

Registry and Natural History of Epilepsy-Dyskinesia Syndromes

NCT06967727

Status RECRUITING
Sponsor Boston Children's Hospital
Enrollment 700 participants

Key Eligibility Criteria

Inclusion (2)

- Having at least one pathogenic or likely pathogenic variant in one of the genes of interest:
- AARS2, ADCY5, ALG13, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, ARX, ATP1A3, CACNA1A, CACNA1E, CACNA2D2, CDKL5, CSTB, DARS2, DLAT, DLD, DNM1, EARS2, EPG5, EPM2A, FARS2, FOXP1, FRRS1L, GABRA1, GABRA2, GABRB2, GABRB3, GABRG2, GNAO1, GRIA2, GRIA4, GRIN1, GRIN2A, GRIN2B, GRIN2D, HARS2, HNRNPU, HTT, IQSEC2, IRF2BPL, KCNA2, KCNB1, KCNC1, KCNMA1, KCNQ2, KCNQ3, KCNT1, LARS2, MECP2, MEF2C, MTND5, MTTK, MTTL1, NARS2, NHLRC1, PCDH12, PCDH19, PDE10A, PDE2, PDHA1, PDHB, PDHX, PDK3, PDP1, PIGA, PIGN, PIGP, PIGQ, PIGS, PLCB1, POLG, PRRT2, PURA, RHOBTB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SETBP1, SETD5, SLC13A5, SLC1A2, SLC25A22, SLC2A1, SMC1A, SNX14, SPTAN1, ST3GAL3, STXBP1, SYNGAP1, SYNJ1, SZT2, TARS2, TBC1D24, UBA5, UBE3A, VAMP2, VARS2, WARS2, WDR45, WWOX, YIF1B, YWHAG, and other genes associated with epilepsy-dyskinesia syndromes.

Exclusion (1)

- Not having a pathogenic or likely pathogenic variants in the genes of interest

Locations (1 total)

Boston Children's Hospital, Boston, Massachusetts, United States