



Children's Hospital Boston

## RESEARCH CONSENT FORM

**Use Plate or Print:**

**Protocol Title:** Genetic studies of patients and their families with disorders of eye and eyelid movement.

**MRN#:**

**DOB:**

**Pt Name:**

(Consent B: For patients in whom DNA Diagnostic Laboratory testing does not identify a causative gene mutation)

**Gender:**

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**Doctor Directing Research: Elizabeth C. Engle, MD**  
**Phone: 617-919-4030; email: engle@enders.tch.harvard.edu**

### Introduction

You are being asked to participate in a research study. In order to decide whether or not you want to be a part of this research study, you should understand enough about its risks and benefits to make an informed judgment. This consent form gives detailed information about the research study that will be discussed with you. Once you understand the study, you will be asked to sign this form if you wish to participate.

### Description and Explanation:

I (Dr. Elizabeth Engle) and members of my research laboratory at Children's Hospital Boston study the genetic causes of congenital eye and eyelid movement disorders. Our research is federally funded by the National Institute of Health (NIH). We expect to enroll 340 patients in this research study per year, and anticipate about 120 of these will be from Children's Hospital. This research is ongoing and will be undertaken in the Engle Laboratory, located in the Enders building of Children's Hospital.

To date, we have discovered the disease genes for several forms of these disorders, and we are continuing our research to find additional disease genes. Because our discoveries are research-based, when we determine that one of our research participants harbors a gene mutation that causes their movement disorder, federal law does not permit us to release these results to the participant or their physician. Instead, the findings must be reported by a laboratory licensed to report results for clinical and diagnostic purposes. Therefore, we have arranged for the Children's Hospital DNA Diagnostic Laboratory (referred to below as the "Diagnostic Lab") to provide clinical genetic testing for the disorders we study. The Diagnostic Lab is a federally licensed clinical lab and can provide reports to requesting physicians.

Your physician is requesting that the Diagnostic Lab perform clinical diagnostic testing for you or one of your family members. We would like to offer you the opportunity to enroll in our research study at the same time that your physician orders this diagnostic genetic testing. If the Diagnostic Lab identifies a mutation, this will be reported to your physician and your enrollment will be cancelled. If, however, you enroll in the research study and the Diagnostic Lab does NOT identify a genetic cause for your eye or eyelid movement disorder, the DNA Diagnostic Lab will transfer a portion of your DNA sample along with your identifying information and clinical data to our research laboratory. We will then conduct research and try to determine the genetic cause of your eye or eyelid movement disorder. If we identify

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what we believe to be the cause, you may request that we notify the Diagnostic Lab of our finding. If you choose this option, the Diagnostic Lab will then be authorized to confirm the mutation and release the results to your physician. Your physician would then make these available to you with proper genetic counseling.

If you choose to participate in this research study and your information and DNA sample is transferred to our research laboratory, it may become necessary for us to obtain additional data pertaining to you, your family, and your eye movement disorder. This may include:

### **Medical and family history:**

If you choose to participate, we will ask you to fill out a form with questions about yourself, your children, siblings, grandparents, and possibly other family members. These questions may include age, ethnic background, health status and the biological relationship between individuals. You may be asked to release medical records to the Engle laboratory pertaining to the eye movement disorder and associated medical conditions. Your physician may examine your eye movements and write down their findings. It may be necessary for us to contact you to obtain additional information from you. Please indicate below if we may contact you in the future to obtain additional information if necessary:

**Yes, I do**     **No, I do not** **wish to be contacted in the future in order to provide obtain additional information.**

### **Photographing and videotaping eye movements:**

If you agree, we may photograph and/or video-record your eye movements, or request that your physician do so for us. These recordings would be used to review your eye movements in the future.

**Yes, I do**     **No, I do not** **agree to have my eye movements photographed and/or videotaped.**

Also, with your permission, we may use these recordings in medical teaching and medical publications. These recordings would not be used except as described above, and will not be released to anyone else. Please indicate your decision below:

**Yes, I do**     **No, I do not** **give permission for these photographs and/or videotapes to be used in medical teachings, and/or publications.**

### **Sample collection:**

If you agree to participate in this study, a portion of your DNA sample will be transferred from the Children's Hospital Boston DNA Diagnostic Lab to the Engle research lab. If there is an inadequate

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amount of DNA, we may need to request an additional blood sample or we may take a small swab of cells from your inner cheek, a sample of mouthwash that you have swished in your mouth, or some cut fingernails from which we can isolate a smaller amount of DNA. In some cases we may use blood that has already been drawn to grow your blood cells in a dish. Blood cells grown in this manner can survive indefinitely, providing a greater source of genetic material. We may also use your blood to examine your chromosomes for abnormalities that may cause the eye movement disorder. Finally, if you or your child is scheduled to undergo surgery for the eye movement disorder, we may ask your surgeon if any muscle tissue normally removed during the surgery is available for examination and study.

**Confidentiality:**

Samples and medical information obtained for this research study will be accessible only by Dr. Engle and her associates, and to our collaborators in the Diagnostic Lab. We will do our best to keep information confidential and only with your permission will we make this information available to others.

**Cost/time commitment:**

Your participation in the study should take no more than an hour and should coincide with your physician's request for diagnostic testing. There is no fee for you to participate, as the costs associated with this study are covered by research funds. You will not be paid or otherwise compensated for your participation.

**Recontact for additional data or participation in future studies:**

Over time we may wish to obtain updated information from participants. In addition, other studies may arise as a direct result of this study. Please indicate at the end whether we are permitted to contact you in the future.

**Yes, I do**     **No, I do not** wish to be contacted in the future in order to provide additional clinical information.

**Yes, I do**     **No, I do not** wish to be contacted if future studies arise.

**Risks and Discomforts:**

Risks associated with a blood draw are minor discomfort and bruising. When possible, we will use an aliquot of DNA from the Diagnostic Laboratory so that you will not need to have blood drawn only for research purposes.

**There is a chance that participation in this study could cause psychological distress. Some people involved in genetic studies have felt anxious about the possibility of carrying an altered gene that**



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**places them at risk or that may be passed on to children. If these feelings arise at any time during the study, you may contact us and we will arrange for you to speak with a genetic counselor.**

You should also be aware that there might be social and economic disadvantages, which can be associated with the gathering of genetic information. You should understand that our testing might find an inherited defective gene, which puts you at risk for a genetic disorder in the future. Genetic information divulged to the wrong source, could affect you and your family (if an insurance company or employer acquired this genetic information) or socially. We will do our best to keep all information confidential and only with your permission would we make this information available to others.

### **Potential Benefits:**

If we identify a genetic mutation in the research laboratory, you may request that this be confirmed by the Diagnostic Lab and released to your physician. These results would then have direct benefit to you. If we do not identify a mutation, you and your family may not directly benefit by participating in this research. We hope, however, that information obtained from this study will help us to understand the genetic causes of eye and eyelid movement disorders.

### **Reporting of Research Results:**

Because we are a research laboratory and not a clinical laboratory with certified procedures for patient result reporting, we cannot directly release results from the testing to you. This is why it is necessary to have our results confirmed by the Diagnostic Laboratory. It is important to remember that results from genetic tests performed for research purposes may take months or years to complete. If you wish to inquire into the progress of this research, you are welcome to do so at any time. If we do identify a genetic mutation that is the cause of your eye movement disorder and you request confirmation, we will have these results confirmed by the Diagnostic Lab. The Diagnostic Lab is then authorized to release results from patient tests for clinical and diagnostic purposes.

### **Storage of Research Samples:**

We will store any remaining DNA or biopsy samples indefinitely for future studies on eye and eyelid movement disorders. These samples will be used for the future studies of genetic causes of your disease and will be stored in the Division of Genetics at Children's Hospital. Samples are stored without your name or medical record. Your sample will be given a unique number. Only the investigator will have a list to know which sample is linked to a patient identifier. This list will be kept confidential in a secure location. If Dr. Engle distributes your sample to other individuals who have an interest in the genetic causes of your disease, it will be released with the unique identifiers without your name or medical record number. If at any time you would like to have your sample removed from storage, please let us know and it will be destroyed.

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### **Alternatives:**

Participation in the research is completely voluntary. You should not feel any pressure to participate. If you do not want to participate it will not interfere with obtaining the DNA Diagnostic test your physician is ordering or any future care you or your family receives at this institution.

### **Research at Children's Hospital**

Children's Hospital has recently developed a web-based, interactive educational program for parents called "A Parent's Guide to Medical Research." To find out more about research at Children's Hospital, please visit the program at [www.researchchildren.org](http://www.researchchildren.org)

Children's Hospital is interested in hearing your comments, answering your questions and responding to any concerns regarding clinical research at Children's hospital. If you would like further information about the type of clinical research performed at the hospital or have suggestions, questions or concerns regarding clinical research you may send an email to [cci@childrens.harvard.edu](mailto:cci@childrens.harvard.edu) or call 617 355-7052 between the hours of 8:30 and 5:00.

### **What information do I need to know about the Health Insurance Portability and Accountability Act (HIPAA)?**

During this research, information about your or your child's health will be collected. In general, under federal law, information about patients is private, but there are exceptions and you should know who will have access to this information and might see it.

Researchers may be collecting information about you or your child from medical records. They may also learn things from procedures that are part of the research itself such as tests, office visits, questionnaires and interviews.

The following people will be able to see this information:

- Medical and research staff at Children's Hospital, including people listed on your informed consent.
- Medical staff who are directly involved in your care that is related to the research or arise from it.
- People who oversee, advise or conduct research at Children's Hospital, and people who oversee or evaluate research and care, including the Committee on Clinical Investigation, staff working on quality improvement, and other clinicians and administrative staff of Children's Hospital..
- People from agencies and organizations that provide independent accreditation and oversight of research
- Sponsors or others involved in funding the research
- Federal agencies that oversee or review research information.
- Government agencies and sponsors.
- If some law or court requires us to share the information, we would have to follow that law or final ruling

You/your child should be aware that the federal privacy rule does not cover all of these possible uses. This means that once some of the above mentioned users receive your/your child's health information they do not have to follow the same rules. Other laws may or may not protect sharing of private health information. If you have a

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question about this you may contact the Children's Hospital Privacy Officer at 617-355-5502

There is no set time for destroying this information and no time limit for its use. Researchers continue to analyze data for many years and it is not possible to know when they will be done.

You or your child do not have to sign this form. If the form is not signed, however, you won't be able to participate in the study. Not signing will not affect your care or your child's care at Children's Hospital in any way now or in the future. Also, there will be no penalty or loss of benefits if you choose not to sign and participate.

You or your child also have the right to withdraw from this study at any time. You have the right to end your permission for Children's Hospital to use or share the protected information about you or your child that was collected as part of the research.

Researchers may also continue to use information already collected to protect the integrity of the study. This means that your withdrawal won't make the whole study useless. Once you remove your permission and you or your child is no longer in the study, no more private health information will be collected. If you wish to withdraw you will need to do so in writing. Your investigator will have a form for you to use. If you or your child decide to share private information with anyone not involved in the study, the federal law designed to protect privacy may no longer apply to this information.

Although there are some legal limitations, you or your child have the right to get protected information resulting from this research that relates to your treatment or to payments. This information is available after the study analysis is done. To request the information, please contact the Hospital's Privacy Officer at 617-355-5502. If you have questions, please be sure to ask for answers.

### **INVESTIGATOR'S AND/OR ASSOCIATE'S STATEMENT:**

I have fully explained to \_\_\_\_\_ (insert name) the nature and purpose of the participant/parent/guardian above-described procedures and the risks involved in its performance. I have provided the subject/family with the Privacy Rule if requested. I have answered and will answer all questions to the best of my ability. I will inform the participant of any changes in the procedures or the risks and benefits if any should occur during or after the course of the study. I have given a copy of the consent/authorization form to the subject/family.

\_\_\_\_\_  
Date                      Investigator's and/or Associate's Signature

### **CONSENT/AUTHORIZATION:**

I have been satisfactorily informed of the above-described procedure with its possible risks and benefits. I have been provided with the applicable Privacy Rule provisions under the Health Insurance Portability and Accountability Act. I give permission for my/my child's participation in this study and for use of the associated protected health information as described above.

