Supplement

Table S1. Whole exome sequencing report of donor and patient

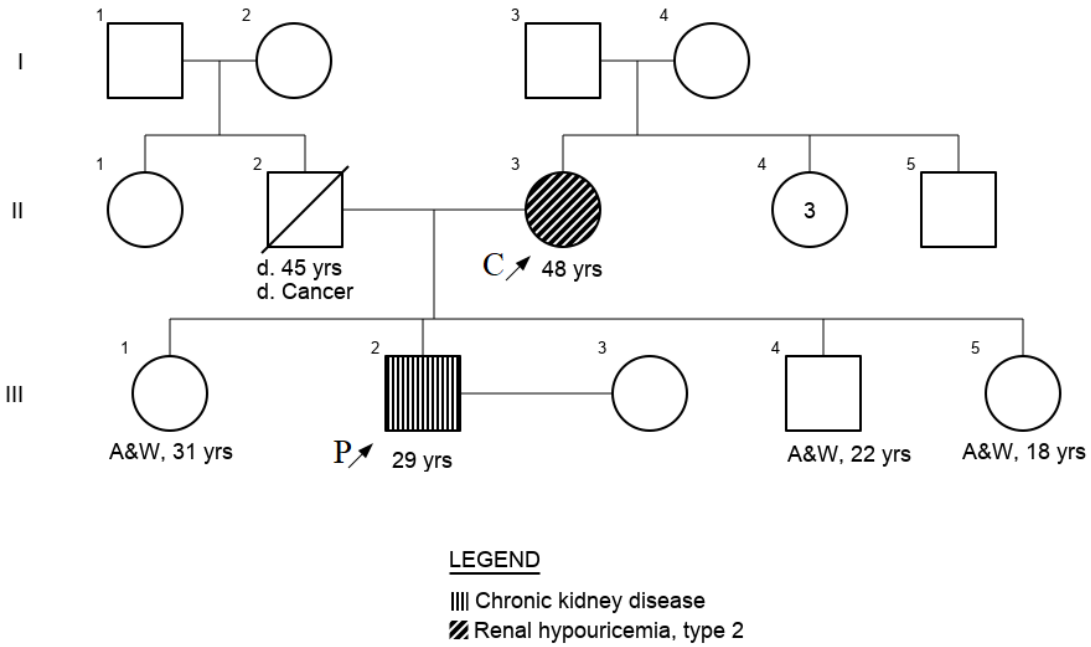
Donor

Gene (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
SLC2A9	Exon 8	c.1004T>A (p.Ile335Asn)	Likely compound Heterozygous	Renal hypouricemia-2 (OMIM#612076)	Autosomal recessive	Uncertain significance (PM2, PP3)
	Exon 9	c.1138C>T (p.Arg380Trp)				**Pathogenic** (PS4, PS3, PM2, PP3, PP5)

Patient

Gene (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
SLC2A9	Exon 8	c.1004T>A (p.Ile335Asn)	Heterozygous	Renal hypouricemia-2 (OMIM#612076)	Autosomal dominant; Autosomal recessive	Uncertain significance (PM2, PP3)
FN1	Exon 14	c.1944A>T (p.Lys648Asn)	Heterozygous	Glomerulopathy with fibronectin deposits 2 (OMIM#601894)	Autosomal dominant	Uncertain significance (PM2, PP3)

Figure S1. Pedigree chart



Supplementary references

S1. Matsuo H, Chiba T, Nagamori S, Nakayama A, Domoto H, Phetdee K, et al. Mutations in Glucose Transporter 9 Gene SLC2A9 Cause Renal Hypouricemia. Am J Hum Genet. 2008 Dec;83(6):744–51.

S2. Chiba T, Matsuo H, Nagamori S, Nakayama A, Kawamura Y, Shimizu S, et al. Identification of a Hypouricemia Patient with SLC2A9 R380W, A Pathogenic Mutation for Renal Hypouricemia Type 2. Nucleosides Nucleotides Nucleic Acids. 2014 Apr 4;33(4–6):261–5.