AI discovers new autism-linked mutations in 'junk' DNA

Many mutations in DNA that contribute to disease are not in actual genes but instead lie in the 99% of the genome once considered "junk." Even though scientists have recently come to understand that these vast stretches of DNA do in fact play critical roles, deciphering these effects on a wide scale has been impossible until now.

Using artificial intelligence, a Princeton University-led team has decoded the functional impact of such mutations in people with autism. The researchers believe this powerful method is generally applicable to discovering such genetic contributions to any disease.

[Publishing May 27](#) in the journal Nature Genetics, the researchers analyzed the genomes of 1,790 families in which one child has autism spectrum disorder but other members do not. The method sorted among 120,000 mutations to find those that affect the behavior of genes in people with autism. Although the results do not reveal exact causes of cases of autism, they reveal thousands of possible contributors.

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When the researchers then looked at what genes were affected by these mutations, they turned out to be genes strongly associated with brain functions. These newly discovered mutations affected similar genes and functions as do previously identified mutations.

Read full, original post: [Artificial intelligence detects a new class of mutations behind autism](#)