

CNKSR2 Natural History Study

NCT06500260

Status	RECRUITING
Sponsor	University of California, San Francisco
Enrollment	15 participants

Key Eligibility Criteria

Inclusion (3)

- Age between 6 and 21 years (inclusive) at time of consent.
- Confirmed CNKSR2 mutation, as demonstrated by genetic testing and confirmed by the investigators.
- Confirmed intellectual disability or developmental delays, as defined by the American Academy of Pediatrics (Moeschler, J, et al. 2014).

Exclusion (3)

- Known pathogenic or clinically suspected mutation in a seizure-associated gene besides CNKSR2.
- Confirmed mutation in a gene besides CNKSR2 that is known to increase the severity of the seizure phenotype.
- Known central nervous system structural abnormality confirmed by imaging scan of the brain that is not consistent with the clinical phenotype of CNKSR2 EAS / ID.

Locations (1 total)

University of California, San Francisco (UCSF), San Francisco, California, United States