

Table S1: Participant survey demographics summary

Participant demographic	Count	Percentage (%)
<i>Participant sex</i>		
- Male	6	18.8
- Female	25	78.1
- Unknown	1	3.1
<i>Survey completed by parent / carer</i>	18	56.3
<i>Age bracket of person unable to complete the form themselves</i>		
- 0 – 17	13	72.2
- 18+	5	27.8
<i>Age bracket of participant</i>		
- 18-34	11	34.4
- 35-44	5	15.6
- 45-54	7	21.9
- 55+	8	25.0
- Skipped question	1	3.1
<i>Ethnicity</i>		
- British	8	25.0
- Northern Irish	18	56.3
- Other	5	15.6
- Skipped question	1	3.1
<i>Medical specialism(s) attended</i>		
- Cardiology	4	13.8
- Dermatology	5	17.2
- Ear, nose and throat	5	17.2
- Gastroenterology	5	17.2
- Genetics	18	62.1
- General practitioner	22	75.9
- Haematology	1	3.4
- Immunology	3	10.3
- Nephrology/Urology	4	13.8
- Neurology	14	48.3
- Oncology	2	6.9
- Ophthalmology	4	13.8
- Paediatrics	11	37.9
- Psychiatry	4	13.8
- Respiratory	5	17.2
- Rheumatology	1	3.4
- Other	7	24.1

Table S2: Themes and associated codes from semi-structured interviews of healthcare professionals

Theme	Associated codes
Healthcare professionals had a largely positive experience of the 100KGP	<ul style="list-style-type: none"> - Felt the project ran smoothly / enjoyed participation - Discussed the significant benefits for the patients <i>e.g.</i> ending the diagnostic odyssey / modifying care plans - Reported that they felt the participants also had a good experience of the 100KGP - Emphasised willingness to participate in future similar projects - The multi-disciplinary approach was useful - Felt that WGS was the future of healthcare
Facilitating WGS was a significant workload burden	<ul style="list-style-type: none"> - Time-consuming - Involved extra hours - Difficult to manage on top of regular workload - Capacity was limited
Interviewees found that participants expressed some concerns about additional findings and time to results	<ul style="list-style-type: none"> - Concerns about additional findings - Concerns about time to results / management of expectations - Generally good understanding - Consent process could be streamlined / excess paperwork
There is a need for additional training	<ul style="list-style-type: none"> - Assumptions were made about genetics understanding beyond specialists - Complicated field to understand - Mainstreaming beyond genetics is needed - Needs to be more straight-forward - Training for non-specialists needed - Concerns patients attending non-specialists may be at a disadvantage - Flow chart of next steps may be useful

Table S3: Themes and associated codes from discussion workshop

Themes	Codes
Theme 1: Resource constraints hinder collaborative rare disease research	<ul style="list-style-type: none"> • Lack of collaboration • Lack of funding • Lack of political will to provide resources • Lack of incentive for clinical research • Cross border relationships in research enable better utilisation of limited resources on whole island • Incentives needed to encourage research, both as a career and within clinical environments
Theme 2: Collaborative rare disease research is hindered by ineffective communication	<ul style="list-style-type: none"> • Lack of progress • Lack of collaboration • Lack of follow up and dissemination of results • Multidisciplinary approach is needed • Importance of communicating with patients effectively • Need to establish better communication procedures to maintain contact between patients and healthcare professionals • Wider support services need to be available to aid patients/families in processing a diagnosis

<p>Theme 3: Rare disease awareness, support and information services are insufficient.</p>	<ul style="list-style-type: none"> • Lack of community rare disease awareness / outreach • Need for more communication and engagement on social media • More comprehensive support services need to be available to aid patients/families in processing a diagnosis • Need for more awareness in educational settings • Need for rare disease events as social, educational, and networking opportunities for patients, families, HCP's, and researchers • Need to explore multiple avenues of dissemination of research opportunities and communication. • Lack of true accessibility in facilities deemed accessible • Need for more awareness among HCPs
<p>Theme 4: Current administrative systems are barriers to collaborative rare disease research.</p>	<ul style="list-style-type: none"> • Ethical procedures require streamlining to facilitate research • Hospital ethics procedures are particularly slow compared to institutions • Northern Ireland is slower compared to other parts of the UK
<p>Theme 5: Interprofessional collaboration was regarded as beneficial for rare diseases.</p>	<ul style="list-style-type: none"> • Benefits of the multi-disciplinary approach discussed • Need for cross-border collaboration • International collaboration helpful for conditions where only a small number of people in one location are affected