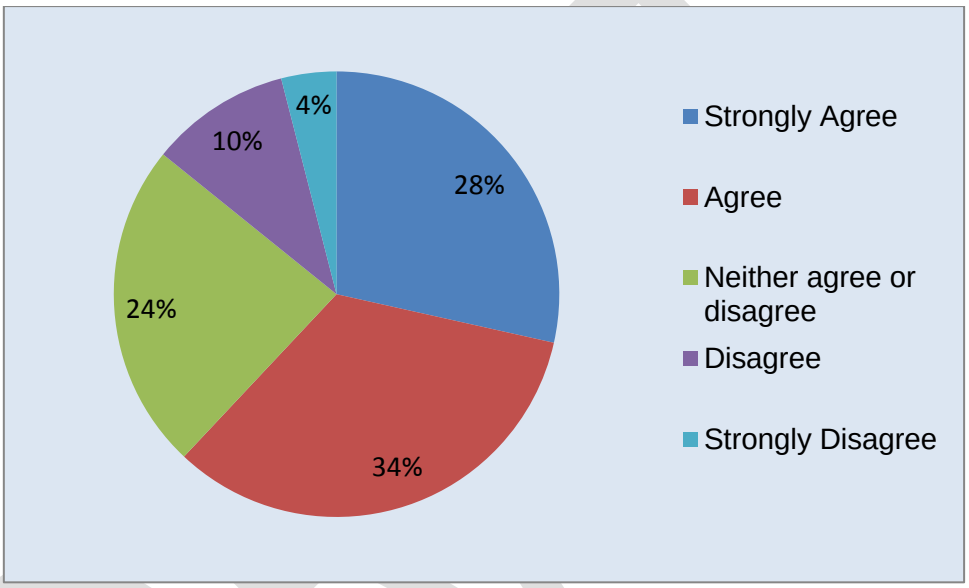


Figure 10: Linking data anonymously to other databases **(Question 12)**

The response about linking to other databases was overall positive. Over 60% would agree to share their data anonymously, while only 14% would be against their data linked anonymously with other registries.

Additional comments from participants include the following:

"I think reassurance about privacy will be crucial to the success of this. A lot of people with children on the spectrum have had disputes with HSE/education etc. about service provision and the thought of these agencies having access to data on the data base which is identifiable to an individual will put people off".

"I agree as one person's information can still solve another person's problem".

"You are making people with ASD vulnerable by doing this - they need to be accepted not something that needs to be cured".

"There is a need to know how many cases there are in order to better provide serviced and support to these areas. Schools are struggling for resource hours and the extent of the support needed is growing. I think it's crazy that this information is not compiled already".

"Unsure of what current database such as NIDD actually does".

Section 3: Biobank

Section 3 of the survey was related to the development of a national autism and neurodevelopmental disorders biobank. The following questions were targeted at individuals with ASD or their family members only and concerned the placement of biological materials into a biobank for research purposes. Thirty-three percent of respondents reported that their child had, at some point, provided blood or saliva samples for DNA testing. For a proportion of respondents who had received genetic testing and subsequent diagnosis, there were elements of uncertainty as to what happened with the blood sample or if it was stored (Figure 12).

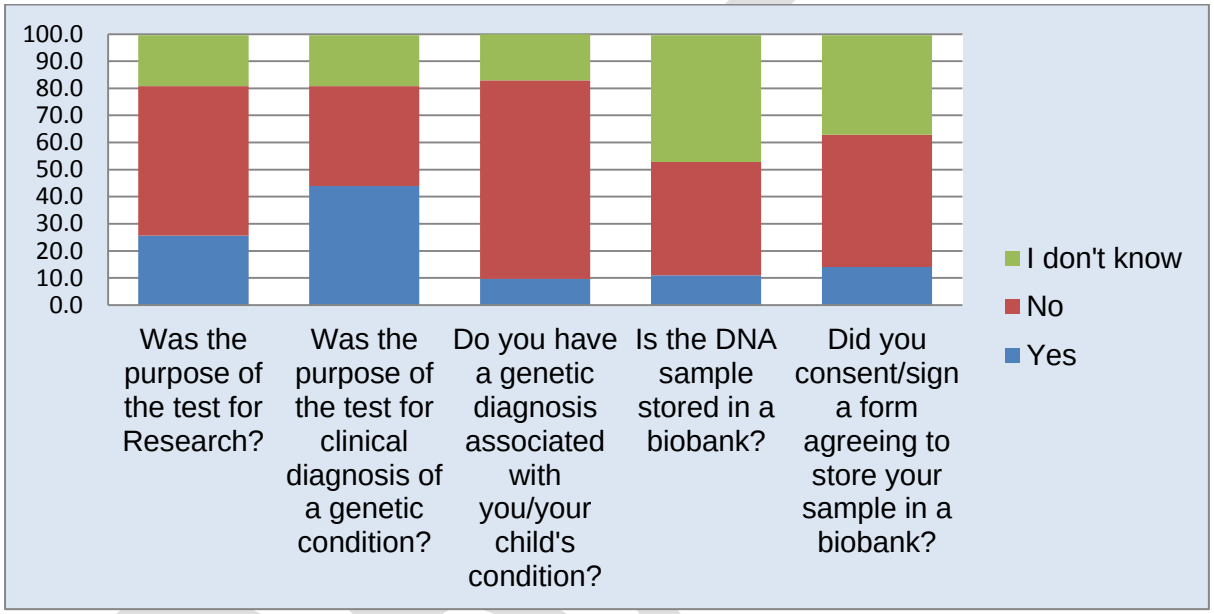


Figure 11: Previous experience with biobanks (%) **(Question 13)**

The following is a selection of comments highlighting a need for greater information about biobanks and clarification about the process for those who receive testing:

"DNA extraction was taken and held but nobody explained this was being done I found out as it was stated on a report".

"Bloods were taken at Tallaght hospital to be sent away I think I may have consented to research and biobank storage of data but cannot be certain".

"The collection of biological materials is a very disturbing prospect as there would be no guarantee of their security or what purposes they were used for".

"We had genetic screening done & have taken part in a few which were saliva based but can 't remember if they were stored but have no issue as would love a cure"

"I signed consent forms but can't remember if a biobank was mentioned. I would have no problem with samples being stored in a biobank".

Comments highlighting need for urgent and efficient genetic diagnostic testing:

"I think the whole diagnosis process lacked medical and scientific input".

"Fragile X testing in Ireland is completely outdated and inadequate and my family has been waiting years for a proper test. This is not acceptable"

"Our son had blood sample taken to test for genetic conditions as part of his diagnosis. We weren't asked for his sample to be entered into a biobank at the time".

"Would be prepared to have samples taken and stored".

"I think the criteria for diagnosing ASD should be much tighter than currently in use nationally"

Only a small number of respondents commented with confirmation in which biobank their samples were stored in:

"Immunological and genetic samples maintained since the birth of our first son was born and died with the Autism Genome Project"

"Samples taken 16 years ago around the time of diagnosis, sent to Cambridge"

"Trinity College Dublin Genome Study"

"Genetic study donation whilst at TCD years ago, relating to neural tube defects".

Question 14 specifically targeted those who have given DNA samples and if they knew who could access the samples hence the low number who answered this question. Twenty one individuals reported that clinicians and researchers in Ireland could access their samples; 12 reported that researchers outside of Ireland could access their samples; and two reported that their samples had been accessed by the Gardai.

Question 15 asked about the scope and aims of biobank. The majority (84%) of respondents supported a national biobank and 77% agreed they would be willing to contribute biological samples to the biobank.

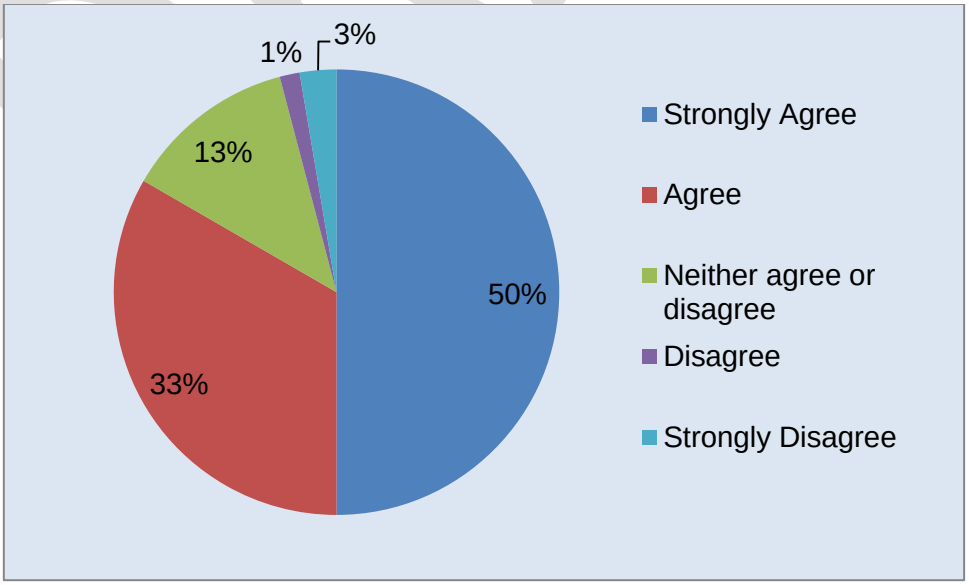


Figure 12: Development of a national biobank **(Question 15)**

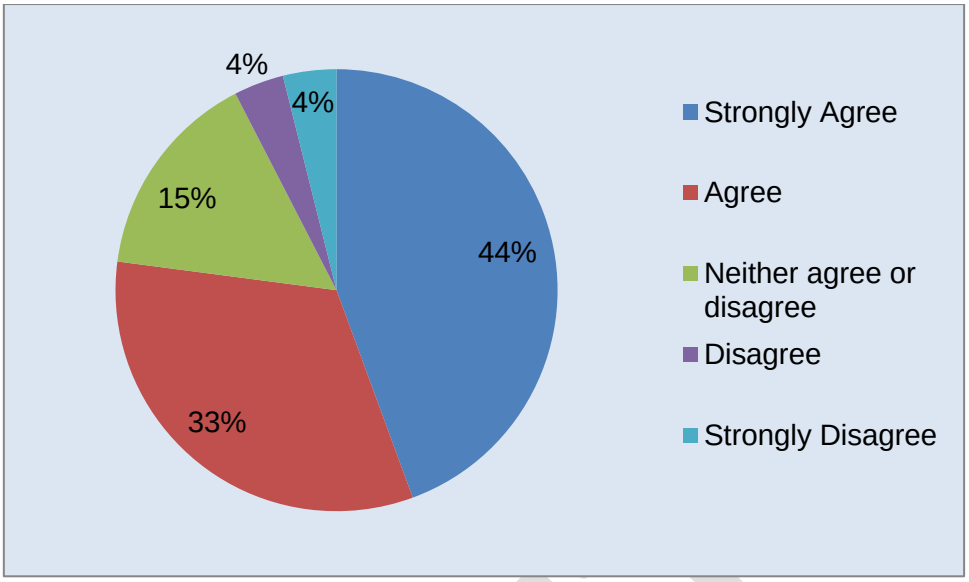


Figure 13: Willingness to contribute biological samples to the biobank

Question 15 also asked respondents about the objective of a biobank.

	Strongly agree	Agree	Neither agree nor disagree	Disagree	Strongly Disagree
A biobank should exist to provide more information on the genetic causes of ASD/ NDD	54	33	10	1	2
A biobank should exist to support biomedical research in ASD/ NDD	53	33	11	1	2
A biobank should exist to identify individuals with specific genetic subtypes for targeted research, e.g. stem cell research or clinical trials	46	30	16	4	4

Table 4: Objectives of an Irish Biobank **(Question 15)**

Qualitative themes in support of a biobank included the following:

"It could provide very interesting results".

"I think that it is essential to include individuals who have autism at every step of the research."

Comments indicating insufficient knowledge highlighted the need for an awareness and information campaign:

"I would want more information on this if I was to be involved".

"Probably don't know enough to support my answers".

"Would need more information about it. Undecided at present".

Comments indicating concern included the following:

> *"Need legislation to prevent discrimination by life insurance etc. like happens with cancer genes".*
>
> *"The biobank should only exist for research not by private pharma companies".*
>
> *"I think genetics is an important area however, I would be concerned with pregnant women terminating a pregnancy on the basis of having a child with Autism".*
>
> *"I have concerns about the misuse of such information".*
>
> *"I agree under the provision that all samples be anonymous and access to all information on test participants remain confidential at all times".*

Comments from respondents who do not support a biobank included:

> *"I would have concerns if biobank was used for searching for a 'cure'".*
>
> *"I understand that a bio bank would utilise data and material samples to identify the cause or causes of ASD / NDD. I believe that is a worthwhile endeavour in order to help professionals, caregivers and service users achieve a better understanding of how to support those with ASD / NDD. I do not wish to have a bio bank using samples of my child's DNA in order to isolate and eliminate the autism 'gene'. My child deserves her place in the world and I will not participate in any study, database, biobank, or research trial that proposes to render her and others with ASD / NDD 'obsolete' in the future".*

Question 16 probed the issue of who would have access to the biobank. Over 85% were in agreement that Irish researchers should have access. Close to 70% felt that international researchers should have access. Only a small number of respondents felt that private companies should have access; over 50% disagreed completely with private companies having any access.

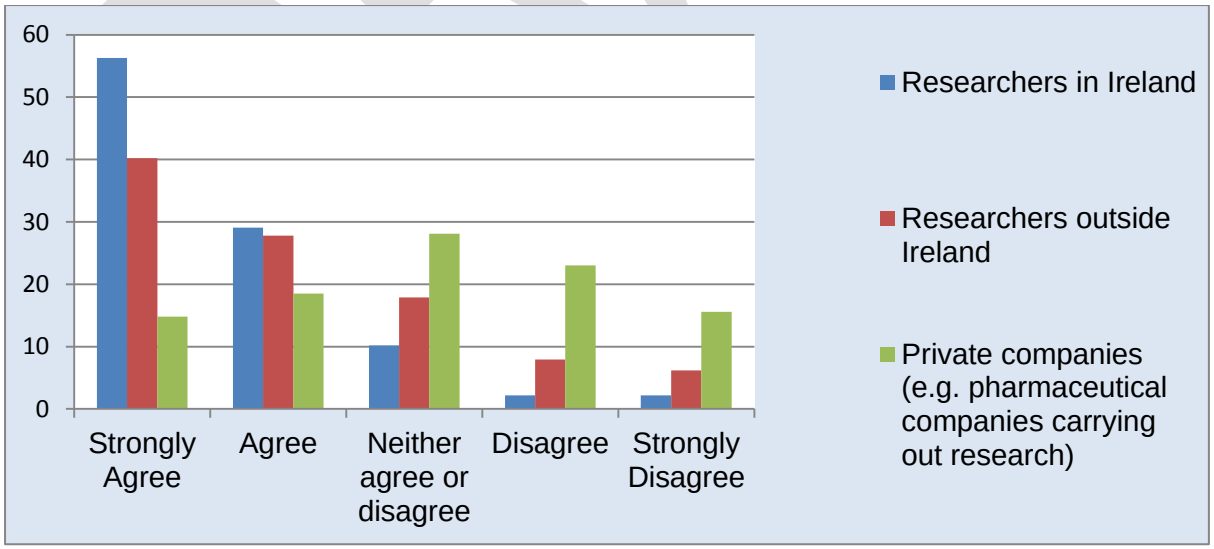


Figure 14: Access to biobank data **(Question 16)**

Question 17 was included to draw out if respondents were concerned about specific organisations accessing the biobank.

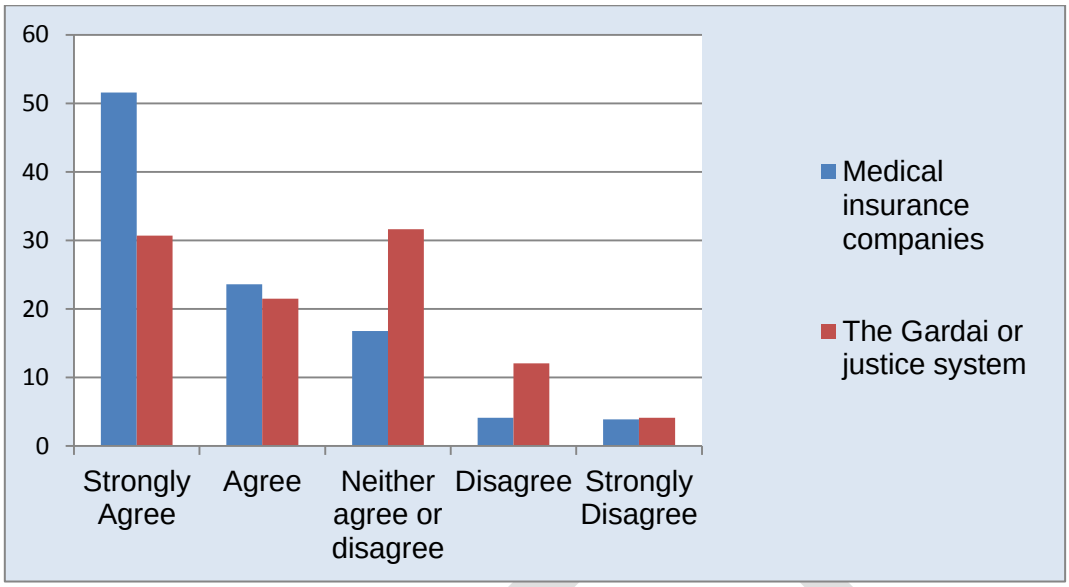


Figure 15: Concerns about specific groups accessing biobank (%) **(Question 17)**

Respondents were asked to specify if there were other groups they felt should not have access to the biobank. Responses included: *"social services, banks, Dept of Education, companies selling autism aids of any kind, pharmaceutical companies, employers, media".* Some have no issue with the Garda while others feel *"Gardai have a habit of jumping to conclusions assuming negative behaviour attributed to autism, rather than fact".*

Section 4: Linking the Registry and Biobank

Question 18 asked about linking data anonymously from the registry with the biobank samples. The majority (79%) agreed data should be linked. **Questions 19** and **20** sought feedback on possible uses of a registry and biobank, respectively.

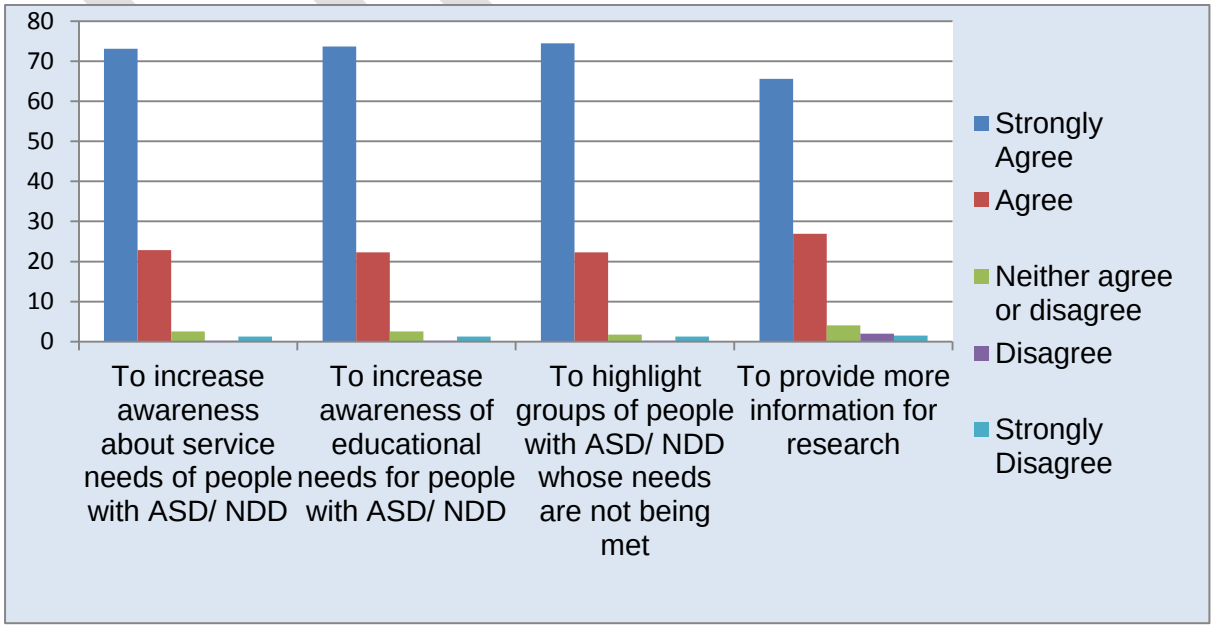


Figure 16: Uses of registry (%) **(Question 19)**

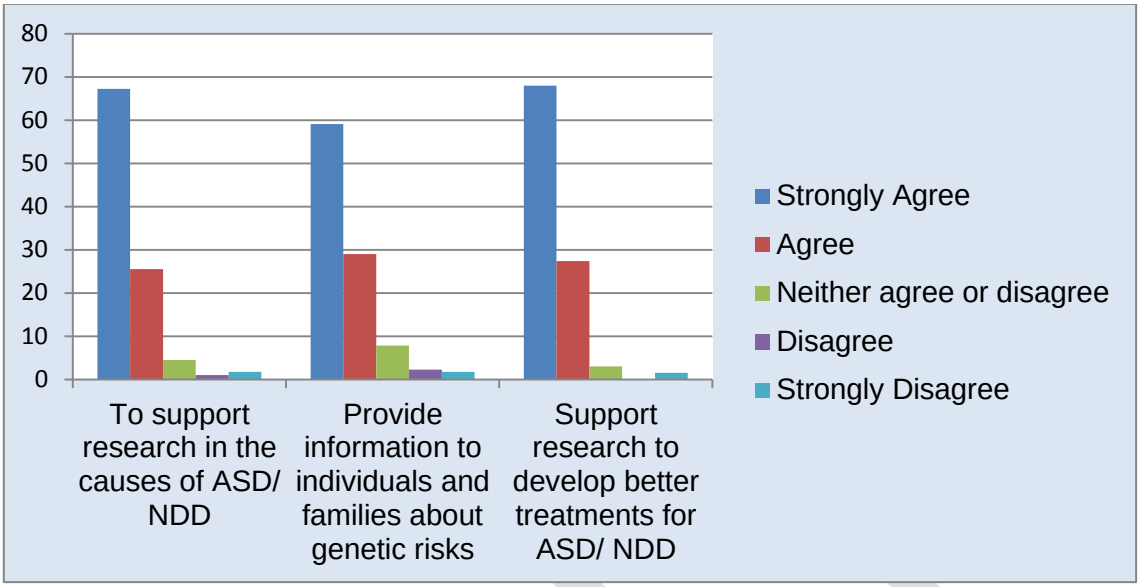


Figure 17: Uses of biobank (%) **(Question 20)**

A number of respondents gave additional input to the uses of a registry and biobank:

> *"To highlight the stages at which further supports and interventions are required during the lifetime of individuals and the impact the lack of these supports can have on the individuals(and further higher costs incur from illnesses which occur from lack of understanding and support)".*
>
> *"To help people transitioning from childhood to adulthood, with more understanding that ASD children do become ASD adults".*
>
> *"To provide data to assist planning for service provision in particular when grown up".*
>
> *"I think it would be also important to look at the talents (the positive achievements of individuals with ASD)".*
>
> *"Identify environment factors that may contribute to presentation of neurodevelopment disorders e.g. pre-term birth etc."*
>
> *"To give an indication as to what professionals are required to treat people in the future and to help teenagers when searching for careers"*
>
> *"To note the prevalence of autism. It will give a scientific picture of how many people are affected by the condition and allow for whole of government planning"*

Question 21 related to ethical considerations and governance. The creation of a registry and a biobank will require the establishment of ethical guidelines concerning how the data may be accessed and utilized. Typically a governance structure will oversee the activities of a registry/biobank and ensure that ethical guidelines concerning data access and storage are fully maintained.

Respondents ranked the following individuals and their involvement in the governance of a registry/biobank as shown in Table 7 below. Strongest support was for individuals or relatives, followed by advocacy groups and researchers.

	Strongly Agree	Agree	Neither agree or disagree	Disagree	Strongly Disagree
University/ Higher education based researchers	47	33	15	4	2
Representatives of advocacy groups	48	26	17	6	2
Representative of health service provider	37	37	13	8	4
Representative from Government Department or Agency (e.g. Dept of Healt, Education, HSE, NCSE)	38	35	15	9	3
Representative from Advocacy groups supporting individuals with ASD/ NDD	54	29	10	5	2
Individuals with ASD/ NDD or their family members	60	27	8	2	2

Table 5: Governance of the registry and biobank **(Question 21)**

Question 22 asked how findings from the ASD/ NDD registry and biobank be communicated. There was strong agreement that data should be communicated back to families and service providers through both online and written communication medium, and conferences/workshops.

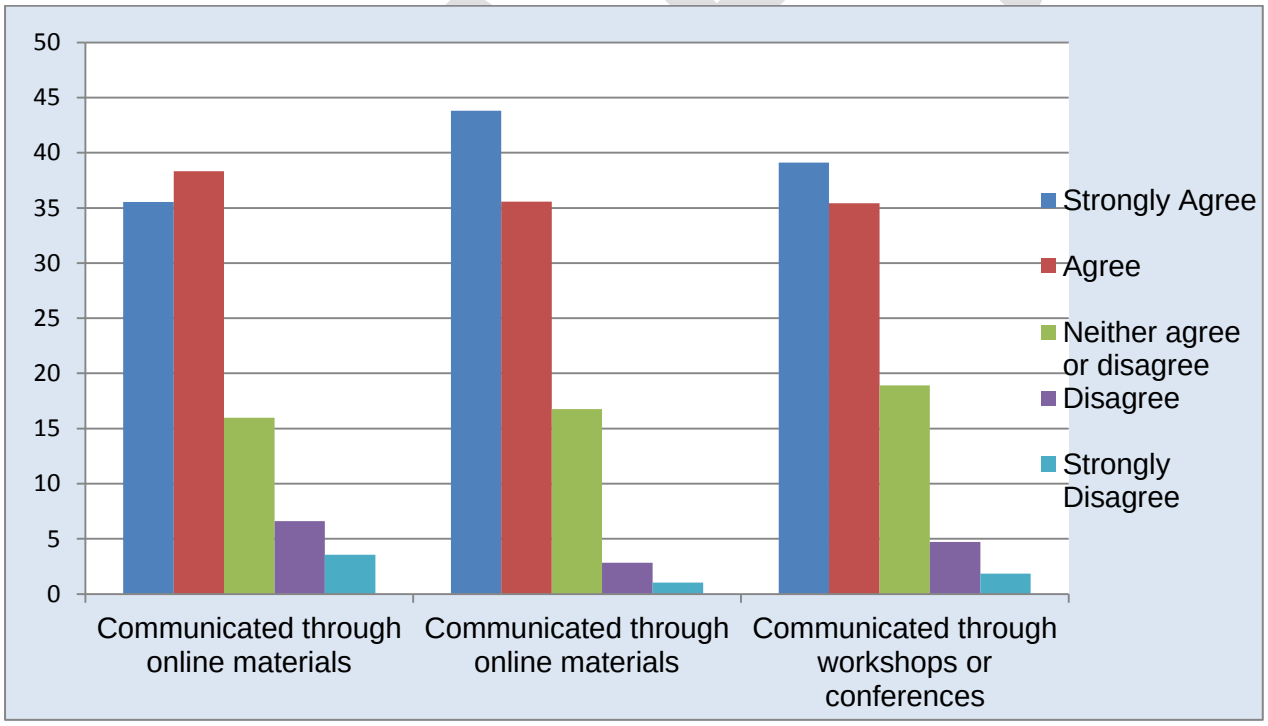


Figure 18: Communication of findings from the registry and biobank (%) **(Question 22)**

The final section of the survey invited respondents to provide any further thoughts they might have about a national registry or biobank. Here follows an overview of the comments received. Qualitative themes in support of the development of an Irish registry and biobank for ASD/NDD included:

"Thank you for starting this immensely important project".

"I feel this needs to be set up asap as ASD is not being addressed scientifically or medically more on the basis of psychologist reports. My child never had bloods taken to see what is going on biologically with him".

"Research and new ideas and methods of helping and assisting people and their families and friends with ASD are very important'.

"It seems like you good people have thought of everything. Well done".

"I really hope this works as it would be a fantastic tool".

"Information about autism and communication with others in a similar situation are key to learning how to live with autism. The services such as SLT and OT required by these children are not provided by the HSE. I feel much of the information we have come across over the last three years has happened by accident and come from such a variety of sources and services. There is no one central location to head to for information, services seem to be disconnected and provided in a haphazard fashion without an overall direction".

"I think this is a great idea for national registry and bio bank.it appears very positive for children and adults with autism for the future".

"Should have been established years ago, would gladly volunteer info and time! Good luck".

"Hope the registry is put to good use. One often wonders about the NIDD as it would appear that the Authorities don't use it for planning purposes!!"

"Possibly break it down to areas in the country-it might show patterns, numbers etc that might help research, as environmental factors could be as high as genetic ones in contributing to ASD/NDD".

"My son's autism is as a result of rubella during pregnancy. Would welcome research into this".

"As a parent, I am very supportive of this. More information needs to be collected in order to carry out research into possible causes and treatments and equally important to determine which health therapies and educational supports provide the best outcomes".

"Thank you for caring"

"I am very excited as a parent of a young child with autism that this biobank will be set up as I think it will be a fantastic resource".

"I personally wouldn't have any faith in a registry if Government officials were involved, especially this current government. Well done though, this is a great idea".

Comments showing feelings of concern included:

"The NIDD hasn't worked and is not being used. Why will this be any different?"

"Security of information concerns".

"No power to the government".

"Anonymity is paramount. If it is breached people will not consent".

"There is an ethical question to consider of whether autism needs to be cured".

"I would be concerned that if genetic risks were identified that it might encourage people to terminate pregnancies based on risk, also my concern would be that this information in the wrong hands could be used badly. An extreme example would be the register of those considered mentally impaired in Nazi Germany which was used

to kill undesirables. Anonymous research is great, but targeted and linked data could potentially be hacked and used against people".

"I do not like the idea of a national registry of people with ASD. As it stands, in my experience, children with ASD diagnosis are being "labelled" in school, their needs are not being catered for, they are being dealt with as per their "label" as opposed to their needs. Service provision is extremely poor unless you have the money to pay for private therapies yourself. Diagnosis is of a poor standard also. Parents are being told what their child cannot/will not be able to do, based on the "label" that is applied to the child. Why should the child continue to carry this label and have it linked to them for life".

Comments with recommendations from respondents:

"A new biobank should work closely with the National Centre for Medical Genetics as to date in Ireland this is where patient samples/DNA are currently stored".

"The main thing I have to suggest is for the registry. If it is to be used for service provision purposes, every child who is diagnosed needs to be included in this unless a parent 'opts-out'. It should be something that you need to opt out of instead of choosing to opt-in. Any clinician who is diagnosing a child or an adult should be required to obtain consent or give an opt-out opportunity to the family. It should be part of the diagnosis procedure (public & private). A lot of children are diagnosed privately because of the huge waitlists for the public process. If it's not possible to make sure that public and private professionals will do this, I would suggest that the involvement with the registry happens at the school age, perhaps with the schools themselves or with the NCSE and the SENO who will be given all the reports concerned and can then at that stage update the registry. Whatever about public or private diagnosis, every child at the moment will need to access the school system in Ireland I think initial school or SENO contact is the best to access the families with the 'system' in its present form".

4.3 Findings from Public Consultation Meetings

Response to the creation of an Irish Registry and Biobank was overall positive at each of the four regional meetings. The passion and frustrations of parents, families and service providers who attended was palpable.

In summary, those in favour of the registry and biobank were keen for developments in research and keen for the registry to help alleviate issues such as huge gaps in diagnosis, service provision and education for individuals with ASD. They welcomed the initiative and felt it was long overdue. However, there were some common concerns and questions around data protection and privacy that arose at each meeting. There was an element of suspicion and cynicism amongst a number of attendees with regard to who would have access to their data and fear that welfare benefits could be removed. There was a sense that people have been so worn down by empty promises in the past that their default reaction to anything new is suspicion and doubt.

To give an insight and direct feel for the perspectives of the public, here follows a compendium of comments from the meetings:

Comments from attendees **in favour of** the Registry and Biobank:

"There is a huge appetite for research across rare disorders so participation should not be an issue. It is difficult to justify putting money into project that might only answer one question on one rare disorder and be relevant to 10% of the people. We

can however make a persuasive approach and make a powerful case with this registry. We cannot achieve anything on our own".

"Autism services and autism specific needs aren't being dealt with. Future planning and placements are breaking down in secondary schools. The projections from this registry will create the economic argument that front loaded investment in early intervention will help the economy. This data is timeless so we are very pleased that it is happening".

"A registry and biobank would uncover more and more research. I place my trust in the individuals running this project and have no problem sharing. I am on several registries already".

"Very important to have generational information".

"I hope with this research attitudes will become more positive. The individuals have such skills and values that contribute to the economy and this should be put out front, they are not kids to be put in a little room or aside. They have a wonderful future and thanks to people like our panel members with autism and Asperger's here tonight, parents can see the positive value".

"The larger amount of clinical data that we can feed into the better – information is power".

"I see it as a very powerful tool. This will generate data to tell the government this is how many there are, this is what's needed, there is the data now do something about it. Need to get something done about this now".

"All I wanted when I got my son's diagnosis was a cure, I wanted him to be able to speak and everything that impaired his life and our lives to be gone. I am totally for research if there is anything that can be done"

"Effective research that will impact on everyday lives which requires large groups of participants is needed. If you have questions to be answered you need a lot of population data so I am a keen advocate".

"When given the diagnosis it can feel like a life sentence and you search for answers. Accurate information and well documented therapies are so important...this biobank and registry give me hope for a positive future for this generation and my child growing up and hope that help and proof will come through this registry".

"We need evidence and research. It is wonderful that it has been started. We have many questions like why some of your children are affected and not others. By having a biobank it is the only way. People power is important. A biobank would help identify if there is a genetic influence which could reoccur. It will inform policy development. Lots can be done by the people involved and who it will help by sharing information and people power".

"People should know that the information given will be in safe hands and should not be afraid reaching out to people on the spectrum itself. Even more severe aspects of ASD such as such as hypersensitivity to sounds, gag refluxes when eating foods, motor coordination, anxiety, depression – that is where I see the research from the registry and biobank making huge improvements".

"I think the biobank is really positive and informed feedback will be very good. Getting an understanding of gene cluster from samples will be great to find biomarkers and develop treatment".

Concerns and questions around the process to be addressed:

"Parents have had to fight for what they want. It is very hard for parents constantly being beaten down by the public system. I do welcome the initiative. There needs to be more open engagement but people affected are so downtrodden and being dictated to as what is available. The families need to know they are in partnership and not working against a system so be aware there is that mentality".

"Respect and anonymity are the crucial things to me".

"Parents are the voice of their children with special needs. I could fill a room with all the information I have, where does it all get you. I think this is a wonderful idea setting up a database. I have given information so many times, if we fill in this database can it go somewhere and not just be left there?"

"I have gotten no feedback from previous studies I participated in; will there be a feedback mechanism?"

"My biggest concern is about the biobank, proper diagnosis and difficulty pulling data. In Ireland the current problem is resources are linked to labels and parents are under pressure to get a diagnosis and a label. If you don't have diagnosis you don't have resources so there is bias. There is a huge variety of diagnosis especially now with the DSM5. My feeling is with the variety of diagnosis can it be scientific? Will everyone be rescreened for a validated diagnosis?"

"I am aware of genetic research but think it is a lot to put children through and then deal with the stigma that can be attached to the children following a diagnosis".

"I am supportive but making that decision for a child who doesn't know the implications and not knowing what kind of information can be taken 10 years down the line. Is it a fair decision?"

"I have Asperger's and am also a researcher so I can see things from both sides. Privacy is a huge concern for me".

There was a small number of attendees at one particular meeting who were concerned that genetic research would "wipe out" people with autism. These individuals were diagnosed on the autism spectrum and also had children with autism. They supported the registry but were against the development of a biobank. *"For someone with autism hearing talk of a cure makes them feel like they are not important".* They were assured the goal was to support and not eliminate people with autism.

Public consultation meeting held in Trinity College Dublin

Meeting with the Health Research Board (HRB)

A discussion with the HRB was held on 27 February 2014. The HRB manages two disability databases and engagement was sought with them to enhance possible areas for collaboration and to avoid duplication of effort. It was also agreed that engagement with the HRB was useful because of their long-standing experience in developing and managing national databases.

Meeting with the Department of Health and Department of Education

A discussion with Senior Principal Officers of the Department of Health and Department of Education was held on 28 February 2014 in the Department of Health headquarters in Dublin. The officers were interested in the initiative but said they would have to submit the presentation and information to various internal reviews. They are currently reviewing frameworks for new systems and future strategies. Budgetary constraints were a concern for implementing any new system.

Analysis of other Registry's and Biobanks

A selection of other registries were analysed to establish criteria for what makes the best successful model and to identify and pre-empt the challenges that arise with registries and biobanks. The analysis includes models specifically in Ireland, as well as European and multi-national collaborative models.

Registry	**National Intellectual Disability Database (NIDD)**
Function/Aim	National service-planning
Country & year established	Ireland Established in 1995
Ownership & Funding	Administered by Health Service Executive (HSE) but managed by the HRB on behalf of the Department of Health and Children
Registration	25,500 Registration is voluntary
Data collected	Demographics, current service use and future service need
Strengths	Provides information on trends in demographics, current service use and future service need.
Challenges/ Weaknesses	Changes on individuals circumstances may not always be communicated in the data With extremely long wait lists for services the data collection process and reporting cannot be completely accurate. Participation is voluntary so it does not capture all individuals who need services and support.
Data access	Regional HSE & Service Providers
Technical aspects	The NIDD is hosted in the Local Government Computer Services Board. The HRB develops and maintains software to facilitate the collection and reporting of NIDD data. NIDD software is a web enabled centralised system through which Health Service Executive areas and services providers can access their relevant information. The HRB designs and delivers a standardised training programme and provides frontline support to all NIDD software users. Any issues outside of this remit are dealt with by external software developers and are channelled through the HRB.
Communications	Annual report published by HRB

Registry	**National Physical and Sensory Disability Database (NPSDD)**
Function/Aim	Aims to provide a comprehensive and accurate information base for decision making in relation to the planning of specialised health and personal social services for people with intellectual, physical or sensory disabilities
Country & year established	Ireland Established in 2002
Ownership & Funding	Managed by the HRB on behalf of the Department of Health and Children.
Registration	27,000 Registration is voluntary
Data collected	Demographics, current service use and future service need
Strengths	Collection and reporting of data on people with a physical, sensory or speech and language disability in the Republic of Ireland, who wish to register
Challenges/ Weaknesses	Cannot provide any definitive epidemiological statement on the number of people with a particular type of disability. Therefore the database may not cover a proportion of people living in Ireland who have a physical or sensory disability. Furthermore, the database does not currently include those who are aged 66 years or over.
Data access	Regional HSE & Service Providers
Technical aspects	The NPSDD is a web-enabled centralised database hosted by the Local Government Computer Services Board (LGCSB). The HSE accesses the NPSDD via the Government Virtual Private Network (VPN) and staff have access only to the data relevant to their region. The HRB provides full software training for all users and is also responsible for frontline software support. Any software issues outside of this remit are dealt with by an external software developer.

Communications	Annual Report

Registry	**Autism Genetic Resource Exchange (AGRE)**
Function/Aim	Largest private, open-access repository of clinical and genetic information dedicated to help autism research
Country & year established	Established in the United States in1997
Ownership & Funding	AGRE is a science programme of Autism Speaks. It is managed by a Steering Committee composed of experts in genetics and neurological disorders who set standards and regulations for the AGRE's clinical and genetic data. They ensure that the data is being used for good science within the autism researcher community. AGRE is funded by Autism Speaks and National Institute of Mental Health
Registration	2,000 families
Data collected	Biomaterials and data on families with members on the autism
Strengths	Enables an unlimited number of scientists to join in the search for the factors that influence autism
Challenges	AGRE had to discontinue in-home data collection in order to expand the resource with collaboration data so role changed to that of a Data Coordinating Center (DCC), receiving data and biomaterials primarily from outside researchers.
Data access	Over 150 research groups worldwide with access to extensive clinical data, tissue, and biospecimens from over 15,000 individuals with autism spectrum disorder (ASD) and their relatives
Technical aspects	An infrastructure that provides a centralized hub to manage data that maintains the flexibility to incorporate new studies and new results such as genetic, EEG and imaging data. The Research Exchange Database allows for data analysis across collections to assist in the AGRE DCC's mission to accelerate the pace of autism research. In addition, they will also develop a web-based system that will allow research participants to more easily participate in studies via user-friendly web-based technologies.
Communications	Scientific publications, Website, Newsletters for families,

Registry	**Interactive Autism Network (IAN)**
Function/Aim	Facilitate research that will lead to advancements in understanding and treating ASD through IAN Research and IAN Community.
Country & year established	United States Established 2006
Ownership & Funding	IAN's Scientific Advisory Board is made up of researchers, physicians, scientists and other professionals with special expertise in the fields of autism spectrum disorder and research. IAN's Parent Advisory Committee is composed of parents of children with ASD. Committee members also have experience as advocates or volunteers for autism-related organizations. IAN is funded by Autism Speaks and the Simons Foundation. IAN also has a grant from the National Institutes of Health.
Registration	Over 30,000 individuals registered
Data collected	Longitudinal data on variety of topics from diagnosis to comorbidity, from family history to treatments and outcomes, and bio-specimens and related clinical information
Strengths	"IAN Research" is a strong tool for recruiting participants for research solving one of the major difficulties that autism research projects face by connecting researchers with individuals with ASD and their families who can take part in appropriate local and national research projects. "IAN Community" is an online learning environment that where everyone concerned with ASD can learn more about autism research. The tool allows individuals with ASDs, their families, and friends to connect with researchers, therapists, educators, and other professionals in the autism field thereby increasing learning and knowledge for all stakeholders.

	Another advantage of IAN is that it incorporates requires measurement of non-biological phenomena including behaviour, communication and social styles which is essential for autism research.
Challenges	Ensuring participants fully complete all study-related questionnaires
Data access	Researchers and families of participants
Technical aspects	Based on mdlogix Clinical Research Management System which is a suite of pure web application modules that support and integrate a comprehensive set of health research processes, including subject recruitment, administrative management of the research, protocol execution, financial workflow, specimen tracking and banking, and data mining.
Communications	All communications and research feedback is via a web-based platform.

Registry	**International Collaboration for Autism Registry Epidemiology (iCARE)**
Function/Aim	Collaboration to create repositories so that you can combine samples and get much larger numbers
Country & year established	Involves use of national health registry data from six countries including Australia, Denmark, Finland, Norway, Sweden and Israel
Ownership & Funding	All member sites have a defined role in the collaboration. The project lead is Denmark who must provide general coordination and oversight of consortium and responsible for general iCARE communication The analytical oversight committee (AOC) provides iCARE administrative and scientific oversight on an ongoing basis and is comprised of representatives. with voting authority from all member sites. Funded by Autism Speaks ($1.2 million) 2009-2013
Registration	36736 individuals with an ASD
Data collected	Autism geographical and temporal heterogeneity, phenotype, family and life course patterns, etiology Birth weight, birth order, age of diagnosis
Strengths	(1)cost efficiency through use of existing resources; (2) flexible infrastructure accommodating current research needs and future network growth and data upgrades; (3) flexibility in study designs to suit particular analyses (e.g., cohort, case-cohort, multigenerational or sibling designs); (4) large sample sizes based on federated data that would enhance statistical precision, especially in individual strata based on ASD phenotype characteristics or risk factors; (5) ability to characterize population trends in reported diagnoses over time (e.g., by age at reporting, birth cohort or time period), aswell as changes over the life course of affected individuals; and (6) enhanced comparison and interpretation of between-site results based on data harmonization and application of uniform analytic methods to multi-site data. (Schendel et al., 2013)
Challenges	Logistical and technical challenges Harmonising the different datasets Challenges in multinational collaboration concerning data access security , confidentiality and management Strict regulations that prohibit information being transported outside the countries of origin
Data access	Researchers. Researcher's access to the registry data based on an approved application does not require individual consent. Under these circumstances, to preserve confidentiality, no personal identifying information is permitted in any iCARE harmonized analytic datasets.
Technical aspects	Analyses performed using database federation via a computational infrastructure with a secure, web-based, interface. Pooled dataset then disappears without being saved to the researchers computer or altered at any original site allaying ethical and data privacy concerts Rigourous harmonisation and quality control processes Icare created a computational infrastructure with a secure web-based, interface to facilitate analysis of the federated harmonised icare research datasets
Communications	Annual report

Registry	National Cancer Registry Ireland
Function/Aim	To identify, collect, classify, record, store and analyse information relating to the incidence and prevalence of cancer and related tumours in Ireland; To collect, classify, record and store information in relation to each newly diagnosed individual cancer patient and in relation to each tumour which occurs; To promote and facilitate the use of the data thus collected in approved research and in the planning and management of services.
Country & year established	Ireland Established 1991
Ownership & Funding	The Registry was set up and is fully funded by the Department of Health, but is independent of it. The Registry is governed by a Director and Board. Four functional areas which consist of corporate services, research and analysis, data management and tumour registration officers and information technology report to the Director.
Registration	Over 200,000 cases
Data collected	Cancer incidence, treatment and survival
Strengths	Promoting the use of the data collection in research and in the planning and management of services reduce the cancer burden. Formed a cross border co-operation to produce an All-Ireland cancer statistics report.
Challenges	Data can be two years behind and there are several contributing factors to this including resource constraints. As cancer cases are identified through a number of sources such as pathology reports, scans and x-rays, and from a number of health care providers, it is essential that all the reports be put into a single record containing the best and most accurate information available which is time consuming. Strict quality control and best practice is carried out and data cannot be published until case reporting is estimated to be at least 95% complete with the last 10% is the most difficult to complete (www.ncri.ie).
Data access	Only Consultants can access data which would identify an individual. Data is anonymised for reports and datasets that are issued openly.
Technical aspects	The NCR provides de-identified data from its database for researchers via unique biobank codes.
Communications	Annual Report, Website, Research publications

Registry	The Danish National Biobank
Function/Aim	Create a unique platform for tomorrow's research making it possible to study its population from cradle to grave (biobankdenmark 2012)
Country & year established	Denmark Established 2012
Ownership & Funding	The Novo Nordisk Foundation and the Ministry of Science Technology and Innovation. Statens Serum Institut is the host institution. In 2009–2010, the Foundation awarded the Biobank a 10-year grant of €15.9 million. Other contributors to establishing the centre include Denmark's Ministry of Science, Innovation and Higher Education (€4.8 million) and the Lundbeck Foundation (€3.4 million).
Registration	15 million biological specimens
Data collected	detailed information on samples from blood, tissue and DNA amongst other
Strengths	Allows scientists access to over 15 million biological samples creating new research opportunities; Allows linkage of disease development to a number of parameters such as familial, medicinal and educational; High level of necessary data protection, while also allowing researchers to use private data. Combined with top quality health register data.
Challenges	The Danish model faced less challenges than the other models as it had full support from the Danish Government and substantial funding to cover human resources and state-of-the-art biobanking.
Data access	Researchers (before the biological material from an individual can be linked with information from national registries the research projects are assessed

	by research ethics committees and he Danish Data Protection Agency
Technical aspects	The physical Biobank is 2400-m² and includes a gigantic freezer room in which robots store, retrieve and deliver the biological samples
Communications	Website

Registry	**UK Multiple Sclerosis Register**
Function/Aim	To build a working prototype MS Register, for use in 5 centres across the UK, capable of being scaled to a national deployment.
Country & year established	UK Established 2011
Ownership & Funding	Commissioned by, and is supported by a grant from, the MS Society of Great Britain and Northern Ireland.
Registration	7786 adults with MS Registration is voluntary
Data collected	Demographic data Baseline information about MS including date of diagnosis of MS, type of MS and age at onset, plus information on education, employment and living arrangements for people with MS
Strengths	Captures data from three main sources and is able to anonymously link these data at the individual level whilst retaining privacy. (Jones et al., 2012)
Challenges	The information used in this study was self-reported and the respondents are not necessarily a fully representative sample of people with MS in the UK.
Data access	Potentially an open resource for the use of all professionals and government bodies, whose access could improve and enhance MS treatment and provision.
Technical aspects	Register is populated via a web-based portal, NHS neurology and clinical systems; and administrative data sources. “The data are de-identified and linked at the individual level (Ford et al., 2012) The Register is based on the proven technologies and robust Information Governance arrangements in place in the Secure Anonymised Information Linkage (SAIL) system developed by the Health Information Research Unit (HIRU)” (Jones et al., 2012)
Communications	People who register are sent an email every 3 months inviting them to return to the portal and update their questionnaires to build up a longitudinal data source.

Registry	**Autism Spectrum Database - UK**
Function/Aim	To facilitate research and help families
Country & year established	UK Established 2011
Ownership & Funding	The database is run by researchers at Newcastle University. Governance is by a steering committee composed of clinicians, researchers and parents who oversee data security and ethics. Funded by Autistica, a UK charity.
Registration	1700 families contacted
Data collected	Child’s name and date of birth, parents/carers names and dates of birth, address and contact details, the type of ASD diagnosis and other medical conditions, information about communication, developmental skills and behaviour. Schooling information and data about family members, such as siblings names and dates of birth and other family members with a diagnosis of ASD.
Strengths	Engagement with parents. Make effective use of nationwide clinical teams.
Challenges	Refusal to consent or families who don’t return completed forms with data. Ethical challenges. Validity of diagnosis for older individuals.
Data access	Data is password protected and only available to researchers.
Technical aspects	Web-based portal. Also linked with another UK register, Daslne.

Communications	Newsletter for families. Newsletter for professionals. Site visits and conference presentations. Website.

Registry	**Phelan-McDermid Syndrome International Registry**
Function/Aim	Provide family support, accelerate research and raise awareness.
Country & year established	United States 2011
Ownership & Funding	Phelan-McDermid Syndrome Foundation (PMSF) is governed by a Board of Directors. The registry is funded by PMSF
Registration	640+ from 43 countries
Data collected	demographic, genetic, clinical and developmental data on patients diagnosed with PMS
Strengths	The Registry is structured so that it can be filled directly by the patient's relatives or tutors. Researchers have access to data and biosamples but data is de-identified. Caregivers can get data about symptoms of the syndrome. Returns aggregate data to families.
Challenges	Translation challenges for the international community
Data access	Families, Researchers, Clinicians
Technical aspects	Hosted by Patient Crossroads. Multiple data feeds will be established to extract and link data from well-characterized patient and population cohorts from electronic health records into the backbone informatics architecture provided by the open source, i2b2 based, tranSMART platform with rich phenotype extraction via Apache cTAKES. Such datasets must meet the needs of researchers (including support for diverse scientific collaborations with networks that are funded by PCORI or others, while simultaneously preserving security and maintaining appropriate privacy and ethical safeguards.
Communications	Website, quarterly newsletter, Facebook, Twitter, regional gatherings, and a biennial international conference.

Table 6: Analysis of other registries and biobanks

Summary & Conclusion

Key research findings shows evidence that Ireland needs and will benefit from a registry and biobank for ASD/NDD to accelerate clinical, biomedical and environmental research, to inform service requirements and drive policy making. Delivery of best practice healthcare requires effective use of valid information therefore it is now timely to act and invest resources to build this registry and biobank with the potential to alleviate the rising financial and resource challenge on Irish public health system and align with upcoming government health strategies.

This report focused on a stakeholder consultation seeking input from all those concerned with ASD/NDD. The purpose of this process was to gain diverse insights and to ensure adequate involvement and input with all stakeholders, such an analysis is theoretically and empirically founded upon sound organization and project management theory. The consultation process was not just to ask questions of a balanced representation of stakeholders but to ensure they were informed and understood the objectives and benefits of creating a registry and biobank. This was particularly evident at the consultation meetings where members of the audience came with concerns about ethical issues and data protection however their worries were allayed after being informed by the presentation and open floor discussion.

The impact that an ASD registry and biobank would have in Ireland would be far reaching and of immense long-term benefit. All stakeholders and the public were asked what they think should be the scope and aims of this initiative of establishing an ASD/NDD registry and biobank. As informed by the consultation process, research was identified as a priority and agreement was unanimous that the following elements should be included as part of the scope: biomedical research to support identification of genetic or other biological causes of ASD/NDD; environmental research to identify environmental factors; and clinical research to identify clinical aspects associated with ASD/NDD. There was strong agreement that the registry should inform health, education and social care services throughout life transition stages as the consultation process revealed serious lack of support and interventions for individuals as they transition from childhood to adulthood. It should also function as an important educational ICT system and provide information about ASD and genetic risks that can affect individuals and families. It should give accurate data as to the extent of ASD in Ireland for future planning and decision-making. The biobank would support research to develop better treatments for ASD. It was highlighted that it is also important to develop research into behavioural therapies as well as medical treatments.

Collaboration with government stakeholders was important at the early planning stage for ICT strategy so no aspects of the registry would be unnecessarily replicated. It is important that this registry and biobank complements the strategic initiatives of government bodies particularly the DoH and the HRB. Governments need data and evidence upon which to act and operate.

This is Ireland's opportunity to propel world-class research and its application for the public good with an innovative unique registry and biobank for a major public health challenge.