

Supplemental Table S1. Candidate mutations fitting compound heterozygous model. SKDP 6.1 is the father, SKDP the mother, and SKDP 6.3 and 6.4 the affected children.

Mutation (HGVS nomenclature)	Gene	SIFT prediction	SKDP 6.1	SKDP 6.2	SKDP 6.3	SKDP 6.4
NM_001009944.2:c.3502C>T, p.Pro1168Ser	<i>PKD1</i>	Tolerated	X	ND	X	X
NM_001009944.2:c.5611G>A, p.Ala1181Thr	<i>PKD1</i>	Tolerated	ND	X	X	X
NM_182924.3:c.2660A>C, p.Lys675Thr	<i>MICALL2</i>	Tolerated	X	ND	X	X
NM_182924.3:c.1885G>T, p.Ala604Ser	<i>MICALL2</i>	Tolerated	ND	X	X	X
NM_000093.3:c.3562A>G, p.Ile1188Val	<i>COL5A1</i>	Tolerated	X	ND	X	X
NM_000093.3:c.574G>A, p.Asp192Asn	<i>COL5A1</i>	Tolerated	ND	X	X	X