National Strategy for Accelerating Genetic and Genomic Medicine in Ireland

Minutes of Implementation Steering Group for Genetics and Genomics Meeting			
Date	23/11/2023	**Time**	10:30-11:15
Location	MS Teams		

Attendees	
Dr Colm Henry (CH)	Dr Mark Bale (MB)
Dr Richard Hagan (RH)	Eleanor Masterson (EM)
Valerie Walshe (VW)	Christopher Ryan (CR)
Prof Risteárd Ó Laoide (ROL)	Ailish Kelly (AK)
Martina Burns (MBs)	Marie Culliton (MC)
Prof Eileen Treacy (ET)	Margaret Cuddigan (MC)

Apologies	
Eilísh Hardiman (EH)	Deirdre McNamara (DMcN)
Philippa Ryan Withero (PRW)	Dr. Eppie Jones
Emma McCann (EMcC)	Avril Daly (AD)
Dr Cliona Murphy (CM)	Prof Mary Day (MD)
Oonagh Ward (OW)	

HSE Staff	
Pauline Sargent (PS)	Aisling Cusack (AC)
Maeve Smith (MS)	James Kelly (JK)
Christine Daly (CD)	

Notes

1. Apologies, Minutes and Matters Arising

- MB welcomed members to the meeting and gave an overview of the agenda for the meeting.
- MB noted the apologies
- MB commented that Avril Daly would be joining the ISG as a patient representative and taking over from Karen Morgan who had to leave due to other commitments.

2. Genetics and Genomics Operating Model – Update

- MB provided an update on the Operating Model consultations and noted that they were well engaged and interactive sessions. All the feedback was taken and grouped into six main themes and aligned to the Sláintecare principles. This feedback will be held by the NGGO for future development of an operating model.
- Recruitment update – MB welcomed new staff members JK & CD and indicated that a new Programme Manager will be starting in December.
- Frontline Post Recruitment Update –MB discussed the challenges brought by the HSE recruitment embargo and gave an update on the posts being withdrawn for now and that unfortunately the Genetic counsellor positions and Genetic Research Associates are all on hold as well.
- Education – MB informed the group that 3 webinars/masterclasses have been completed to date. The materials are due to remain active on HSELanD following the completion of the programme through RCSI and RCPI channels.

Testing Guidance & Directory Update

- RH – Gave update on the Testing Guidance – identified 3 areas in particular, Pre-Testing, during testing and post testing. NGGO have held a number of working group meetings with a lot of discussion around the request form and consent form. RH expressed thanks to the group for the effort and time given to this activity.
- Next meeting with the Testing guidance working group is the 6th of December. RH will inform the group of feedback raised by the Communications and PPI Working Group around the topic of informed consent in relation to Testing Guidance for clinicians.
- RH informed the group that the development of a Testing Directory is the key priority for remainder of the year and early 2024.
- RH described the proposed phased development of a draft Testing Directory. The first phase to commence immediately will involve the development of a comprehensive list of conditions, tests and associated methodology, to be delivered by Q2 2024. This will initially be in draft form not for publication until the tests can be operationalised.
- In early 2024, the NGGO will begin a parallel second phase which will involve identification and selection of a subset of specialties for the phased development of a test directory which will be operationalised through an iterative process via current networks of laboratories and overseas suppliers.
- CH stated that this is an important piece of work and that he has been given a very clear description of the work that is planned and that this is one of the important deliverables for this programme.
- RH said that for the EMT meeting in December we will have the project plan put together for the development of a Test Directory for Rare and Inherited Disease. The Plan will involve engaging with the specialities and with the clinicians. By end of Q2 in 2024, the draft version will be completed, noting that it will not be ready for operationalisation.
- CR said thanks for the update. And that he thinks it would be useful to have these slides so that we can go through it in more detail. CR asked whether the process is to go through the ISG group before the EMT and noted that it would be useful to take time to digest it before and provide feedback.
- CH agrees should reviewed by the group before it goes to the EMT
- MB agreed that the NGGO will circulate the slides and explained timelines are very tight.
- CH pointed out ISG function is to mark themselves against the strategy and that if that requires issuing another document before EMT then that should be actioned.

- ET commented that she has had a number of meetings with Europe. What has come out from other countries is that it is a missed opportunity to not use ORPHA codes. ET raised the point that the absence of an Interim National Bioinformatics Director is a concern. MB explained that the NGGO did try to backfill the post to cover leave, but the HSE recruitment embargo has meant that all applicants were ineligible. MB noted that any support on that from ET and CR would be welcomed.
- ET we have to deliver on the RD ORPHA code project.
- CH actioned that members of this group need to see this Test Directory plan and provide feedback on it as soon as possible.

3. Communications & Stakeholder Engagement Update

- AC discussed the new communications research project, its role and objectives. The project aims to increase the genetic and genomic health literacy among patients who have been referred to genetic and genomic services.
- AC informed the group that a research agency has been selected and the project will kick-off in the coming weeks with a planned completion of February-March 2024. CH commended the work for its patient-focus and thanked AC. MC commented that it was very positive and shows a detailed approach and emphasised the pivotal role genetic counsellor's play in the patient experience.
- CH patient stories were a strength of the strategy and we need to mark that.

4. 1+MG Project & National Mirror Group Update

- CR provided an update on the recent 1+MG special group meeting in Barcelona he attended. The main purpose of the meeting was to move into a deployment phase from 2022-2027. They showed a Roadmap that he found very useful particular for where we are currently at and how we ensure Ireland can draw on European best practise.
- CR had a clearer idea of the 1+MG initiative after the meeting. It is designed to try to build a data infrastructure for genomic and clinical data across Europe. Where reference population but also clinical reference. There are 2 parts there is the Genome of Ireland project. The application went in from RCSI yesterday.
- The other aspect is to build the clinical side of things and the implementation of the strategy. They are looking at cancer, rare diseases, infectious, Covid samples and others. This reinforces the need for staff recruitment and lab capacity as set out in the Strategy. What capacity is available on the market and what is appropriate to deliver the goal of a database of whole genomes.
- This is focused on researchers but also how benefits are communicated to individuals (and via the Communications and PPI group should be considered.
- One of the areas they are still working on is the level of industry involvement. That is something we will have to navigate and embrace and maybe an opportunity for Ireland to take a lead on.
- CH spoke about the recruitment embargo and said lesson learnt when you get funding, recruit quickly.
- RoL requested a 2 pager on the 1+ MG, GDI and Genome of Europe/Ireland to provide an overall view. CR agreed to action.
- MB thanked CR for the helpful update and said that the clinical impact needs to be addressed, for example, what happens to large Panel data and Exome data, and how it connects into rare diseases and cancer?
- ET highlighted that Ireland is very involved in EU Rare Diseases. In addition, that 50% of rare diseases are undiagnosed. All the documents are there. Would be helpful if a paper is written about it so that people will understand how it is all linked up. There are other initiatives such as ERDERA as well. A simple paper that we could all understand would be helpful.
- RH asked CR if the roadmap has been published?
- 5. Closing Remarks/AOB

MB talked through the next steps for the implementation plan and mentioned the upcoming testing directory meeting and that the slides from today and the extract from the EMT report would be sent on to members for their feedback on the Testing directory.

- CH thanked members for their time and encouraged them to provide feedback on the Test Directory.

New Actions				
#	**Action**	**Action Owner**	**Date Raised**	**Action Date**
1.	Testing Directory extract to be sent to members for their review and feedback	NGGO (PS)	23/11/2023	
2.	ISG members to review extract and submit any comments by cob on the 29th Nov	ISG Members	23/11/2023	
3.	2 Page overview document of 1+MG/GDI/Genome of Europe to be prepared for ISG members	DoH (CR)	23/11/2023	
4.	Slides from ISG meeting to be shared with ISG members	NGGO (PS)	23/11/2023	