FOR GENETIC COUNSELORS, PHYSICIANS & CARE TEAMS

16p11.2 GENETIC FOUNDATION

No 16p11.2 family should walk alone.

We are a parent-led nonprofit supporting individuals and families with **16p11.2 deletions and duplications**, at every breakpoint. When you diagnose a 16p11.2 CNV, please pass this along.



~3 in 10,000
births with a 16p11.2 deletion

3–5 in 10,000
people with a 16p11.2 duplication

Highly variable
features differ, even within families

We speak in breakpoints (BP)

Chromosome 16p11.2 · schematic, not to scale



16p11.2 BP4–BP5
~600 kb · ~25–30 genes. The most common 16p11.2 CNV.

16p11.2 BP2–BP3
~220 kb · ~9 genes, including *SH2B1*.

Larger & atypical
e.g. BP1–BP5 or BP2–BP5. Every 16p11.2 family is welcome.

For clinicians

Care guidelines. Primary-care guidance for 16p11.2 deletion and clinical resource summaries.

Research. We connect families to Simons Searchlight and active clinical studies.

Education. Quarterly 16p11.2 seminars with the UW and University of Iowa IDDRCs.

Refer at diagnosis. Share this sheet or our website when you report a 16p11.2 CNV.

For families

Newly diagnosed? Plain-language guides and a family roadmap for what comes next.

Community. An online parent group, family conferences and expert webinars.

Every age. The Curious Kid book series and an occupational therapy handbook.

Advocacy. Pushing for a designated ICD code and coordinated centers of care.

SCAN OR VISIT

16pfoundation.org

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November 16 is 16p11.2 Genetic Variants Day

16p11.2 Genetic Foundation is a 501(c)(3) nonprofit · Tax ID 88-3066433