**WHAT IS 16p11.2?**
A 16p11.2 copy number variant is a small missing or duplicated piece of DNA on chromosome 16. It comes in four forms, a proximal or distal deletion or duplication, all affecting genes that control how brain cells connect and communicate. The result can touch development, learning, speech, movement, behavior, and physical health at once, differently in every child.
*It is one of the most common single genetic causes of neurodevelopmental disability, yet most physicians have never been trained to recognize or manage it.*

## 16p11.2 Facts & Figures

| | |
|---|---|
| **1 in 600**<br>combinedestimated prevalence of all four 16p11.2 copy number variants | **560,000+**<br>Americansareestimated to be living with one of the four variants today |
| **~6,000**<br>children are born in the U.S. each year with a 16p11.2 variant, about 16 a day | **0**<br>the number of federally recognized standards of care that currently exist for it |
| **250+**<br>individual clinicians a single affected child may see over a lifetime, with no coordinated team | **1,000+**<br>caregivers connected through our Foundation's community, across all 50 states and more than 25 countries |

### The Four Variants

| | |
|---|---|
| **~1 in 2,000**<br>Proximaldeletion(BP4-BP5) | **~1 in 2,500**<br>Proximalduplication (BP4-BP5) |
| **~1 in 4,100**<br>Distaldeletion(BP2-BP3) | **~1 in 1,500**<br>Distalduplication(BP2-BP3) |

## What It Can Look Like

Documented effects include developmental delay, autism, ADHD, speech and motor planning difficulties, seizures, weight issues, sleep disruption, anxiety, scoliosis, precocious puberty, and mental health disorders including schizophrenia. No two children present the same way, and most families see many specialists before anyone connects the cause.

## Who We Are

The 16p Genetic Syndrome Foundation is a parent-led 501(c)(3) closing the gap between what science knows about 16p11.2 and the families and clinicians who don't yet have access to it, through published clinical guidelines, a growing caregiver community, and an emerging coordinated care and research program.

16p11.2 GENETIC FOUNDATION