chrom	position	ref	alt	Variant type	Gene refGene	Omim Inheritance	Amino acid	avsnp144	ExAC ALL	CADD phred	MaxEnt Scan diff	CLINSIG	Intervar	HGMD	Variant category	Phenotype group								
																1 (n=14)			2 (n=13)			3 (n=18)		4 (n=19)
																NG299	NG445	NG534	NG318	NG327	NG383	NG512	NG521	NG386
1	235766255	G	A	ns	*LYST*	AR	T1982I	rs146591126	0.00630	17.1					2									
3	149162256	G	A	ns	*HPS3*	AR	G739R	rs78336249	0.00960	22.9					2									
5	78015546	C	T	ns	*AP3B1*	AR	V999M	rs146503597	0.00380	16.4	-0.79				2									
6	15523217	G	A	ns	*DTNBP1*	AR	P272S	rs17470454	0.04360	19.9	0.14				2									
6	35505739	A	C	sp	*TULP1*	AR				23.8	7.64		P		1									
9	12694076	C	G	ns	*TYRP1*	AR	P27R	rs373327120	0.00002	24.5					2								C	
9	12702394	C	G	ns	*TYRP1*	AR	P346R	rs377679582	0.00007	31.0					2								C	
11	18281986	G	A	ns	*HPS5*	AR	T1098I	rs61884288	0.02360	18.6		P			1		C							
11	18306204	C	T	ns	*HPS5*	AR	G252E	rs755846129	0.00003	32.0					2		C							
11	**89284793**	**G**	**A**	**ns**	***TYR***	**AR**	**R402Q**	**rs1126809**	**0.17700**	**34.0**		**P**			**1**									
13	23330159	T	G	ns	*SACS*	AR	N4573H	rs34382952	0.00320	25.5					2					R				
13	23332844	G	C	ns	*SACS*	AR	P3678A	rs17078601	0.03970	25.9					2					R				
13	23340137	G	T	ns	*SACS*	AR	Q1247K			21.8					2				R					
13	23354532	C	T	ns	*SACS*	AR	A694T	rs17325713	0.02330	15.0					2									
13	23354671	C	A	ns	*SACS*	AR	K647N	rs201021919		23.6					2				R					
15	27845050	T	C	ns	*OCA2*	AR	N781D			26.7	-0.06				2							C		
15	28014795	T	C	ns	*OCA2*	AR	Y342C		0.00020	24.3				DM	1							C		
15	28022592	T	C	sp	*OCA2*	AR			0.00620		0.71			DM	1									
15	52343197	T	A	ns	*MYO5A*	AR	R1320S	rs61731219	0.03370	21.7	0.93				2									
19	13286647	G	C	ns	*CACNA1A*	AD	P1137A	rs199793367	0.00040	23.1					2			C						
19	13298593	C	T	ns	*CACNA1A*	AD	E1014K	rs16024	0.00260	16.8					2				R					
19	13298659	C	T	ns	*CACNA1A*	AD	E992K			25.7					2									C
19	13300637	T	G	ns	*CACNA1A*	AD	E731A	rs16019	0.01010	24.8					2					R				
X	132077973	C	T	ns	*FRMD7*	XL	G682S			28.8					2						C			
X	132082478	A	C	ns	*FRMD7*	XL	C264G			25.5					2	R								