chrom	position	ref	alt	Variant type	Gene refGene	Omim Inheritance	Amino Acid	avsnp144	ExAC_ALL	CADD_phred	MaxEnt Scan_diff	CLINSIG	Intervar	HGMD	Phenotype group																
															1 (n=18)				2 (n=15)		3 (n=20)		4 (n=28)								
															NG335	NG433	NG477	NG528	NG315	NG381	NG280	NG540	NG195	NG340	NG391	NG394	NG395	NG416	NG498	NG543	NG551
2	237493530	G	A	ns	*MLPH*	AR	R35Q			33.0				DM																	
6	35505739	A	C	sp	*TULP1*	AR				23.8	7.65		P																		
11	18310740	C	T	sp	*HPS5*	AR				27.2	8.18		P													C					
11	31800832	G	A	ns	*PAX6*	AD	R142C	rs121907918		34.0		P		DM																	C
11	31801623	C	G	ns	*PAX6*	AD	A113P			27.8				DM									C								
11	31806401	C	T	sp	*PAX6*	AD				27.2	8.18		P	DM											C						
11	**89284793**	**G**	**A**	**ns**	***TYR***	**AR**	**R402Q**	**rs1126809**	**0.17700**	**34.0**		**P**																			
11	89284805	C	T	ns	*TYR*	AR	P406L	rs104894313	0.00350	32.0		P		DM				C									C				
11	89284879	C	A	ns	*TYR*	AR	P431T	rs368604842		28.3				DM				C													
11	89284924	G	A	ns	*TYR*	AR	G446S	rs104894317	0.00002	31.0		P		DM													C				
13	23339410	T	C	ns	*SACS*	AR	N1489S	rs147099630	0.00910	0.0				DM																	
15	27983383	T	C	ns	*OCA2*	AR	N489D	rs121918170	0.00030	28.2		P		DM								C						C			
15	27985101	C	T	ns	*OCA2*	AR	V443I	rs121918166	0.00280	34.0		P		DM					C		C	C		C				C			
15	27990579	G	A	s	*OCA2*	AR	G371G		0.02060					DM																	
15	28014795	T	C	ns	*OCA2*	AR	Y342C		0.00020	24.3				DM										C							
15	28022554	G	A	ns	*OCA2*	AR	P198L		0.00010	29.0				DM							C										
15	28022592	T	C	sp	*OCA2*	AR			0.00620		0.71			DM					C		C										
15	48121951	T	G	sg	*SLC24A5*	AR	Y72X	rs142056637	0.00004	35.0				DM																	
19	13317310	C	T	ns	*CACNA1A*	AD	A453T	rs41276886	0.00480	28.2				DM																C	
X	49222720	T	G	ns	*CACNA1F*	XL	N746T		0.00170	26.1	1.35			DM														C			
X	49226037	C	T	ns	*CACNA1F*	XL	R519Q		0.03040	33.0	-			DM			C												C		
X	49226936	C	G	sp	*CACNA1F*	XL				23.2	8.27		P							C											
X	132080053	G	A	sg	*FRMD7*	XL	R335X	rs137852208	0.00001	39.0		P	P	DM	C																
X	132100704	C	T	ns	*FRMD7*	XL	G24R	rs137852210		29.6		P		DM		C															