

Clinical, Pathological, and Genetic Characteristics of Patients with Digenic Alport Syndrome

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Supplemental Table 1. List of 68 podocyte-related genes included in targeted sequencing analysis within a clinically approved gene panel test developed in our laboratory.

| | | | | | |
|-----------------|---------------|----------------|---------------|-----------------|---------------|
| <i>ACTN4</i> | <i>COQ2</i> | <i>ITSN1</i> | <i>MAGI2</i> | <i>PAX2</i> | <i>TPRKB</i> |
| <i>ADCK4</i> | <i>COQ6</i> | <i>ITSN2</i> | <i>MYH9</i> | <i>PDSS2</i> | <i>TRIM8</i> |
| <i>ANKFY1</i> | <i>CRB2</i> | <i>KANK1</i> | <i>MYO1E</i> | <i>PLCE1</i> | <i>TRPC6</i> |
| <i>ANLN</i> | <i>CUBN</i> | <i>KANK2</i> | <i>NPHS1</i> | <i>PODXL</i> | <i>TTC21B</i> |
| <i>ARHGAP24</i> | <i>DLC1</i> | <i>KANK4</i> | <i>NPHS2</i> | <i>PRDM15</i> | <i>WDR4</i> |
| <i>ARHGDI1A</i> | <i>EMP2</i> | <i>KIRREL1</i> | <i>NUP85</i> | <i>PTPRO</i> | <i>WDR73</i> |
| <i>AVIL</i> | <i>FAT1</i> | <i>LAGE3</i> | <i>NUP93</i> | <i>SCARB2</i> | <i>WT1</i> |
| <i>CD2AP</i> | <i>GAPVD1</i> | <i>LAMA5</i> | <i>NUP107</i> | <i>SGPL1</i> | <i>XPO5</i> |
| <i>CKD20</i> | <i>GON7</i> | <i>LAMB2</i> | <i>NUP133</i> | <i>SMARCAL1</i> | |
| <i>COL4A3</i> | <i>INF2</i> | <i>LMNA</i> | <i>NUP160</i> | <i>TNS2</i> | |
| <i>COL4A4</i> | <i>ITGA3</i> | <i>LMX1B</i> | <i>NUP205</i> | <i>TBC1D8B</i> | |
| <i>COL4A5</i> | <i>ITGB4</i> | <i>MAFB</i> | <i>OSGEP</i> | <i>TP53RK</i> | |

Supplemental Figure 1

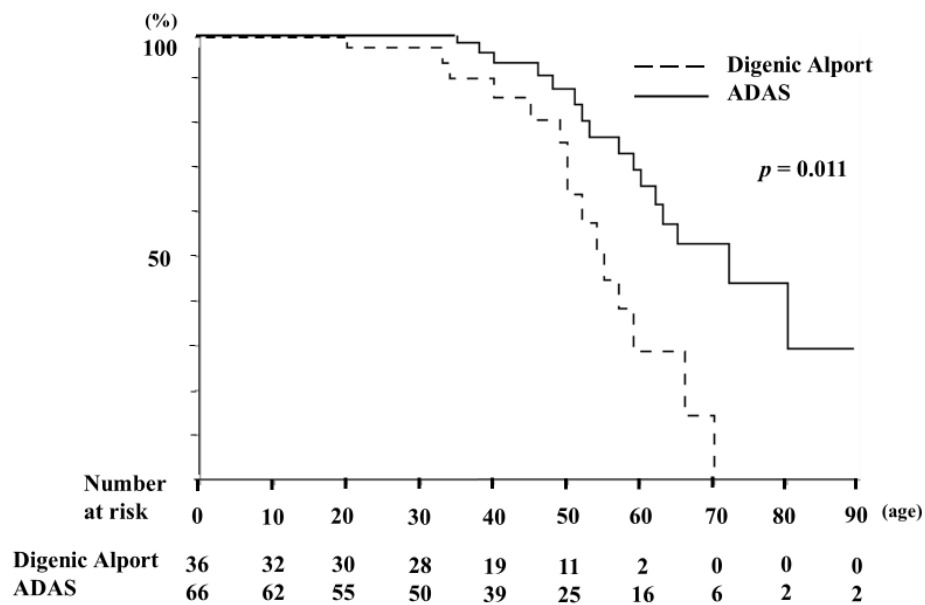


Figure S1. Probability of developing ESKD in Alport syndrome cases with digenic *COL4A3* and *COL4A4* variants including previously reported¹ cases compared with that in autosomal dominant Alport syndrome (ADAS) cases in our cohort ($P = 0.011$).

1. Savige J, Renieri A, Ars E, et al. Digenic Alport syndrome. *Clin J Am Soc Nephrol*.

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