

Table S1. Primers for identification of variants in known OCA genes

Primers name	Primers Sequence	Purpose	conditions		
TYR-EXON1A-F	ccagttcctgcagacctgt	Screening TYR mutation			
TYR-EXON1A-R	tcattggccataggtccct				
TYR-EXON1B-F	gggaccaaactgcacagaga				
TYR-EXON1B-R	ggctgcaatgagtggtcagg				
TYR-EXON2-F	tgatggatttctcagaacatatccct				
TYR-EXON2-R	acaacacatatcttggtcaactca				
TYR-EXON3-F	tgggataatcacataggtttcagt				
TYR-EXON3-R	ggtgacaacctgatcacagaca				
TYR-EXON4F	ccatgtctccagatttaatatatgcc				
TYR-EXON4R	cactttcaggatttaaagtggtcagga				
TYR-EXON5-F	gccttcaaaccagggtgtct				
TYR-EXON5-R	ggaacctggacattactttgagt				
SLC45A2-EXON1F	ataacttgatcttagaggcagga			Screening SLC45A2 mutation	All primers were amplified using a touch-down program beginning at 64°C, decreasing by 0.5°C each cycle, until finishing at a final annealing temperature of 57°C.
SLC45A2-EXON1R	ggaagttcattgtaacctaggagag				
SLC45A2-EXON2F	ggtggaaattacaaacgcgg				
SLC45A2-EXON2R	cagaaactgcagcactagtca				
SLC45A2-EXON3F	tgcatgaggaaaatgaacgat				
SLC45A2-EXON3R	accccatgaaactcttctcgt				
SLC45A2-EXON4F	tccatgtccaggaaaagtgac				
SLC45A2-EXON4R	caggctccgtagtgctctct				
SLC45A2-EXON5F	gacttggatgacagcatca				
SLC45A2-EXON5R	caagagcaaagtaatcagtgagga				
SLC45A2-EXON6F	caaaagccagctgtttcaac				
SLC45A2-EXON6R	gtgagcatgcctgtaccagat				
SLC45A2-EXON7F	tgtgccctaaatgacagtcc				
SLC45A2-EXON7R	caatgggcagaccctaagatt				
OCA2-EXON2F	actcccaaggtgcaaacgft	Screening OCA2 mutation			
OCA2-EXON2R	cacaacaaactgtgggggaac				
OCA2-EXON3F	tggtgtcttctaaaactcagcc				
OCA2-EXON3R	gtcaaggatctggcagaggft				
OCA2-EXON4F	ccattcccaccagtatgaga				
OCA2-EXON4R	cctggccagatgagacaaaac				
OCA2-EXON5F	tgtgggttactggtcactgat				
OCA2-EXON5R	cctcagcatctcctcaaaga				
OCA2-EXON6F	taatcagggcccattgtagct				
OCA2-EXON6R	ctccatctctcatcagacatcct				
OCA2-EXON7F	gggacatggggttctcctg				
OCA2-EXON7R	cctgtggctccccatcaaat				
OCA2-EXON8F	ggtgtctcaggtgaaaagcc				
OCA2-EXON8R	agccttgagctcactgtcta				
OCA2-EXON9F	ctcactgttcattgtcgggtg				
OCA2-EXON9R	gagagagggacacgctaact				
OCA2-EXON10F	tagctgtatgtgtctgtggg				
OCA2-EXON10R	tctagcatggttctgggcaa				
OCA2-EXON11F	ccaacttcaaaggcaagtga				

OCA2-EXON11R	atagccccattccattcctcc
OCA2-EXON12F	gtgacggctttctgcatctc
OCA2-EXON12R	tagcaagtataccctgccctg
OCA2-EXON13F	gactccgtgggtctatgtctt
OCA2-EXON13R	gtactgcagtcacacaaagca
OCA2-EXON14F	aggctcactctggaaaggaat
OCA2-EXON14R	ggtggcgtgatgatcttgatt
OCA2-EXON15F	ctgcaagtccgcctctc
OCA2-EXON15R	agcagccctgaaaacagtgta
OCA2-EXON16F	tgttgaaattgcattactcgct
OCA2-EXON16R	cacaccaagcacagtctgag
OCA2-EXON17F	ccatgactagtcctgccagc
OCA2-EXON17R	ttgctttccacatcctcaca
OCA2-EXON18F	cctgaatctgtacgggttcc
OCA2-EXON18R	gatgcatctctctccctcc
OCA2-EXON19F	aggttaaagaaatgaatcgggtg
OCA2-EXON19R	gtcagaaatctctcagtgctaag
OCA2-EXON20F	ggttctaaactgattctcaccac
OCA2-EXON20R	cctgcagcactcattactga
OCA2-EXON21F	tcgtgatgggtaagaggaagg
OCA2-EXON21R	cttaggaagggagggtgagc
OCA2-EXON22F	ccgtataagagccacaacagc
OCA2-EXON22R	atcagaaccagtcaccccag
OCA2-EXON23F	cttgcggtgtgtgtgtttcc
OCA2-EXON23R	aatctcccctacaccacagtc
OCA2-EXON24F	ttgcacacaatggaggatgtc
OCA2-EXON24R	ggagaaaggcacacagagga
TYRP1-EXON2F	ttcctctacgtctcagctc
TYRP1-EXON2R	agagccttgagtctcatgcag
TYRP1-EXON3F	ggataaatgggtcggtgtg
TYRP1-EXON3R	acgaaatgccaagacaggtt
TYRP1-EXON4F	aggtttaactcctctgggcc
TYRP1-EXON4R	gcactttctgtttcccctgg
TYRP1-EXON5F	tctacttgattcaaagaaatgggac
TYRP1-EXON5R	gaaaagagctcatggtgactc
TYRP1-EXON6F	cataggtacagagaagcagtc
TYRP1-EXON6R	aagtgactgatattacagctatgc
TYRP1-EXON7F	ttctcctgaatattggatgcct
TYRP1-EXON7R	agaataccgtgattactctactga
TYRP1-EXON8F	ccaatagggccactcattaaaga
TYRP1-EXON8R	agtatgcctagaactttaatgtaac

Screening
TYRP1
mutation

SLC24A5-EXON1F	agcagagccagtatgaagaca		
SLC24A5-EXON1R	aaactggagacttggcctact		
SLC24A5-EXON2F	gcgctttatacattttcagtg		
SLC24A5-EXON2R	cgaccagttccttaagacacg		
SLC24A5-EXON3-4F	attcccttagtctcctggcc		
SLC24A5-EXON3-4R	ttagtgccctgtgtccatcc		
SLC24A5-EXON5F	ccacaaatagctcttggtcaga		
SLC24A5-EXON5R	gaaggaaaagtgggtgtctcc	Screening	
SLC24A5-EXON6F	gctgtaatcaagtctggacaaatcta	SLC24A5	
SLC24A5-EXON6R	tgcgaaatagacttaataatgggcaa	mutation	
SLC24A5-EXON7F	tcacatcttaccattgtctgcc		
SLC24A5-EXON7R	atgcaactcacaactccagt		
SLC24A5-EXON8F	tctcactggcagaattttagct		
SLC24A5-EXON8R	agcctttagaatgaccattgc		
SLC24A5-EX9F	gtgactccagaggaaaagttac		
SLC24A5-EX9R	tcatgaaaagaaagggctcaga		
LRMDA-EXON1F	ataaagctgtggcatggagt		
LRMDA-EXON1R	gagacctgcggtgttct		
LRMDA-EXON2F	gattgatcagggaaacagtcac		
LRMDA-EXON2R	cacgcatctcagagccttct		
LRMDA-EXON3F	atttatcctgcaggccgtatc		
LRMDA-EXON3R	gccagaagacagaatgcttg		
LRMDA-EXON4F	accgtatacccacagctgaat	Screening	
LRMDA-EXON4R	ggaaaccactgatatgtacca	LRMDA	
LRMDA-EXON5F	accttgagccaagtgtacca	mutation	
LRMDA-EXON5R	gacccttcatgtgagattcca		
LRMDA-EXON6F	ttctccaagctcagatcccaa		
LRMDA-EXON6R	ttgcctcctgaatagactgct		
LRMDA-EXON7F	ggagcttcatccactttctgc		
LRMDA-EXON7R	aaggatcatcttgacaggcct		
WT.OCA2_c.808-3C>G-FW	gaccggatcccagagatgtgagagctcaat		
WT.OCA2_c.808-3C>G-RV	gaccacgcgtgctcactgtctacacagca		
WT.OCA2_c.2140-2A>G-FW	gaccggatccgcccctggcttcacacctatgt	Construction	
WT.OCA2_c.2140-2A>G-RV	gaccacgcgtccccaggacgtgttcca	of wildtype	
WT.SLC45A2_c.1032+1G>t-FW	gaccggatcctccatggaaacctagttccatg	vectors for	
WT.SLC45A2_c.1032+1G>t-RV	gaccacgcgtcaggctccgtagtgtcctct	splicing assay	
WT.OCA2_c.2245-11T>G-RV	gaccggatccccgtataagagccacaacagc		
WT.OCA2_c.2245-11T>G-RV	gaccacgcgtatcagaaccagtcaccccag		
MUT.OCA2_c.808-3C>G-FW	gctttagaggtcactcacaactg	Mutant-direct	
MUT.OCA2_c.808-3C>G-RV	gtgacctctacaagccaaagcataagttatg	site with	
MUT.OCA2_c.849C>A-FW	gagaagagagcactcagtgatgagcag	wildtype	60 degree

MUT.OCA2_c.849C>A-RV	gtgctctcttctcctcggatttaaatacac	vectors for splicing assay	
MUT.OCA2_c.2140-2A>G-FW	cttccggatggtcccagaggagcag		
MUT.OCA2_c.2140-2A>G-RV	ggaccatccggaaggaggacaatagcagct		
MUT.SLC45A2_c.1032+1G>t-FW	catgggccagttaatgaacgtgtctgtgca		
MUT.SLC45A2_c.1032+1G>t-RV	gttcattaactggcccatgaaatctgtgaaga		
MUT.OCA2_c.2245-11T>G-FW	gtcctcgtcctccacagattcccgt		
MUT.OCA2_c.2245-11T>G-RV	tggaggacgaggacattgatgccacgtc		
Oligo pCAS-KO1 -F	tgacgtcgccgcccatcac	RT-PCR of splicing assay	57degree
Oligo pCAS - 2R	attggtgttgagttggtgtgc		
pCAS-Seq-F	gggtcaatagcagtgagagg	Sequencing of vector to confirm	60 degree
pCAS-SeqRV	gctccatttcacaggtagaga		