

Supplementary table 1 : Genotype of probands and parents.

Supplementary table 1 : Genotype of probands and parents.										
Proband			Mother			Father				
ID	Gender	Gene	Variant	Zygoty	Gene	Variant	Zygoty	Gene	Variant	Zygoty
1	M	TYR	c.575C>A: p.(Ser192Tyr) c.1205G>A: p.(Arg402Gln) c.1217C>T: p.(Pro406Leu) rs147546939 A > G	Heterozygous Heterozygous Heterozygous Heterozygous	TYR	c.1217C>T: p.(Pro406Leu)	Heterozygous	TYR	c.1205G>A: p.(Arg402Gln) c.575C>A: p.(Ser192Tyr) rs147546939 A > G	Heterozygous Heterozygous Heterozygous
2	F	SLC45A2 TYR	c.593G>A: (p.Gly198Asp) c.575C>A: p.(Ser192Tyr)	Homozygous Homozygous	SLC45A2 TYR	c.593G>A: (p.Gly198Asp) c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous	SLC45A2 TYR	c.593G>A: (p.Gly198Asp) c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous
3	F	TYR	c.238T>C: p.(Trp80Arg) c.575C>A: p.(Ser192Tyr) c.1205G>A: p.(Arg402Gln) rs147546939 A > G	Heterozygous Heterozygous Homozygous Heterozygous	TYR	c.575C>A: p.(Ser192Tyr) c.1205G>A: p.(Arg402Gln) rs147546939 A > G	Heterozygous Heterozygous Heterozygous	NP	NP	NP
4	M	NP	NP	NP	NP	NP	NP	NP	NP	NP
5	F	OCA2 OCA2 TYR	c.1324_1326del:p.(442_442del) c.1117-20A>G <sup>a</sup> c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous Heterozygous	OCA2	c.1324_1326del:p.442_442del	Heterozygous	NP	NP	NP
6	F	OCA2 TYR	c.1327G>A: (p.Val443Ile) c.575C>A: p.(Ser192Tyr)	Homozygous Homozygous	OCA2 TYR	c.1327G>A: (p.Val443Ile) c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous	OCA2 TYR	c.1327G>A: (p.Val443Ile) c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous
7	F	OCA2	c.1327G>A: (p.Val443Ile) Exon 10 to exon 18 deletion	Heterozygous Heterozygous	NP	NP	NP	NP	NP	NP
8	M	TYR	c.74dupT: p.(V25fs) c.575C>A: p.(Ser192Tyr) c.1205G>A: p.(Arg402Gln) rs147546939 A > G	Heterozygous Heterozygous Heterozygous Heterozygous	NP	NP	NP	NP	NP	NP
9	M	OCA2 TYR	c.2159G>A: p.(Arg720His) <sup>a</sup> c.575C>A: p.(Ser192Tyr) c.1205G>A: p.(Arg402Gln)	Heterozygous Heterozygous Heterozygous	NP	NP	NP	NP	NP	NP
10	M	OCA2 OCA2 TYR	c.1465A>G: (p.Asn489Asp) c.1239+5G>A c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous Homozygous	OCA2 TYR	c.1239+5G>A c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous	OCA2 TYR	c.1465A>G: (p.Asn489Asp) c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous
11	M	OCA2 OCA2 OCA2 TYR	c.1327G>A: (p.Val443Ile) c.2051T>G: p.(Phe684Cys) <sup>a</sup> c.2055delT: p.(F685fs) c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous Heterozygous Heterozygous	OCA2 OCA2 TYR	c.2051T>G: p.(Phe684Cys) c.2055delT: p.(F685fs) c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous Heterozygous	OCA2	c.1327G>A: (p.Val443Ile)	Heterozygous
12	F	NP	NP	NP	NP	NP	NP	NP	NP	NP
13	F	NP	NP	NP	NP	NP	NP	NP	NP	NP
14	F	NP	NP	NP	NP	NP	NP	NP	NP	NP
15	M	TYR	c.229C>T: p.(Arg77Trp) c.575C>A: p.(Ser192Tyr) c.1205G>A: p.(Arg402Gln)	Heterozygous Homozygous Heterozygous	TYR TYR	c.229C>T: p.(Arg77Trp) c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous	TYR	c.575C>A: p.(Ser192Tyr) c.1205G>A: p.(Arg402Gln) rs147546939 A > G	Heterozygous Heterozygous Heterozygous
16	M	OCA2 TYR	c.1327G>A: (p.Val443Ile) c.1465A>G: (p.Asn489Asp) c.575C>A: p.(Ser192Tyr)	Heterozygous Heterozygous Heterozygous	OCA2 TYR	c.1327G>A: (p.Val443Ile) c.575C>A: p.(Ser192Tyr)	Heterozygous Homozygous	OCA2 TYR	c.1465A>G: (p.Asn489Asp) c.1205G>A: p.(Arg402Gln)	Heterozygous Heterozygous

NP = not performed

<sup>a</sup>These three variants are novel OCA2 variants.

Accession IDs for transcripts:

TYR = NM\_000372

OCA2 = NM\_000275

SLC45A2 = NM\_016180