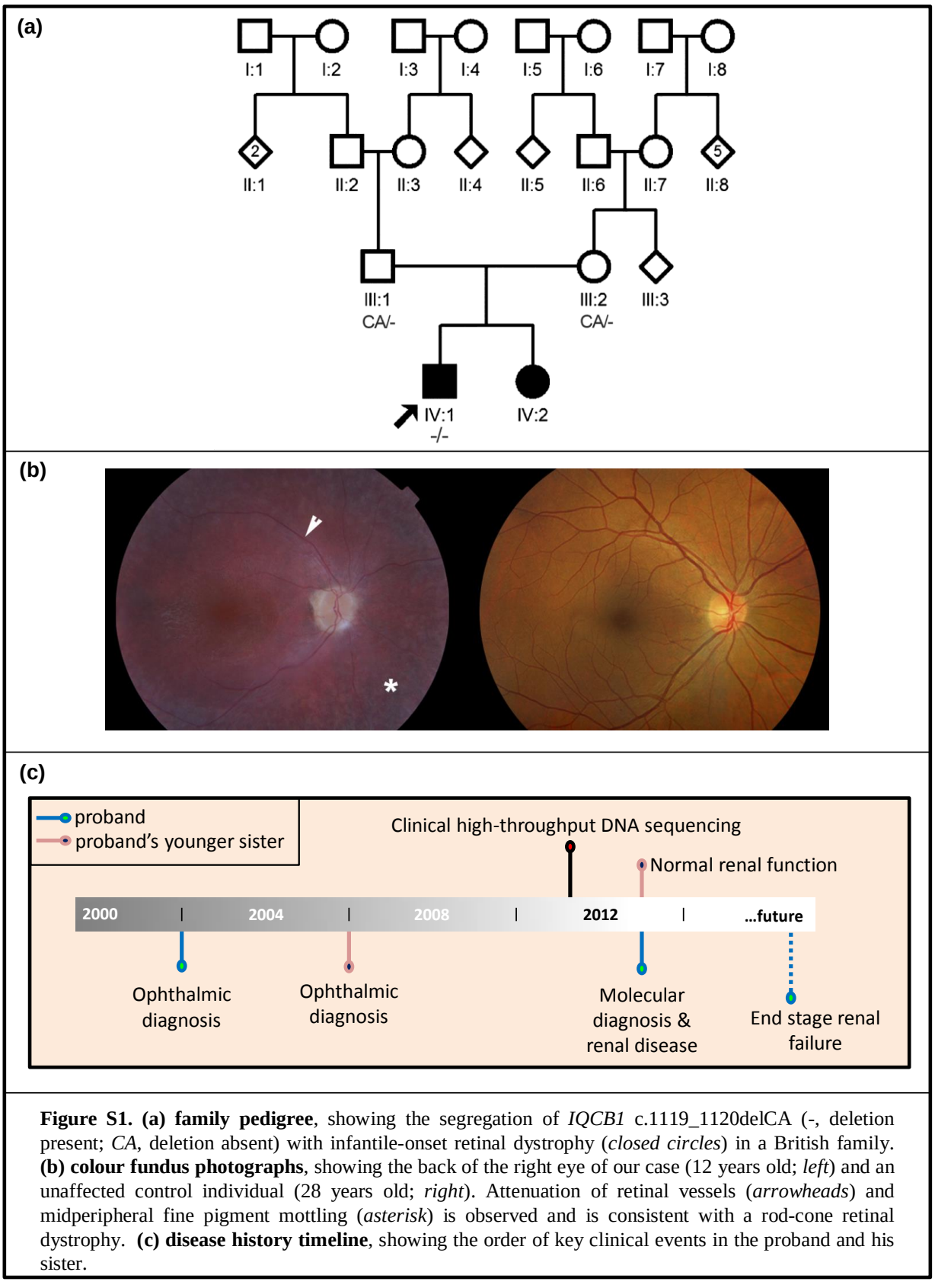


Figure S1. (a) family pedigree, showing the segregation of *IQCB1* c.1119_1120delCA (-, deletion present; *CA*, deletion absent) with infantile-onset retinal dystrophy (*closed circles*) in a British family. **(b) colour fundus photographs**, showing the back of the right eye of our case (12 years old; *left*) and an unaffected control individual (28 years old; *right*). Attenuation of retinal vessels (*arrowheads*) and midperipheral fine pigment mottling (*asterisk*) is observed and is consistent with a rod-cone retinal dystrophy. **(c) disease history timeline**, showing the order of key clinical events in the proband and his sister.

Table S1. Specified transcripts for 105 genes analysed in the proband's DNA after referral to the clinical molecular diagnostic service for inherited retinal disease.

HGNC	RefSeq Transcript ID	HGNC	RefSeq Transcript ID	HGNC	RefSeq Transcript ID
ABCA4	NM_000350.2	PRPF31	NM_015629.3	IMPG2	NM_016247.3
ADAM9	NM_003816.2	PRPF6	NM_012469.3	KCNV2	NM_133497.3
AIPL1	NM_014336.3	PRPF8	NM_006445.3	KLHL7	NM_001031710.2
ARL6	NM_032146.3	PRPH2	NM_000322.4	IMPG2	NM_016247.3
BBS1	NM_024649.4	RAX2	NM_032753.3	KCNV2	NM_133497.3
BBS10	NM_024685.3	RBP3	NM_002900.2	KLHL7	NM_001031710.2
BBS12	NM_001178007.1	RD3	NM_183059.2	LCA5	NM_181714.3
BBS2	NM_031885.3	RDH12	NM_152443.2	LRAT	NM_004744.3
BBS4	NM_033028.3	RDH5	NM_001199771.1	LRP5	NM_002335.2
BBS5	NM_152384.2	RGR	NM_002921.3	MERTK	NM_006343.2
BBS7	NM_176824.2	RGS9	NM_001165933.1	MKKS	NM_018848.2
BBS7	NM_018190.3	RGS9	NM_003835.3	MKS1	NM_001165927.1
BBS9	NM_198428.2	RHO	NM_000539.3	MKS1	NM_017777.3
BEST1	NM_004183.3	RIMS1	NM_014989.4	MYO7A	NM_000260.3
C1QTNF5	NM_015645.3	RIMS1	NM_001168407.1	NDP	NM_000266.3
C2orf71	NM_001029883.1	CRB1	NM_201253.2	RIMS1	NM_001168410.1
CA4	NM_000717.3	CRX	NM_000554.4	RLBP1	NM_000326.4
CACNA2D4	NM_172364.4	DFNB31	NM_015404.3	ROM1	NM_000327.3
CDH23	NM_022124.5	DHDDS	NM_024887.2	RP1	NM_006269.1
CDHR1	NM_001171971.1	EFEMP1	NM_001039348.2	RP1L1**	NM_178857.5
CDHR1	NM_033100.2	ELOVL4	NM_022726.3	RP2	NM_006915.2
CEP290*	NM_025114.3	EYS	NM_001142800.1	RP9	NM_203288.1
CERKL	NM_001030311.2	FAM161A	NM_001201543.1	RPE65	NM_000329.2
CHM	NM_000390.2	FSCN2	NM_001077182.2	RPGR***	NM_001034853.1
CLRN1	NM_052995.2	FZD4	NM_012193.3	RPGRIP1	NM_020366.3
CLRN1	NM_001195794.1	GNAT2	NM_005272.3	RS1	NM_000330.3
CNGA1	NM_001142564.1	GPR98	NM_032119.3	SAG	NM_000541.4
CNGA3	NM_001298.2	GUCA1A	NM_000409.3	SEMA4A	NM_022367.3
CNGB1	NM_001297.4	GUCA1B	NM_002098.5	SNRNP200	NM_014014.4
CNGB3	NM_019098.4	GUCY2D	NM_000180.3	SPATA7	NM_018418.4
NR2E3	NM_014249.2	IDH3B	NM_006899.2	TEAD1	NM_021961.5
NRL	NM_006177.3	IDH3B	NM_174855.1	TIMP3	NM_000362.4
OTX2	NM_021728.2	IMPDH1	NM_000883.3	TOPORS	NM_005802.4
PCDH15	NM_001142763.1	IMPG2	NM_016247.3	TRIM32	NM_012210.3
PCDH15	NM_001142769.1	KCNV2	NM_133497.3	TTC8	NM_144596.2
PCDH15	NM_001142771.1	KLHL7	NM_001031710.2	TULP1	NM_003322.3
PCDH15	NM_001142770.1	LCA5	NM_181714.3	UNC119	NM_005148.3
PDE6A	NM_000440.2	LRAT	NM_004744.3	UNC119	NM_054035.2
PDE6B	NM_000283.3	LRP5	NM_002335.2	USH1C	NM_005709.3
PDE6C	NM_006204.3	MERTK	NM_006343.2	USH1C	NM_153676.3
PDE6G	NM_002602.3	MKKS	NM_018848.2	USH1G	NM_173477.2
PITPNM3	NM_031220.3	MKS1	NM_001165927.1	USH2A	NM_206933.2
PRCD	NM_001077620.2	MKS1	NM_017777.3	ZNF513	NM_144631.5
PROM1	NM_006017.2	MYO7A	NM_000260.3		
PRPF3	NM_004698.2	NDP	NM_000266.3		

* Testing of the intronic mutation c.2991+1655A>G in the *CEP290* gene is included in this analysis

** Analysis of the coding region of exon 4 of the *RP1L1* gene is not included

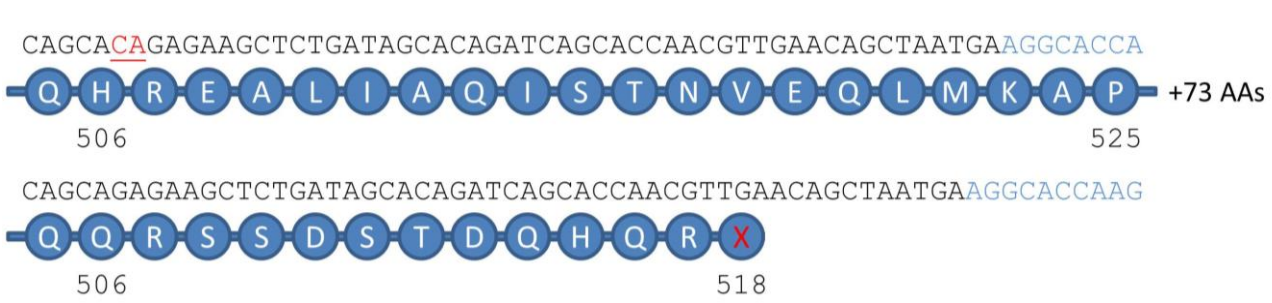
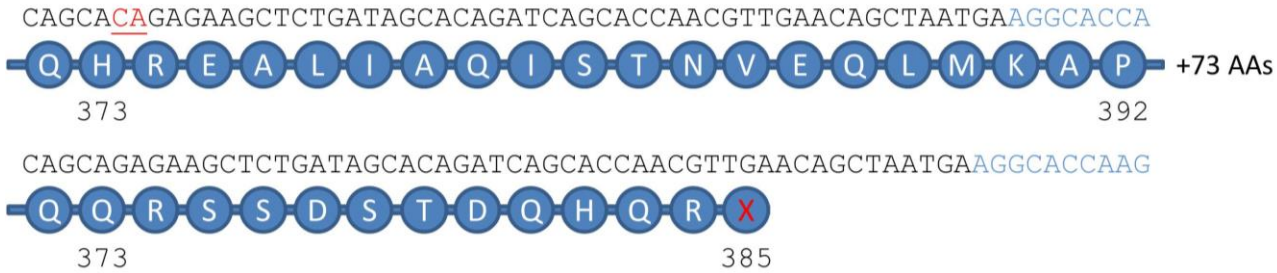
*** Analysis of the coding region of the final exon (*orf15*) of the *RPGR* gene is not included

Table S2. Selected previously reported cases with the homozygous two nucleotide deletion in *IQCB1* (chr3:121491452_121491453delCA, *hg19*) detected in the proband's DNA sample during the analysis of whole exome sequencing data.

Origin	Current age	Parent consanguinity	Affected siblings	Age of RD	Age of ESRD	Reference
British	13	No	Yes	2 months	kidney dysfunction detected before ESRD *expected within 6 months*	This study
USA	-	Not reported	Not reported	Present, age not reported	15 yrs	[7]
Brazil	-	Not reported	Not reported	Present, age not reported	12 yrs	[7]
Germany	-	Not reported	Not reported	Present, age not reported	>10 yrs	[7]
USA	-	Not reported	Not reported	Present, age not reported	7 yrs	[7]
Germany	-	No	Yes	<2 months	12 yrs	[8]
Germany	-	No	Not reported	<2 months	<24 yrs	[8]
Germany	-	No	Not reported	Not reported	10 yrs	[8]
Germany	-	No	Not reported	<6 months	15 yrs	[8]
USA	-	Not reported	Not reported	<6 months	No manifest renal disease at 7 yrs	[9]

Abbreviations: *RD*, retinal disease; *ESRD*, end stage renal disease.

Table S3. Predicted consequence of the *IQCB1* mutation found in the proband's DNA sample in all transcripts for the *IQCB1* gene included in the National Centre for Biotechnology Information Reference Sequence database (NCBI RefSeq). For each transcript the bases deleted from the reference genetic sequence (*top*) in the proband's DNA sample (*bottom*) is indicated by red underlined text. The alteration between black and blue text indicates a change in coding exons. Numerical values refer to the relative position of amino acids (single letter amino acid codes included within solid circles: *X*, stop codon) within the primary structure of the *IQCB1* protein.

RefSeq ID	coding region co-ordinates (*hg19*)	coding region size	number of coding region exons	Variant annotation	Exonic location of variant	Expected premature termination
NM_001023570	chr3:121,489,192-121,547,807	58,616	13	c.1518_1519delCA p.His506Glnfs*13	12	in exon 12
						
NM_001023571	chr3:121,489,192-121547,807	58,616	10	c.1119_1120delCA p.His373Glnfs*13	9	in exon 8
						

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