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Supplemental Data

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

Fatema Zahrani, Mohammed A. Aldahmesh, Muneera J. Alshammari, Selwa Al-Hazzaa, and Fowzan S. Alkuraya

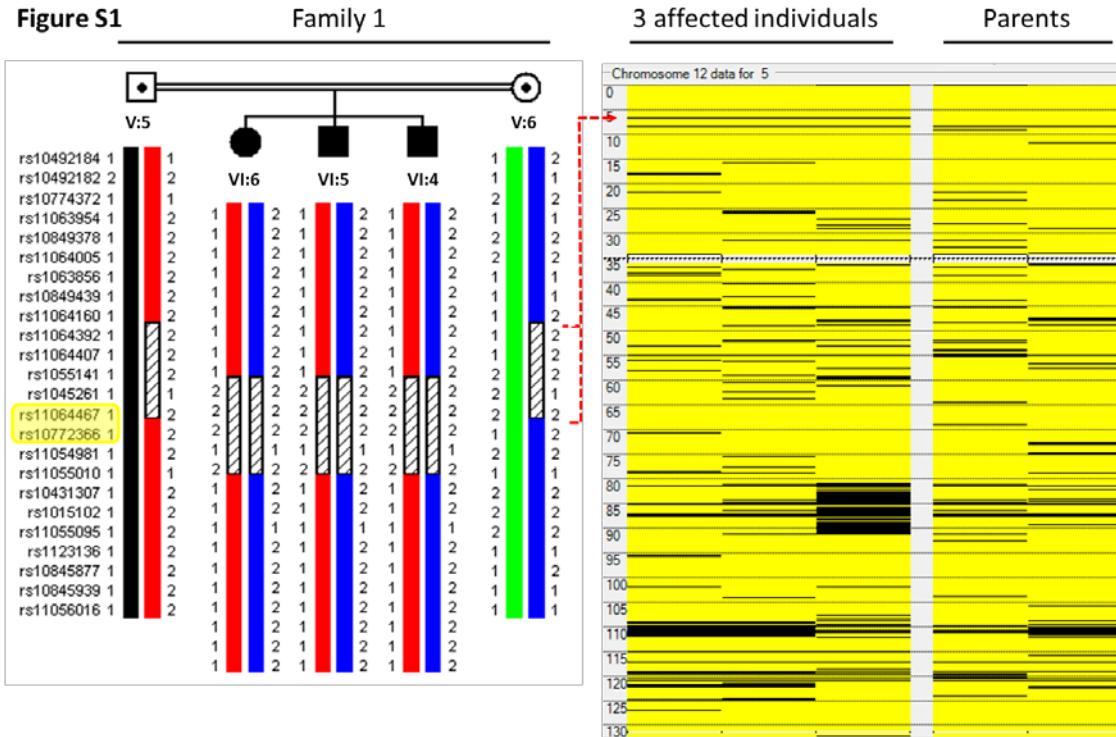
Figure S1

Figure S1. Autozygosity analysis of a family with a syndromic form of colobomatous microphthalmia. 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

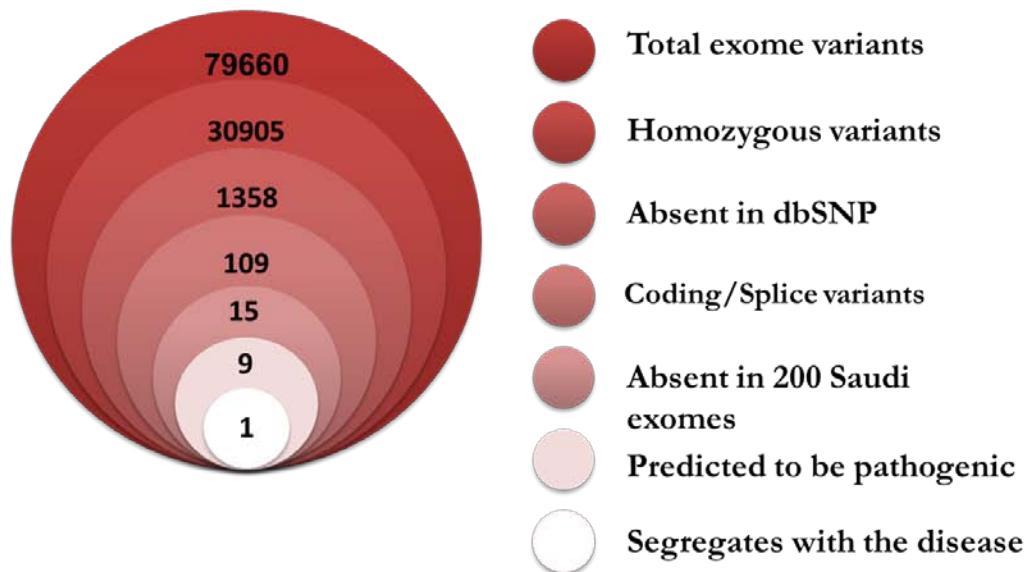


Figure S2. Filtration strategy used in analyzing exome data in Family 1.

Figure S3

	c.152T>A (p.Leu51Gln)
Human	MASASTQPAALSAEQAKVVLAEVIQAFSAPENAVRMDEARDNAACNDMGKMLQFVLPVATQIQQEVIKAYGFSCDGE
Cat	MASASAQPAAALSAEQAKVVLAEVIQAFSAPENAVRMDEARDNAACNDMGKMLQFVLPVATQIQQEVIKAYGFSCDGE
Mouse	MASASAQPAAALSAEQAKVVLAEVIQAFSAPENAVRMDEARDNAACNDMGKMLQFVLPVATQIQQEVIKAYGFSCDGE
Cow	MASASAQSAAALSAEQAKVVLAEVIQAFSAPENAVRMDEARDNAACNDMGKMLQFVLPVATQIQQEVIKAYGFSCDGE
Pig	MASASAQAALSAEQAKVVLAEVIQAFSAPENAVRMDEARDNAACNDMGKMLQFVLPVATQIQQEVIKAYGFSCDGE
Horse	MASSSAQPAALSAEQAKVVLAEVIQAFSAPENAVRMDEARDNAACNDMGKMLQFVLPVATQIQQEVIKAYGFSCDGE
Macaque	MASASAQPAAALSAEQAKVVLAEVIQAFSAPENAVRMDEARDNAACNDMGKMLQFVLPVATQIQQEVIKAYGFSCDGE
Dog	MASAPAPPAALSAEQAKVVLAEVIQAFSAPENAVRMDEARDNAACNDMGKMLQFVLPVATQIQQEVIKAYGFSCDGE
ZF	MASAPAQQPTLTVQARVVLSEVIQAFSVPENAAARMEEARESACNDMGKMLQLVLPVATQIQQEVIKAYGFNNEGE
Fugu	FFADMASAPAQQPTLTVQTRVVLSEVIQAFSVPDNAARMEEARESACNDMGKMLQLVLPVATQIQQEVIKSYGFNNEGE
Xenopus	SSLQRSPASQNVSPLLEQVKEALGEVNLALQSPPTGSARLEEARENSGNDLGKVQLLLLPAAVQIQQEVLQNYGFSPDGE

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