



Alport Syndrome Genetic Panel

Results in 1 Week (48 hours STAT, M-F)

Machaon Diagnostics is a clinical reference laboratory, specializing in rare disease genetic testing and the diagnosis, treatment and monitoring of hemostatic, thrombotic conditions. We strive to be your fastest resource for the laboratory testing.

Draw kits with free priority overnight shipping available. Call for kits.

Background

Alport Syndrome (AS) is a progressive inherited kidney disease (1, 2). It is caused by mutations in genes *COL4A3*, *COL4A4* and *COL4A5*, which are involved in the synthesis of type IV collagen.

Patients with AS gradually develop kidney failure, hearing loss and eye abnormalities. Renal dysfunction progresses from microscopic hematuria to proteinuria, progressive renal insufficiency, and end-stage renal disease (ESRD) in all males with X-linked AS (XLAS), and in all males and females with autosomal recessive (AR) AS.

About 50% of untreated males with XLAS (80% of patients) develop kidney failure by age 25, increasing to 90% by age 40 and nearly 100% by age 60. Both males and females with ARAS (15% of patients) develop kidney failure, usually by their teens or young adult years.

The current diagnostic tests rely on evaluation of the patient's signs, symptoms, blood and urine tests, kidney and skin biopsy, renal ultrasonography, family history.

These approaches can be time-consuming, invasive or provide uncertain results, requiring further investigation.

Machaon Diagnostics is the only laboratory in the US currently offering noninvasive and accurate Alport Syndrome genetics testing with a turnaround time of a week or less, compared with a four week turnaround time at other laboratories.

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/>.

1-week
Turnaround
Time

(fastest in the US)

Genes sequenced:

COL4A3*, *COL4A4* and *COL4A5

Test covers all the exons of these genes and all of the known mutations

- Available across US (FedEx Service Area)
- Draw Kits (includes free shipping)
- Clinical Consultation
- Results called to referring physician

Turnaround Time:

1 week

STAT Turnaround Time:

48 hours (M-F)

Methodology:

Next Generation Sequencing (NGS)

Price:

\$2,978

Note: please call with all insurance billing requests as benefits must be verified by Machaon. Medicare and Medicaid programs do not reimburse for this test.

Additional Information

Specimen: 3mL EDTA whole blood

Alternative Specimen: buccal swab

Stability: >1 month at 2-25°C

CPT Code: 81479x3

Days Run: Monday - Friday

Reflex: COL4A5 CNV testing



TOLL FREE
1-800-566-3462

Phone : 510-839-5600
Fax: 510-839-6153
E-mail: info@machaondiagnosics.com

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Machaon Diagnostics

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Alport Genetic Panel Order Form

Routine Turnaround Time: 1 week

www.MachaonDiagnostics.com

1-800-566-3462

PATIENT INFORMATION <i>(complete or attach)</i>			SUBMITTING FACILITY		
Patient's Name: (Last, First, M.I.)		Gender: (circle one) M / F	Facility Phone Number: _____ Fax Number for Results: _____		
Specimen Date: (MM/DD/YY) Time: AM / PM		Date of Birth: (MM/DD/YY)			
Medical Record / ID#:		Patient's Social Security #:			
ORDERING PHYSICIAN INFORMATION			BILLING INFORMATION		
Physician's Name: (Last, First, M.I.)		Physician's NPI:	Bill to: <input type="checkbox"/> Facility <input type="checkbox"/> Insurance <input type="checkbox"/> Patient <i>All inpatient testing is billed to facility</i>		<small>Currently, there is no payable outpatient benefit from Medicare or Medicaid for this test.</small>
Contact Phone Number:		Fax Number for Results:	Patient status: <input type="checkbox"/> Inpatient <input type="checkbox"/> Outpatient		
Physician's direct phone number to call results: <i>(highly encouraged)</i>			<input type="checkbox"/> STAT <i>Samples shipped for weekend arrival must be ordered STAT. Mark FedEx Airbill for 'SATURDAY Delivery.'</i>		
CLINICAL INFORMATION <i>(if available)</i>			TEST SELECTION		
On-going Pregnancy? <input type="checkbox"/> Yes <input type="checkbox"/> No		Has a biopsy been performed? <input type="checkbox"/> Yes <input type="checkbox"/> No	<input type="checkbox"/> Alport Genetic Panel		Methodology: <i>Rapid Next Generation sequencing of 3 genes</i>
Creatinine Level:	Proteinuria? <input type="checkbox"/> Yes <input type="checkbox"/> No	Hematuria? <input type="checkbox"/> Yes <input type="checkbox"/> No	Genes included: COL4A3, COL4A4, COL4A5		
SPECIMEN COLLECTION, PROCESSING AND SHIPPING / <i>call for draw kits (free shipping)</i>					
Specimen Collection and Processing			Specimen Shipping		
1. Draw 1 lavender top tube (EDTA).			6. Place all forms into document sleeve of the biohazard bag.		
2. Mix tube by inversion, gently to ensure proper mixing.			7. Ensure that no patient-specific information is visible.		
3. Label tube with patient first and last name, draw date and DOB.			8. Place biohazard bag into the provided FedEx Clinical Pak.		
4. Enclose tube in Styrofoam box and seal in biohazard bag.			9. Complete the provided FedEx Airbill and affix Airbill pouch to Pak.		
5. Complete this form and attach insurance billing information.			10. Call FedEx 800-238-5355 for a pick-up.		
Note: Samples can be shipped room temperature or refrigerated. Please call for guidance.					
OUTPATIENT ONLY: INSURANCE BILLING INFORMATION <i>(complete or attach)</i>					
Insurance Company: <i>(Medicare patients must sign ABN on reverse)</i>			Patient Address:		Patient Phone Number:
Insurance Policy / Medicare Number:		Insurance Group Number:	Patient City:		State: Zip Code:
Insurance Company Address:			Authorization Number:		
Insurance Company City:		State:	Zip Code:		
DIAGNOSIS CODE(S):			OUTPATIENT ONLY: PATIENT SIGNATURE		
ICD-10 Code: ICD-10 Code: ICD-10 Code:			MachaonDiagnosticsmayneedtoobtainadditionalinformationfromyouphysiciarto completetheseservicesherebyauthorizethereleaseofmedicalinformationrelatedtothe services described herein and authorize payment directly to Machaon Diagnostics. This test is currently not covered or reimbursed by Medicare or Medicaid. The Alport Syndrome Genetic Panel is \$2,978; shipping charges may apply. I agree to assume responsibility for payment of all charges not covered by my healthcare insurer.		
Patient's Signature: _____			Date: _____		
ADDITIONAL INFORMATION					
Machaon Diagnostics is a specialized coagulation, platelet and genetic laboratory providing comprehensive clinical evaluations of bleeding and clotting patients. Most evaluations can be completed within 24 hours, 7 days a week. Machaon Diagnostics is a California-licensed, CLIA-accredited, CAP-accredited, clinical laboratory approved to provide high-complexity testing services. This test is not covered or reimbursed by Medicare or Medicaid. All patients are considered OUT-OF-NETWORK and will be billed for services not covered by their insurance provider. Medicare patients must sign an ABN, either located on the reverse side of this form or downloaded from the Machaon Diagnostics website. Patient insurance billing services are provided in accordance with the Machaon Insurance Billing Policy. HMO or medical group covered patients may need a prior authorization if they seek full reimbursement. For more information please visit www.MachaonDiagnostics.com or call (510) 839-5600.					