Adverse Events in Episodic Ataxia Natural History Study

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Study Update

Consortium for Investigation of Neurological Channelopathies
  ❖ Episodic Ataxia (EA)
  ❖ Non-dystrophic Myotonias (NDM)
  ❖ Andersen-Tawil Syndrome (ATS)

Study design
  ❖ Natural History
  ❖ Yearly neurological exams
  ❖ Genetic testing
  ❖ EA--EEGs
Study sites

- UCLA--EA
- University of Kansas--NDM
- University of Rochester--ATS
- UCSF
- University of Texas, Southwestern
- Brigham and Women’s Hospital
- London Health Sciences, London, Ontario
- National Hospital for Neurology and Neurosurgery, London, UK
- National Institutes of Health
Episodic Ataxia

- **EA-1-mutations in KCNA1**
  - Age of onset: childhood
  - Interictal myokymia
  - Attacks: seconds to minutes

- **EA-2-mutations in CACNA1A**
  - Age of onset: childhood or teens
  - Interictal nystagmus
  - Attacks: minutes to hours, sometime days

- Hallmark of all EAs: attacks triggered by stress, alcohol, caffeine
Participants to date

- EA--41
- NDM--56
- ATS--0
- UCLA enrolled 10 EA participants
  - 5 with EA-2
  - 5 with EA-other (non EA1 or EA2)
Case study

- Patient MD, 52 year old RH man
- Episodic ataxia since age 14
- Progressive ataxia
- Family history of episodic ataxia
- Mutation in CACNA1A causing EA2
Event

Participant, sister and nephew enrolled in 11-21-06
Drove down from San Jose
Went to Disneyland day before appointment
Arrived for 8:30am EEG
Movie
Adverse events

- 1 person had attack triggered before enrollment
- 2 people had attacks triggered after enrollment
- 1 person had attack triggered during EEG photic stimulation
Amendments

- Allow investigators to travel to participants’ homes for enrollment
- Shorter visits—finish enrollments over the phone
- Encourage participants to plan rest periods before and after visit. Avoid excessive activity.