

## Supplementary Information

**Title: The correlation between *CRBI* variants and the clinical severity of Brazilian patients with different inherited retinal dystrophy phenotypes**

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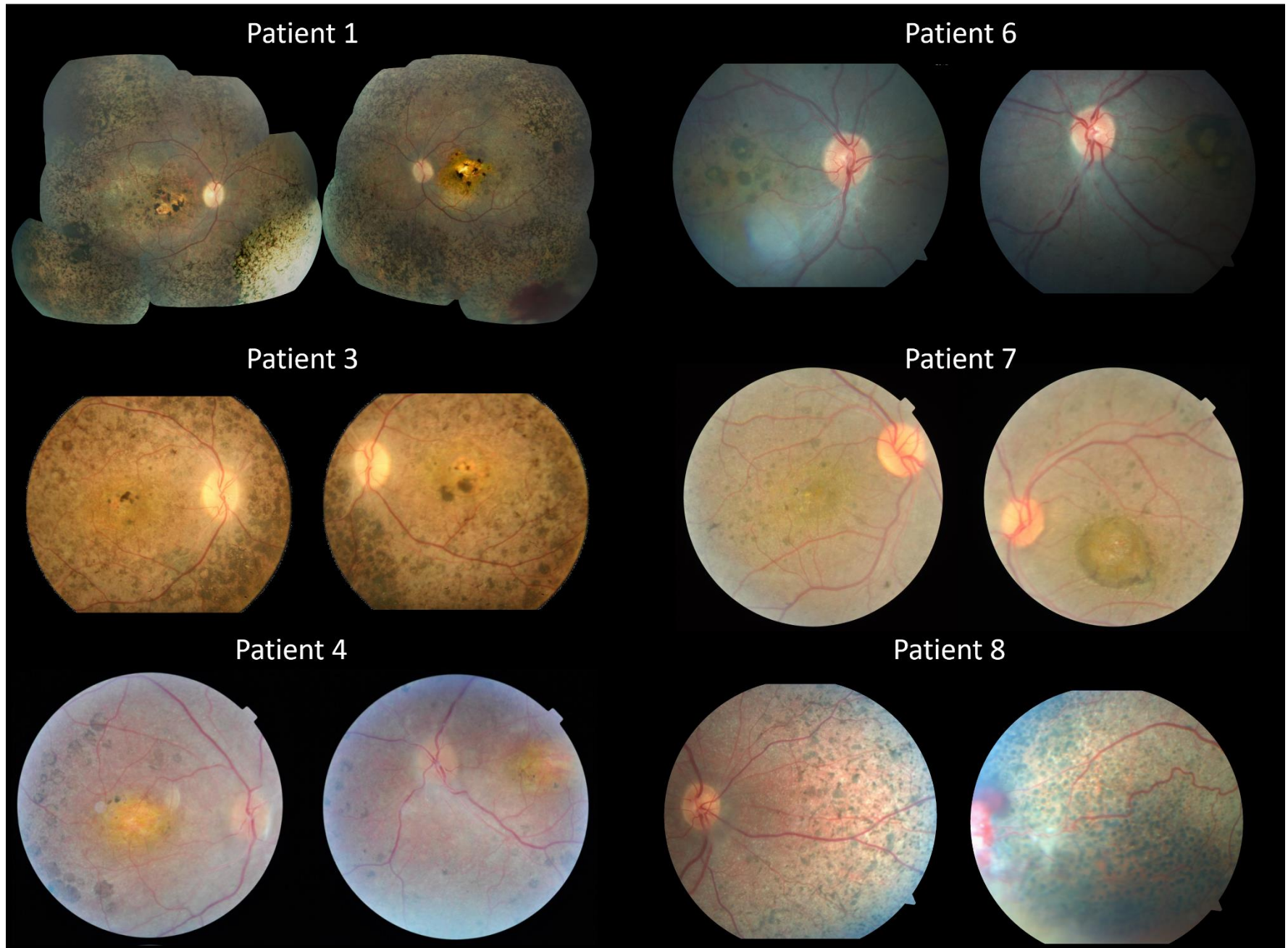
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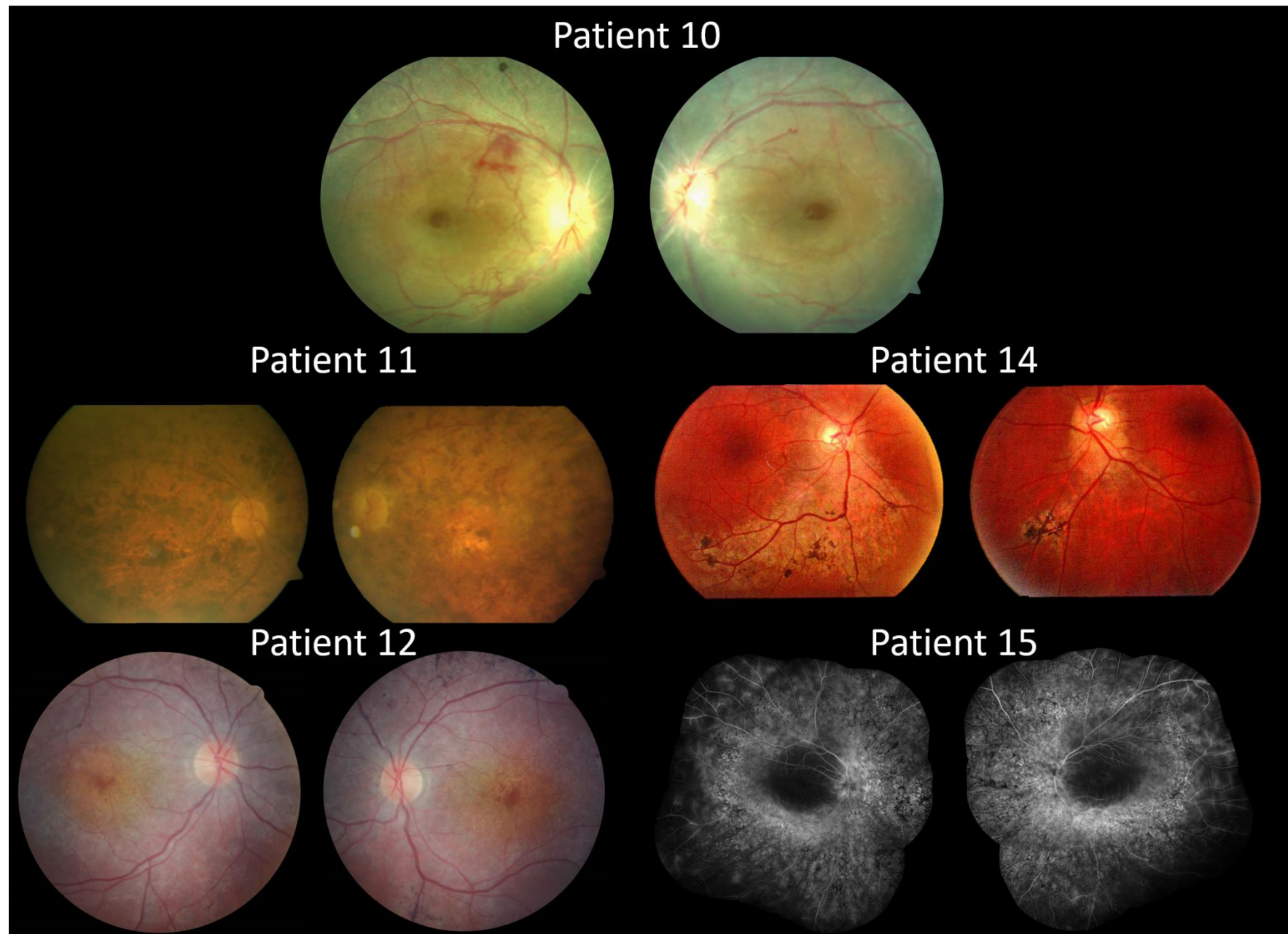
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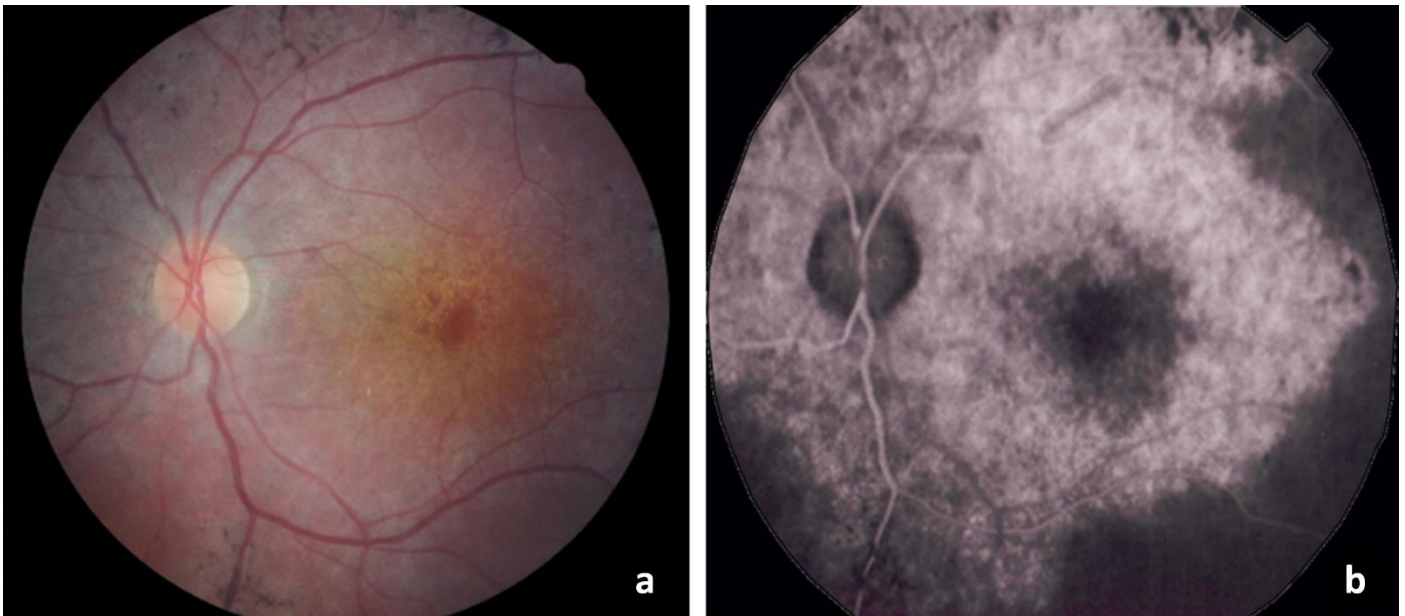
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**Supplementary Figure S1. Fundus appearance of LCA patients.**



**Supplementary Figure S2. Fundus appearance of EORD (10), CRD (11 and 12) and RP (14 and 15) patients.**



**Supplementary Figure S3. Fundus appearance of patient 12. (A) Color fundus photograph showing macular atrophy. (B) Fluorescein angiography showing hyperfluorescence of posterior pole and alteration of RPE pigmentation.**

PolymiRTS Database 3.0 - <http://compbio.uthsc.edu/miRSNP> (Bhattacharya et al. 2014)

RefSeq ID: [NM\\_001193640](#)  
 Gene Symbol: CRB1  
 Description: Homo sapiens crumbs homolog 1 (Drosophila) (CRB1), transcript variant 2, mRNA.  
 Gene Location: chr1(+):197237333-197447585

SNPs and INDELs in miRNA target sites

Location	dbSNP ID	Variant type	Wobble base pair	Ancestral Allele	Allele	miR ID	Conservation	miRSite	Function Class	Exp Support	context+ score change
197447021	<a href="#">rs199582748</a>	SNP	N	C	C	<a href="#">hsa-miR-4475</a>	3	ttgtGCCCTTcg	D	N	No Change
					C	<a href="#">hsa-miR-7515</a>	3	ttgtGCCCTTcg	D	N	No Change
					T	<a href="#">hsa-miR-7845-5p</a>	4	ttgtGCCCTTcg	D	N	No Change
					T	<a href="#">hsa-miR-1252-5p</a>	3	ttgtGCCCTTcg	C	N	No Change
197447037	<a href="#">rs41302107</a>	SNP	N	T	T	<a href="#">hsa-miR-3691-5p</a>	5	tggggATCCACAc	D	N	-0.112
					T	<a href="#">hsa-miR-376b-5p</a>	5	tggggATCCACAc	D	N	-0.123
					T	<a href="#">hsa-miR-376c-5p</a>	5	tggggATCCACAc	D	N	-0.123
					T	<a href="#">hsa-miR-3911</a>	5	tggggATCCACAc	D	N	-0.234
					T	<a href="#">hsa-miR-4752</a>	5	tggggATCCACAc	D	N	-0.109
					T	<a href="#">hsa-miR-8056</a>	5	tggggATCCACAc	D	N	-0.111
					C	<a href="#">hsa-miR-6748-5p</a>	5	tggggACCCACAc	C	N	-0.134
					C	<a href="#">hsa-miR-6759-5p</a>	5	tggggACCCACAc	C	N	-0.167
					C	<a href="#">hsa-miR-6793-5p</a>	5	tggggACCCACAc	C	N	-0.144

Function Class: **D**: The derived allele disrupts a conserved miRNA site (ancestral allele with support >= 2).  
**C**: The derived allele creates a new miRNA site.

TargetScan (v7.0) - [www.targetscan.org](http://www.targetscan.org) (Agarwal et al. 2015)

**Poorly conserved**

	Predicted consequential pairing of target region (top) and miRNA (bottom)	Site type	Context++ score	Context++ score percentile	Weighted context++ score	Conserved branch length	PCT
Position 18-29 of CRB1 3' UTR	5' ...UCCUUCGAGAUAGGGG-4UCCAC... 3' GACUUUCUCUACCCCUACUCG	non-canonical	N/A	N/A	0.00	0	N/A
Position 18-29 of CRB1 3' UTR	5' ...UCCUUCGAGAUAGGGG-4UCCAC... 3' GACUUUCUCUACCCCUACUCG	non-canonical	N/A	N/A	0.00	0	N/A
Position 27-33 of CRB1 3' UTR	5' ...CCCUUCGAGAUAGGGG-4UCCAC... 3' UUGUAUCUCCUUAUAGGUGC	7mer-A1	-0.16	76	-0.16	0.031	N/A
Position 27-33 of CRB1 3' UTR	5' ...CCCUUCGAGAUAGGGG-4UCCAC... 3' UUGUAUCUCCUUAUAGGUGC	7mer-A1	-0.11	76	-0.11	0.031	N/A
Position 27-33 of CRB1 3' UTR	5' ...CCCUUCGAGAUAGGGG-4UCCAC... 3' CAUGGCUCAGAGGUAGUAGGUGA	7mer-A1	-0.16	75	-0.16	0	N/A
Position 27-33 of CRB1 3' UTR	5' ...CCCUUCGAGAUAGGGG-4UCCAC... 3' UACGUAGGUCUGUAGGUGC	7mer-A1	-0.07	59	-0.07	0	N/A
Position 27-33 of CRB1 3' UTR	5' ...CCCUUCGAGAUAGGGG-4UCCAC... 3' UCGUGUAGGAACUCUAGGUGUU	7mer-m8	-0.02	29	-0.02	0	N/A
Position 28-35 of CRB1 3' UTR	5' ...CCCUUCGAGAUAGGGG-4UCCAC... 3' ACGGAGGAGGUCCUAGGUGUGU	8mer	-0.20	88	-0.20	0	N/A

**Supplementary Figure S4. Result of miRNA in silico analysis.**