LEGACY SPECIAL REPORTS

The genetics of autism

BY **SPECTRUM**

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Autism is a complex condition, with a wide range of features that vary in their severity. Autism's **genetic roots** are equally complex. The condition runs in families, but most of the relevant mutations identified so far arose spontaneously in the sperm or egg or after fertilization.

In the past 10 years, scientists have identified some 65 genes tied to autism, and **the list continues to grow**. Many of these genes play key roles in the brain. Some scientists say the time has come to stop searching for **autism genes** and focus instead on **understanding their function** .

Some research teams are expanding their search to **mitochondrial DNA**, which is inherited from mothers, and to the 'dark genome' — the 98 percent that **doesn't contain genes**. Elements **buried within the dark genome** regulate the expression of genes. **Environmental factors and experiences** can also alter gene expression by changing the patterns of chemical tags on DNA.

Genetic variants that are common in the general population may work together in groups to increase the risk of autism. To find these variants, researchers must gather DNA from **tens of thousands of people with autism** — a feat made possible by the **participation of families** in autism studies and groups. Advocacy groups that **unite people with the same rare mutations** are helping to reveal how autism's genetic heterogeneity gives rise to its wide spectrum of features.

You can hear more about gene discovery in autism in our **Spectrum Stories podcast**. Articles from our archives round out this special report on the genetics of autism.