



knowledge changing life



**Human Ocular Disorders  
Program: *Investigating the  
genetic causes of eye  
conditions***

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### **What eye conditions qualify for this study?**

Conditions that qualify for this study include but are not limited to **anterior segment dysgenesis** (Aniridia, Axenfeld-Rieger, Peters anomaly, etc.), **congenital/childhood cataracts or glaucoma, microphthalmia/anophthalmia, coloboma**, or other childhood eye conditions suspected to have a genetic cause.

### **What is the purpose of this study?**

The goal of this study is to identify and understand the genetic causes of eye disorders.

### **Will the study help me/my child?**

We may be able to find an explanation for your/your child's eye condition. We hope that better understanding of the genetics of various eye conditions will lead to improved diagnosis and treatment in the future for all individuals.

### **What will happen if I/my child participate(s)?**

We will collect a saliva or blood sample from the affected person and other family members (if possible).

### **Will I get results?**

If the study is able to identify a specific genetic cause for your/your child's eye condition, you will be informed.

### **How is this study different than clinical testing?**

Clinical testing analyzes a specific set of genes for specific types of variants. The research study will use broader types of analyses and may be able to identify new genes involved

in eye disorders or new types of variants not detected by current clinical testing.

### **Is there any cost to participate?**

No- all research analysis is covered by the study.

### **Why do you need samples from family members?**

Each person has hundreds of rare genetic variants. Including samples from family members in the analysis helps us to interpret your/your child's results.

### **How can I/my child participate?**

If you/your child qualify for the study, we are able to enroll in-person or remotely via a phone/Zoom call with samples collected at home. Enrollment takes about 20-30 minutes.

### **How will my privacy be protected?**

We will assign a unique study ID number to the samples and use the coded samples for analysis.

**To learn more**, please call our study team at (414) 955-7645 or email us at [seminagenetics@mcw.edu](mailto:seminagenetics@mcw.edu). You can also volunteer for the study through our website:

