Longitudinal Study of Phenotypic and Developmental Severity in Patients With Dravet Syndrome With SCN1A Gene Mutation

NCT07251673

Status	RECRUITING
Sponsor	Assistance Publique - Hôpitaux de Paris
Enrollment	50 participants

Key Eligibility Criteria

Inclusion (7)

- The patient or his/her legal representative must be able to give informed consent for participation in the study.
- The participant or legal representative are able (in the opinion of the investigator) to comply with the research protocol.
- Patient (male/female) between 6 months and 21 years of age inclusive at the time of consent.
- The patient has a confirmed pathogenic or probably pathogenic variant of the SCN1A gene demonstrated by a genetic test.
- The patient had normal development prior to the onset of the first seizure.

... and 2 more (see full listing online)

Exclusion (10)

- The patient has a copy number variation of the SCN1A gene affecting other genes, including a microdeletion of SCN1A.
- The patient has a mutation in the SCN1A gene on both alleles.
- The patient has a known or clinically suspected pathogenic mutation in a gene associated with epilepsy other than the SCN1A gene.
- The patient has a concomitant genetic mutation or clinical comorbidity deemed likely to disrupt the typical phenotype of Dravet syndrome.
- The patient has a known gain-of-function mutation, defined by functional studies, including p.Thr226Met.

... and 5 more (see full listing online)

Locations (1 total)

Robert Debré Hospital, Paris, Ap-hp / DRCI, France