ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform Alport Syndrome testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR ALPORT SYNDROME TESTING

Patient's Name _____ Date of Birth _____/_____/_____ Gender [ ] F [ ] M

Physician _____ Physician Phone (_____)_____Practice Specialty_____

Genetic Counselor _____ Counselor Phone (_____)_____

Patient's Ethnicity (check all that apply)

[ ] African American [ ] Ashkenazi Jewish [ ] Asian [ ] Caucasian
[ ] Hispanic [ ] Middle Eastern [ ] Native American [ ] Other_____

Does the patient have SYMPTOMS of Alport Syndrome? [ ] No [ ] Yes (check all that apply)

[ ] Hematuria [ ] Corneal erosion [ ] Proteinuria
[ ] Hypertension [ ] Renal insufficiency [ ] End stage renal disease (Age_____)
[ ] Anterior lenticonus [ ] Maculopathy [ ] Sensorineural hearing loss (Age_____)
[ ] Other _____

Does the patient have a FAMILY HISTORY of Alport Syndrome? [ ] No [ ] Yes [ ] Unknown

If yes, **please attach PEDIGREE or** specify the RELATIONSHIP of the family member(s) to the patient and detail the symptoms/age of onset in each symptomatic relative.

_____

Please attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Has the patient undergone previous testing for Alport Syndrome? [ ] No [ ] Yes

If yes, please describe test(s) and results_____

Circle the test you intend to order.

2002398 Alport Syndrome, X-Linked (*COL4A5*) Sequencing and Deletion/Duplication Detects >80% of mutations in males and females with X-linked Alport syndrome.

0051786 Alport Syndrome, X-linked *(COL4A5)* Sequencing Offer to patients who have a family history of X-linked renal failure or to individuals with unexplained hematuria or chronic kidney disease. Clinical sensitivity > 80% in males and >70% in females with X-linked Alport syndrome.

0051710 Alport Syndrome, X-linked *(COL4A5)* 3 Mutations Identifies three common mutations found only in adult type, X-linked Alport syndrome (C1564S, L1649R and R1677Q). Test not useful for juvenile or non X-linked forms of Alport Syndrome. Clinical sensitivity approximately 90%.

2002394 Alport Syndrome, X-Linked (*COL4A5*) Deletion/Duplication For patients with negative *COL4A5* sequencing result. Clinical sensitivity 10%. **Also order for familial *COL4A5* large deletion or duplication**.

2001961 Familial Mutation, Targeted Sequencing Targeted sequencing for a *COL4A5* mutation previously identified in a family member; copy of relative's lab result is REQUIRED.

For questions, contact an ARUP genetic counselor at (800) 242-2787, ext. 2141

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