ONLINE SUPPLEMENTARY MATERIAL

Clinical and genetic variability in children with partial albinism

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Supplementary Table 1: Gene panels used in this study

Optic nerve disorders panel (40 genes; including foveal hypoplasia & ocular/oculocutaneous albinism panels)

Gene (HGNC)	Transcript
ACO2	NM_001098.2
AP3B1	NM_003664.4
ATOH7	NM_145178.3
BLOC1S3	NM_212550.4
C10orf11	NM_032024.3; NM_001305581.1
C12orf65	NM_152269.4
CACNA1F	NM_005183.3; NM_001256790.2
CISD2	NM_001008388.4
DHCR7	NM_001360.2
DTNBP1	NM_032122.4; NM_183040.2
FRMD7	NM_194277.2
GJA1	NM_000165.3
GPR143	NM_000273.2
HMX1	NM_018942.2
HPS3	NM_032383.4
HPS4	NM_022081.5; NM_152841.2
HPS5	NM_181507.1
HPS6	NM_024747.5
LARGE	NM_004737.4
LYST	NM_000081.3
OCA2	NM_000275.2
OPA1	NM_015560.2; NM_130837.2
OPA3	NM_025136.3; NM_001017989.2
PAX6	NM_001604.4
PLDN (BLOC1S6)	NM_001311255.1; NM_012388.3
RAB18	NM_021252.3
RAB3GAP1	NM_001172435.1
RAB3GAP2	NM_012414.3
RTN4IP1	NM_032730.4
SIX6	NM_007374.2
SLC24A5	NM_205850.2
SLC38A8	NM_001080442.1
SLC45A2	NM_016180.4; NM_001012509.3
SLC4A11	NM_032034; NM_001174089.1; NM_001174090.1
SOX2	NM_003106.3
TBC1D20	NM_144628.3
TMEM126A	NM_032273.3
TYR	NM_000372.4
TYRP1	NM_000550.2
WFS1	NM_006005.3

Foveal hypoplasia & nystagmus panel (26 genes; including ocular/oculocutaneous albinism panel)

Gene (HGNC)	Transcript
ACO2	NM_001098.2
AP3B1	NM_003664.4
ATOH7	NM_145178.3
BLOC1S3	NM_212550.4
C10orf11	NM_032024.3; NM_001305581.1
CACNA1F	NM_005183.3; NM_001256790.2
DTNBP1	NM_032122.4; NM_183040.2
FRMD7	NM_194277.2
GPR143	NM_000273.2
HMX1	NM_018942.2
HPS3	NM_032383.4
HPS4	NM_022081.5; NM_152841.2
HPS5	NM_181507.1
HPS6	NM_024747.5
LYST	NM_000081.3
OCA2	NM_000275.2
PAX6	NM_001604.4
PLDN (BLOC1S6)	NM_001311255.1; NM_012388.3
RTN4IP1	NM_032730.4
SIX6	NM_007374.2
SLC24A5	NM_205850.2
SLC38A8	NM_001080442.1
SLC45A2	NM_016180.4; NM_001012509.3
SLC4A11	NM_032034; NM_001174089.1; NM_001174090.1
TYR	NM_000372.4
TYRP1	NM_000550.2

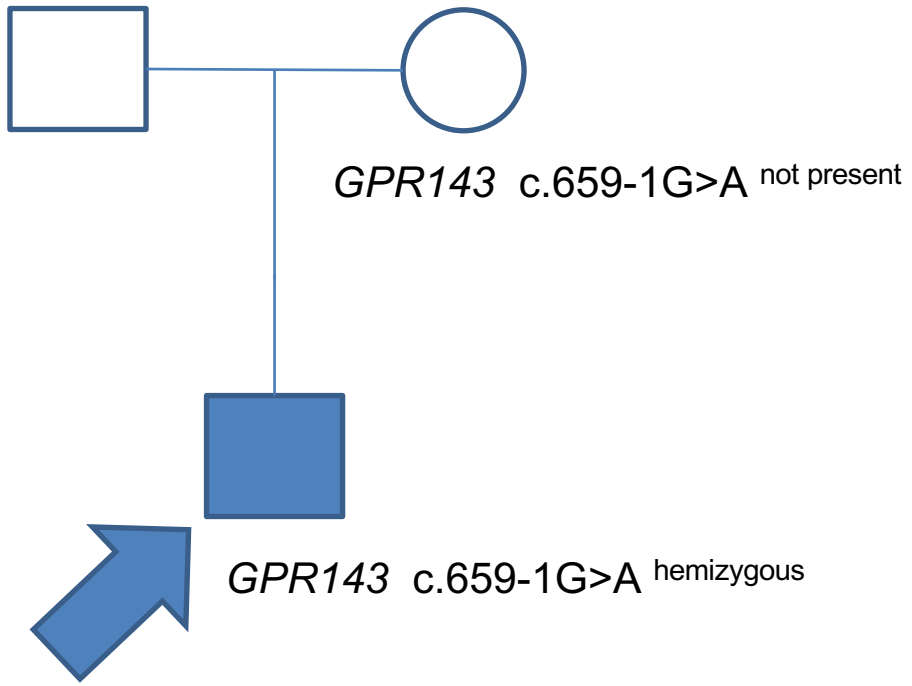
Ocular/oculocutaneous albinism panel (18 genes)

Gene (HGNC)	Transcript
AP3B1	NM_003664.4
BLOC1S3	NM_212550.4
C10orf11	NM_032024.3; NM_001305581.1
CACNA1F	NM_005183.3; NM_001256790.2
DTNBP1	NM_032122.4; NM_183040.2
GPR143	NM_000273.2
HPS3	NM_032383.4
HPS4	NM_022081.5; NM_152841.2
HPS5	NM_181507.1
HPS6	NM_024747.5
LYST	NM_000081.3
OCA2	NM_000275.2
PLDN (BLOC1S6)	NM_001311255.1; NM_012388.3
SLC24A5	NM_205850.2
SLC45A2	NM_016180.4; NM_001012509.3
SLC4A11	NM_032034; NM_001174089.1; NM_001174090.1
TYR	NM_000372.4
TYRP1	NM_000550.2

It was not possible to gain acceptable coverage of *HPS1* and it was therefore not included in these clinical diagnostic panels. *AP3D150*, *MITF51*, and *GNAI3* were also not included.

Supplementary File: Family trees of children with partial albinism

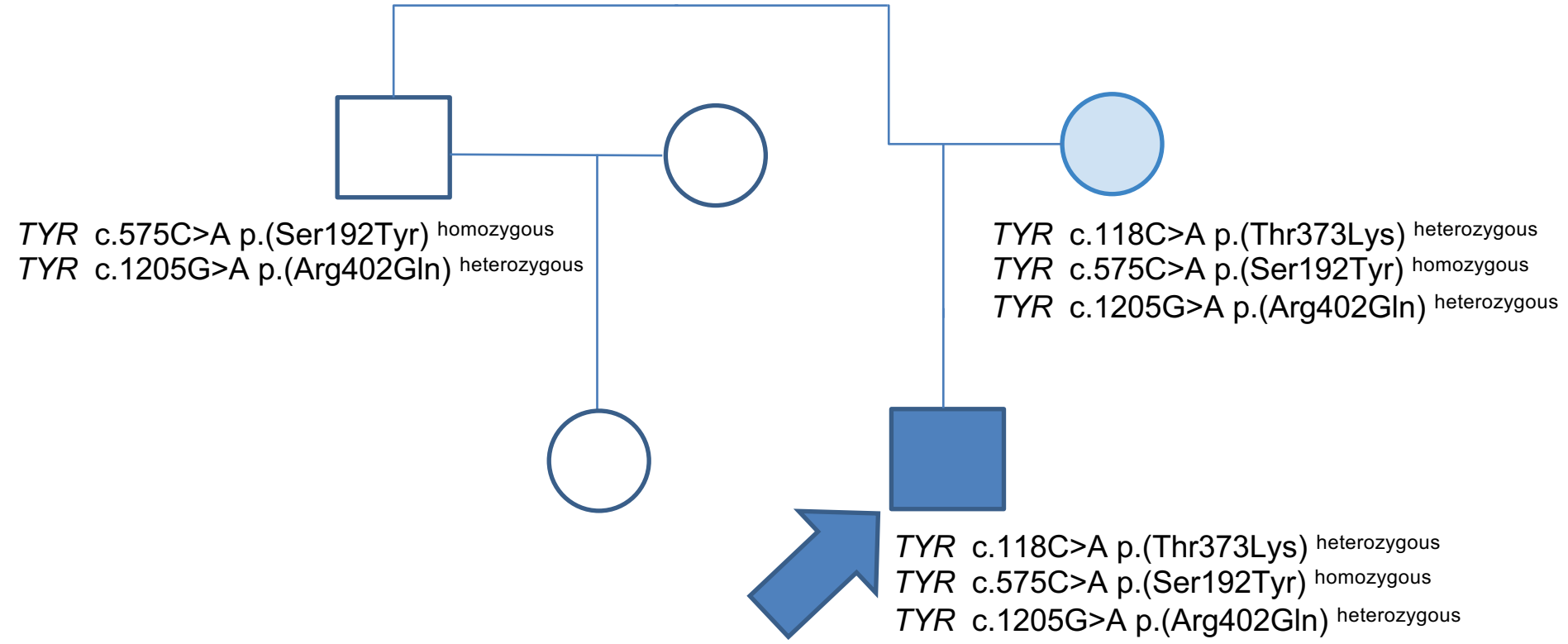
Proband 1



affected

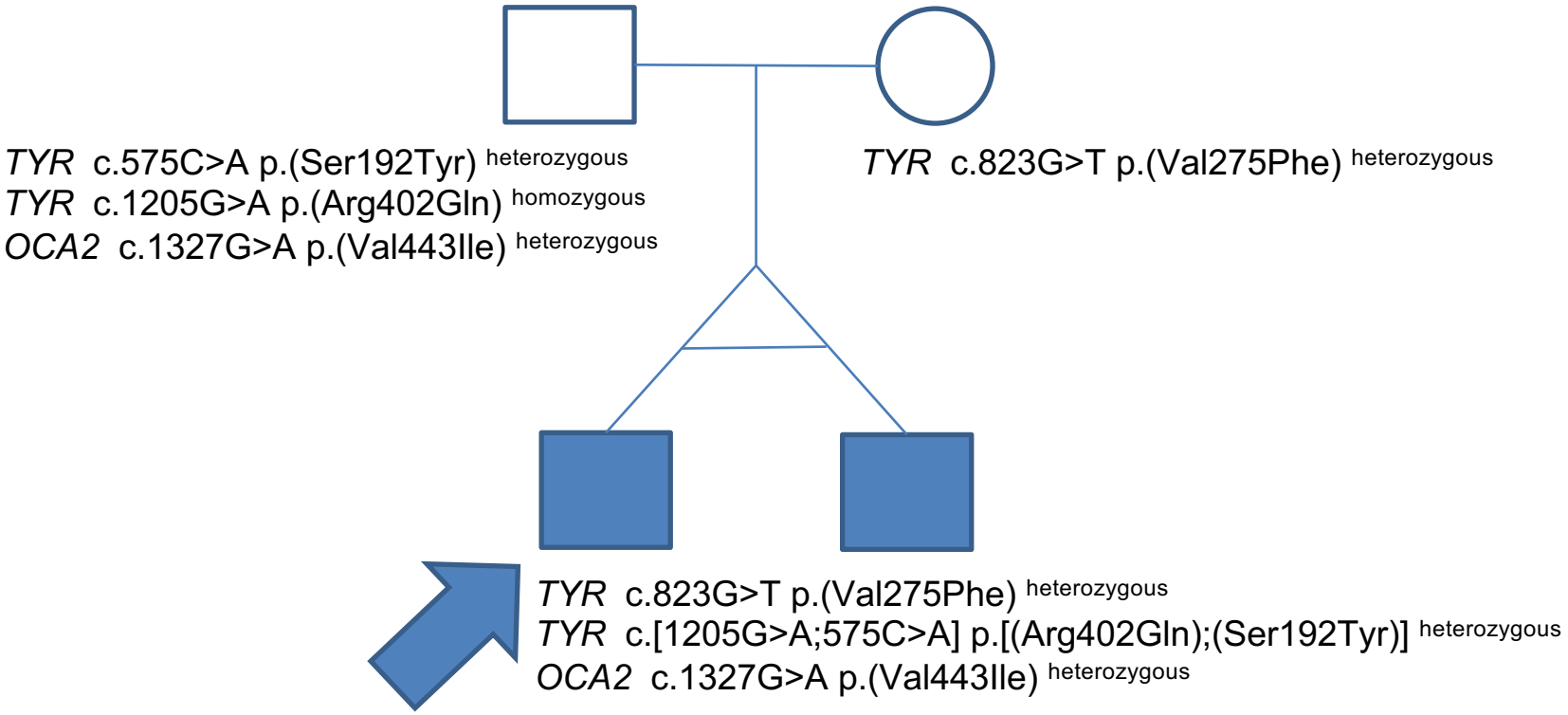
unaffected

Proband 4

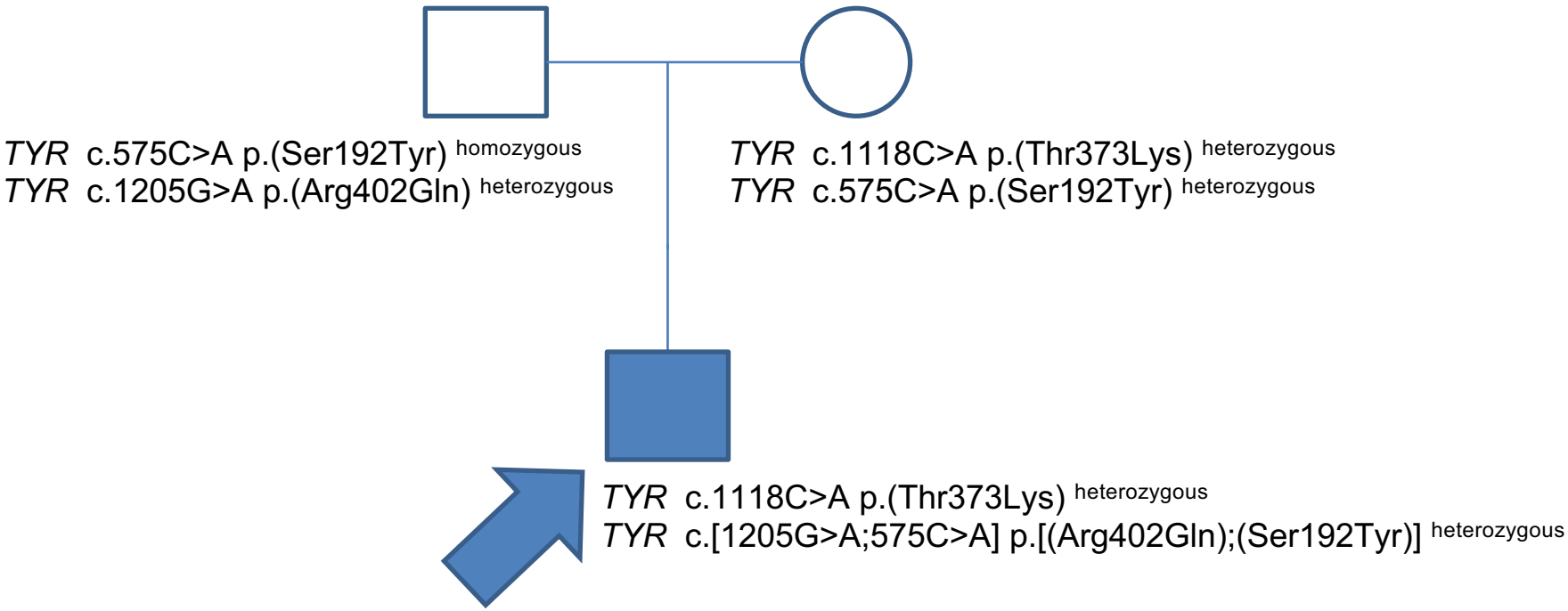


intermediate phenotype
(mild transillumination, foveal hypoplasia but no nystagmus)

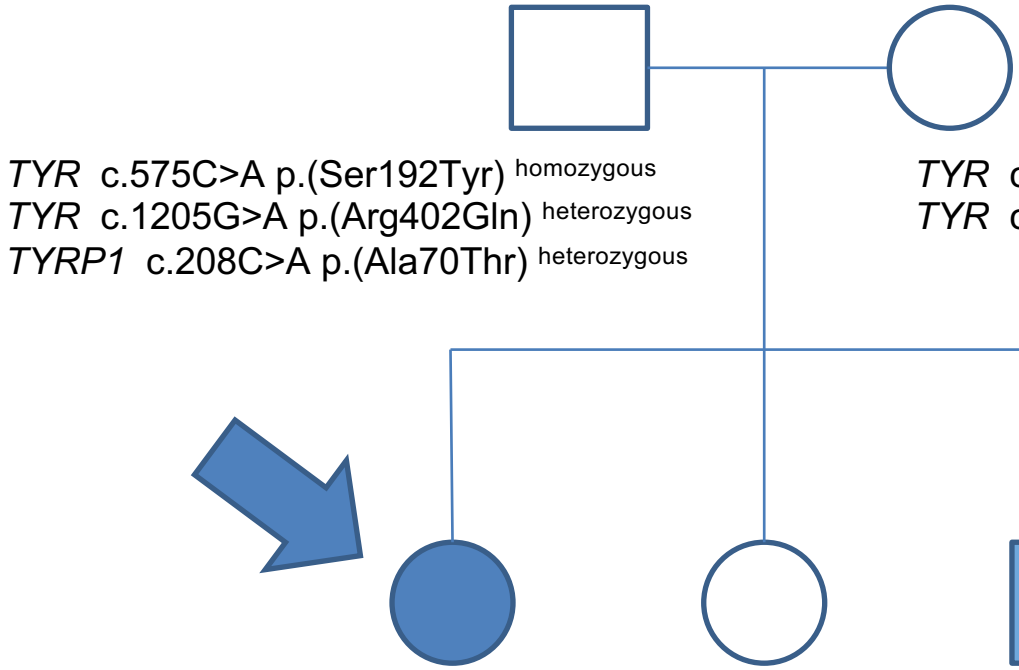
Proband 5


TYR c.575C>A p.(Ser192Tyr) heterozygous
TYR c.1205G>A p.(Arg402Gln) homozygous
OCA2 c.1327G>A p.(Val443Ile) heterozygous
TYR c.823G>T p.(Val275Phe) heterozygous
TYR c.823G>T p.(Val275Phe) heterozygous
TYR c.[1205G>A;575C>A] p.[(Arg402Gln);(Ser192Tyr)] heterozygous
OCA2 c.1327G>A p.(Val443Ile) heterozygous

Proband 6


TYR c.575C>A p.(Ser192Tyr) homozygous
TYR c.1205G>A p.(Arg402Gln) heterozygous
TYR c.1118C>A p.(Thr373Lys) heterozygous
TYR c.575C>A p.(Ser192Tyr) heterozygous
TYR c.1118C>A p.(Thr373Lys) heterozygous
TYR c.[1205G>A;575C>A] p.[(Arg402Gln);(Ser192Tyr)] heterozygous

Proband 8



TYR c.575C>A p.(Ser192Tyr) homozygous
TYR c.1205G>A p.(Arg402Gln) heterozygous
TYRP1 c.208C>A p.(Ala70Thr) heterozygous

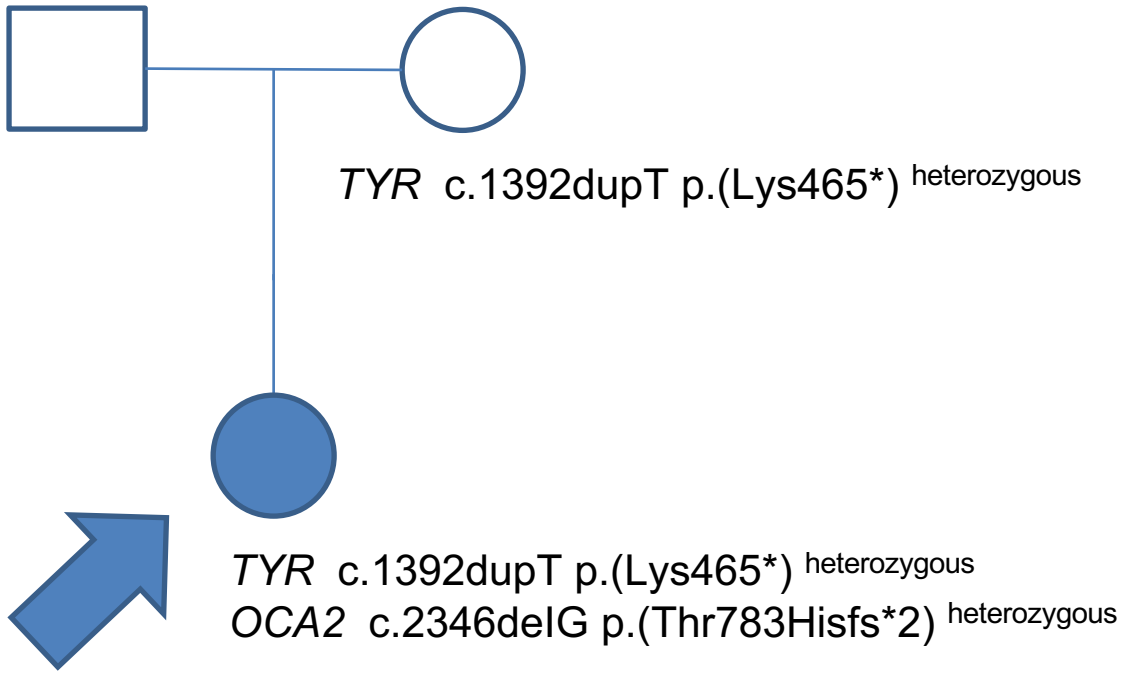
TYR c.575C>A p.(Ser192Tyr) heterozygous
TYR c.1205G>A p.(Arg402Gln) heterozygous

TYR c.575C>A p.(Ser192Tyr) homozygous
TYR c.1205G>A p.(Arg402Gln) homozygous
TYRP1 c.208C>A p.(Ala70Thr) heterozygous

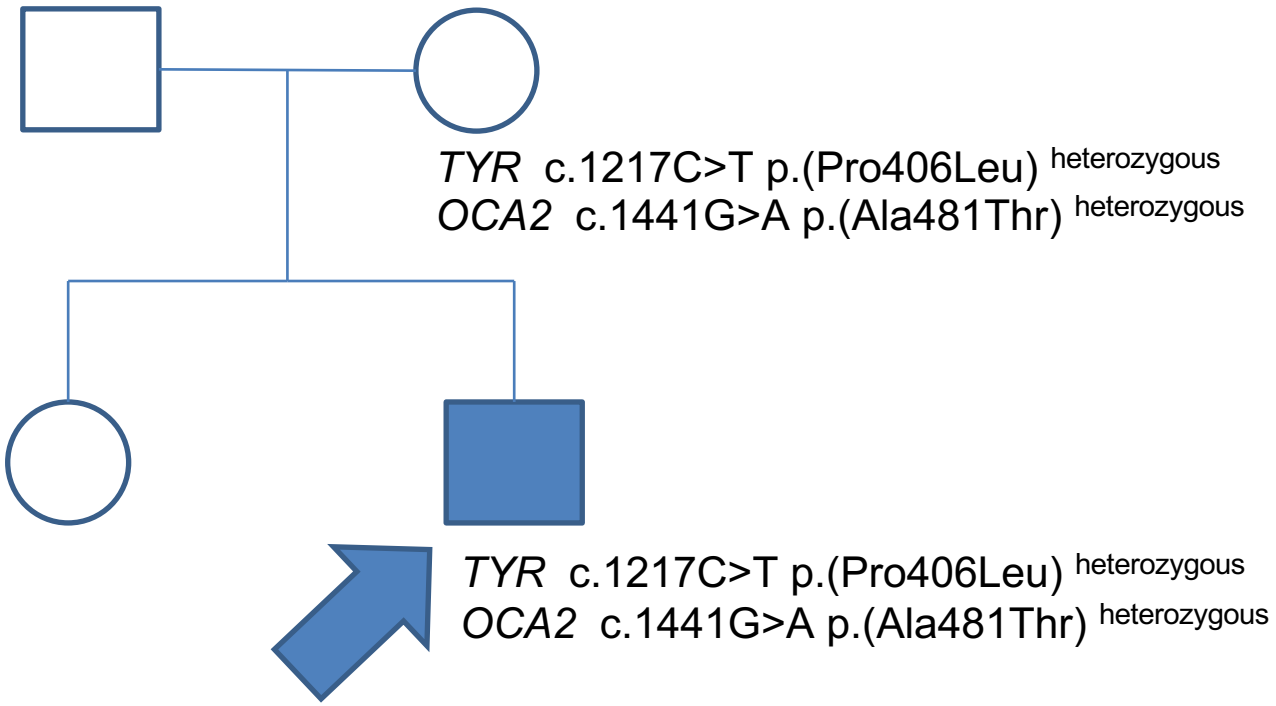
TYR c.575C>A p.(Ser192Tyr) homozygous
TYR c.1205G>A p.(Arg402Gln) homozygous

Likely ocular albinism phenotype (nystagmus present but no clinical tests performed yet as <1y old)

Proband 10


TYR c.1392dupT p.(Lys465*) heterozygous
TYR c.1392dupT p.(Lys465*) heterozygous
OCA2 c.2346delG p.(Thr783Hisfs*2) heterozygous

Proband 11


TYR c.1217C>T p.(Pro406Leu) heterozygous
OCA2 c.1441G>A p.(Ala481Thr) heterozygous
TYR c.1217C>T p.(Pro406Leu) heterozygous
OCA2 c.1441G>A p.(Ala481Thr) heterozygous