

ALPORT SYNDROME (AS) CAN LEAD TO THE DEVELOPMENT OF END-STAGE KIDNEY DISEASE (ESKD)



X-Linked

Males



risk of ESKD

Females



risk of ESKD

Autosomal Recessive

Males & Females



risk of ESKD

Autosomal Dominant

Males & Females



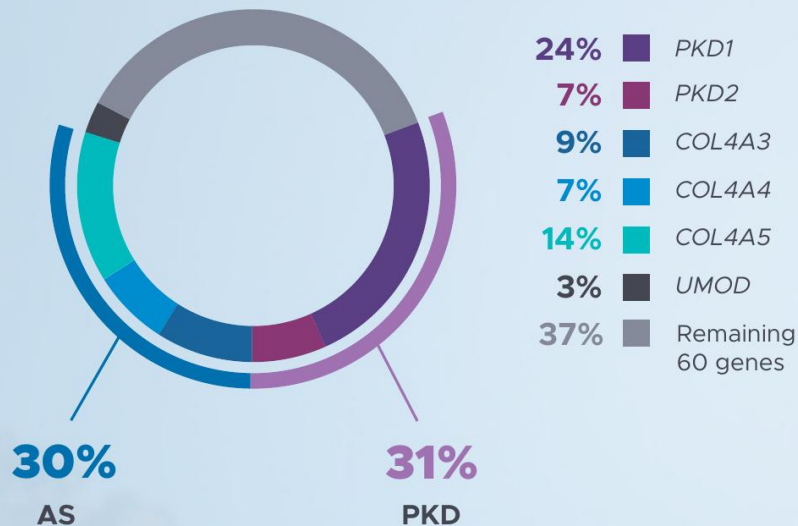
risk of ESKD

Estimated
Risk of ESKD¹

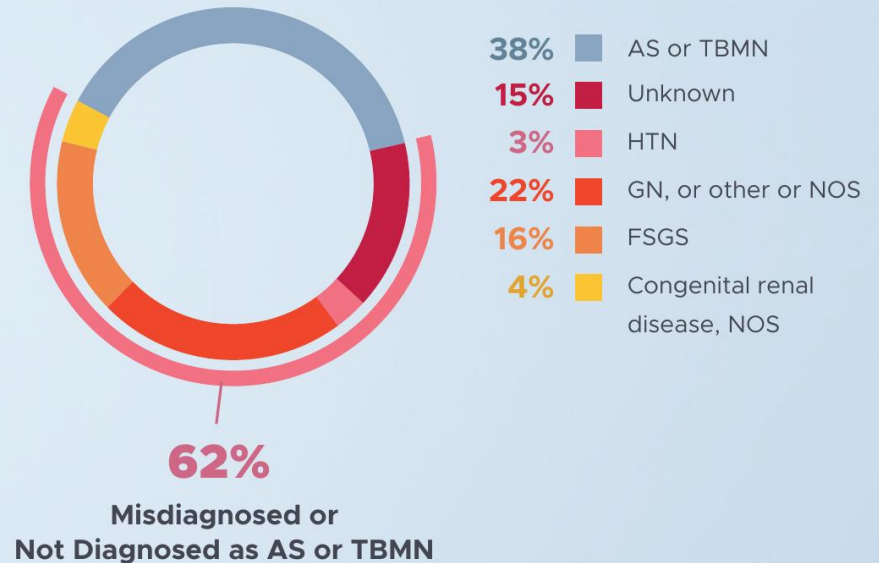
COL4A3/4/5 Mutations Representing AS Are as Common as PKD Mutations

In a recent exome sequencing analysis of 3315 patients with CKD, ~10% (307/3315) had a genetic cause.²

Abnormalities in COL4A3/4/5 Represent 30% of Patients With a Genetic Finding



Of COL4A3/4/5 Mutations, 62% Were Misdiagnosed or Not Diagnosed as Having AS or TBMN



CKD, chronic kidney disease; FSGS, focal segmental glomerulosclerosis; GN, glomerulonephritis; NOS, not otherwise specified; PKD, polycystic kidney disease; TBMN, thin basement membrane nephropathy.

1. Kashtan CE et al. *Kidney Int.* 2018;93(5):1045-1051.
2. Groopman EE et al. *N Engl J Med.* 2019;380(2):142-151.