**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\(^1\) typically 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?**

- Unique charity n=276 (11%)
- ECHO research study n=158 (6%)
- IMAGINE ID website n=53 (2%)
- Other patient support groups n=186 (7%)
- Social media n=35 (1%)

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

**Individuals recruited into IMAGINE ID through UK Regional Genetics Centres**

- 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.

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 Barriers 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**

- Enrollment 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**

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