

GENETIC TEST: Stargardt disease

FULL NAME:	Stargardt disease
DESCRIPTION:	ABCA4 coding sequence + 34 pathogenic deep intronic variants (Khan et al. Genet Med 2020; PMID: 32307445)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Sequence analysis: entire coding region Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS) MLPA based techniques
RIZIV CODE:	565471-565482

ACCREDITATION (ISO 15189):	2021-09-11 / 2026-09-10
TURNAROUND TIME (MAXIMUM):	3-4 months
CREATED:	02 Aug 2019 - 08:18
CHANGED:	14 Dec 2022 - 16:51

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