Recruitment of >2600 families to Intellectual Disability and Mental Health: Assessing the Genomic Impact on Neurodevelopment (IMAGINE ID) Study

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Background: Using genetics to ascertain an ID cohort

- IMAGINE ID is a cohort study that aims to identify the genetic contribution to mental health outcomes in ~3000 children and young adults with intellectual disability (ID).
- Genetic testing using a microarray (aCGH) is offered in the NHS if a child has developmental delay. ~14% of children will be diagnosed this way[1,2] often revealing large structural chromosome anomalies; copy number variations (CNVs).
- For some individuals, more detailed analysis of single genes or the whole of the genetic code is required to identify the genetic cause of ID, uncovering single nucleotide variants (SNVs).
- Systematic sampling and assessment to determine why some, but not all, ID-related CNVs and SNVs are associated with poor mental health outcomes is required to help support better care and early interventions in the future.

So, what does this *mean* for my child?

Methods: who joined and what did they do?

✓ **Individuals aged 4 and over**

✓ **Clinically significant CNV(s) and/or SNV**

✓ **Generalised ID or developmental delay**

- Individuals who met the above inclusion criteria were recruited through referrals by 22 NHS Regional Genetics Centres (RGCs), charities and self-referrals by carers over a 2-4 year period.
- Primary carers reported on the individual's behavioural adjustment and mental health via the DAWBA (Development and Well-Being Assessment) used in previous national and international surveys of mental health in both typical and ID populations.
- Face-to-face interviews were also conducted with ~10% of the cohort with specific neurosusceptibility loci CNVs to validate all online data collection.

Results: n=2635 where do we recruit from?

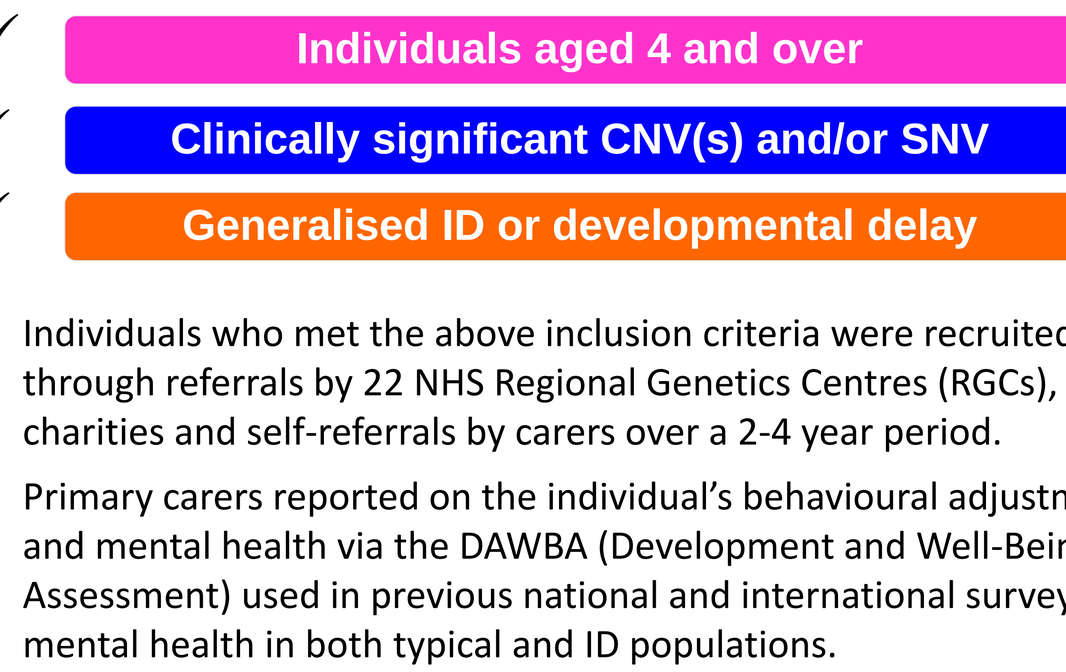

Figure 1: Chart illustrating recruitment into IMAGINE ID and the number of individuals recruited per pathway

- 1927 (73%) of individuals were recruited through RGCs from October 2014 to September 2018.
- The remaining 708 (27%) were recruited through charities, social media and other research studies.

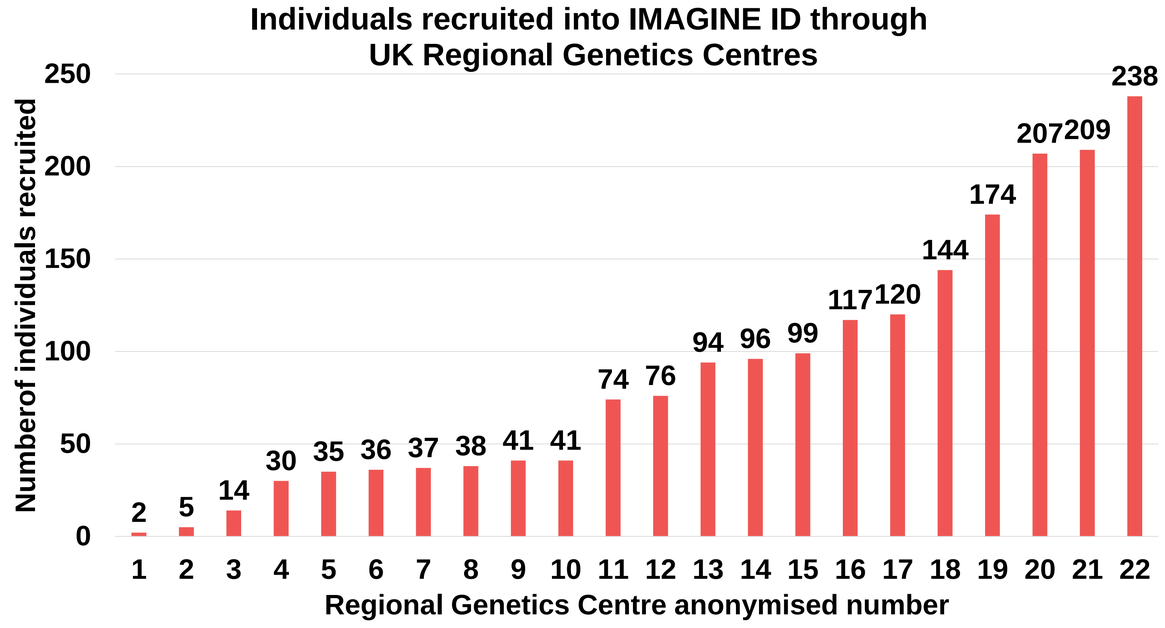

Figure 2: Graph demonstrating range of numbers of individuals recruited from UK Regional Genetics Centres over a 2-4 year period

- A significant recruitment disparity existed across UK RGCs.
- Workshops in 2016/17 were held for RGC staff (n=39 attended) to encourage recruitment, share good practice and feedback to the study team on the barriers and facilitators to ID/genetic research.

Research Barriers in NHS:

- Lack of recruiting staff and resources
- Research saturation for staff
- Poor communication in busy clinic environments or lack of understanding of the research inhibiting informed consent

Research Facilitators in NHS:

- Simple consent processes for staff and families
- Support from clinical and research management teams
- Hearing positive research participant feedback to motivate further invites

Conclusions: supporting accessibility of research

- Enrolment into large-scale genetic and ID studies relies heavily on staffing and infrastructure within clinical services.
- Inequity of access to genetic and ID research exists across the UK.
- Allocation of grant funds to supporting recruitment within NHS services, particularly when focusing on post-genomic diagnoses, is strongly recommended.
- Further work and recruitment is continuing on the IMAGINE ID study to enable additional conclusions to be made on a significantly powered cohort with ID and a pathogenic genetic cause.

References and acknowledgments

Thank you to all the patients and their families for their time and effort in participating in the IMAGINE ID study. Thanks also to all the staff at 22 NHS Regional Genetics Centres that have worked tirelessly in recruiting individuals into the project and those that were able to attend the workshops, thank you for your insightful contributions. Thanks to all the charities and patient support groups that have promoted our study to their members.

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