

[Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome.](#)

Bassett AS, Lowther C, Merico D, Costain G, Chow EWC, van Amelsvoort T, McDonald-McGinn D, Gur RE, Swillen A, Van den Bree M, Murphy K, Gothelf D, Bearden CE, Eliez S, Kates W, Philip N, Sashi V, Campbell L, Vorstman J, Cubells J, Repetto GM, Simon T, Boot E, Heung T, Evers R, Vingerhoets C, van Duin E, Zackai E, Vergaelen E, Devriendt K, Vermeesch JR, Owen M, Murphy C, Michaelovsky E, Kushan L, Schneider M, Fremont W, Busa T, Hooper S, McCabe K, Duijff S, Isaev K, Pellecchia G, Wei J, Gazzellone MJ, Scherer SW, Emanuel BS, Guo T, Morrow BE, Marshall CR; International 22q11.2DS Brain and Behavior Consortium.

Am J Psychiatry. 174(11):1054-1063, 2017.

doi: 10.1176/appi.ajp.2017.16121417.

The authors found that rare copy number variants (CNVs) that overlap protein-coding sequences can contribute to an increased risk of schizophrenia in individuals with 22q11.2 deletions. These CNVs include both rare duplications that overlap genes involved in neuronal function, as well as rare deletions that overlap more genes in individuals who develop schizophrenia.