ENGLE LABORATORY GENETIC RESEARCH ON CRANIAL NERVE FUNCTION

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

CONDITIONS WE STUDY

Conditions linked to the cranial nerves and associated ocular & facial muscles not working properly are called Congenital Cranial Dysinnervation Disorders (CCDDs). Symptoms exist since birth, though may be diagnosed later. Specific CCDDs include:

- **Congenital Fibrosis of the Extraocular Muscles (CFEOM)**
- **Marcus Gunn Jaw Winking Syndrome**
- **Facial Palsy/Weakness**
- **Moebius syndrome**
- **4th Nerve Palsy**
- **Duane syndrome**
- **Brown syndrome**
- **Ptosis**

Some conditions we study and enroll also affect other parts of the body, such as **Horizontal Gaze Palsy with Progressive Scoliosis (HGPPS)** and **Duane Radial Ray Syndrome (DRRS).**

GENES IDENTIFIED

We have collaborated with clinicians and other researchers around the world to identify many genes linked to CCDDs. These include *KIF21A, TUBB3, CHN2, SALL4, ROBO3, MAFB* and several others-though there are still more to find! Our investigations into known genes and our ongoing search for additional CCDD genes helps us to learn more about the conditions we study, and hopefully may guide potential treatments and therapies in the future.

WHO CAN PARTICIPATE & WHAT IS INVOLVED?

People of any age 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 medical and family histories & available ophthalmology, neurology, genetics and surgical records. If head or orbital imaging has been performed, copies of those MRI images can be very helpful and we may request face and eye movement photos. 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**
You may inquire about our studies at Boston Children's:
http://www.childrenshospital.org/clinical-trials/nct03059420