Project 8p Foundation, *of the* Commission on Novel Technologies for Neurodevelopmental Copy Number Variants

Project 8p Foundation
project8p.org

Ring14 USA
ring14usa.com

Dup15q Alliance
dup15q.org

To empower a unified community for chromosome 8p heroes for a meaningful life today while accelerating treatments for tomorrow.

Chromosome 8p disorders affect approximately 550 patients around the world, with most cases de novo. Symptoms are wide-ranging and vary in severity, and include intellectual disability, congenital heart defects, epilepsy, autism, GI dysfunction, orthopedic and muscular conditions, agenesis of corpus callosum, and sensory processing disorders. There are no current treatments.

The leaders of the Commission for Neurodevelopmental Copy Number Variants (CNVs), left to right: Yssa DeWoody, Bina Maniar, Vanessa Vogel-Farley -attend the Chan Zuckerberg Initiative Rare As One Network 2022 Meeting.

During the Grant Period

Impact Spotlight
When Project 8p Foundation Founder Bina Maniar considered applying for Rare as One Cycle 1, she saw the opportunity as a chance to make progress not just for those with chromosome 8p disorders, but for the broader neurodevelopmental copy number variants (CNVs) community, and ultimately, beyond.

Maniar joined forces with Yssa DeWoody of Ring14 USA, and Vanessa Vogel-Farley of the Dup15q Alliance, and approached CZI's grant to the Project 8p Foundation with a shared question and goal: could first-of-its-kind collaboration drive progress for these complex disorders?

Toward generating proof positive, the three leaders launched the Commission for Neurodevelopmental Copy Number Variants (CNVs), with the hypothesis that, though their respective brain development diseases affected different chromosomes, collaborative study would reveal converging biology, and in turn, opportunities for common research progress toward treatments and cures.

By the end of the grant, the Commission's work had begun to bear fruit. What would have been disparate, disease-specific data streams have been consolidated into one centralized repository, a wide-ranging resource that has attracted interest from industry and other disease efforts, alike, and all with patient interest and need at the center.

Key research and research infrastructure achievements

- Built the collaborative Copy Number Variants (CNVs) Data Portal, a workspace where approved investigators can access biospecimens, clinician- and patient-reported data and sequenced genomes.
- Launched a collaborative partnership with Global Genes/RARE-X to host the CNVs Data Portal for longitudinal data collection and support regulatory and operational needs.
- Advanced open science partnership with Illumina to provide long read sequencing, RNA seq, DNA methylation and analysis to support molecular characterization of variants and the identification of potential therapeutic targets associated with neurodevelopmental chromosome disorders.

Key publications

American Journal of Human Genetics (2022): a roadmap to improving outcomes for patients with neurodevelopmental copy-number variants (authored by the Commission on Novel Technologies for Neurodevelopmental Copy Number Variants).

Key operational achievements

- Developed a fundraising model for support of the Commission for Neurodevelopmental CNVs.
- Executed a Charter among all Commission members, aimed at supporting a Team Science approach for effective collaboration.

Key community achievements

Hosted two conferences: a virtual conference and a joint in-person Family & Science conference, bringing together Project 8p, Ring14 USA and Dup15q Alliance families and stakeholders.

After the Grant

The Commission and its leaders are working to identify additional research opportunities and approaches, all in the close patient-centered partnership that has been so central to their progress thus far. The Commission will be co-hosting the "Uniting Chromosomal Disorders for Translational Breakthroughs in Rare Disease THINK TANK" in 2025 aimed at advancing its foundational goals.

In summer 2021, Project 8p, Ring14 USA, and Dup15q Alliance hosted the Moving Mountains joint conference, the first time the three nonprofits collaborated to reflect similar clinical phenotypes and goals. The CNV Commission brought together scientists actively co-authoring a roadmap published in the American Journal of Human Genetics*, doctors, patients, and their families to mix and mingle, share sorrows, and collect data for the database.*

"When you truly collaborate in an open science format and leveraging team science, it's powerful for innovative breakthroughs."

Bina Maniar, MBA
Founder and CEO, Project 8p Foundation