MACHAON DIAGNOSTICS

TEST SUMMARY

Alport Syndrome Genetic Panel

Alport Syndrome (AS) is a progressive inherited kidney disease. It is caused by mutations in genes COL4A3, COL4A4 and COL4A5, which are involved in the synthesis of type IV collagen. Sanger sequencing may be used to confirm variants as needed. Genetics can help confirm a diagnosis of Alport syndrome and may be especially helpful when the clinical phenotype is not clear-cut. Genetics can also inform family planning (for example, does an affected male have X-linked or autosomal Alport and is an affected female heterozygous or homozygous?).

Please visit Machaondiagnostics.com for further information.

Limitations: This test will not detect variants located outside of the targeted DNA regions. This test is not optimized to detect chimerism or somatic mosaicism. This test will detect small indels but may miss larger deletions or duplications. Balanced structural variants will not be detected unless specifically targeted by a custom PCR assay.

References: 1. Alport, A. C., Hereditary familial congenital haemorrhagic nephritis. Brit. Med. J. 1: 504-506, 1927.2. Kashtan, C. E., Alport Syndrome and Thin Basement Membrane nephropathy http://www.ncbi.nlm.nih.gov/books/NBK1207/.

SPECIMEN REQUIRMENTS
3mL EDTA Whole Blood

STABILITY
Room Temp: 1 month
Refrigerated: 1 month

CPT CODE
81407x2
81408x2

METHODOLOGY
NGS

TURNAROUND TIMES
Routine TAT: <5 days (M-F)
STAT TAT: <48 hours (M-F)

ALTERNATIVE NAME
COL4A3
COL4A4
COL4A5

DRAW KITS AVAILABLE

V:04MAR2025

WHY CHOOSE US?
- Fastest turnaround in the US
- Draw kits (includes free shipping)
- Clinical consultation
- Critical Results called to physician

ABOUT US:
Machaon Diagnostics is a clinical reference laboratory, specializing in coagulation, platelets, complement, genetics and rare disease.

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