

Publication

The genetic architecture of human brainstem structures and their involvement in common brain disorders

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Brainstem regions support vital bodily functions, yet their genetic architectures and involvement in common brain disorders remain understudied. Here, using imaging-genetics data from a discovery sample of 27,034 individuals, we identify 45 brainstem-associated genetic loci, including the first linked to midbrain, pons, and medulla oblongata volumes, and map them to 305 genes. In a replication sample of 7432 participants most of the loci show the same effect direction and are significant at a nominal threshold. We detect genetic overlap between brainstem volumes and eight psychiatric and neurological disorders. In additional clinical data from 5062 individuals with common brain disorders and 11,257 healthy controls, we observe differential volume alterations in schizophrenia, bipolar disorder, multiple sclerosis, mild cognitive impairment, dementia, and Parkinson's disease, supporting the relevance of brainstem regions and their genetic architectures in common brain disorders.

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