



CCDD Family Information Sheet
For genetic studies of the congenital cranial dysinnervation disorders:
Congenital fibrosis of the extraocular muscles (CFEOM)
Congenital ptosis
Duane syndrome
Related ocular motility disorders

The Engle Laboratory

Division of Medicine, Program in Genomics, Enders 5, Children's Hospital Boston
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The Engle Laboratory at Children's Hospital Boston investigates the genetic basis of congenital eye movement disorders.

How is the research done?

DNA is extracted from blood samples, saliva or buccal swabs collected from participating family members. If more than one family member is affected by the eye movement disorder, we may use a technique called "linkage analysis" to identify regions of DNA shared by the affected relatives. These shared regions of DNA define where the eye movement gene must be located, and by studying many large families, we are able to narrow down the choices to a single region of DNA. Once we have a single region for a specific eye movement disorder, we then begin to search the genes within that region for errors (mutations) that cause the eye movement disorder. Once the mutated gene is identified, we can study small families and families with only one affected member to determine if they too have mutations in the newly identified gene. We can also then study the function of this gene in normal and abnormal development.

What are the laboratory's results so far?

With the help of many families, the Engle laboratory has been able to identify the genes responsible for CFEOM1 (congenital fibrosis of the extraocular muscles type 1), CFEOM2, Duane syndrome with radial ray anomalies and Horizontal gaze palsy with progressive scoliosis. We have also localized the regions in which the genes for CFEOM3 and

congenital ptosis are located, and are actively searching for the genes mutated in CFEOM3, congenital ptosis, and several forms of Duane syndrome.

What kinds of families and individuals are needed for this research?

We continue to enroll families and individuals with CFEOM, congenital ptosis, Duane syndrome, and other related ocular motility disorders. Each new participating family may hold the key to unlock the precise location of each of these unidentified genes, and thus the contribution of each family is critical to the success of this project. Please contact Ms. Caroline Andrews if you have any questions about whether you would be appropriate for enrollment in our studies.

What should you do if you would like to participate in this research?

Please contact, or ask your physician to contact, Caroline Andrews at the above address. Caroline will answer any questions you or your physician may have, and will help organize your family's participation in the study.

What does participation mean?

Participation means filling out a short information questionnaire, reading and signing a consent form, undergoing an eye examination (or giving us permission to access your eye examination records), and donating a small sample of blood. The blood sample can be collected close to your home. Your blood sample will be used only for this study and your identity will be kept confidential.

Who will have access to your information?

All the information obtained is confidential. The results of this study are considered research and cannot be part of your medical record. We have, however, established diagnostic testing at the Children's Hospital Boston for some of the mutations we have found in the CFEOM1 gene as well as confirmatory testing for some of the other mutations. This information can be released to your physician. Please contact Ms. Caroline Andrews for additional information about this testing.

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