

Genetic Study

Study objective:

The underlying cause of motor stereotypies is unknown, however, a positive family history is identified in 20% of patients. In this study we use whole exome testing, on DNA isolated from saliva, to identify possible genetic abnormalities.

This is a joint effort with Dr. Thomas Fernandez, a geneticist at Yale University.

Participant eligibility:

Participants can be any age. We are currently recruiting families that contain more than one individual with primary complex motor stereotypies.

Requirements:

1. **Introductory phone screening** – Screening is completed by the Study Coordinator to confirm eligibility
2. **Online forms** – Several online questionnaires must be completed before saliva kits are provided
3. **Saliva test** – Samples are collected from both parents and the affected child. Sample collection involves spitting into a provided cup and mailing to Dr. Fernandez (postage is provided)