

WHO SHOULD GET TESTED?



Individuals with a family history of Ophthalmological disease

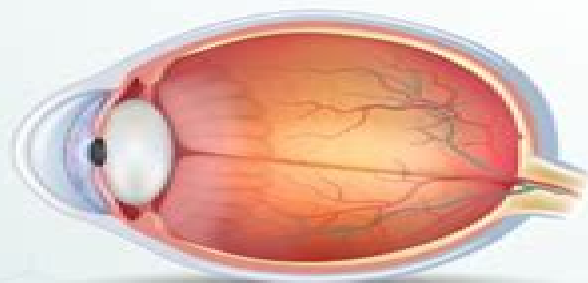


Individuals presenting with an Ophthalmological disease with a pending diagnosis

DIAGNOSTIC TESTS

Confirmation of a clinical diagnosis through genetic testing will likely change medical management and allows for genetic counseling.

	GENES	COVERAGE
Clinical exome sequencing	~ 6,700	>99% ≥ 20x
Whole exome sequencing	~ 22,000	>99.45% ≥ 20x



Test Range

Nx GEN CLINICAL EXOME SEQUENCING, BASIC
(does not includes sanger sequencing)

Nx GEN CLINICAL EXOME SEQUENCING, REFLEX
CONFIRMATION WITH PARENTS
(includes sanger sequencing for Trio)

Nx GEN CLINICAL EXOME, ADVANCED
(includes sanger sequencing)

Nx GEN WHOLE EXOME

SANGER SEQUENCING, SINGLE VARIANT

Retina Volume 1 Of A 3 Volume Set Basic Science And Inherited Retinal Disease Tumors

J Spring



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