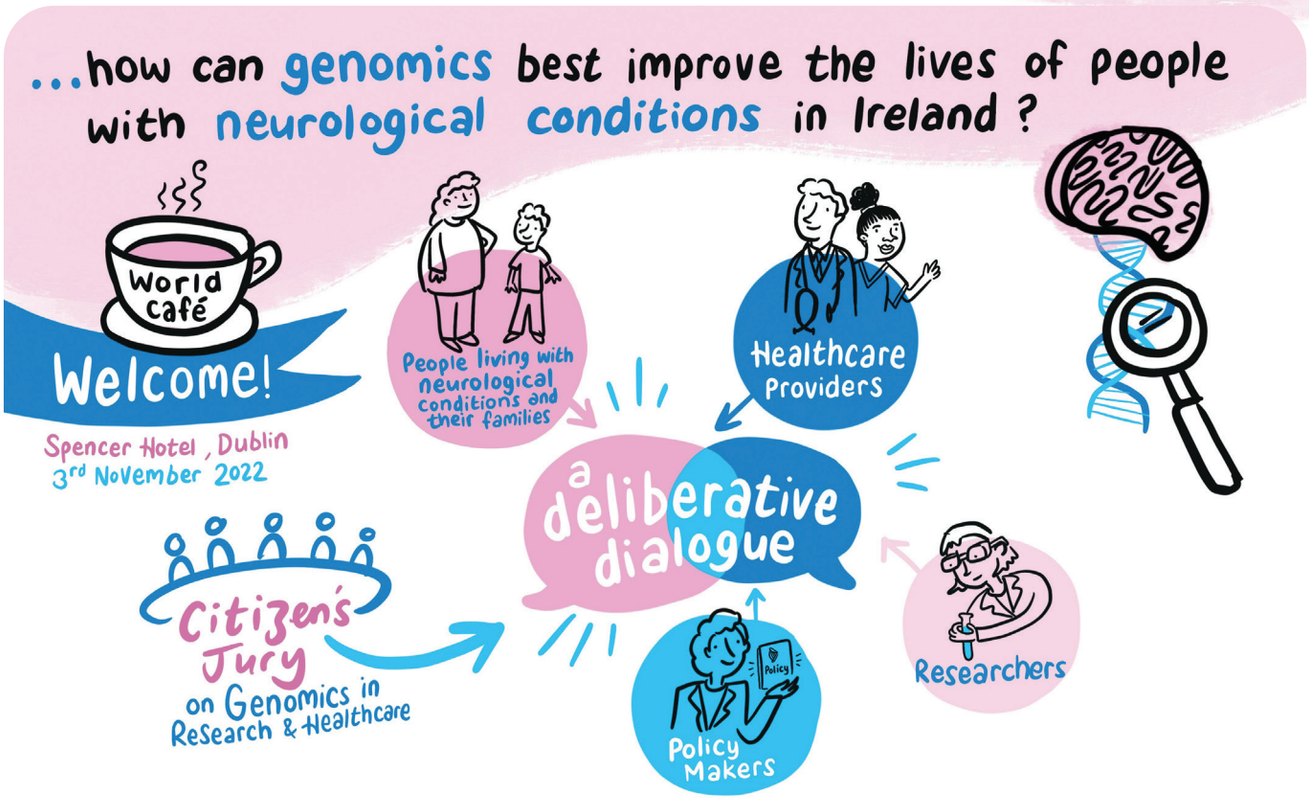
Deliberative Dialogue on Genomics in Neurological Healthcare & Research

Dublin, Ireland, 2023

Representing the contribution of patients, advocates of people affected by neurological disease, clinicians and researchers

Contents



Contributors to the Deliberative Dialogue meeting

Introduction

This document provides a summary of the Deliberative Dialogue on Genomics in Neurological Healthcare and Research which took place on November 3rd, 2022. The event was designed such that recommendations emerging from this dialogue could inform the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland. This document summarises the contribution of 35 stakeholders – representing patients, advocates of people affected by neurological disease, clinicians, and researchers – who met to address the question:

"How best to design a national genomics research and healthcare programme for Ireland from the perspective of neurological patients, families, and clinical stakeholders?"

Background and Context

RCSI University of Medical and Health Sciences is a Lead Site of the National PPI Ignite Network, established in 2021 to advance public patient involvement (PPI) in health and social care research in Ireland (www.ppinetwork.ie).

In 2022, as one of its commitments to this network, RCSI PPI led a 'deliberative dialogue' on genomics in collaboration with FutureNeuro (the Science Foundation Ireland Research Centre for Chronic and Rare Neurological Disease) and IPPOSI (The Irish Platform for Patient Organisations, Science and Industry). RCSI and IPPOSI had previously partnered on a national Citizen's Jury on Genomics in Research and Healthcare https://www.ipposi.ie/our-work/policy/health-information/2022-citizens-jury-on-genomics/

Given FutureNeuro's expertise on neurological disease https://www.futureneurocentre.ie/, the deliberative dialogue was designed to dynamically explore the key recommendations emerging from the Citizen's Jury from the multiple perspectives of 1) patients living with neurological conditions and their families 2) healthcare providers 3) researchers with expertise in neurogenomics.

Methodology:

The deliberative dialogue event used a 'World Café' methodology (www.theworldcafe.com) to deeply explore the dialogue questions in small group rounds and through iterative reflection involving the full room of participants. This ensured that a multiplicity of stakeholder perspectives informed the evolving dialogue and shaped the collaborative recommendations for this report. The overall event question was: **How best to design a national genomics research and healthcare programme for Ireland from the perspective of neurological patients, families, and clinical stakeholders?** The 5 Dialogue questions formulated were:

Dialogue 1 When has genomics helped you or helped your family/patients/research?

Dialogue 2 How can we support people to safely contribute their genomic data to research and healthcare, if they choose to?

Dialogue 3 Imagine you are invited to be involved in the development and implementation of a new national genomics programme. As someone interested in neurological conditions, what matters most to you to focus on? What is most important to you?

Dialogue 4 What next steps could we take as a neurological community to collaborate on the design and implementation of a genomics programme?

Dialogue 5 How might you like to contribute to the development and implementation of a new national genomics programme? What support would you need to contribute well in that case?

A detailed description of the methodology can be found in Appendix I. Note that due to the iterative nature of the methodology employed, the reader may note that some points were repeated by participants across dialogues. This serves to underscore the pervasive and important nature of these issues.

Contributors

The contributors comprised of representatives from research, clinical and patient advocacy organisations in the area of neurology (or brain health). A full list of contributors is available in Appendix II and participants in Appendix III.

Recommendations

- **Stakeholder Engagement and Consultation**
  - Build trust through a consultative, collaborative process with a diverse and multidisciplinary neurological stakeholder group, assembled to inform the implementation, evaluation and governance of the National Genomics programme.
- **Development of Supporting Infrastructure**
  - Create a secure central genomics database that enables impactful research and diagnosis for people with neurogenetic conditions.
  - Implement the individual health identifier (Health Identifiers Act 2014) in daily clinical practice
  - Improve coordination of research ethics processes at national level and consider legislation to facilitate and support genomics.
- **Access to Services**
  - Ensure equity of access to an appropriately resourced national genomics service.
- **Education and Awareness Raising:**
  - Motivate individual participation, and public and political support through targeted approaches on the value of genomics research to healthcare.

Dialogue 1

When has genomics helped you or your family/patients/research

Emerging themes

Theme 1.1 For diseases of the genome, genetic testing is essential to early diagnosis, clinical treatment, informed decision making, and patient hope.

Theme 1.2 Access to genetic testing in the clinic is a healthcare equity issue.

Theme 1.3 The interplay between genomics research and clinical genetic testing is key to improving diagnostics and developing new therapies.

Theme 1.4 Neurological patients want more involvement in genomics research both as participants and as PPI partners to improve the research process.

Theme 1.5 Patient participants hope for respectful treatment, reduced stigma, improved communication and further education for the public and for healthcare professionals.

Dialogue/perspectives contributing to themes: Stories from patients shared in the whole room discussion included the following:

1.1 Genetic testing & early diagnosis

A patient participant shared how genomics helped them by providing a molecular diagnosis for their cancer, which in turn guided therapy. They were able to avoid standard chemotherapy and instead be prescribed a specific therapy tailored to the genetic profile of the cancer. Another participant shared how a relative was able to continue working during treatment for breast cancer, as genetic profiling of their tumour allowed her to receive a targeted therapy. This individual avoided the very visible signs of cancer treatment – hair loss, sickness etc. – which allowed her a better quality of life during her treatment than she might otherwise have had. Another experience was shared by the parent of a child with a neurological condition, whereby genomic testing provided their child with a genetic diagnosis for a rare form of epilepsy. Although there was no precision treatment available, the parents were able to join a global support group (of ~250 families). This was an experience that was echoed by other participants, who agreed that access to a patient/carer community was hugely beneficial.

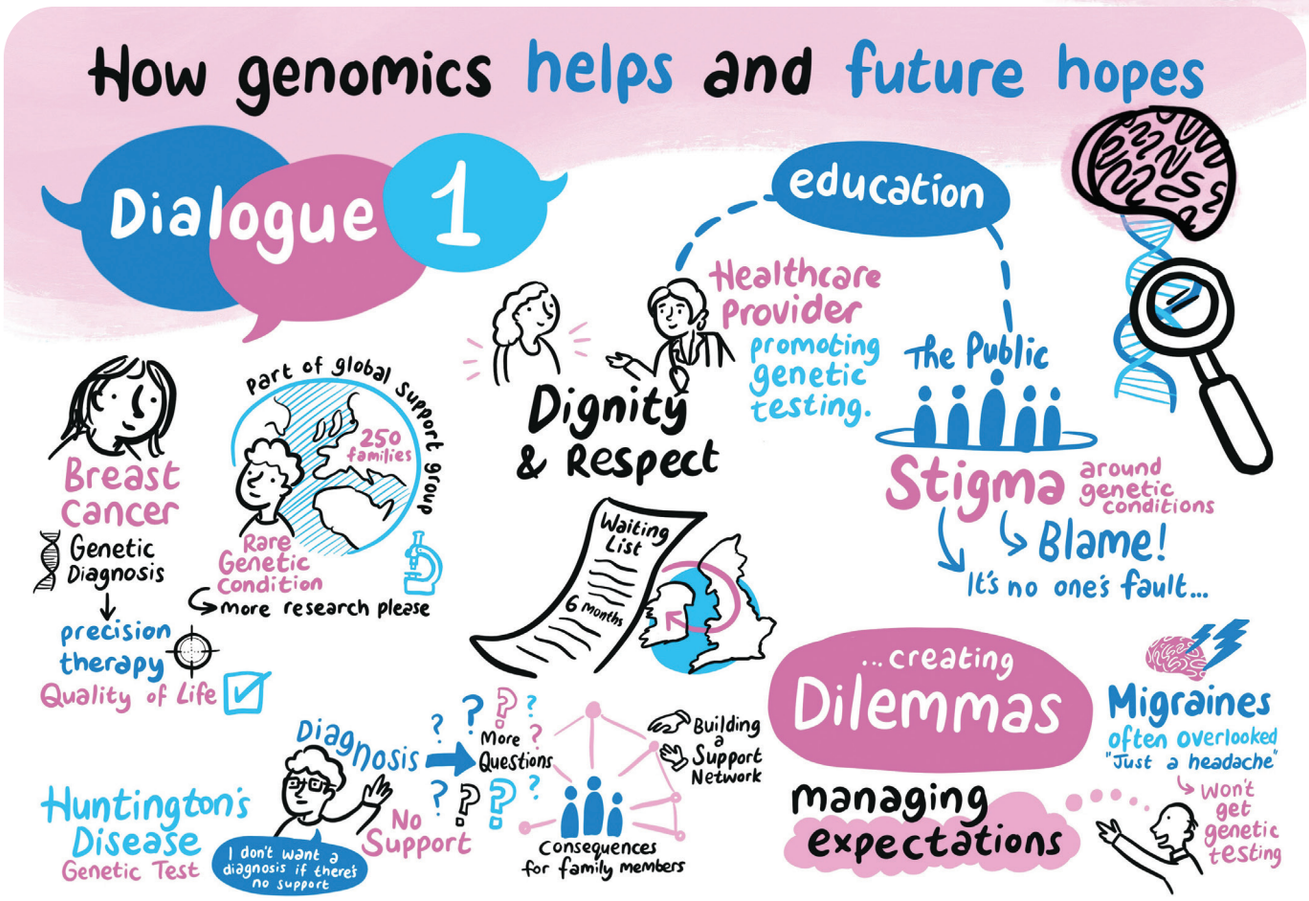
A healthcare professional shared how early genomic testing helped a family with Spinal Muscular Atrophy (SMA). The genetic diagnosis informed how the clinical teams worked with the family. For example, they could provide prenatal testing for a later pregnancy and were able to prepare a treatment plan before the child was born. This prenatal diagnosis led to a much better outcome for the second child as an early intervention was possible.

1.2 Healthcare equity issue

Conversely, in another case, a lack of service capacity resulted in a long delay in genetic diagnosis, that in turn resulted in a lack of timely access to services and worse health outcomes for the child. There was agreement among healthcare professionals that there is huge variation in genomics services across the country. For example; in one Irish hospital there is a highly administrative system in place that unintentionally but unfortunately places bureaucratic barriers to the ordering of genetic tests by hospital consultants. This greatly reduces the number of genetic tests that are ordered.

It was agreed that while, in Ireland, there are pockets of excellence in research and clinical use of genomics for the diagnosis and treatment of neurological disease, it was noted the ongoing challenge of lack of resources such as Clinical Nurse Specialists and Genetic Counsellors really undermines the efficacy of the service.

Infographic Summary of Dialogue 1



1.3: The interplay between genomics research and clinical genetic testing

Participants discussed how genomics research is central to understanding the causes of neurodevelopmental disorders, and how our knowledge of genetic causes of neurodevelopmental (and indeed all) disease comes from the research domain.

1.4: Patients want more involvement as research participants and as PPI partners

Participants articulated the need for people with lived experience of health conditions to be involved as partners in genomics research but specified the need for Public Patient Involvement in research to be supported: *"It's wonderful that PPI is recognised as critical for scientific breakthroughs & research but support is important & training"*. The importance of ensuring that patients are offered the opportunity to contribute genomic data in life and post-mortem was shared by a participant living with a rare neurological condition: *"I would be devastated if I die from disease and not have opportunity to contribute to research"*.

1.5: Patient hopes for respectful treatment, reduced stigma, improved communication

Patient participants expressed their hope that stigma would be reduced through public education about genomics and how genetic conditions are not the 'fault' of individuals. Patients also spoke about the need for further professional development for healthcare professionals around communicating in appropriate ways when inviting for tests, sharing diagnostic findings, and making referrals. The need for support in the form of genetic counselling was also highlighted.

Dialogue 2

How can we support people to safely contribute their genomic data to research and healthcare, if they choose to?

Emerging themes

Theme 2.1 Ensure timely supported consultation with neurological patients and healthcare stakeholders to build trust in genomics research and healthcare services.

Theme 2.2 Seek informed consent through a clear, understandable and respectful process that is opt in and that outlines how data will be generated and used.

Theme 2.3 Provide strong and trustworthy data security around a central/national genomics database, that enables collaboration and impactful discoveries from the research community.

Theme 2.4 Invest in appropriate resources to facilitate genomics in research and healthcare.

Theme 2.5 Conduct a national campaign on the value of genomics research to healthcare to enable informed and considered decision making around participation.

Dialogue/perspectives contributing to themes:

2.1 Building Trust

There was consensus in the room regarding the vital importance of building public, patient and stakeholder trust. There is a need for meaningful consultation with stakeholders to discuss the benefits and risks of sharing data for research and potential risk management strategies. Patients want to see how their data is being used and to have trust in the system that their data is being used for public good, with patients being the ultimate beneficiaries.

Where public-private partnership is being considered, the government should negotiate clear benefits for the citizen/state from this collaboration e.g. reduced costs of medication/interventions. A participant shared the current situation whereby "*Cystic Fibrosis patients in Ireland gave their data for pharmaceutical trials but now they can't afford the medicine that was developed*".

2.2 Consent

A patient representative described the time of consent as very stressful, was not sure what she even signed. It is important that consent forms are understandable and all patients receive a copy. Ideally, the researcher/clinician would revisit the consent with participants to check their understanding. A researcher noted the current process, which involves the Health Research Consent Declaration Committee (HRCDC), is not suitable or sustainable (as it in effect acts as a barrier, even if intended to enable) and changes in legislation may be required - this issue is particularly relevant for neurological research.
A clinician noted that consideration needs to be given to enabling people with Intellectual Disability to participate. Handling the transition from paediatric to adult services was also raised, as this can be a particularly challenging period for patients (and parents), to understand which particular adult service they will attend and the resulting impact. Transition of care in Ireland is poorly developed although it is being addressed by the European Reference Networks, who are producing clinical guidelines to help with transition and fostering research.

2.3 Genomic Database

There was consensus in the room regarding the need for a centrally controlled, sustainable national genomics database, which would facilitate both diagnostics and research. The central database should ensure that genomics data is studied as widely as possible, including by international research consortia, but with a transparent access route and clear controls/governance in place.

One participant noted that while there were concerns about data security in Electronic Health System infrastructure, the current practice of paper records also had security issues. A patient commented that they wanted their data to be shared widely but articulated


How do you imagine genomics might
support you (or your family/patients/
research) in the future?

the need to identify and eliminate risks so people feel safe to contribute data. A patient representative outlined their positive experience of having a diagnosis facilitated through participation in an international database. A clinician described the importance of the infinity loop model whereby data goes back to the clinic for diagnostics and trial stratification purposes - this currently exists but needs to be better resourced. A researcher also requested that consideration is given to the treatment of incidental findings.

Participants were concerned about insurance companies being able to access their families' genomic data and other identifiable data and deny healthcare insurance if there is vulnerability to certain conditions.

2.4 Investment in Genomic Services

From the researcher perspective, it was noted that considerable investment is required in genomics services in Ireland to ensure both genetic testing and counselling are available and accessible. It is vital to adequately staff and resource genomic services. It is also important to establish dedicated research time for healthcare professionals to collaborate with clinical geneticists and neurogenomics researchers. It was acknowledged that hospitals were already overburdened and that investment is also required to facilitate the consenting process including space in clinics.

2.5 Education

There is a need for a national educational campaign to increase knowledge about genomics and the benefits (and risks) it brings. A clinician commented that genomics currently has a negative profile in the media which needs to be addressed through education. One participant felt that the Irish population needs to be informed of the value of genomics research using real patient stories. The role of the GP as a source of trustworthy information was acknowledged. Another participant highlighted the need for education for the next generation, e.g., inclusion of genomics education in the school curriculum to ensure that we have an educated public ready to debate the issues and make informed decisions. A clinician felt that education is also required for healthcare professionals and to further promote the value of genomics research to healthcare.

Infographic Summary of Dialogue 2


Kevin
Mitchell

Dialogue 3

Imagine you are invited to be involved in the development and implementation of a new national genomics programme. As someone interested in neurological conditions, what matters most to you to focus on? What is most important to you?

Emerging themes

Theme 3.1 Address the implementation of the individual health identifier (Health Identifiers Act 2014) in daily practice, as it will enable linking of data across sites.

Theme 3.2 Consider the need for an integrated, resourced, sustainable and secure database to facilitate diagnosis and inform related research.

Theme 3.3 Consent by patients with neurological conditions in clinic settings needs to be supported by patient advocates and healthcare professionals, with appropriate training.

Theme 3.4 Health Care Professional capacity/training is required to provide support pre and post diagnosis.

Theme 3.5 There must be equity of access to the national genomics service, for all healthcare conditions, as well as geographic locations.

Dialogue/perspectives contributing to themes:

3.1 Individual health identifier (IHI)

A patient advocate noted how a unique patient ID is key to joining up datasets in our health system, yet today, the health system struggles to identify where patients are. The implementation (in daily clinical service) of the IHI can help enable national studies such as that emerging from the genetics and genomics strategy.

3.2 Integrated Database

One researcher pointed out how having access to genomics data drives progress and how the proposed national genomics database (from IPPOSI Citizens Jury) is not an end point but a starting point; a living clinical tool that informs research on an ongoing basis. It was deemed essential that such a database would not be a 'dead repository', but rather actively used and studied to enable maximum benefit for all. A patient representative noted how their child benefited from being part of an international research study (and associated database), through which they finally received a diagnosis, years after an initial diagnostic test.

3.3 Consent and ways to support the consent process

A parent of a child with a rare disease commented that the diagnostic journey can be a very stressful time and that it would help to be able to re-access consent provided. A researcher commented on the resources required to enable consenting (in the clinic and beyond), including space in clinics and the development of robust processes and that reference to resources for the consent process are lacking in the national strategy. It was also noted how for neurological diseases, it is extremely important to have efficient systems to enable inclusion in research of those with intellectual disabilities. Advocates could help ensure a tailored consent process.

3.4 Healthcare Practitioner

A participant called for focus on training of healthcare Practitioners on person-centred care/empathy. Specifically, around the impact (on the individual) of a genetic diagnosis, especially for rare diseases. It was also noted that patient involvement should be a key component of this training, so that healthcare Practitioners can learn from peoples' experiences to inform their clinical decision-making.

3.5 Equity

A healthcare professional noted how equity in access to genetic testing and services for all conditions was needed: "*We've heard so much are access based on location and local hospital, but access based on conditions should also be addressed by national strategy*". A patient representative noted how understandably, acute conditions that may lead to death are seen as more important. However, debilitating chronic conditions such as migraines can be brushed aside - with a perception that "*they're just headaches*". How can there be equity when perception of disease differs?

Infographic Summary of Dialogue 3

Dialogue 4

What next steps could we take as a neurological community to collaborate on the design and implementation of a genomics programme?

Emerging themes

Theme 4.1 We need to identify leadership and seek representation and consultation to directly inform implementation.

Theme 4.2 We need to inform/shape the implementation of the national genomics service, including resource allocation and data security considerations.

Theme 4.3 We need to improve the research ethics process at the national level and consider legislation to facilitate and support genomics.

Theme 4.4 We need to develop a Model of Care for neurogenetics.

Theme 4.5 We need to increase public awareness of the benefit of genomics among people living with neurological conditions (as well as other conditions).

Dialogue/perspectives contributing to themes:

4.1 Leadership, representation & consultation

The neurological stakeholder community needs to grasp this time limited opportunity and engage with the national genomics office, to help shape implementation. Stakeholders recommended a collective voice from patient representative bodies from the neurological sector, a collaborative approach to implementation/service design, and "equitable representation across stakeholders" in the implementation planning group. The neurological community needs to address practical issues like influencing local, national and international politicians.

Further consultation with neurological stakeholders via a community-wide forum would usefully and transparently inform the design of the implementation plan for the national strategy. The forum should include broad representation of the neurology community and would be tasked with identifying the priorities of the community, to inform implementation. There should be ongoing communication with the broad neurological community throughout the process of implementation design, initial phases, evaluation etc.

4.2 Inform the design of the national genomics service

Specialist clinicians, researchers, and patient advocates should inform the design of services based on their expertise and lived experience. The resourcing, training, recruitment, staffing, and retention of clinical and Health Care Practitioners with genomics expertise to form Multi-Disciplinary Teams (MDTs) capable of dealing with populations with particular needs requires significant planning.

There needs to be access to genetics testing, and crucially resourcing of the services that are required once that genetic diagnosis occurs. Health Care Practitioners will require additional clinical time for referrals along with physical space for confidential communications.

It is critically important that the national genomics service/biobank would apply high-standard security and IT systems. Considerations include piloting and testing potential failure scenarios to mitigate risks; addressing safe access for patients to their own data as well as access for healthcare professionals and researchers; and safely and clearly adding data already collected from local and overseas service labs.

4.3 Ethics process and legislation

Participants recommended that the current health research ethics process and possibly legislation needs to be reviewed and improved to facilitate genomics research as

Infographic Summary of Dialogue 4

there are currently significant obstacles (for example the time it often takes to get ethics approval for a study, and to initiate the study, especially when multiple study sites are involved). Equity needs to be a core ethical principle both in terms of access to genetic testing, genomics services and participation in research. The National Research Ethics Committee needs to enable genomics research and high-quality biobank-related collections.

There is a fear of potential genetic discrimination if a person's genomic data is inappropriately shared or linked to other elements of their life (e.g. their employer, health insurer etc), which, it was noted, may discourage participation in a national genomics programme. Is legislation sufficiently robust to prevent this? One participant noted that legislation was drafted around 17 years ago to protect against this type of discrimination, but it hasn't been implemented. Stakeholders recommended there is a focus on "*protection from the insidious use of genetic/familial information*".

4.4 Model of Care for neurogenetics

Health Care Practitioners called for a national model of care for neurogenetic disease, for it to be provided to the National Office for Genetics and Genomics, with a proposal to work together on implementation. This could be in the shape of a neurology genomics clinic for suspected monogenic forms of disease, with multidisciplinary team input.

4.5 Public awareness

The neurological community needs to be more involved in the public engagement aspect of the implementation of the National Genomics strategy. The neurological community should focus on political awareness and media engagement on economics including the cost benefit to the State of such a genomics service. The neurological community could also actively increase awareness of the role of genomics among neurology patients who are often unaware of the genomic connection to their healthcare condition and relevant research opportunities.

Dialogue 5

How might you like to contribute to the development and implementation of a new national genomics programme? What support would you need to contribute well in that case?

Emerging themes

Theme 5.1 Organise a consultative, collaborative process with a diverse and multidisciplinary neurological stakeholder group to inform the implementation, evaluation and governance of the National Genomics programme.

Theme 5.2 Conduct a targeted advocacy campaign for TDs and MEPs, to raise political awareness of the value of genomics to society and to align (where appropriate) healthcare and research funding.

Theme 5.3 Resources are required for protected HCP time and bioinformatic expertise to realise the potential of the National Genomics Strategy for people impacted by neurological disease.

Dialogue/perspectives contributing to themes:

5.1 Consultative process

In this final dialogue, key themes of neurological stakeholder representation, consultation, communication, and collaboration emerged. Stakeholders expressed their desire for collaboration stating that "*there should be genuine collaboration with the neurological community, not just consultation.*" Equally issues of funding, time, public and political awareness, education, and ethical approval need to be addressed.

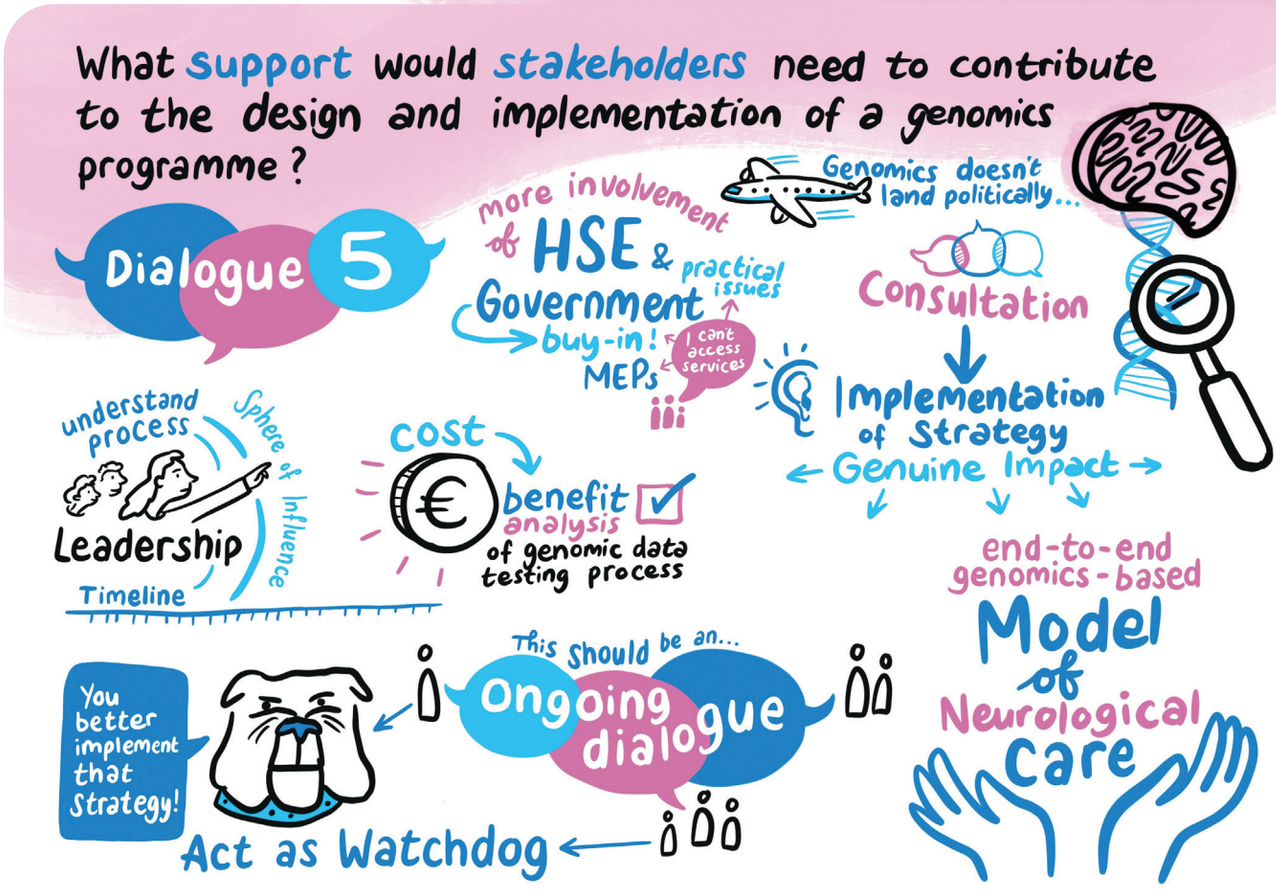
One participant commented: "*Today has been important in bringing the neuro community together, but we think there's an important ongoing role. When the implementation plan comes out, the community should come together again. We should act almost as a watchdog. It's important that we can continue to feed in* ". While another participant commented: "*As a member of the FutureNeuro PPI Panel, I am of course the exception to other patients. Other patients know nothing about this new (genomics) programme, and I would like to see them given information about it, so that they too can provide support and give feedback and contribute as it is designed and implemented. I feel lucky today but I cannot be the voice for everyone with epilepsy.*"

5.2 Targeted advocacy campaign

While it was mentioned in the programme for government under rare disease, as one stakeholder noted "*Genomics usually doesn't land politically so we need to raise political awareness*". It was noted how genomics at a political level in Ireland can be challenging, as the level of awareness of genomics among elected representatives is low. Ideally this campaign would communicate the cost-benefit of genomics in the healthcare system.

5.3 Resources

To engage effectively in implementation, stakeholders require space and time. For example, Healthcare Practitioners need clinical genetics, bioinformatics and genetic counselling resources to support neurogenetics multidisciplinary team meetings. Healthcare Practitioners require more protected time for research. Stakeholders working in both research and healthcare settings noted that a more efficient system for research ethical approval and related governance agreements would enable more efficient and impactful genomics research. Significant investment is required in IT infrastructure for clinical bioinformatics.



Infographic Summary of Dialogue 5

Appendix I

Methodology

The dialogic learning methodology of 'World Café' was chosen to underpin this Deliberative Dialogue as it seeks to create "living knowledge through conversations that matter" (Brown, 2001). There are four key design principles informing the design of world café events including (1) Exploring questions that matter (2) Creating hospitable space (3) Connecting diverse people and ideas (4) Listening together for patterns, insights and deeper questions. These principles were actualised in event planning, data collection and data analysis as follows:

To explore questions that mattered to the neurological community, FutureNeuro established a multi-stakeholder committee comprising specialist genomics and neurology clinicians, academic researchers, patient advocates and people with lived experience of living with or caring for people with neurological conditions.

This group identified the over-arching question for the dialogue, which emerged as: '*How can genomics improve the health of people living with neurological conditions in Ireland?*' Five dialogic questions were subsequently developed with the support of the international world café consultant to encourage participants to deeply explore this overall theme in conversational rounds. Each question used a different lens building on the previous question, including personal experience, community benefit, implementation steps and ultimately policy recommendations. These questions were then sense checked with individual representatives of the four different constituent groups – clinician, researcher, patient advocate and policy makers. This robust design process ensured the final questions were of definite interest to all participating stakeholders, would generate lively, inclusive dialogue and findings would have the potential to inform genomics policy.

FutureNeuro used a *'snowballing networking'* technique to identify who plays a key role in the areas of neurology and genomics in Ireland, with the initial support of the Advisory Panel. Over 50 potential participants were contacted, and 35 participants were ultimately confirmed for participation in the deliberative dialogue. Efforts were made to have double the amount of patient advocates to clinical consultants and specialist researchers so that the inherent power imbalance between patients and hospital consultants/academics would be somewhat redressed.

A hospitable space for all participants was created by hosting the event in an accessible city center hotel location with parking, catering and accessibility supports available. To support those travelling, the event began later in the morning at 11am. All patient representatives travelling more than 2 hours were offered overnight hotel accommodation. To reduce the power imbalance that honorific academic or medical titles can generate, badges were prepared using only first names. To ensure participants' comfort, refreshments were served on arrival, at lunch and post-event, with diverse dietary needs catered for. This also meant people had plenty of time to informally network. Flowers decorated tables and colourful stationary encouraged doodling and notetaking.

Active participation in the event was encouraged by the Lead and Table Facilitators in an encouraging way, articulating that every voice matters, encouraging deep listening, validating all individual contributions and ensuring to invite contributions for those who had not yet spoken. This conscious creation of a hospitable, safe space was intended to help participants feel welcome, valued and supported to contribute.

Diverse perspectives were connected through event design and inclusive facilitation. Participant lists were designed pre-event to ensure that every table of four participants had two patient advocates, one healthcare provider and one researcher. Participants moved to a different table for each of the 5 discussions so all participants met at least 25 out of the 35 participants in discussions. This ensured multiple opportunities to hear diverse perspectives.

The Lead Facilitator ensured to use connecting phrases during the large room group discussions such as *'so that links back to what X said in our previous conversation when she said Y'* or *'I wonder if anyone would like to build on what has been shared by B'* etc. The graphic harvester also focused on the connecting threads between the dialogue rounds. She presented her graphic storytelling of the emerging dialogue work just after lunch and at the end of the day. This enabled participants to become more aware of all of the connections from multiple dialogues feeding into the emerging bigger picture.

To facilitate collective listening, participants were encouraged to deeply listen to each participant at their table, and to note their own key points and responses to the conversations on post-it's in each round. These post-it's were then collected after each of the five rounds, typed up and then hung up on the wall under the question title. Participants were invited to go for a gallery walk before/after lunch, at coffee break and before the end of the event to view these notes.

The Lead Facilitator used a *'mini-harvest'* technique of around 20 minutes after each of the 5 dialogue rounds to open up full room discussion. Participants were asked open-ended questions like *'so what is sitting with you now'*, *'what has surprised you in this dialogue'*, *'what learning or understanding new perspective are you taking with you'*. For the final large room discussion, participants were asked *'where to from here as a community of people interested in neurological research and healthcare?'* and *'what are your top recommendations for successful implementation of the future genomics policy'*. This approach ensured that all participants had the opportunity to contribute and engage in collective listening and learning from each conversation and from each other.

Appendix I

Data Analysis

Data was recorded throughout the event with the support of four researchers who transcribed roundtable notes and large room discussions in real time and stored them in a secure folder in the cloud. Participant feedback sheets were also collected and entered as data as were the graphic illustrator's images developed during the event.

The dialogic data was in the form of hundreds of post-it notes from small table discussion grouped under the 5 thematic questions, as well as notes from the Lead Facilitator, capturing key points from the large room discussions, and transcripts documenting the input from the two expert speakers. These were subsequently coded and the raw qualitative data thematically analysed.

Based on either group consensus or expressed importance to individual participants, data were coded according to whether they were 1) essential information to include to ensure understanding of participants' perspectives and lived experiences, 2) they contained issues of concern that participants wanted policy makers to be aware of and 3) for suggestions and recommendations for policy makers/healthcare management to consider and respond to. Thematic analysis was subsequently employed as the qualitative data analysis method. This involved reading through the data set for each dialogue round and identifying patterns in meaning across the data to derive themes (Brown & Clarke 2006). This resulted in the identification of themes for each dialogue supported by direct quotations by participants.

Since thematic analysis involves 'reflexivity' where a researcher's subjective experience plays a central role in meaning making from the data, this draft report was then reviewed by others in conjunction with the raw data set to reduce bias. The emerging themes were distilled to produce overall key recommendations. The expert Advisory Panel then reviewed the summary and draft report. Their comments and edits informed the report.

This draft report was sent to all event participants for their sense checking to ensure the report correctly reflected the input of participants. Any corrections were incorporated at the final stage of report preparation.


Aislinn
Cooper

Appendix II

Contributors and Acknowledgements

It takes a village to organise and participate in a genuinely inclusive deliberative dialogue about a topic as complex as neurogenomics. Sincere thanks to all those listed below who made the time to plan and engage in the dynamic event. We would like to thank in particular Lorna Kerin for her role in the design of the event, for acting as Lead Facilitator on the day and for her critical contributions to the analysis of the data and drafting of the report.

This event was supported by Science Foundation Ireland (SFI) under Grant Number 16/RC/3948 and co-funded under the European Regional Development Fund and by the Health Research Board and the Irish Research Council under grant PPI-221-001.

We also greatly appreciate the policy makers, patient organisations and health service providers who we hope will now consider and implement the recommendations generated in partnership with the neurological community.

Deliberative Dialogue Advisory Panel:

- Sally Ann Lynch, Consultant Geneticist, CHI at Crumlin
- Mark Bale, Independent consultant in genomics, policy and bioethics
- Gary Boyle, FutureNeuro PPI Advisory Panel member
- Laura Brady, BESTS Programme Manager RCSI
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- Nuala Ryan FutureNeuro PPI Panel member

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Analysis: Lorna Kerin; Gianpiero Cavalleri
Report writing: Lorna Kerin, Bridget Doyle, Susan Crawford, Gianpiero Cavalleri
Graphic Illustrator: Hazel Hurley

Event Facilitation Team

Appendix III

Deliberative Dialogue Participants:

Name	Organisation	Stakeholder Group/Specialization
Aislinn Cooper	Mater Hospital	Researcher (Medical Genomics)
Andrew Green	Children's Health Ireland (CHI) at Crumlin	Clinician/Researcher (Consultant Clinical Geneticist)
Anne O'Shea Clarke	Huntington's Disease Association of Ireland	Patient Advocate (Huntington's Disease)
Bridget Doyle	FutureNeuro	Research Centre Manager
Colin Doherty	St. James' Hospital	Clinician, Researcher (Consultant Neurologist)
Deirdre Ní Mhórdha	Migraine Association of Ireland	Patient Advocate (Migraine)
Derick Mitchell	Irish Platform for Patient Organisations, Science and Industry (IPPOSI)	Patient Advocate (National Organization inclusive of all conditions)
Ed Gilbert	FutureNeuro/RCSI	Researcher (Population Genetics)
Eleanor Molloy	Trinity College Dublin	Clinician/Researcher (Consultant Neonatal Paediatrician)
Eloise Cowie	Brain Tumour Ireland	Patient Advocate(Brain Tumour)
Emma Dorris	UCD	Researcher (Biomedics/PPI)
Geraldine Dunne	Epilepsy Ireland	Patient Advocate (Epilepsy)
Gianpiero Cavalleri	FutureNeuro/ RCSI	Researcher (Genomics)
Helena Quaid	The Alzheimer Society of Ireland	Patient Advocate (Alzheimer's)
Hugh Kearney	St. James' Hospital	Clinician/Researcher (Consultant Neurologist)
Joe Condon	FutureNeuro PPI Panel	Patient Advocate (Parkinson's)
Katarzyna Whysall	NUI Galway	Researcher (Epigenetics)
Kathleen Gorman	Children's University Hospital, Temple Street	Clinician/Researcher (Consultant Paediatric Neurologist)
Kevin Quaid	The Alzheimer Society of Ireland	Patient Advocate (Alzheimer's)
Laura Brady	RCSI	Researcher (Digital Health/Clinical Trials)
Laura O' Philbin	The Alzheimer Society of Ireland	Non Profit Representative (Alzheimer's)

Name	Organisation	Stakeholder Group/Specialization
Lillian McGovern	Irish Motor Neurone Disease Association (IMNDA)	Patient Advocate (Motor Neuron Disease)
Lorna Lopez	Maynooth University	Researcher (Neuropsychiatric Genetics)
Mags Rogers	Neurological Alliance of Ireland	Patient Advocate (Neurological)
Marguerite Keating	The Alzheimer Society of Ireland	Patient Advocate (Alzheimer's)
Maria Carty-Mole	FutureNeuro PPI Panel	Patient Advocate (Epilepsy)
Nicola Kehoe	Dravet Syndrome Ireland	Patient Advocate (Dravet Syndrome)
Norman Delanty	Beaumont Hospital	Clinician/ Researcher (Consultant Neurologist)
Nuala Ryan	FutureNeuro PPI Panel	Patient Advocate (Epilepsy)
Omar Mamad	FutureNeuro/RCSI	Researcher (Neuroscience/Epilepsy)
Patricia Towey	Huntington's Disease Association of Ireland	Patient Advocate (Huntington's)
Petra Bencurova	FutureNeuro/RCSI	Researcher (Genetics of Epilepsy/ Dravet)
Susan Byrne	Children's Health Ireland (CHI) at Crumlin	Clinician, Researcher (Consultant Paediatric Neurologist)
Susan Crawford	FutureNeuro/ RCSI	Patient Advocate (Autism)
Vicky McGrath	Rare Diseases Ireland	Patient Advocate (Rare Diseases)

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