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Supplemental Data

Mutations in *C12orf57* Cause a Syndromic Form of Colobomatous Microphthalmia

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Figure S1 Family 1 3 affected individuals Parents

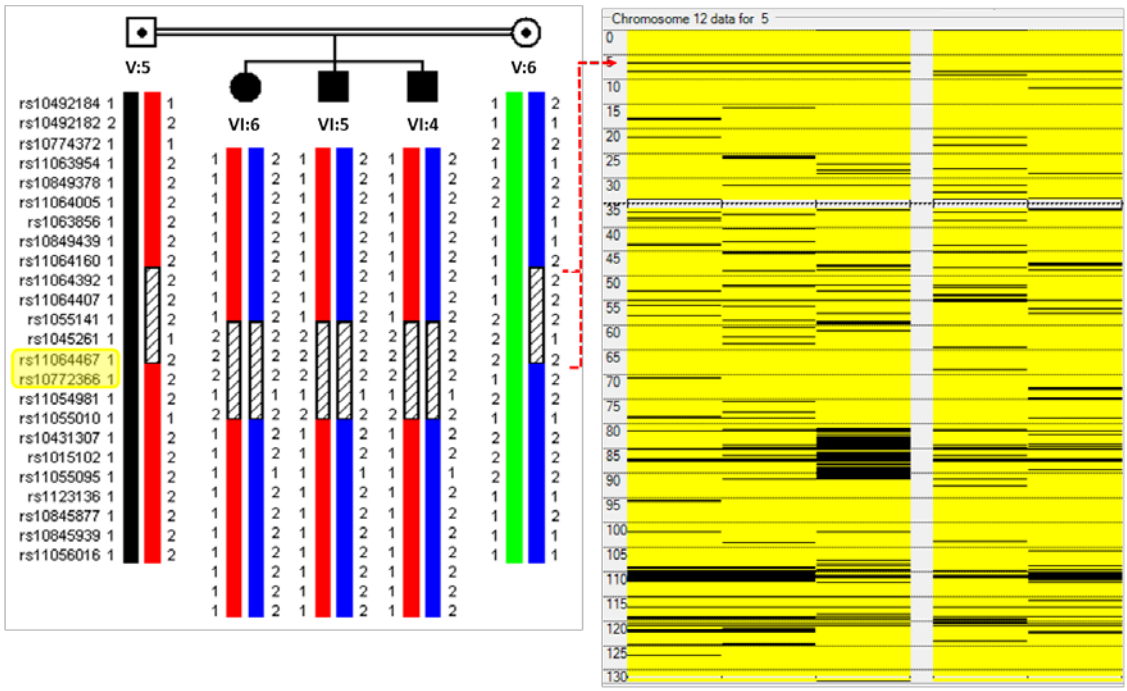

Figure S1. Autozygosity analysis of a family with a syndromic form of colobomatous microtphthalmia. Right: Homozygosity scan in Family 1 shows no apparent ROH at the *C12orf57* locus. Please note that each column represents one family member and each row represents a SNP on chromosome 12. Homozygous SNPs are shown in black and heterozygous in yellow. Left: Haplotype analysis revealed a very small ROH delimited by rs11064160 and rs10772366. *C12orf37* is flanked by rs11064467 and rs10772366 (highlighted in yellow).

Figure S2

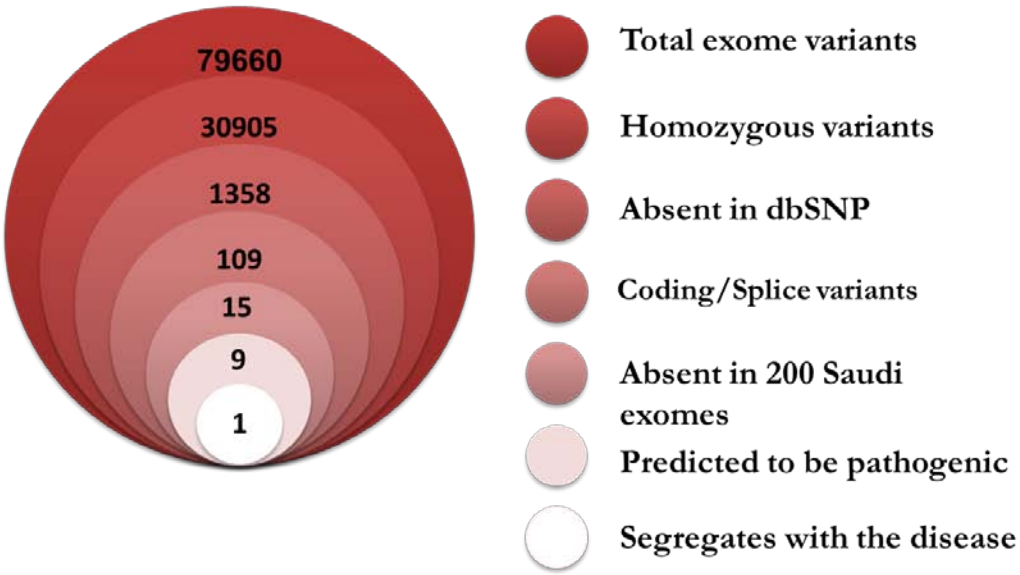

Figure S2. Filtration strategy used in analyzing exome data in Family 1.

Figure S3

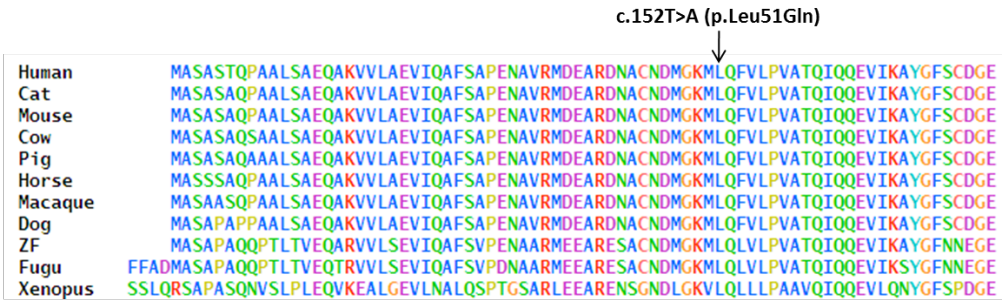

Figure S3. High level of conservation across species is shown for the Leu51 residue