



Ethylin Jabs, MD. Professor and Vice Chair of Genetic and Genomic Sciences, Professor of Pediatrics and Developmental and Regenerative Biology, Icahn School of Medicine at Mount Sinai

Elizabeth C. Engle MD. Professor of Neurology and Ophthalmology, Boston Children's Hospital & Harvard Medical School. Investigator, Howard Hughes Medical Institute.

Eirini Manoli MD, PhD. Staff Clinician, National Human Genome Research Institute, National Institutes of Health

To the Moebius Syndrome Foundation:

We are pleased to announce a new collaborative effort amongst a team of researchers at Mount Sinai School of Medicine, Boston Children's Hospital and the National Institutes of Health, that aims to characterize the clinical phenotype of children and adults with Moebius syndrome and associated congenital facial weakness disorders and use genetic studies to identify disease-causing genes.

Participants are eligible if between 2 and 80 years old and have an established diagnosis of Moebius syndrome or other disorders associated with congenital facial weakness, based on clinical history and findings. Investigators will apply rigorous selection criteria to choose about 30 families per year for an evaluation at the National Institutes of Health Clinical Center in Bethesda, Maryland. During a three- to five-day outpatient stay, participants will undergo a physical examination, meet many specialties, including geneticists, ophthalmologists, neurologists and rehabilitation medicine doctors, and have a blood sample drawn to extract DNA (genetic material).

The NIH research team will use advanced technologies for brain imaging and to test the patient's DNA for possible genetic causes of these disorders. It is important to also obtain DNA from both the patient's parents, but the parents would not necessarily have to travel to NIH in person.

Combining our unique expertise and resources will maximize our ability to make new discoveries that will help understand what causes Moebius syndrome and provide more accurate diagnosis and genetic counseling for individuals with facial weakness and their families.

For more information regarding the study, please respond with your contact information to the phone numbers/email address below:

Carol Van Ryzin 301-827-1071

Eirini Manoli 301-402-2543

Email: moebius@mail.nih.gov