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 RETINITIS PIGMENTOSA/LEBER CONGENITAL AMAUROSIS 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: _____

**Suspected diagnosis:**

☐ Adult vitelliform macular dystrophy
☐ Bothnia dystrophy
☐ Choroideremia
☐ Cone-rod dystrophy
☐ Leber congenital amaurosis
☐ Macular atrophy
☐ Macular dystrophy
☐ Oguchi disease
☐ Retinitis pigmentosa
☐ Retinitis punctata albescans
☐ S-cone syndrome
☐ Senior-Loken syndrome
☐ Stargardt disease
☐ Usher syndrome

☐ Other: _____

**Does the patient have symptoms?** ☐ No ☐ Yes (check all that apply and describe) ☐ bilateral or ☐ unilateral

Age of onset: _____

☐ "Night blindness"
☐ "Oculo-digital sign"
☐ Abnormal ERG
☐ Cystoid macular edema
☐ Dust-like particles in the vitreous
☐ Exudative vasculopathy
☐ Hyaline bodies
☐ Hyperopia
☐ Keratoconus
☐ Loss of peripheral vision (tunnel vision)
☐ Macular degeneration
☐ Macular lesions
☐ Nystagmus
☐ Photophobia
☐ Posterior subcapsular cataracts
☐ Retinal vessel attenuation
☐ RPE granularity
☐ Sensorineural hearing loss
☐ Sinopulmonary infection
☐ Sluggish pupillary response
☐ White spots in the retina

☐ Other symptom(s): _____

**Has the patient undergone previous genetic testing for this condition?** ☐ No ☐ Yes ☐ Unknown

If yes, describe the test performed 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. List their symptoms and age of onset: _____

_____

_____

**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.**

☐ **2007085 Retinitis Pigmentosa/Leber Congenital Amaurosis Panel, Sequencing and Deletion/Duplication:** Next generation sequencing and microarray coverage of select genes associated with RP and LCA.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Targeted testing for sequence variants previously identified in a family member; a copy of the relative's lab result is REQUIRED.

Master Label

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