

SMA Research Update – Providing Hope!

4-Aminopyridine (4-AP) in SMA: A Pilot Study in Ambulatory Patients

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Fatigue is a common symptom in SMA with a resultant impact on physical function and quality of life; the precise mechanisms of fatigue in SMA are yet not understood. Currently, there is no treatment for SMA. There is evidence that 4-AP improves function in SMA animal models. Furthermore, in patients with multiple sclerosis, 4-AP was found to improve walking ability and diminish fatigue. This study is being conducted to see if 4-AP improves walking ability and endurance in adult patients with SMA Type 3. The purpose of the study is to determine whether treatment with 4-AP is associated with an increase in walking speed and endurance compared to placebo and whether the duration of treatment affects outcome.

The Columbia University SMA Clinical Research Center is currently recruiting SMA Type 3 individuals who are 18 years or older, have genetically confirmed SMA, and are able to walk without assistance for at least 25 meters.

The study will last for 21 weeks, and will require that subjects come for visits at the Columbia SMA Clinical Research Center 6 times. Each visit will consist of a series of tests that include motor function measures, a physical exam, questionnaires, and a test where the subject is asked to walk as far as they can in six minutes. Additionally at two visits needle EMGs will be performed. The main goal of the study is to see if individuals who take 4-AP (Ampyra) have improved walking speed and endurance.

To be considered for eligibility in the study, potential participants must:

- be 18 years of age or older
- have genetically confirmed SMA 3 (homozygous absence of SMN1 exon 7)
- be free of major orthopedic deformities (i.e. scoliosis, contractures)
- normal Cystatin C clearance (> 80 ml/min)
- live within 75 miles of study site or deemed by staff able to reliably travel to CUMC for study visits
- be able to provide informed consent
- not have a history of seizures
- meet additional study-specific criteria

If you are interested in participating, please contact Jonathan Marra at jdm2132@columbia.edu or 212-342-3022 for more information.