

IMAGINE-ID consortium publications:

Adams, R. L., Baird, A., Smith, J., Williams, N., van den Bree, M. B., Linden, D. E., ... & Linden, S. C. (2023). Psychopathology in adults with copy number variants. *Psychological medicine*, 53(7), 3142-3149.

Baker K, Devine RT, Ng-Cordell E, Raymond FL, IMAGINE-ID consortium , Hughes C. (2021). Childhood intellectual disability and parents' mental health: integrating social, psychological and genetic influences. *The British journal of psychiatry : the journal of mental science*, 218(6), pp. 315-322. doi: [10.1192/bjp.2020.38](https://doi.org/10.1192/bjp.2020.38)

Bartsch U, Moulding H, Eaton C, Marston H, Hall J, Hall J, ... Jones M. (2022). Sleep EEG in young people with 22q11.2 deletion syndrome: A cross-sectional study of slow-waves, spindles and correlations with memory and neurodevelopmental symptoms. *eLife*, doi: [10.7554/elife.75482](https://doi.org/10.7554/elife.75482)

Bass N, Skuse D. (2018). Genetic testing in children and adolescents with intellectual disability. *Current opinion in psychiatry*, 31(6), pp. 490-495. doi: [10.1097/YCO.0000000000000456](https://doi.org/10.1097/YCO.0000000000000456)

Butter, C. E., Goldie, C. L., Hall, J. H., Leadbitter, K., Burkitt, E. M., van den Bree, M. B., & Green, J. M. (2024). Experiences and concerns of parents of children with a 16p11.2 deletion or duplication diagnosis: a reflexive thematic analysis. *BMC psychology*, 12(1), 137.

Chapman G, Alsaqati M, Lunn S, Singh T, Linden SC, Linden DEJ, ... Syed YA. (2022). Using induced pluripotent stem cells to investigate human neuronal phenotypes in 1q21.1 deletion and duplication syndrome. *Molecular psychiatry*, 27(2), pp. 819-830. doi: [10.1038/s41380-021-01182-2](https://doi.org/10.1038/s41380-021-01182-2)

Chawner, S. J., Owen, M. J., Holmans, P., Raymond, F. L., Skuse, D., Hall, J., & van den Bree, M. B. (2019). Genotype–phenotype associations in children with copy number variants associated with high neuropsychiatric risk in the UK (IMAGINE-ID): a case-control cohort study. *The Lancet Psychiatry*, 6(6), 493-505.

Chawner S, Evans A, , Wolstencroft J, Chawner S, Hall J, ... van den Bree M. (2023). Sleep disturbance as a transdiagnostic marker of psychiatric risk in children with neurodevelopmental risk genetic conditions. *Translational Psychiatry*, (1), doi: [10.1038/s41398-022-02296-z](https://doi.org/10.1038/s41398-022-02296-z)

Chawner SJRA, Doherty JL, Moss H, Niarchou M, Walters JTR, Owen MJ, ... van den Bree MBM. (2017). Childhood cognitive development in 22q11.2 deletion syndrome: case-control study. *The British journal of psychiatry: the journal of mental science*, 211(4), pp. 223-230. doi: [10.1192/bjp.bp.116.195651](https://doi.org/10.1192/bjp.bp.116.195651)

Chawner SJRA, Owen MJ, Holmans P, Raymond FL, Skuse D, Hall J, ... van den Bree MBM. (2019). Genotype-phenotype associations in children with copy number variants associated with high neuropsychiatric risk in the UK (IMAGINE-ID): a case-control cohort study. *The lancet. Psychiatry*, 6(6), pp. 493-505. doi: [10.1016/S2215-0366\(19\)30123-3](https://doi.org/10.1016/S2215-0366(19)30123-3)

Chawner SJRA, Doherty JL, Anney RJL, Antshel KM, Bearden CE, Bernier R, ... van den Bree MBM. (2021). A Genetics-First Approach to Dissecting the Heterogeneity of Autism: Phenotypic Comparison of Autism Risk Copy Number Variants. *The American journal of psychiatry*, 178(1), pp. 77-86. doi: [10.1176/appi.ajp.2020.20010015](https://doi.org/10.1176/appi.ajp.2020.20010015)

Chawner SJRA, Owen MJ. (2022). Autism: A model of neurodevelopmental diversity informed by genomics. *Frontiers in psychiatry*, 13, pp. 981691. doi: [10.3389/fpsy.2022.981691](https://doi.org/10.3389/fpsy.2022.981691)

Chawner SJRA, Owen MJ. (2022). Autism: A model of neurodevelopmental diversity informed by genomics. *Frontiers in psychiatry*, 13, pp. 981691. doi: [10.3389/fpsy.2022.981691](https://doi.org/10.3389/fpsy.2022.981691)

Chawner, S. J., Paine, A. L., Dunn, M. J., Walsh, A., Sloane, P., Thomas, M., ... & Procter, A. (2023). Neurodevelopmental dimensional assessment of young children at high genomic risk of neuropsychiatric conditions. *JCPP advances*, 3(2), e12162.

Chawner, S. J., Evans, A., IMAGINE-ID consortium Wolstencroft Jeanne 3 Chawner Samuel JRA Hall Jeremy van den Bree Marianne BM Owen Michael J. Skuse David 3 Raymond F. Lucy 4 5 6, Williams, N., Owen, M. J., Hall, J., & van den Bree, M. B. (2023). Sleep disturbance as a transdiagnostic marker of psychiatric risk in children with neurodevelopmental risk genetic conditions. *Translational Psychiatry*, 13(1), 7

Chi, Z., Devine, R. T., Wolstencroft, J., Skuse, D., Hughes, C., Baker, K., & IMAGINE-ID consortium. (2024). Rare neurodevelopmental conditions and parents' mental health—how and when does genetic diagnosis matter?. *Orphanet Journal of Rare Diseases*, 19(1), 70.

Constable PA, Lee IO, Marmolejo-Ramos F, Skuse DH, Thompson DA. (2021). The photopic negative response in autism spectrum disorder. *Clinical & experimental optometry*, 104(8), pp. 841-847. doi: [10.1080/08164622.2021.1903808](https://doi.org/10.1080/08164622.2021.1903808)

Cunningham A, Delpont S, Cumines W, Busse M, Linden D, Hall J, ... van den Bree M. (2018). Developmental coordination disorder, psychopathology and IQ in 22q11.2 deletion syndrome. *The British Journal of Psychiatry*, (1), doi: [10.1192/bjp.2017.6](https://doi.org/10.1192/bjp.2017.6)

Cunningham AC, Hill L, Mon-Williams M, Peall KJ, Linden DEJ, Hall J, ... van den Bree MBM. (2019). Using kinematic analyses to explore sensorimotor control impairments in children with 22q11.2 deletion syndrome. *Journal of neurodevelopmental disorders*, 11(1), pp. 8. doi: [10.1186/s11689-019-9271-3](https://doi.org/10.1186/s11689-019-9271-3)

Cunningham AC, Fung W, Massey TH, Hall J, Owen MJ, van den Bree MBM, ... Peall KJ. (2020). Movement Disorder Phenotypes in Children With 22q11.2 Deletion Syndrome. *Movement disorders : official journal of the Movement Disorder Society*, 35(7), pp. 1272-1274. doi: [10.1002/mds.28078](https://doi.org/10.1002/mds.28078)

Cunningham AC, Hall J, Owen MJ, van den Bree MBM. (2021). Coordination difficulties, IQ and psychopathology in children with high-risk copy number variants. *Psychological medicine*, 51(2), pp. 290-299. doi: [10.1017/S0033291719003210](https://doi.org/10.1017/S0033291719003210)

Cunningham AC, Hall J, Einfeld S, Owen MJ, IMAGINE-ID consortium , van den Bree MBM. (2022). Assessment of emotions and behaviour by the Developmental Behaviour Checklist in young people with neurodevelopmental CNVs. *Psychological medicine*, 52(3), pp. 574-586. doi: [10.1017/S0033291720002330](https://doi.org/10.1017/S0033291720002330)

Donnelly, N., Cunningham, A., Salas, S. M., Bracher-Smith, M., Chawner, S., Stochl, J., ... & van den Bree, M. B. (2023). Identifying the neurodevelopmental and psychiatric signatures of genomic disorders associated with intellectual disability: a machine learning approach. *Molecular Autism*, 14(1), 19.

Gur, R. C., Bearden, C. E., Jacquemont, S., Swillen, A., van Amelsvoort, T., van den Bree, M., ... & Gur, R. E. (2024). Neurocognitive profiles of 22q11. 2 and 16p11. 2 deletions and duplications. *Molecular Psychiatry*, 1-9.

Hall, J. H., Chawner, S. J., Wolstencroft, J., Skuse, D., Hall, J., Holmans, P., ... & van den Bree, M. B. (2024). Irritability in young people with copy number variants associated with neurodevelopmental disorders (ND-CNVs). *Translational Psychiatry*, 14(1), 259.

Harvey, A., Moreau, C. A., Kumar, K., Huguet, G., Urchs, S. G., Sharmarke, H., ... & Bellec, P. (2024). Challenges in multi-task learning for fMRI-based diagnosis: Benefits for psychiatric conditions and CNVs would likely require thousands of patients. *Imaging Neuroscience*, 2, 1-20.

Lee, I. O., Wolstencroft, J., Housby, H., van den Bree, M. B. M., Chawner, S. J. R. A., Hall, J., ... & Skuse, D. H. (2024). The inequity of education, health and care plan provision for children and young people with intellectual and developmental disabilities. *Journal of Intellectual Disability Research*.

Linden SC, Watson CJ, Smith J, Chawner SJRA, Lancaster TM, Evans F, ... van den Bree MBM. (2021). The psychiatric phenotypes of 1q21 distal deletion and duplication. *Translational psychiatry*, 11(1), pp. 105. doi: [10.1038/s41398-021-01226-9](https://doi.org/10.1038/s41398-021-01226-9)

Linden SC, Watson CJ, Smith J, Chawner SJRA, Lancaster TM, Evans F, ... van den Bree MBM. (2021). Correction: The psychiatric phenotypes of 1q21 distal deletion and duplication. *Translational psychiatry*, 11(1), pp. 372. doi: [10.1038/s41398-021-01296-9](https://doi.org/10.1038/s41398-021-01296-9)

Lynham AJ, Knott S, Underwood JFG, Hubbard L, Agha SS, Bisson JI, ... Walters JTR. (2023). DRAGON-Data: a platform and protocol for integrating genomic and phenotypic data across large psychiatric cohorts. *BJPsych open*, 9(2), pp. e32. doi: [10.1192/bjo.2022.636](https://doi.org/10.1192/bjo.2022.636)

Morrison S, Chawner SJRA, van Amelsvoort TAMJ, Swillen A, Vingerhoets C, Vergaelen E, ... van den Bree MBM. (2020). Cognitive deficits in childhood, adolescence and adulthood in 22q11.2 deletion syndrome and association with psychopathology. *Translational psychiatry*, 10(1), pp. 53. doi: [10.1038/s41398-020-0736-7](https://doi.org/10.1038/s41398-020-0736-7)

Moulding HA, Bartsch U, Hall J, Jones MW, Linden DE, Owen MJ, ... van den Bree MBM. (2020). Sleep problems and associations with psychopathology and cognition in young people with 22q11.2 deletion syndrome (22q11.2DS). *Psychological medicine*, 50(7), pp. 1191-1202. doi: [10.1017/S0033291719001119](https://doi.org/10.1017/S0033291719001119)

Niarchou, M., Cunningham, A. C., Chawner, S. J., Moulding, H., Sopp, M., Hall, J., ... & van den Bree, M. B. (2023). Psychopathology in mothers of children with pathogenic Copy Number Variants. *Journal of Medical Genetics*, 60(7), 706-711.

Niarchou M, Chawner SJRA, Doherty JL, Maillard AM, Jacquemont S, Chung WK, ... Bree MBMVD. (2019). Psychiatric disorders in children with 16p11.2 deletion and duplication. *Translational psychiatry*, 9(1), pp. 8. doi: [10.1038/s41398-018-0339-8](https://doi.org/10.1038/s41398-018-0339-8)

Oliva-Teles N, de Stefano MC, Gallagher L, Rakic S, Jorge P, Cuturilo G, ... Skuse D. (2020). Rare Pathogenic Copy Number Variation in the 16p11.2 (BP4-BP5) Region Associated with Neurodevelopmental and Neuropsychiatric Disorders: A Review of the Literature. *International journal of environmental research and public health*, 17(24), doi: [10.3390/ijerph17249253](https://doi.org/10.3390/ijerph17249253)

Printzlau F, Wolstencroft J, Skuse DH. (2017). Cognitive, behavioral, and neural consequences of sex chromosome aneuploidy. *Journal of neuroscience research*, 95(1-2), pp. 311-319. doi: [10.1002/jnr.23951](https://doi.org/10.1002/jnr.23951)

Rogdaki M, Devroye C, Ciampoli M, Veronese M, Ashok A, McCutcheon R, ... Howes O. (2021). Striatal dopaminergic alterations in individuals with copy number variants at the 22q11.2 genetic locus and their implications for psychosis risk: a [18F]-DOPA PET study. *Molecular Psychiatry*, doi: [10.1038/s41380-021-01108-y](https://doi.org/10.1038/s41380-021-01108-y)

Schaaf CP, Betancur C, Yuen RKC, Parr JR, Skuse DH, Gallagher L, ... Vorstman JAS. (2020). A framework for an evidence-based gene list relevant to autism spectrum disorder. *Nature reviews. Genetics*, 21(6), pp. 367-376. doi: [10.1038/s41576-020-0231-2](https://doi.org/10.1038/s41576-020-0231-2)

Skuse D, Printzlau F, Wolstencroft J. (2018). Sex chromosome aneuploidies. *Handbook of clinical neurology*, 147, pp. 355-376. doi: [10.1016/B978-0-444-63233-3.00024-5](https://doi.org/10.1016/B978-0-444-63233-3.00024-5)

Sønderby IE, van der Meer D, Moreau C, Kaufmann T, Walters GB, Ellegaard M, ... ENIGMA-CNV working group. (2021). 1q21.1 distal copy number variants are associated with cerebral and cognitive alterations in humans. *Translational psychiatry*, 11(1), pp. 182. doi: [10.1038/s41398-021-01213-0](https://doi.org/10.1038/s41398-021-01213-0)

Wolstencroft J, Wicks F, Srinivasan R, Wynn S, Ford T, Baker K, ... Raymond FL. (2022). Neuropsychiatric risk in children with intellectual disability of genetic origin: IMAGINE, a UK national cohort study. *The Lancet. Psychiatry*, 9(9), pp. 715-724. doi: [10.1016/s2215-0366\(22\)00207-3](https://doi.org/10.1016/s2215-0366(22)00207-3)

Wolstencroft J, Srinivasan R, Hall J, van den Bree M, Owen M, , ... Skuse D. (2023). Mental health impact of autism on families of children with intellectual and developmental disabilities of genetic origin. *JCPP Advances*, doi: [10.1002/jcv2.12128](https://doi.org/10.1002/jcv2.12128)