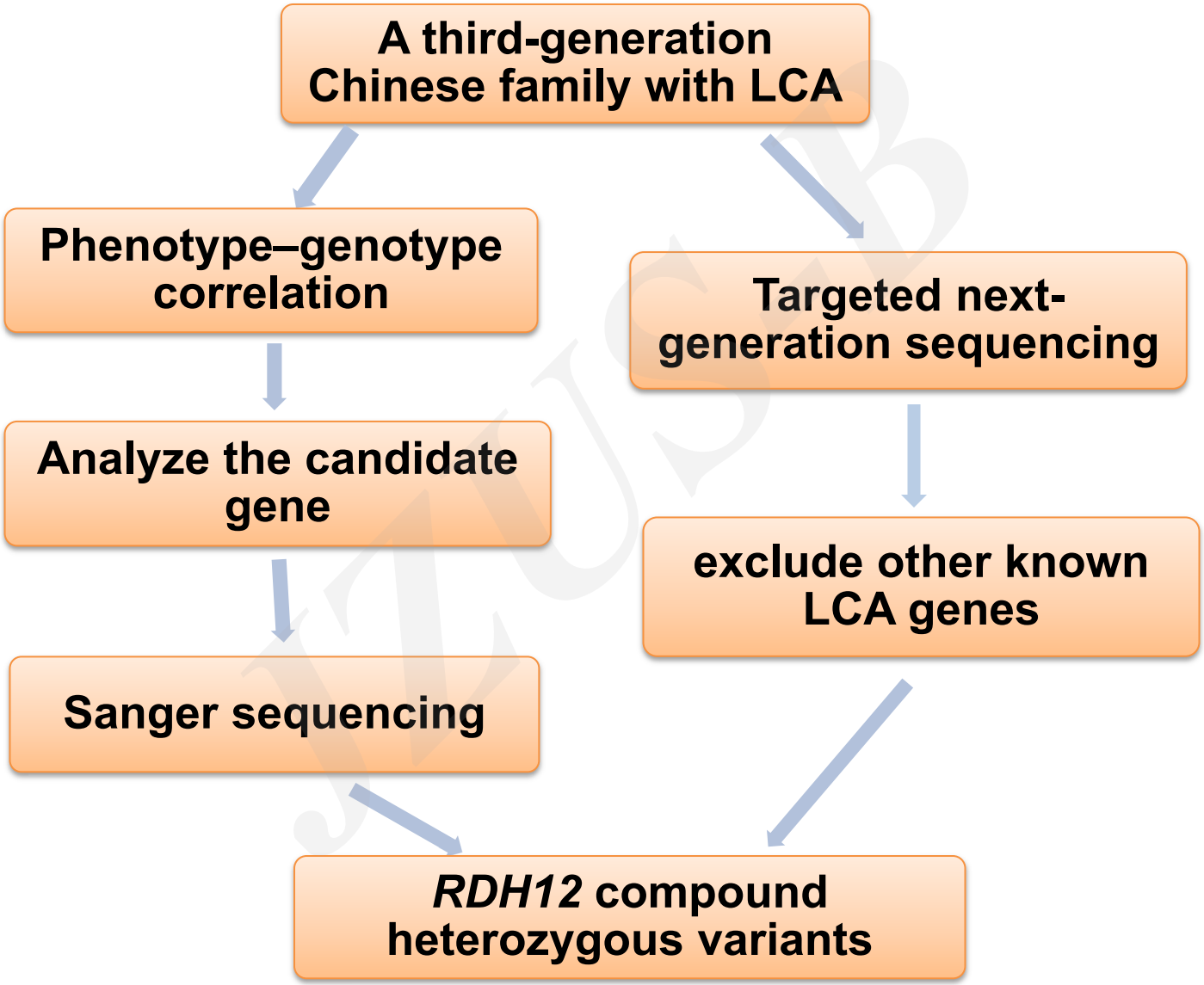
Cite this as: Yun LI, Qing PAN, Yang-shun GU, 2017. Phenotype-genotype correlation with Sanger sequencing identified retinol dehydrogenase 12 (*RDH12*) compound heterozygous variants in a Chinese family with Leber congenital amaurosis. *Journal of Zhejiang University-Science B (Biomedicine & Biotechnology)*, 18(5):421-429. http://dx.doi.org/10.1631/jzus.B1600156

Phenotype-genotype correlation with Sanger sequencing identified retinol dehydrogenase 12 (*RDH12*) compound heterozygous variants in a Chinese family with Leber congenital amaurosis

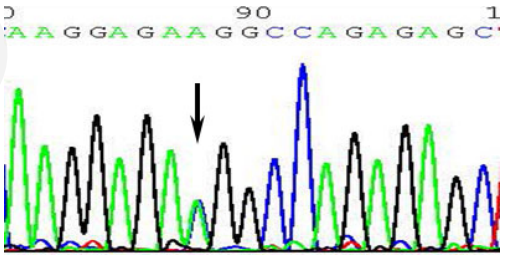
Keywords: Leber congenital amaurosis (LCA), Phenotype-genotype correlations, *RDH12*, Compound heterozygosity

Research Summery

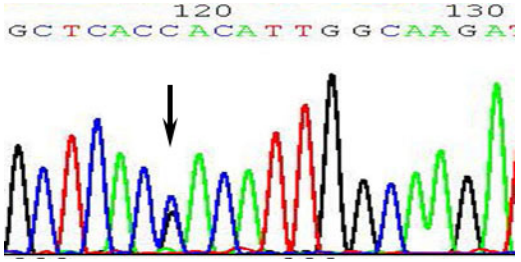


Innovation points

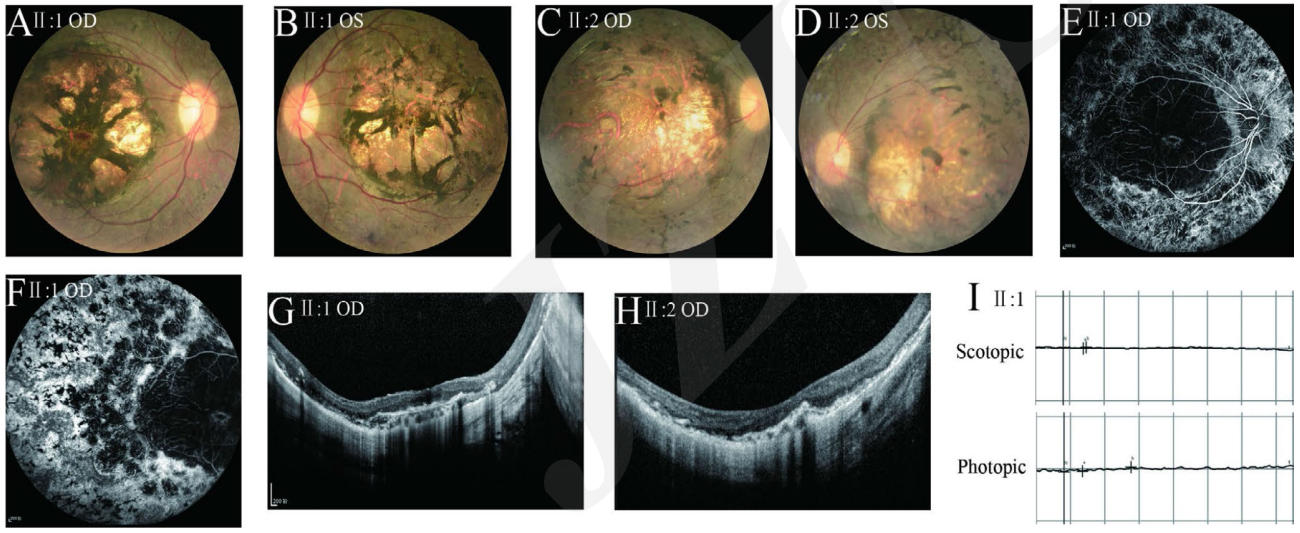
- Based on phenotype–genotype correlation, *RDH12* compound heterozygous variants were identified by Sanger sequencing



c.164C>A



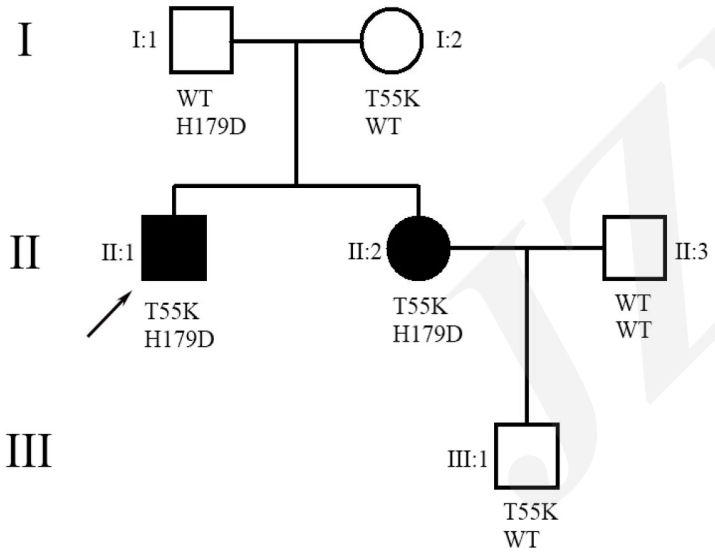
c.535C>G



Ocular phenotype of affected patients

Innovation points

- Targeted next-generation sequencing confirmed the *RDH12* compound heterozygous variants
- The phenotype of the family showed complete cosegregation of the variants with LCA
- Neither of the *RDH12* variants was detected in the 600 normal controls



Gene	Variants	Het/Hom	II:1	II:2
RDH12	p.T55K	Het	+	+
RDH12	p.H179D	Het	+	+
CLRN1	p.G136E	Het	+	+
MITF	p.D491Y	Het	+	+
PROM1	p.E728K	Het	–	+

Candidate variants identified by targeted next-generation sequencing

ZJUS-B