IMAGINE ID

"So what does this mean for my child?"

Rare Disease

- Affects < 5 in 10,000 of the general population
- 1 in 17 people affected at some point in their life
- 80% of rare diseases have a genetic component
- Small structural changes of the genome such as Copy Number Variations (CNV's) or Single Nucleotide Variants (SNV's) can cause or increase the risk of certain diseases

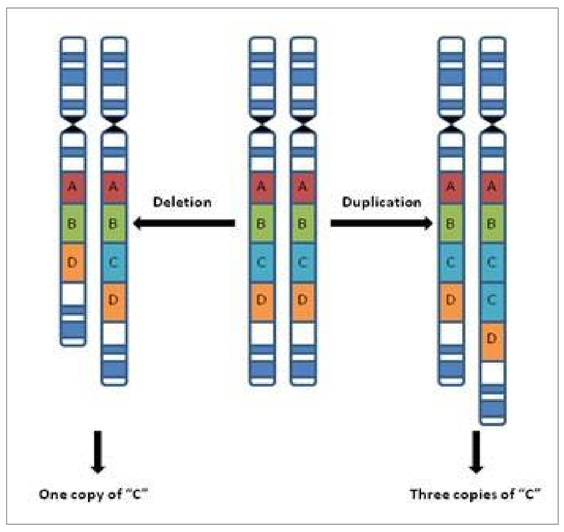


Figure 1: Diagram of copy number variant deletions and duplications

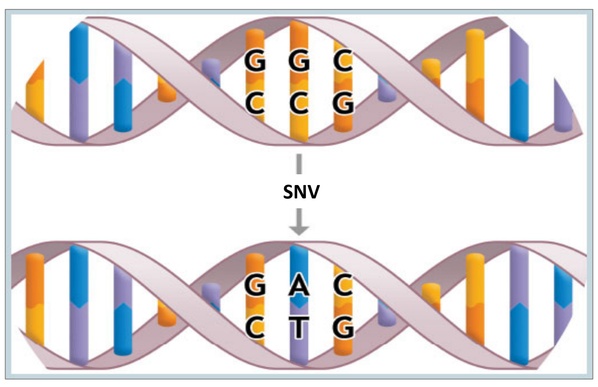


Figure 2: Diagram of single nucleotide variant mutation

Neurodevelopment

- Growth of neurological pathways in the brain that influence performance or functioning
- Defects in this can cause intellectual disability (ID), learning difficulties or developmental delay
  - Significant limitations in cognitive function and adaptive behaviours
  - Failure to meet developmental milestones
- CNV's account for approximately 14% of ID (Cooper et al., 2011)
- CNV's also associated with increased risk of psychiatric illness such as autism spectrum disorders and schizophrenia (Doherty & Owen, 2014).

Aims

- A question parents often ask when their child has a genetic condition is:

  "So what does this mean for my child?"

- To answer this we are:
  - Collecting information about a large group of children with ID and a confirmed genetic change
  - Conducting research to find out how genetic changes affect children and young people's behaviour

Inclusion Criteria

- ✓ Children aged 4 and over
- ✓ Clinically significant CNV(s) or SNV
- ✓ Has intellectual disability, learning difficulties, or developmental delay

Where do we recruit from?

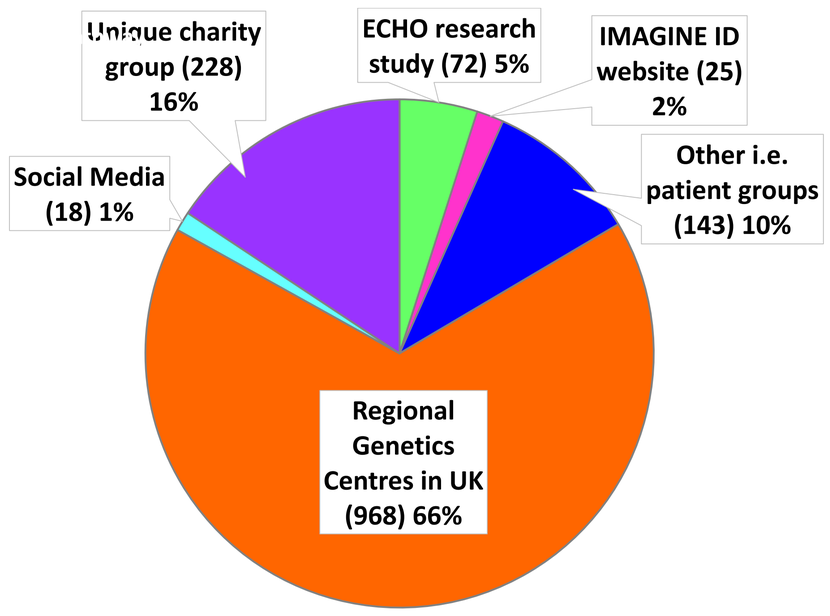


Figure 3: Graph showing recruitment pathway of IMAGINE ID participants

Our recruitment so far…

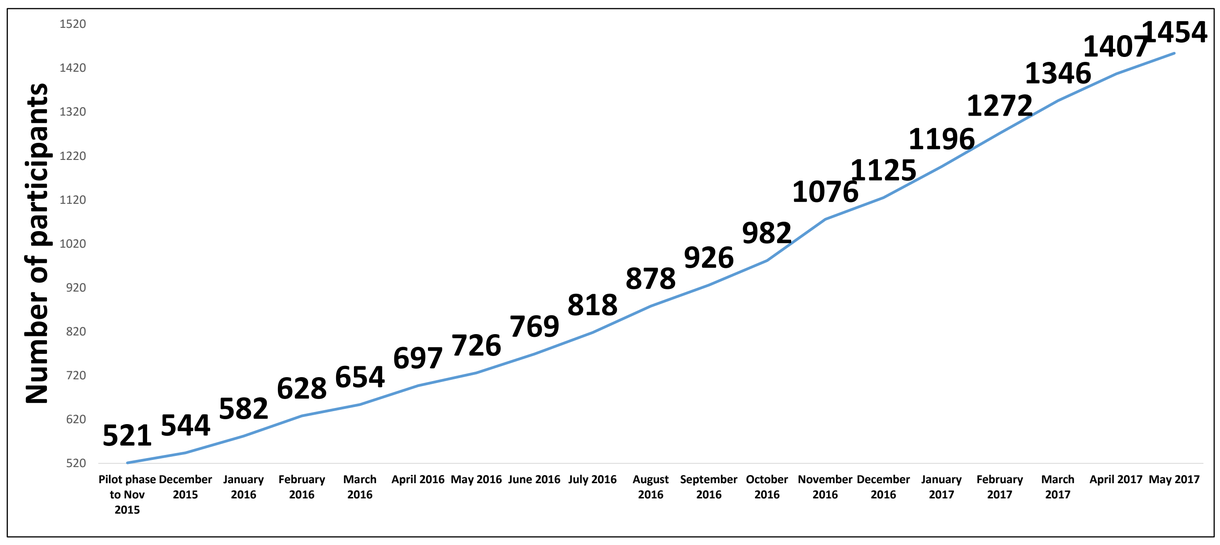


Figure 4: Graph showing cumulative recruitment to date for IMAGINE ID

Age of IMAGINE ID cohort

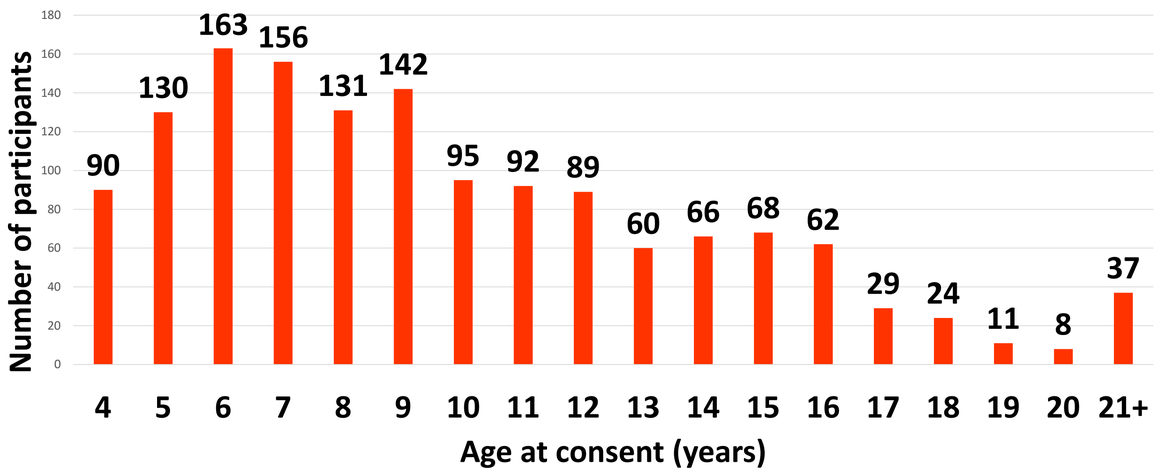


Figure 5: Graph showing age of participants at the time of consent

MRC Medical Research Council | MRC Medical Research Foundation | Cambridge University Hospitals NHS Foundation Trust | University of Cambridge | UCL | Cardiff University Prifysgol Caerdydd | IMAGINE ID

IMAGINE ID

Gender of participants

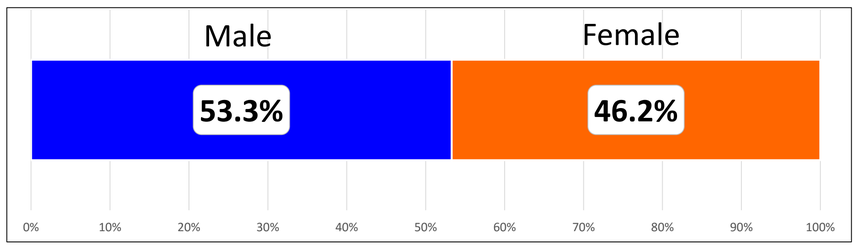


Figure 6: Graph showing proportion of male to female in the cohort

Recurrent CNV's observed in cohort

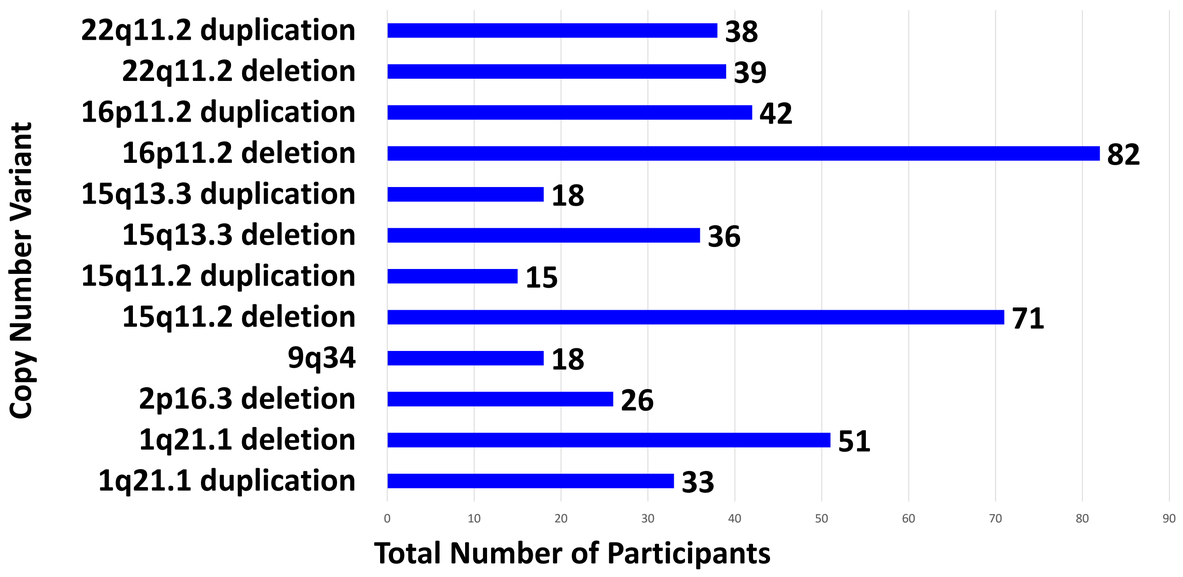


Figure 7: Graph showing recurrent CNV distribution

What does the study involve?

- The primary carers of the participants are invited to complete online questionnaires:
  - Development & Wellbeing Assessment (DAWBA)
  - Medical Questionnaire
  - Adaptive Behaviour Assessment System
- Once DAWBA has been completed, families receive a personalised summary report

DAWBA Report

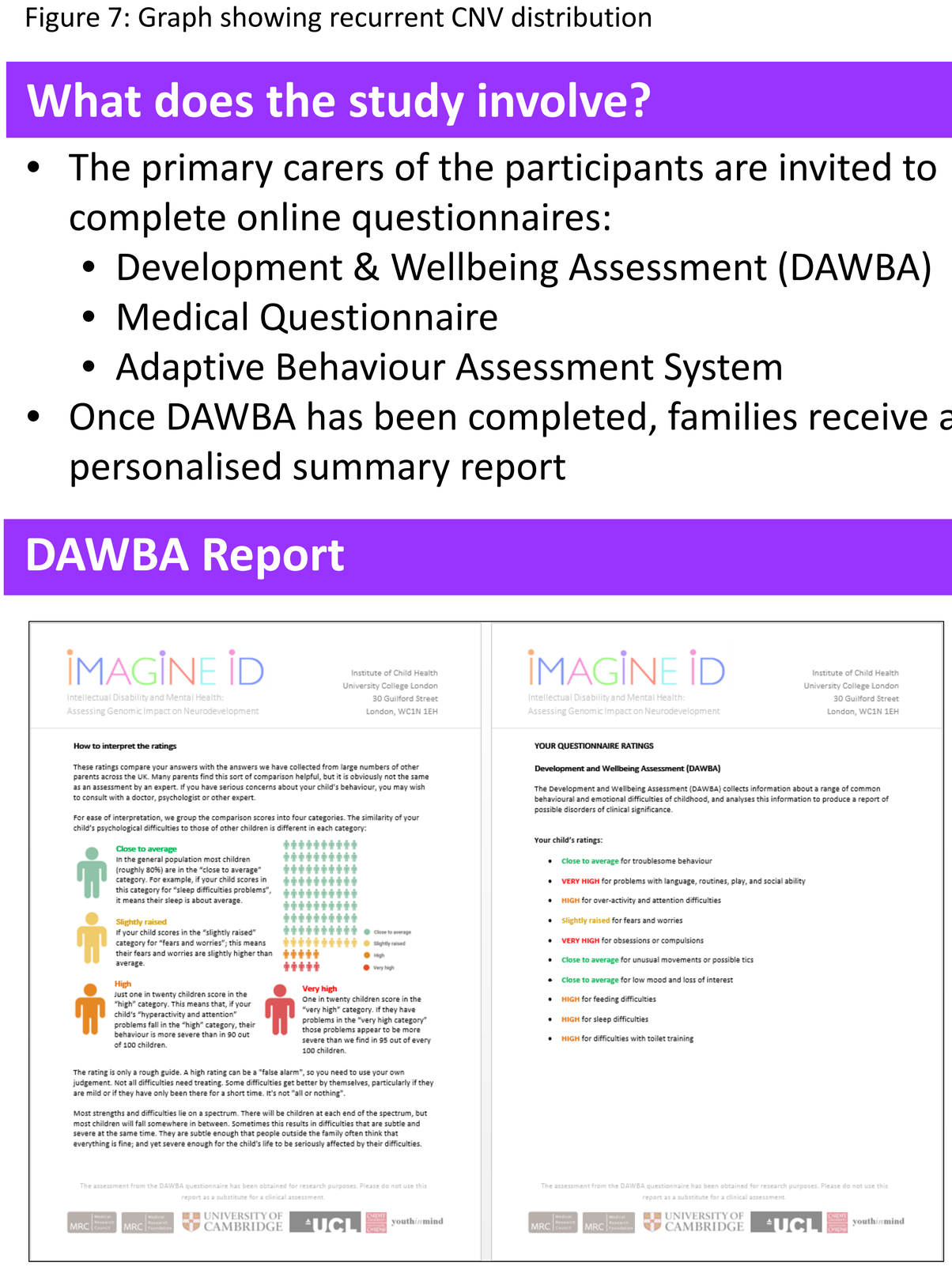


Figure 8: Diagram to show an example DAWBA report

Feedback from families on DAWBA Report

"The format of the questionnaire was so straightforward to follow, allowing me to drop in and out of it as I found the time."

"I was very, very pleased with how it was all set up… really easy to understand and follow"

"Sent it to the educational team – hoping it will help them to discuss providing more support"

"Amazing – helped in education setting and used in his specialist school. They have looked through it and bits they weren't aware of, or bits I haven't mentioned have been really informative for them"

"It gives a record of where she is and how she is now. When I read it in 5 years' time I can look back at how she was and if things have changed."

Draw a Person

- We also ask participants to complete a drawing task to estimate their developmental level

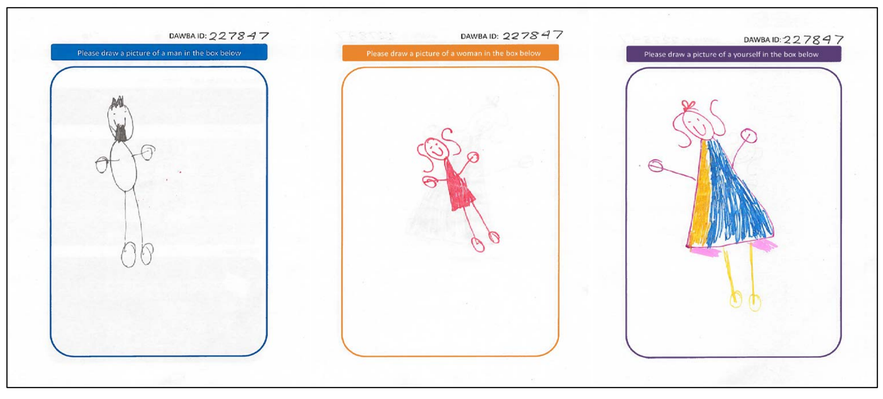
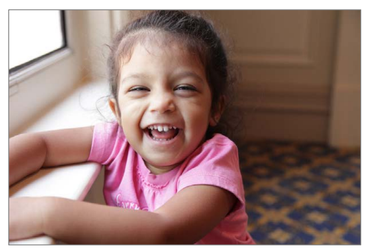


Figure 8: Diagram to show an example Draw a Person task

Face-to-Face Assessment

- A proportion of participants are selected for further in-depth assessments in their home:
  - Child & Adolescent Psychiatric Assessment (CAPA)
  - Wechsler Abbreviated Scale of Intelligence (WASI)
  - CANTAB battery of tests
  - Wisconsin Card Sorting Test
- So far over 235 participants have been assessed



References

- Cooper G.M., Coe, B.P., Girirajan, S. et al. (2011) A copy number variation morbidity map of developmental delay. *Nature Genetics*, 43, 838-846.
- Doherty, J.L. & Owen M.J. (2014). Genomic insights into the overlap between psychiatric disorders: implications for research and clinical practice. *Genome Medicine*, 6, 29.

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