ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR PRIMARY CONGENITAL GLAUCOMA (*CYP1B1*) TESTING

Patient Name _____ Date of Birth _____ Sex ☐ F ☐ M

Physician _____ Physician Phone _____

Practice Specialty _____ Physician Fax _____

Genetic Counselor _____ Counselor Phone _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

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

☐ Abnormally deep anterior chamber

☐ Congenital glaucoma

age of onset: _____
☐ Bilateral
☐ Unilateral

☐ Corneal edema/clouding

☐ Elevated intraocular pressure

☐ Enlargement of the globe (buphthalmos)

☐ Excessive tearing (ephiphora)

☐ Haab stiae (tear in Descemet membrane)

☐ Involuntary eyelid spasms (blepharospasm)

☐ Photophobia (sensitivity to light)

☐ Thinning of the anterior sclera

☐ Other symptom(s): _____

Has Peters anomaly (anterior segment dysgenesis) been ruled out? ☐ No ☐ Yes ☐ Unknown ☐ N/A

Has the patient undergone previous DNA testing? ☐ No ☐ Yes ☐ Unknown

If yes, describe the test(s) and results: _____

_____

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient: _____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's DNA laboratory result. (REQUIRED for familial mutation testing)

Check the test you intend to order.

☐ **0051476 Glaucoma (Primary Congenital) *CYP1B1* 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 mutation previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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