

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform primary congenital glaucoma (CYP1BI) testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR PRIMARY CONGENITAL GLAUCOMA (CYP1BI) TESTING

Patient Name _____ **Date of Birth** ____/____/____ **Gender** F M

Physician _____ **Physician Phone** (____) _____ **Practice Specialty** _____

Genetic Counselor _____ **Counselor Phone** (____) _____

Patient's Ethnicity (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

Does the patient have SYMPTOMS? No Yes (check all that apply)

- Congenital glaucoma (age of onset: _____, Bilateral Unilateral)
- Elevated intraocular pressure
- Enlargement of the globe (buphthalmos)
- Corneal edema/clouding
- Haab stiae (tear in Descemet membrane)
- Thinning of the anterior sclera
- Abnormally deep anterior chamber
- Photophobia (sensitivity to light)
- Excessive tearing (ephiphora)
- Involuntary eyelid spasms (blepharospasm)
- Other _____

Has Peter's anomaly (anterior segment dysgenesis) been ruled out? No Yes Unsure N/A

Does the patient have a FAMILY HISTORY of Primary Congenital Glaucoma? No Yes Unknown

If yes, what is the **RELATIONSHIP** of the affected family member(s) to the patient? _____

Has DNA testing for primary congenital glaucoma been performed for affected family member(s)?

- No Yes Unknown

If yes, please describe test(s) and results _____

Has the patient undergone previous DNA testing for Primary Congenital Glaucoma? No Yes Unknown

If yes, please describe test(s) and results _____

Circle the PRIMARY CONGENITAL GLAUCOMA test you intend to order.

0051476 Glaucoma, Primary Congenital (CYP1BI) Sequencing

Diagnostic testing for symptomatic patients; clinical sensitivity 20-100% in familial cases, 10-15% in isolated cases.

2001961 Familial Mutation, Targeted Sequencing

Tests for a CYP1BI sequence change identified in a family member; copy of relative's lab result is REQUIRED.

For questions, contact an ARUP genetic counselor at (800) 242-2787 ext. 2141

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