

RECOMMENDED FLOWCHART FOR LOWE SYNDROME DIAGNOSTIC TESTING

Families with confirmed diagnosis of Lowe Syndrome- affected male available for DNA testing:

<u>DIAGNOSTIC TEST</u>	<u>AFFECTED MALES</u>	<u>CARRIER FEMALES</u>	<u>PRENATAL*</u>
Clinical	Ophthalmological Exam ↓	Ophthalmological Exam (dilated slit lamp)	N.A.
Biochemical	Enzyme test in affected males (Phosphatidylinositol-4,5-bisphosphate phosphatase; >99% sensitivity) ↓	N.A. (not useful)	Prenatal enzyme test for male fetuses (>99% sensitivity)
DNA	<i>OCRL1</i> Gene Sequencing Analysis of entire coding region ~97% sensitive in males detects point mutations and exon deletions Mutation Positive →	↓ DNA carrier testing for familial mutation →	Combined Prenatal Biochemical + DNA testing if applicable DNA prenatal test for familial mutation

Families without an affected male available for DNA testing:

DNA carrier testing on high-risk females (in conjunction with ophthalmologic exam)
Note: *OCRL1* gene sequencing ~90% sensitive for carrier females; large deletion mutations will not be detected

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***Contact lab in advance for all prenatal cases**