

**THIS IS NOT A TEST REQUEST FORM.**  
**The information below is required to perform retinitis pigmentosa/leber congenital amaurosis testing.**  
**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** \_\_\_\_/\_\_\_\_/\_\_\_\_ **Gender**  F  M

**Physician** \_\_\_\_\_ **Physician Phone** (\_\_\_\_) \_\_\_\_\_ **Practice Specialty** \_\_\_\_\_

**Genetic Counselor** \_\_\_\_\_ **Counselor Phone** (\_\_\_\_) \_\_\_\_\_

**Patient's Ethnicity** (check all that apply)

- African American       Ashkenazi Jewish       Asian       Caucasian  
 Hispanic       Middle Eastern       Native American       Other \_\_\_\_\_

**Suspected diagnosis:**

- Retinitis Pigmentosa       Leber Congenital Amaurosis       Cone-rod dystrophy       Usher syndrome  
 Stargardt disease       Meckel-Gruber syndrome       Nephronophthisis       Joubert syndrome  
 Macular atrophy       Macular dystrophy       Adult vitelliform MD       Retinitis punctata albescans  
 Coroidal sclerosis       S-cone syndrome       Bothnia dystrophy       Oguchi disease  
 Other \_\_\_\_\_

**Is the patient SYMPTOMATIC?**     No     Yes - if yes, are symptoms  bilateral or  unilateral  
Age of onset: \_\_\_\_\_

**Please check all symptoms that apply:**

- "night blindness"       loss of peripheral vision ("tunnel vision")       macular lesions  
 macular degeneration       dust-like particles in the vitreous       cystoid macular edema  
 white spots in the retina       posterior subcapsular cataracts       hyaline bodies  
 exudative vasculopathy       nystagmus       sluggish pupillary response  
 "oculo-digital sign"       photophobia       hyperopia  
 abnormal ERG       keratoconus       RPE granularity  
 retinal vessel attenuation       sensorineural hearing loss       sinopulmonary infection

**Other Symptoms:** \_\_\_\_\_

**Has the patient undergone previous genetic testing for the above disorder?**  No  Yes  Unknown

If yes, please describe test performed, genes tested and results: \_\_\_\_\_

**Does the patient have a FAMILY HISTORY of RP/LCA or a related disorder?**  No  Yes  Unknown

If yes, please attach PEDIGREE or COMPLETE BELOW:

RELATIONSHIP to the patient	Age at Dx	Symptoms	Dx confirmed by DNA testing?	
_____	_____	_____	<input type="checkbox"/> No	<input type="checkbox"/> Yes*
_____	_____	_____	<input type="checkbox"/> No	<input type="checkbox"/> Yes*
_____	_____	_____	<input type="checkbox"/> No	<input type="checkbox"/> Yes*

**Circle the test you intend to order.**

**2007085 Retinitis Pigmentosa/Leber Congenital Amaurosis Panel, Sequencing and Deletion/Duplication, 53 Genes:** Next generation sequencing and microarray coverage of 53 genes associated with RP and LCA.

**2001961 Familial Mutation, Targeted Sequencing:** Targeted sequencing for variants previously identified in a family member; a copy of the relative's lab result is REQUIRED. Contact an ARUP genetic counselor prior to ordering.

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

Master Label