

## INTELLECTUAL DISABILITY AND COPY NUMBER VARIANTS: MENTAL HEALTH IN THE IMAGINE ID COHORT

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**Background:** Increasingly, rare alterations in the genetic code can be found in individuals who have intellectual disability (ID). These alterations range from large copy number variants (CNV) of groups of genes to alterations in single genes. The association of genetic variants with psychiatric morbidity is still poorly understood. IMAGINE ID is a national study of behavioural problems and psychiatric risk in children with ID of known genetic origin. No previous study has systematically evaluated the behaviour of children with ID of genetic aetiology. **Methods:** 902 children and young people were recruited via UK Genetic Services and patient support groups. Caregivers completed the Development and Wellbeing Assessment (DAWBA). It assesses psychiatric symptomatology and provides DSM-5 classifications based on clinical ratings. **Results:** A higher rate of children met criteria for mental health diagnoses in the IMAGINE ID sample (48%) compared to national surveys of children both with and without ID (ID 39%; Non-ID 8.1%). The cohort is 57% male and the mean age is 6.3 years. The mean age of genetic diagnosis after 2010 was 2.9 compared to 9.6 for those diagnosed pre-2007. 39% of the cohort have CNVs associated with known neurosusceptibility loci. 19% of participants had more than one CNV. Where CNV inheritance is known, 49.1% were de novo, 32.5% were maternally inherited and 18.4% were paternally inherited. **Conclusion:** Children with ID of genetic origin have higher rates of mental health and behavioural difficulties than both the general population and children with ID of unknown genetic cause. This suggests that CNVs may confer additional psychiatric risk. Identification and early intervention and support for children and families with ID may improve their long-term mental health outcomes. Further work on the IMAGINE ID cohort will be important in delineating the behavioural phenotypes associated with specific CNVs and single gene disorders. **Keywords:** Intellectual disability, mental health, behaviour, genetics.