

Isolated_LCA	AIPL1	ALMS1	C2ORF71	C8ORF37	CABP4	CCT2	CEP290	CLUAP1	
	CNGA3	CNGB3	CRB1	CRX	CWC27	CYP1A1	CYP4V2	GUCY2D	
	IFT140	IMPDH1	IQCB1	KCNJ13	KCNV2	KRT12	LCA5	LRAT	
	MERTK	MPDZ	NMNAT1	NR2E3	OFD1	OTX2	PDE6G	PRPH2	
	RD3	RDH12	RPE65	RPGRIP1	SPATA7	TULP1			
Syndromic_LCA	AHI1	ALMS1	ARL13B	C21ORF2	C5ORF42	CC2D2A	CEP104	CEP164	
	CEP290	CEP41	CSPP1	IFT140	INPP5E	INVS	IQCB1	KIF7	
	LCA5	NPHP1	NPHP3	NPHP4	POC1B	RPGRIP1L	SDCCAG8	TMEM138	
	TMEM216	TMEM237	TTC8	TUBB4B	VPS13B	WDR19	ZNF423		
Differential_Diagnosis_LCA	ATF6	CACNA1F	CACNA1F	CCT2	CLN1/PPT1	CLN2/TPP1	CLN3	CLUAP1	
	CNGA3	CNGB3	GNAT1	GNAT2	GPR179	GRM6	LRIT3	NYX	
	PDE6C	PDE6H	PRPH2	SLC24A1	SLC38A8	TRPM1			
Rod_Cone	ABCA4	ABHD12	ADIPOR1	AGBL5	AHI1	ARL2BP	ARL6	BBS1	
	BBS2	BBS5	BBS12	BEST1	C2ORF71	C8ORF37	C21ORF2	CA4	
	CACNA1F	CC2D2A	CEP250	CEP290	CEP78	CERKL	CHM	CLN3	
	CLRN1	CNGA1	CNGB1	CRB1	CRX	CWC27	CYP4V2	DHDDS	
	DHX38	EXOSC2	EYS	FAM161A	FLVCR1	FSCN2	GNAT1	GNPTG	
	GPR125	GUCA1B	HGSNAT	HK1	IDH3A	IDH3B	IFT43	IFT140	
	IFT172	IMPDH1	IMPG2	IQCB1	ITM2B	KIAA1549	KIZ	KLHL7	
	LAMA1	LRAT	MAK	MERTK	MFRP	MFSD8	MPDZ	NEK2	
	NEUROD1	NPHP4	NR2E3	NRL	OFD1	OPN1SW	OR2W3	PCYT1A	
	PDE6A	PDE6B	PDE6G	PIK3R4	PITPNM3	POMGNT1	PRCD	PROM1	
	PRPF3	PRPF31	PRPF4	PRPF6	PRPF8	PRPH2	RBP3	RCBTB1	
	RDH11	RDH12	REEP6	RGR	RHO	RIMS1	RIMS2	RLBP1	
	ROM1	RP1	RP1L1	RP2	RP9	RPE65	RPGR	RPGRIP1	
	SAG	SEMA4A	SLC7A14	SLC24A1	SNRNP200	SPATA7	SRD5A3	TOPORS	
	TTC8	TLL5	TTPA	TUB	TULP1	USH2A	WDR19	ZNF408	
	ZNF513								
	Cone_Rod	ABCA4	ABHD12	ACBD5	ADAMTS18	ADAM9	AIPL1	ALMS1	ATF6
		BBS1	BBS5	C2ORF71	C8ORF37	C21ORF2	CABP4	CACNA2D4	CDHR1
		CEP78	CERKL	CNGA3	CNGB3	CNNM4	CRX	FAM161A	GNAT2
		GUCA1A	GUCY2D	KCNV2	LCA5	NMNAT1	NR2E3	PCYT1A	PDE6C
PDE6H		PITPNM3	POC1B	PROM1	PRPH2	RAB28	RASSF8	RAX2	
RDH5		RDH12	RIMS1	RIMS2	RPE65	RPGR	RPGRIP1	SEMA4A	
SPATA7		SRD5A3	TLL5	TULP1	UNC119	USH2A			
Macular_Dystrophy	ABCA4	ATXN7	BEST1	C1QTNF5	CDH3	CNGA3	CRB1	DRAM2	
	EFEMP1	ELOVL4	FSCN2	GUCA1A	GUCA1B	IMPG1	IMPG2	MFSD8	
	PROM1	PRPH2	RLBP1	RP1L1	RPGR	TIMP3			
Achromatopsia	ATF6	CACNA1F	CLUAP1	CNGA3	CNGB3	GNAT2	NDP	PDE6C	
	PDE6H								
Dyschromatopsies	OPN1LW	OPN1MW	OPN1SW						
CSNB	CABP4	CACNA1F	GNAT1	GNB3	GRK1	GPR179	GRM6	HK1	
	LRIT3	NYX	PDE6B	RHO	SAG	SLC24A1	TRPM1		
RS	RS1	CRB1							
Choroideremia	CHM								
Others	PAX2	RBP4	GPATCH11						

Figure S1. Panel of 199 genes involved in retinal dystrophies. LCA: Leber congenital amaurosis; CSNB: congenital stationary night blindness; RS: retinoschisis.

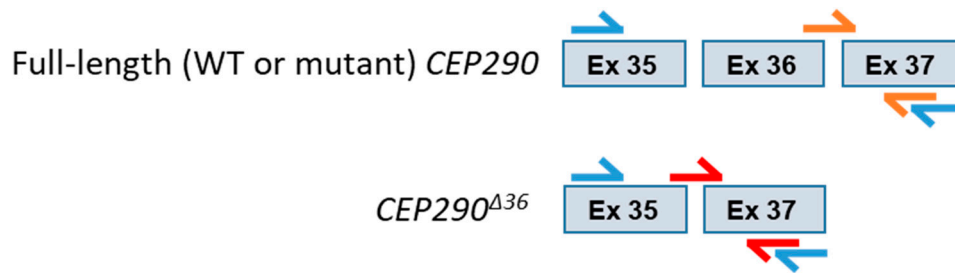


Figure S2. Localization of primers used to amplify the full-length and *CEP290* mRNAs lacking exon 36. In blue, RT-PCR primer pairs used to amplify the full-length and skipped *CEP290* transcripts. In orange, RT-qPCR primer pairs used to amplify the full-length (wild-type, WT or Mutant) *CEP290* transcript. In red, RT-qPCR primer pairs used to amplify the *CEP290* transcript deleted of exon 36 (*CEP290*^{Δ36}), respectively.

Table S1. Sequences and positions of sequencing primers.

Targeted region	Couple	Position	Sequence (5'>3')
<i>CEP290</i> Exon 36	<i>forward</i>	Intron 35	ACAATTTAAGATATAGTTTCG
	<i>reverse</i>	Intron 36	AACAAAAAGGGTAACTTC

Table S2. Genetic and clinical features of individuals.

Individual	Cell line	Gender	Age* (years)	<i>CEP290</i> mutations	Ocular phenotype
Control 1	C1	Female	8	None	No overt pathology
Control 2	C2	Male	10	None	No overt pathology
Control 3	C3	Female	13	None	No overt pathology
Patient 1	P1	Male	34	c.4723A>T (p.Lys1575*) c.4723A>T (p.Lys1575*)	<i>EOSRD</i>
Patient 2	P2	Male	24	c.4723A>T (p.Lys1575*) c.4723A>T (p.Lys1575*)	<i>LCA</i>

EOSRD: Early onset and severe retinal dystrophy; LCA: Leber congenital amaurosis

*age at which the dermal biopsy and the last clinical examination were performed.

Table S3. Sequences and positions of RT-PCR primers.

Targeted region	Couple	Position	Sequence (5'>3')
<i>CEP290</i> Exon 36	<i>forward</i>	Exon 35	CCACTGCAGAAAGAGAAAAGC
	<i>reverse</i>	Exon 37	TTAGTTTGACCAAGAGTGAGGAA

Table S4. Sequences and positions of RT-qPCR primers. "Full-length" refers to the wild-type or mutant *CEP290* transcript, whereas *CEP290*^{Δ36} refers to *CEP290* transcript deleted of exon 36.

Targeted region	Couple	Position	Sequence (5'>3')
« full-length » <i>CEP290</i> (exon 36)	<i>forward</i>	Jonction exon 36/exon 37	AACAAACGGCTTGGGATTTAATGA
	<i>reverse</i>	Exon 37	TTTGACCAAGAGTGAGGAAAGAGA
<i>CEP290</i> ^{Δ36}	<i>forward</i>	Jonction exon 35/exon 37	AAAGCCAGAGAGGATTTAATGAAAC
	<i>reverse</i>	Exon 37	TTGACCAAGAGTGAGGAAAGAG