

5301: Andersen-Tawil Syndrome: Genotype-Phenotype Correlation and Longitudinal Study

Status: Three centers currently recruiting (University of Rochester; London Health Sciences Center, London, Ontario; Institute of Neurology, London, United Kingdom)

For: Andersen-Tawil Syndrome

Study Summary

Please note: The Rare Diseases Clinical Research Network will make every effort to enroll all the patients we can, but we cannot make any guarantees that we will be able to enroll everyone in a particular study who wants to participate.

Background

Andersen-Tawil Syndrome (ATS) is a rare, genetically-caused disease that causes episodes of weakness and potentially life-threatening heart arrhythmias. The purpose of this multi-center study is to collect data from volunteers with the disease in order to learn more about ATS, to find whether symptoms change over time, and to discover whether symptoms are related to the genetic mutation present in each individual.

About this Study

This will be an observational, three year, prospective study in 50 individuals with ATS. Seven centers in the United States, Canada and England will enroll participants. Those participating will be evaluated yearly for 3 consecutive years (total of 3 study visits). The initial visit will require a hospital admission of 1.5 – 3.5 days and the final two evaluations will be done on an outpatient basis. Participants will be evaluated by the following:

- history and physical exam
- blood tests
- muscle strength testing
- heart function testing (EKG, continuous heart monitoring)
- nerve conduction testing
- questionnaire to measure health status
- daily recording of episodes of weakness

Eligibility requirements:

To be eligible to participate:

- You are 10 years of age or older
- You have signed and dated the informed consent
- You have a confirmed diagnosis of ATS that includes:
 - Having the KCNJ2 gene mutation **OR**

- Having **2** of the following **3 features**:
 - (1) Clear-cut episodes of periodic muscle weakness **OR**
Unexplained hypokalemia between episodes of weakness **OR**
Abnormal long-exercise nerve conduction study
 - (2) Heart conduction defects
 - (3) Presence of 2 or more characteristic physical features (low set ears, eyes set wide apart, small lower jaw, inner curving of the 5th finger, webbed or conjoined fingers/toes, small hands/feet)
- OR**
- Having 1 of the above 3 features AND an affected family member meeting 2 criteria

You are not eligible to participate if:

- You are under 10 years of age
- You are unable to stay in the hospital for 1.5 – 3.5 days and attend the other 2 scheduled clinic visits
- You are unable to give informed consent
- You are unwilling to temporarily (during the hospital admission) discontinue any medicines you are taking for weakness – You will **NOT** be asked to stop any heart medication

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