WHO WE ARE: The **CSNK2A1 Foundation is a U.S.-based nonprofit advancing global research and accelerating treatment development for Okur-Chung Neurodevelopmental Syndrome (OCNDS)**, an **ultra-rare genetic condition** first identified in **2016** by Dr. Wendy Chung and Dr. Volkan Okur. At the time of discovery, only five individuals worldwide had been diagnosed and there was no coordinated research effort or patient support infrastructure. Since then, the Foundation has built a global research ecosystem spanning 17 institutions, bringing scientists together to collaborate, share data, publish jointly, and convene quarterly through its Scientific Roundtable. What once had no research pipeline now includes active therapeutic development efforts with the potential to directly address the underlying biological effects of CSNK2A1 gene mutations.

ABOUT OCNDS: OCNDS is a **lifelong neurodevelopmental condition** caused by mutations in the CSNK2A1 gene. Individuals may experience **severe speech delay or inability to speak, global developmental delay, epilepsy, autism-related traits, behavioral challenges, feeding difficulties, and hypotonia**. Symptoms vary widely in severity, but families face daily challenges with communication, mobility, toileting, feeding, education access, and social inclusion. Care typically requires multidisciplinary support including genetic counseling, early intervention services, individualized education plans, speech therapy, occupational therapy, feeding therapy, physical therapy, and developmental pediatric care. Parents and caregivers often become full-time care coordinators, advocates, and case managers navigating fragmented systems of care.

THE DIAGNOSTIC AND CARE GAP: 51 individuals with OCNDS have been reported in the literature to date. Through the CSNK2A1 Foundation's contact registry over 385 cases have been identified in over 40 countries, ranging in age from 11 months old to 62 years old. The **prevalence is estimated as 1:100,000,** which means since its discovery, **over 14,000 children have been born with OCNDS**. **Yet, roughly 385 people have been diagnosed worldwide**. Limited access to genetic testing, shortages of rare disease specialists, insurance coverage barriers, and geographic disparities continue to delay diagnosis and restrict access to appropriate care and treatment pathways.

After consultation with a clinical geneticist and or genetic counselor, the following management recommendations may be appropriate: referral for early intervention programs or evaluation for an individualized education plan, physical therapy for mobility, occupational therapy for fine motor skills, feeding therapy, alternative means of communication for individuals with expressive language difficulties, therapies for social or behavioral concerns, and continued consultation with a developmental pediatrician.

CSNK2A1 Foundation joins rare disease advocates through events such as Rare Disease Week on Capitol Hill to advocate for legislation that would benefit the entire rare disease community. We have joined forces with the Epilepsy Foundation, Genetic Alliance, the Rare Epilepsy Network, the Haystack Project, and the Rare & Ready Coalition to support said change.

In 2026, CSNK2A1 Foundation asks for your support for the following:

- **Improve patient access to rare disease diagnosis, care, and treatment by expanding telehealth flexibilities** and creating exceptions for out-of-state physicians from in-state licensure requirements for remote care.
- **Access to Genetic Counselor Services Act** (S.2323 / H.R. 6280). This bipartisan legislation was reintroduced to expand Medicare coverage for genetic counselors as billable providers and improve patient access to genetic services.
- **PROTECT Rare Act** Introduced in the Senate (S.3551) to expand coverage for off-label rare disease treatments by allowing Medicare, Medicaid, and private insurers to rely on clinical evidence and expert guidance.
- **The Genomic Answers for Children's Health Act** (H.R. 7118): Bipartisan legislation to improve the diagnosis & treatment of kids on Medicaid.
- **Credit for Caring Act** (S. 925/H.R. 2036) Support this Act to help working family caregivers offset the cost of some caregiving expenses.
- Join the **Rare Disease Congressional Caucus**.

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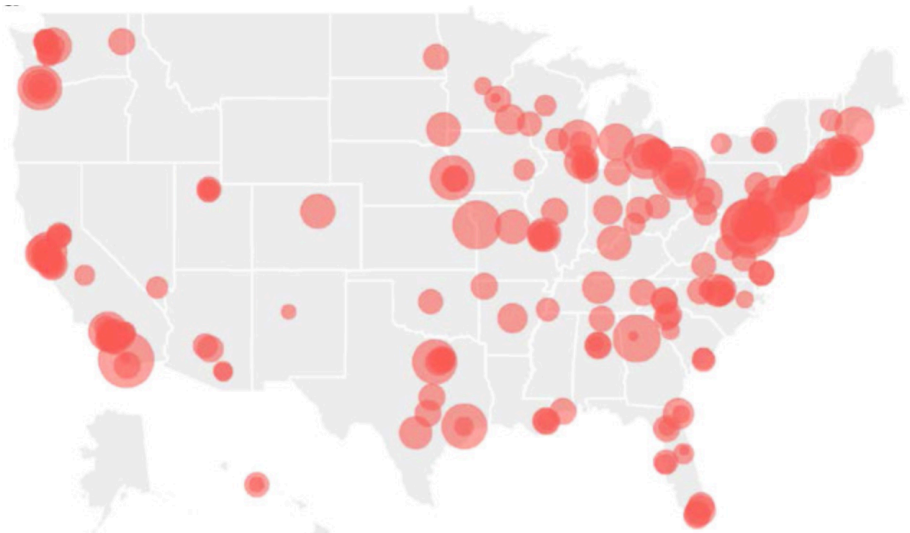
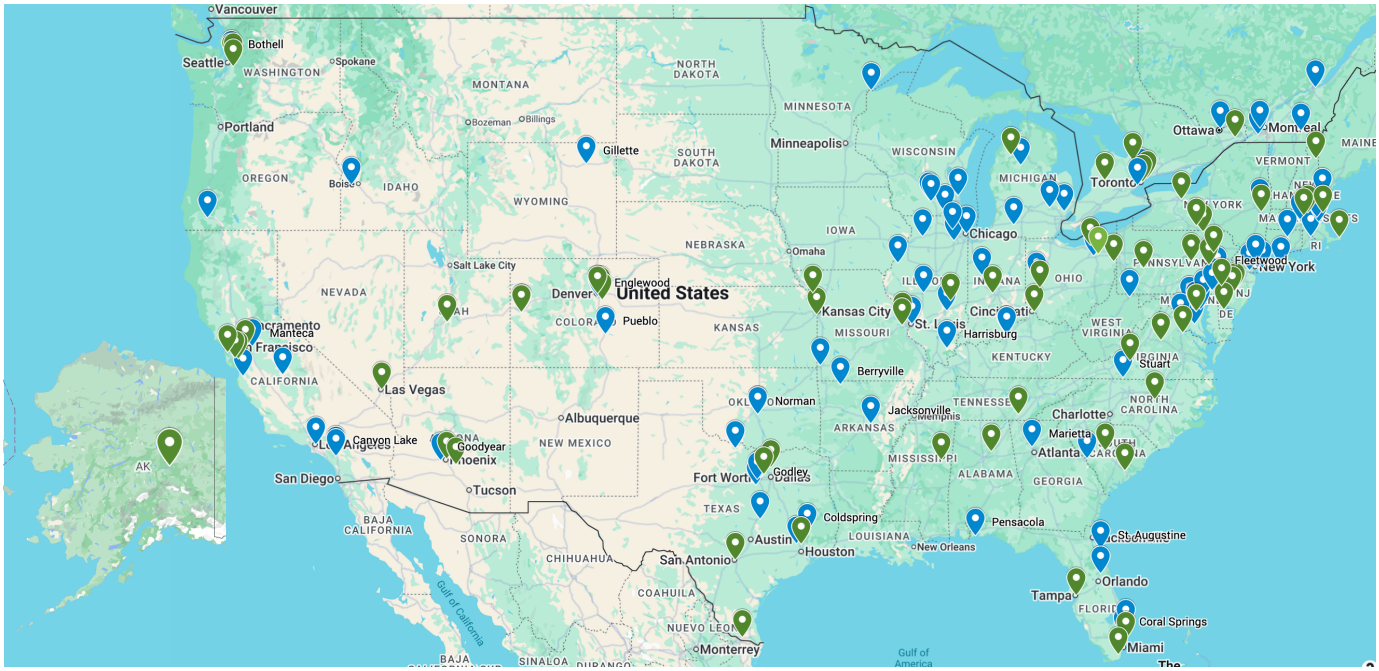
Geographic distribution of clinical geneticist respondents across the United States.



From Genet Med. 2021; 23(8): 1458–1464. Published online 2021 May 3. doi: 10.1038/s41436-021-01162-5

Geographic distribution of OCNDS diagnoses across the United States.



*2024 anecdotal data collected via the Foundation's contact registry