CASE STUDY
NATIONAL INSTITUTE OF HEALTH

Empire Genomics Contributes to First Overnight Video-EEG Evaluation in PMS

Background

Phelan-McDermid Syndrome (PMS) is a rare genetic condition associated with loss of function mutations in the chromosome 22q13 region. This region contains many genes, but the majority of cases involve SHANK3, a gene reportedly associated with seizures. PMS typically presents as intellectual disability and other neurologic deficits. Seizures and abnormal electroencephalograms (EEGs) have also been reported in PMS patients. However, no studies to date have described EEG findings during sleep or the value of abnormal EEG over a longitudinal time period.

Objectives

Prolonged overnight video-EEG recordings with sleep have not been reported in the PMS patient population, and the natural evolution of abnormal EEGs over time has not yet been established. This study consisted of the first and most abundant prolonged awake and sleep EEG data recorded in a PMS cohort, as well as the first prospective longitudinal follow-up on the development of epileptiform abnormalities.

Approach

Sixteen patients with PMS were enrolled as participants and underwent routine and overnight video-EEG, as well as genetic testing, neurodevelopmental assessment, neurological examination, and epilepsy phenotyping. Findings from the routine EEF and the prolonged EEG were compared, and the overnight EEG was repeated in nine of the sixteen participants one or more years later. 22q13 deletion size was confirmed through array Comparative Genomic Hybridization (aCGH) and fluorescence in situ hybridization (FISH), which used a probe specific to the subtelomeric region of chromosome 22q as well as a control probe that hybridizes to the 22q11.21 region, manufactured by Empire Genomics.

Results

All but one of sixteen EEGs were abnormal, and 75% showed epileptiform activity. Of these, only 25% showed definitive epileptiform discharges in the routine study. Despite not elucidating any clinical events consistent with seizures, the overnight EEGs did show more frequent and/or more definitive epileptiform activity in 11 of 16 participants. Follow-up EEG confirmed persistence of these abnormalities, but there was no evolution to electrographic seizures or emergence of epilepsy. This study, utilizing varied techniques and tools including those manufactured by Empire Genomics, reiterate the clinical importance of overnight prolonged EEG studies with sleep in people with PMS.

Prospective longitudinal overnight video-EEG evaluation in Phelan-McDermid Syndrome

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Lead Organization

National Institute of Health

Diseases

- Phelan-McDermid Syndrome

Biomarkers Mentioned

- SHANK3

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