ENGLE LABORATORY GENETIC RESEARCH ON FACIAL NERVE PALSY & MOEBIUS SYNDROME

The Engle Laboratory searches for and studies genes important in the development and function of the nerves (cranial nerves) and muscles (extraocular and facial muscles) important in eye and facial movements. We enroll individuals and families impacted by associated conditions.

CONDITIONS WE STUDY

Conditions linked to the cranial nerves and associated muscles not working properly are called Congenital Cranial Dysinnervation Disorders (CCDDs). We study many conditions, but several current projects are focused on individuals born with **facial movement problems** and **isolated congenital facial palsy**. These diagnoses may be referred to by several names, including:

- **Congenital Facial Palsy (CFP)**
- **Congenital Facial Weakness**
- **Facial Nerve Palsy**
- **7th Cranial Nerve Palsy**
- **Moebius Syndrome**

GENES IDENTIFIED

We have collaborated with clinicians and other researchers to identify multiple CCDD genes. The genes linked to some conditions which include facial palsy have been found, such as TUBB3 and HOXA1 and HOXB1. We continue searching for more genes, including those linked to isolated congenital facial palsy.

WHO CAN PARTICIPATE & WHAT IS INVOLVED?

People of any age with facial palsy and their family members are invited to contact us about enrollment. There is no cost, travel is not necessary & all steps can be arranged remotely. To make sure our study is appropriate, we review available medical records and family history. Copies of previously arranged head MRI images can be very helpful and we may ask for facial movement photos and videos. We collect DNA samples from individuals who have been diagnosed, as well as parents and siblings when possible, usually by saliva sample or cheek swabs. Reviewing with one of our staff and signing a research consent form is required.

EXPECTATIONS AND RESULTS

Major results of our work are made available through scientific publications. Research can be a lengthy process & it may be a very long time before there are any results for specific individuals. It is also possible that we will be unable to identify the gene(s) causing some conditions and no results will be available. US regulations prevent us from directly returning to participants the specific results from our work. However, if there are research findings that may be of possible medical significance, we attempt to contact participating families and/or their physicians to let them know such findings exist. Based on choices participants make when they sign the consent form, re-sampling for confirmatory diagnostic testing in a non-research laboratory and follow-up counseling may be advised. Although the clinical validation testing is not done by or paid for by the Engle Lab, we help guide families and doctors through this process so information can be included in a participant's medical record.

For more information about our research or to coordinate screening & enrollment, please contact Brenda Barry, Genetic Counselor, at **englegc.research@childrens.harvard.edu** or **617-919-2168**.
You may also visit our website: **http://www.childrenshospital.org/research/labs/engle-laboratory**