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Supplemental Data

Mutations in the SPARC-Related Modular
Calcium-Binding Protein 1 Gene, *SMOC1*,
Cause Waardenburg Anophthalmia Syndrome

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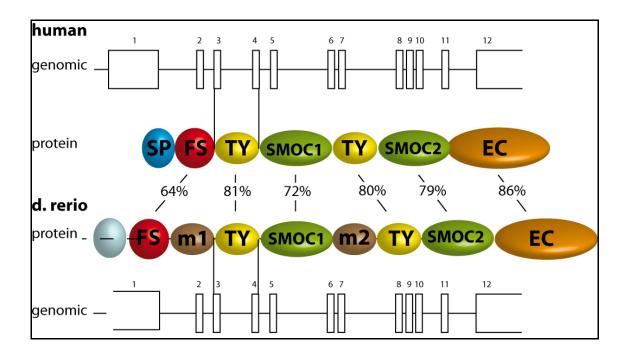


Figure S1. Structure of the Human and Zebrafish SMOC1 Genes and Proteins

Primers were designed from both ends of the zebrafish LOC795519, an entry that was recently removed from Genbank. An amplicon was amplified from RNA extracted from zebrafish embryos after reverse transcription. Sequencing of this clone showed two additional modules when compared to the Genbank zebrafish reported sequence and the human sequence: a first module of 27 amino acids (m1) and a second of 17 amino acids (m2). The overall alignment score between the human and zebrafish *SMOC1* gene was 63%. The percentage of amino acid identity between each domain is indicated in the figure. The identified zebrafish clone is probably missing the sequence of the signal peptide which, together with the two additional modules, reduces the overall alignment score.

Table S1. Summary of Genome-wide Homozygosity Mapping Regions Specific to the Two Patients

Chr	Begin		End		size	Number of homozygous
Chi					(nucleotides)	markers in
	marker	position	marker	position		interval
2	rs842362	135,057,590	rs7578528	136,506,753	1,449,163	165
14	rs11158442	62,198,792	rs4899594	76,031,821	13,833,029	3,123
20	rs6075750	20,953,246	rs6036158	22,529,060	1,575,814	249

(only regions outside of telomers, centromers and heterochromatin regions and larger than 1 Mb are shown)

Table S2. Primers and PCR Conditions for Human SMOC1

Exon	Forward primer (5'-3')	Reverse primer (5'-3')
1 2 3 4 5 6 7 8 9 10 11	ctcccattggctttcacct tccgttcccttcacctttag ggtggagtggttttctcagc cgtatgggtgatgctttgaa tgtggtcaagactgtttccag tgcccttttcttcttttgtttc gaagcctcaatgcctcagac aggcctggtccttgctactt catagatcattgctgggcagt ctggacaagaagggctttga ctttctgggggagtcatac ggatttctcacaagcccaac	catctctctcccctcctc caagcttccctgttgcaaat agaaagtgaaggcgccaag ggttctcccacaagatgcag ccccaggctccttctatctc cttaaacaaacatccaacgttaag gatttttccaaacccacaa actcatctcccaccagacca gaggagacagcttcttctgcat cactctctcaccctccac gcatagcacacttccagcaa tgccaatgcctgtacaaaag

Annealing was done at 58°C + betain for all pairs of primers