

SUPPLEMENTARY DATA

S1. Description of COL4A3-cIgAN families

Abbreviations: CADD, Combined Annotation Dependent Depletion; D, damaging; PP-2, Polyphen-2; gnomAD AF, allele frequency (AF) in gnomAD controls database

†Classification criteria are detailed in the Associated American College of Medical Genetics and Genomics (ACMG) guidelines (doi: 10.1038/gim.2015.30.)

Supplementary figure S1

Family	Gene	C.hgvs	P.hgvs	dbSNP ID	CADD	PP-2	SIFT	MutTaster	gnomAD AF (controls)	Supporting ACMG Criteria†
1	COL4A3	c.3829G>A	p.G1277S	rs190598500	23.5	D	D	D	0.00035	PM1;PM2;PP2;PP3
		c.4510T>C	p.F1504L	rs201671013	24.5	D	D	D	0.00024	PM1;PM2;PM3;PP2;PP3
2	COL4A3	c.3829G>A	p.G1277S	rs190598500	23.5	D	D	D	0.00035	PM1;PM2;PP1;PP2;PP3
3	COL4A3	c.4421T>C	p.L1474P	rs200302125	23.4	D	D	D	0.0023	PM1;PM2;PP2;PP3;PP5
4	COL4A3	c.4510T>C	p.F1504L	rs201671013	24.5	D	D	D	0.00024	PM1;PM2;PP2;PP3