Correction

Correction: Increasing the Yield in Targeted Next-Generation Sequencing by Implicating CNV Analysis, Non-Coding Exons and the Overall Variant Load: The Example of Retinal Dystrophies

The *PLOS ONE* Staff

There is an error in the header of Table 1. The row “Autosomal recessive retinitis pigmentosa, arRP” should not be a separate header for the entire table. The publisher apologizes for this error. A corrected version of Table 1 can be viewed below.

Citation: The *PLOS ONE* Staff (2014) Correction: Increasing the Yield in Targeted Next-Generation Sequencing by Implicating CNV Analysis, Non-Coding Exons and the Overall Variant Load: The Example of Retinal Dystrophies. *PLoS ONE* 9(11): e108840. doi:10.1371/journal.pone.0108840

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Table 1. Causative mutations and putatively pathogenic variants identified in this study.

Patient	Gene	Allele 1				Allele 2				Additional Allele				Gender	Age (years)	Phenotype	Inheritance (family history)	Consang.	Origin
		cds	Protein	db SNP	Ref	cds	Protein	dbSNP	Ref	cds	Protein	dbSNP	Ref						
Autosomal recessive retinitis pigmentosa, arRP																			
2	ABCA4	c.768G>T	p.V256V	-	[1,2,3]	c.5603A>T	p.Asn1868Ile	rs1801466	[4,5]					m	60	RP	air/s	no	Cau
(splice)																			
30	ABCA4	c.1622T>C/	p.Leu541Pro/	rs61751392		c.3210_3211	p.Ser1071Cys			ABCA4:				f	17	RP	air/s	no	Ger
		c.3113C>T	p.Ala1038Val	rs61751374	[6,7,8]	dupGT	fs*14	-	[9]	c.5603A>T	p.Asn1868Ile	rs1801466	[4,5]						
31	ABCA4	c.768G>T	p.V256V	-	[1,2,3]	c.1622T>C/	p.Leu541Pro/	rs61751392	[6,7,8]					f	8	RP	air/s	no	Ger
			(splice)			c.3113C>T	p.Ala1038Val	rs61751374											
53	ABCA4	c.1A>G	p.Met1Val	-	[10]	c.1A>G	p.Met1Val	-	[10]	ABCA4:				f	44	RP	air/s	yes	Ger
			(p.Met1?)				(p.Met1?)			c.6089G>A	p.Arg2030Gln	rs61750641	[10,11]						
										c.6089G>A	p.Arg2030Gln	rs61750641	[10,11]						
3	C20orf71	c.2756_2768del	p.Lys919Thr	-	[12]	c.2756_2768del	p.Lys919Thr	-	[12]					f	44	RP	air/s	yes	Tur
			fs*2				fs*2												
45	CNGA1	c.1086C>T	p.Arg346Trp	-	[a]	c.1166C>T	p.Ser389Phe	rs62625014	[13]	PDE6B:				m	43	RP	air/s	no	Ger
										c.703C>T	p.Arg235Cys	-	[a]						
74	CNGA1	c.2195A>G	p.Glu732Gly	-	[a]	c.2195A>G	p.Glu732Gly	-	[a]	TTCE:				m	50	RP	air/s	no	Ger
										c.1253A>G	p.Gln418Arg	rs142938748	[a]						
5	CRB1	c.1459T>C	p.Ser487Pro	-	[a]	c.1459T>C	p.Ser487Pro	-	[a]	SAG:				m	n.d.	RP	air/s	yes	Pak
										c.374C>T	p.Thr125Met	rs137886124	[a]						
7	CRB1	c.2042G>A	p.Cys681Tyr	rs62636266	[14]	c.2308G>C	p.Gly770Arg	-	[a]	FSCN2:				f	28	RP	air/s	no	Pol
										c.805C>T	p.Arg269Cys	-	[a]						
48	CRB1	c.2367T>A	p.Asn789Lys	-	[a]	c.2401A>T	p.Lys801*	-	[15]					f	37	RP	air/s	no	Ger
8	EYS	c.604T>C	p.Cys202Arg	-	[a]	c.4350_4356del	p.Ile1451Pro	-	[16]	MERTK				m	52	RP	air/s	no	Au
							fs*3			c.791C>G	p.Ala264Gly	-	[a]						
										SAG:									
										c.473C>A	p.Thr158Lys	-	[a]						
34	EYS	c.7055+1G>T	splice	-	[a]	c.7055+1G>T	splice	-	[a]					f	40	RP	air/s	yes	Syr
57	EYS	c.4045C>T	p.Arg1349*	-	[a]	deletion of	?	-	[a]					f	32	RP	air/s	no	Ger
						exon 1													

Table 1. Cont.

Patient	Allele 1		Allele 2				Additional Allele				Gender		Age (years)	Phenotype	Inheritance (family history)	Consang.	Origin		
	Gene	cds	Protein	db SNP	Ref	cds	Protein	dbSNP	Ref	cds	Protein	dbSNP						Ref	
88	EYS	c.67delA	p.Thr23His	-	[a]	c.162C>A	p.Tyr54*	-	[a]				m	48	RP	air/XI	no	Ger	
fs*19																			
93	EYS	c.4350_4356del	p.Ile1451Pro	-	[16]	c.-448+5G>A	splice	-	[a]	RD3:			m	23	RP	air/s	no	Ger	
fs*3																			
9	MERTK	c.345C>G	p.Cys115Trp	-	[a]	c.1530delIT	p.Cys510Trp	-	[a]	c.584A>T	p.Asp195Val	rs143207434	[a]	m	19	RP	air/XI	no	Ger
fs*5																			
46	MERTK	c.1786G>A	p.Gly596Arg	-	[a]	c.1786G>A	p.Gly596Arg	-	[a]				f	36	RP	air/s	n.d.	n.d.	
76	MERTK	c.1450G>A	p.Gly484Ser	-	[a]	c.1450G>A	p.Gly484Ser	-	[a]	RP2:			m	30	RP	air/s	no	Italy	
c.844C>T																			
p.Arg282Trp																			
rs1805147																			
[17/18]																			
RHO:																			
c.310G>A																			
p.Val104Ile																			
rs144317206																			
[19]																			
20	PDE6B	c.669T>A	p.Tyr223*	-	[a]	c.669T>A	p.Tyr223*	-	[a]	RP3:			f	30	RP	air/s	yes	Iran	
c.2092C>A																			
p.Arg698Ser																			
-																			
[a]																			
21	PDE6B	c.1699C>T	p.Gln567*	-	[a]	c.1699C>T	p.Gln567*	-	[a]	ABC14:			f	36	RP	air/s	yes	Ger	
c.5603A>T																			
p.Asn1868Ile																			
rs1801466																			
[45]																			
62	PDE6B	c.2193+1G>A	splice	-	[20]	c.2193+1G>A	splice	-	[20]	ABC14:			m	47	RP	air/s	yes	Ger	
c.2184C>A																			
p.Ser728Arg																			
-																			
[a]																			
75	PDE6B	c.2193+1G>A	splice	-	[20]	c.2047G>A	p.Val683Met	-	[a]	PDE6B:			m	45	RP	air/s	no	Ger	
c.2249T>G																			
p.Val750Gly																			
-																			
[a]																			
55	PRDM1	c.642T>A	p.Tyr214*	-	[a]	c.1209_1229del	p.Gln403_Ser	-	[a]	RP1:			m	26	RP	air/s	no	Ger	
c.6304C>T																			
p.Gln2102*																			
-																			
[a]																			
GRB1:																			
c.2042G>A																			
p.Cys681Tyr																			
rs62636266																			
[14]																			
PPG/RP1:																			
c.1767G>T																			
p.Gln589His																			
rs34067949																			
[21/22]																			
ABC14:																			
-																			
[a]																			
89	RDH12	c.226G>C	p.Gly76Arg	-	[23]	c.869T>G	p.Val290Gly	rs61740289	[a]	ABC14:			f	34	RP	air/s	yes	DRC	
c.618C>G																			
p.Ser206Arg																			
rs61748536																			
[24]																			

Table 1. Cont.

Patient	Allele 1		Allele 2				Additional Allele				Gender	Age (years)	Phenotype	Inheritance (family history)	Consang.	Origin				
	Gene	cds	Protein	db SNP	Ref	cds	Protein	dbSNP	Ref	cds							Protein	dbSNP	Ref	
C20R71:																				
25	RP1	c.597C>A	p.Tyr199*	-	[a]	c.3157delT	p.Tyr1053Thr	-	[26]	CDH23:	c.6322G>T	p.Glu2108*	-	[27]	f	38	RP	air/s	no	Ger
C9B1:																				
26	RP1	c.4342_4243del	p.His1414Gln	-	[a]	c.4474G>T	p.Glu1492*	-	[a]	c.3122T>C	p.Met1041Thr	rs62635656	-	[28]	f	47	RP	air/s	yes	Iran
27	RP1	c.1012C>T	p.Arg338*	-	[a]	c.1012C>T	p.Arg338*	-	[a]	ABCA4:	c.5603A>T	rs1801466	-	[4,5]	f	37	RP	air/s	yes	Iran
28	RP1	c.5278_5287del	p.Asn1760Cys	-	[a]	c.5278_5287del	p.Asn1760Cys	-	[a]	c.5603A>T	p.Asn1868Ile	rs1801466	-	[4,5]	f	12	RP	air/s	yes	Tur
36	RP1	c.607G>A	p.Gly203Arg	-	[a]	c.607G>A	p.Gly203Arg	-	[a]						m	n.d.	RP	air/s	yes	Iran
101	RP1	c.3843delT	p.Pro1282Leu	-	[a]	c.3843delT	p.Pro1282Leu	-	[a]						f	37	RP	air/s	yes	Tur
fs*12																				
29	SAG	c.577C>T	p.Arg193*	-	[29]	?									m	16	RP	air/s	no	Ger
33	TULP1	c.371_394del	p.Asp124_	-	[a]	?				C9K:	c.122G>A	p.Arg41Gln	rs61748436	[30]	m	52	RP	air/s	no	Ger
132delinsAla																				
C9K:																				
82	TULP1	c.371_394del	p.Asp124_	-	[a]	?				c.425A>G	p.Tyr142Cys	rs61748442	-	[21]	f	35	RP	air/s	no	Ger
132delinsAla																				
37	TULP1	c.1047T>G	p.Asn349Lys	-	[a]	c.1047T>G	p.Asn349Lys	-	[a]	PDE6A:	c.923C>T	p.Pro3308Leu	-	[a]	f	n.d.	RP	air/s	yes	Iran
38	TULP1	c.1198C>T	p.Arg400Trp	-	[31]	c.1198C>T	p.Arg400Trp	-	[31]	ABCA4:	c.2353C>T	p.Arg785Cys	-	[a]	f	n.d.	RP	air/s	yes	Iran

Table 1. Cont.

Patient	Gene	Allele 1				Allele 2				Additional Allele				Ref	Gender	Age (years)	Phenotype	Inheritance (family history)	Consang.	Origin
		cds	Protein	db SNP	Ref	cds	Protein	dbSNP	Ref	cds	Protein	dbSNP	Ref							
F508C2:																				
13	USH2A	c.10421A>G	p.Tyr3474Cys	-	[a]	c.13257_13263 del	p.Phe4419Leu fs*2	-	[a]	c.325C>T	p.Arg109Cys	-	[a]		m	26	RP	air/s	no	Ger
18	USH2A	c.6925T>C	p.Cys2309Arg	-	[a]	c.10561T>C	p.Trp3521Arg	rs111033264	[32,33]	ABC44:					f	50	RP	air/s	no	Ger
c.5603A>T																				
p.Asn1868Ile																				
rs1801466																				
[4,5]																				
ALPL1:																				
c.401A>T																				
p.Tyr134Phe																				
rs16955851																				
[34]																				
68	USH2A	c.1256G>T	p.Cys419Phe	rs121912600	[35]	c.10342G>A	p.Glu3448Lys	-	[a]					m	44	RP	air/s	no	Ger	
Autosomal dominant RP, adRP																				
22	PRPF31	c.1048C>T	p.Gln350*	-	[a]					RPE65:					f	38	RP	air/s	no	E-Eur
c.963T>G																				
p.Asn321Lys																				
rs149916178																				
[36]																				
23	PRPF31	c.1067_1073+8 del	Splice	-	[a]										f	36	RP	air/s	no	Ger
43	PRPF31	c.217A>T	p.Lys73*	-	[a]					ABC44:					m	18	RP	ad	no	Ger
113	PRPF31	deletion of exons 1-14	haplo- insufficiency	-	[37,38]					c.1928G>A	p.Val643Gly	rs114572202	[39]		f	55	RP	ad	no	Ger
c.5603A>T																				
p.Asn1868Ile																				
rs1801466																				
[4,5]																				
RPGRIPI:																				
c.2555G>A																				
p.Arg852Gln																				
rs181758389																				
[21]																				
116	PRPF31	deletion of exons 1-5	haplo- insufficiency	-	[37,38]										f	14	RP	air/s	No	Cau
47	PRPH2	c.920delT	p.Leu307Arg	-	[40]					F508C2:					m	33	RP	ad	no	Ger
fs*17																				
p.His138Tyr																				
rs143796236																				
[a]																				
32	RHO	c.35C>G	p.Pro12Arg	-	[a]					RLBP1:					f	71	RP	ad	no	Ger
c.545T>G																				
p.Phe182Cys																				
rs142244640																				
[a]																				
44	RHO	c.541G>A	p.Glu181Lys	-	[41]										f	47	RP	s	no	Ger

Table 1. Cont.

Patient	Gene	Allele 1				Allele 2				Additional Allele				Ref	Gender	Age (years)	Phenotype	Inheritance (family history)		Consang.	Origin	
		cds	Protein	db SNP	Ref	cds	Protein	dbSNP	Ref	cds	Protein	dbSNP	Ref									
92	RHO	c.180C>A	p.Tyr60*	-	[a]					ROM1:					f	31	RP	ad	no		Ger	
										c.178C>A	p.Pro60Thr;	-	[42,43]									
										c.323C>T	p.Thr108Met	rs146358003	[42,43]									
										ABCA4:												
										c.1654G>A	p.Val552Ile	rs145525174	[44]									
										c.5714+5G>A	splice	rs61751407	[45]									
										RP2:												
										c.844C>T	p.Arg282Trp	rs1805147	[17,18]									
										RPGRIP1:												
										c.2510C>G	p.Ala837Gly	-	[22]									
121	RHO	c.937-1G>T	splice	-	[46,47]					ABCA4:					m	25	RP	ad	no		Gau	
										c.5603A>T	p.Asn1868Ile	rs1801466	[45]									
103	TOPORS	c.2554_2557del	p.Glu852Gln	-	[a]										f	30	RP	ad	no		Ger	
			fs*13																			
108	TOPORS	c.2550_2553del	p.Asp850Glu	-	[a]					ABCA4:					f	59	RP	ad	no		Ger	
			fs*15							c.4771G>A	p.Gly1591Arg	rs113106943	[a]									
										PDE6A:												
										c.298C>T	p.Arg100Trp	-	[a]									
X-linked RP																						
77	RP2	c.226G>T	p.Asp76Tyr	-	[a]					RP2:					m	15	RP	air/s	no		SE-Eur	
										c.844C>T	p.Arg282Trp	rs1805147	[17,18]									
										ABCA4:												
										c.5882G>A	p.Gly1961Glu	rs1800553	[7,48]									
100	RPGR	c.1853_1856dup	p.Glu621Lys	-	[a]										f	54	RP	ar	no		Ger	
			fs*10																			
119	RPGR	c.1544T>G	p.Leu515*	-	[a]										m	5	LCA	ar	no		KSA	

Table 1. Cont.

Patient	Gene	Allele 1				Allele 2				Additional Allele				Ref	Gender	Age (years)	Phenotype	Inheritance (family history)	Consang.	Origin
		cds	Protein	db SNP	Ref	cds	Protein	dbSNP	Ref	cds	Protein	dbSNP	Ref							
Leber congenital amaurosis, LCA																				
1	AIP1	c.834G>A	p.Trp278*	rs62637014	[49]	c.834G>A	p.Trp278*	rs62637014	[49]	RD3: c.584A>T	p.Asp195Val	rs143207434	[a]		m	2	LCA	ar	possible Ger	
SAG:																				
83	AIP1	c.50T>C	p.Leu17Pro	-	[50]	c.50T>C	p.Leu17Pro	-	[50]	c.374C>T	p.Thr125Met	rs137886124	[a]		f	10	LCA	ar	possible Tur	
C8B1: c.3397G>A																				
p.Val1133Met																				
SEMA4A:																				
4	CEP290	c.3640dupG	p.Glu1214Gly	-	[a]	c.6604delA	p.Ile2202Leu	-	[51]	c.1301T>C	p.Met434Thr	rs146822426	[a]		f	2	LCA	ar	no	Au
fs*7																				
80	CEP290	c.2578G>T	p.Glu860*	-	[a]	c.3758G>A	p.Arg1253His	-	[a]	CEP290: c.5254C>T	p.Arg1752Trp	-	[a]		f	9	LCA (early CRD)	ar	no	Tur
FSQV2:																				
94	CEP290	c.6012-2A>G	splice	-	[a]	c.6870delT	p.Gln2291Lys	-	[52]	ABC44: c.1140T>A;	p.Asn380Lys	rs61748549	[4]		f	5	LCA	ar	no	KSA
fs*10																				
c.5642C>T;																				
p.Ala188Val																				
c.5882G>A																				
p.Gly1961Glu																				
[748]																				
RP1:																				
c.1380G>C																				
p.Lys460Asn																				
rs143494598																				
[a]																				
6	CRB1	c.2842+2T>A	splice	-	[a]	c.2842+2T>A	splice	-	[a]						m	20	LCA	no	yes	Tur
63	CRB1	c.1180T>C	p.Cys394Arg	-	[a]	c.1180T>C	p.Cys394Arg	-	[a]						m	6	LCA	ar	yes	KSA
110	CRX	deletion of exon 4 NMD?	-	-	[a]	c.899A>G	p.(*)300Trp	-	[a]	RP1: c.3191G>T	p.Ser1064Ile	-	[a]		f	9	LCA	ar	yes	Tur
ext*118)																				
SPATA7:																				
c.1112T>C																				
p.Ile371Thr																				
rs150364664																				
[a]																				

Table 1. Cont.

Patient	Gene	Allele 1				Allele 2				Additional Allele				Ref	Gender	Age (years)	Phenotype	Inheritance (family history)	Consang.	Origin
		cds	Protein	db SNP	Ref	cds	Protein	dbSNP	Ref	cds	Protein	dbSNP	Ref							
56	<i>GUCY2D</i>	c.1093_1106del	p.Arg365Trp fs*77	-	[a]	c.1093_1106del	p.Arg365Trp fs*77	-	[a]						m	4	LCA	ar	yes	Tur
71	<i>GUCY2D</i>	c.1401dupT	p.Leu468Ser fs*89	-	[a]	c.1401dupT	p.Leu468Ser fs*89	-	[a]	<i>RDH12</i> : c.917T>G	p.Val306Gly	-	[a]		m	5	LCA	ar	n.d.	KSA
73	<i>GUCY2D</i>	c.2766C>G	p.Tyr922*	-	[a]	c.2766C>G	p.Tyr922*	-	[a]	<i>GUCY2D</i> : c.2927G>T	p.Arg976Leu	rs61750184	[54]		f	18	LCA	ar	yes	Tur
										c.2927G>T	p.Arg976Leu	rs61750184	[54]							
118	<i>GUCY2D</i>	c.2080C>T	p.Gln694*	-	[a]	?									m	5	LCA	ar	no	Ger
123	<i>GUCY2D</i>	c.389delC	p.Pro130Leu fs*36	rs61749670	[55]	c.389delC	p.Pro130Leu fs*36	rs61749670	[55]						f	1	LCA	ar	no	Mor
125	<i>GUCY2D</i>	c.1401dupT	p.Leu468Ser fs*89	-	[a]	c.1401dupT	p.Leu468Ser fs*89	-	[a]	<i>RDH12</i> : c.917T>G	p.Val306Gly	-			f	5	LCA	ar	yes	KSA
107	<i>LCA5</i>	c.763C>T	p.Arg255*	rs151017794	[a]	c.763C>T	p.Arg255*	rs151017794	[a]	<i>ABCA4</i> : c.1927G>A	p.Val643Met	rs61749417	[10]		f	1	LCA	ar	n.d.	KSA
										<i>PDE6B</i> : c.704G>A	p.Arg235His	-	[a]							
84	<i>LRAT</i>	c.233_242del	p.Leu78Arg fs*85	-	[a]	c.233_242del	p.Leu78Arg fs*85	-	[a]						m	19	LCA	ar	yes	KSA
87	<i>LRAT</i>	c.449dupG	p.Phe151Leu fs*33	-	[a]	c.449dupG	p.Phe151Leu fs*33	-	[a]						f	11	LCA	ar	yes	Tur
70	<i>MERTK</i>	c.1744_1751 delinsT	p.Ile582* delinsT	-	[a]	c.1744_1751 delinsT	p.Ile582* delinsT	-	[a]	<i>RBP2</i> : c.2789T>C	p.Ile930Thr	-	[a]		m	16	LCA	ar	yes	Tur
24	<i>RDH5</i>	c.602C>T	p.Ser201Phe	-	[a]	c.602C>T	p.Ser201Phe	-	[a]						f	15	LCA (early CRD)	ar	yes	Pak
49	<i>RDH12</i>	c.133A>G	p.Thr45Ala	-	[56]	c.133A>G	p.Thr45Ala	-	[56]	<i>GUCY2D</i> : c.2359T>G	p.Cys787Gly	-	[a]		m	12	LCA	ar	no	Ger
										<i>SPATA7</i> : c.3G>A	p.Met11Ile (p.Met17)	-	[57]							

Table 1. Cont.

Patient	Gene	Allele 1				Allele 2				Additional Allele				Gender	Age (years)	Phenotype	Inheritance (family history)	Consang.	Origin
		cds	Protein	db SNP	Ref	cds	Protein	dbSNP	Ref	cds	Protein	dbSNP	Ref						
120	RDH12	c.188G>T	p.Gly63Val (splice)	-	[a]	c.188G>T	p.Gly63Val (splice)	-	[a]	ABC44: c.3482G>T	p.Arg1161Leu	-	[a]	m	7	LCA	ar	yes	KSA
95	RPE65	c.271C>T	p.Arg91Trp	rs61752871	[58]	c.271C>T	p.Arg91Trp	rs61752871	[58]	CDHR1: c.1202G>C, c.1202G>C LC45:	p.Gly401Ala, p.Gly401Ala;	-	[a]	f	28	LCA	ar	n.d.	KSA
35	RPGRIPI	c.2608_2609InsA	p.Leu870Tyr fs*7	-	[a]	c.2608_2609InsA	p.Leu870Tyr fs*7	-	[a]	RPE65: c.394G>A	p.Ala132Thr	rs61752878	[a]	f	9	LCA	ar	no	Dubai
50	RPGRIPI	c.1107delA	p.Glu370Asn fs*5	rs61751266	[59]	c.1107delA	p.Glu370Asn fs*5	rs61751266	[59]					m	6	LCA	ar	yes	KSA
81	RPGRIPI	c.3565C>T	p.Arg1189* fs*5	-	[a]	c.3565C>T	p.Arg1189* fs*5	-	[a]	TTC8: c.512A>T	p.Asp171Val	-	[a]	m	11	LCA	ar	n.d.	KSA
90	RPGRIPI	c.1107delA	p.Glu370Asn fs*5	rs61751266	[59]	c.1107delA	p.Glu370Asn fs*5	rs61751266	[59]	MERTK: c.2435A>C	p.Tyr812Ser	rs141361084	[a]	m	1	LCA	ar	n.d.	KSA
109	RPGRIPI	c.2662C>T	p.Arg888* fs*5	-	[a]	c.2662C>T	p.Arg888* fs*5	-	[a]	C2ORF71: c.1844T>A	p.Val615Asp	rs140776870	[25]	f	6	LCA	ar	yes	KSA
111	RPGRIPI	c.1107delA	p.Glu370Asn fs*5	rs61751266	[59]	c.1107delA	p.Glu370Asn fs*5	rs61751266	[59]	RBP3: c.490G>A	p.Ala164Thr	-	[a]	f	6	LCA	ar	yes	KSA
115	RPGRIPI	c.1107delA	p.Glu370Asn fs*5	rs61751266	[59]	c.1107delA	p.Glu370Asn fs*5	rs61751266	[59]	PHOM1: c.2094C>A	p.Ser698Arg	-	[a]	f	2	LCA	ar	yes	KSA
96	TULP1	c.901C>T	p.Gln301* fs*5	-	[60]	c.901C>T	p.Gln301* fs*5	-	[60]	C2ORF71: c.1844T>A	p.Val615Asp	rs140776870	[25]	m	7	LCA	ar	yes	KSA
112	TULP1	c.901C>T	p.Gln301* fs*5	-	[60]	c.901C>T	p.Gln301* fs*5	-	[60]					m	19	LCA	ar	no	KSA
114	TULP1	c.1604T>C	p.Phe535Ser	-	[a]	c.1604T>C	p.Phe535Ser	-	[a]					f	7	LCA	ar	n.d.	UEA
117	TULP1	c.901C>T	p.Gln301* fs*5	-	[60]	c.901C>T	p.Gln301* fs*5	-	[60]					f	8	LCA	ar	yes	KSA
122	TULP1	c.901C>T	p.Gln301* fs*5	-	[60]	c.901C>T	p.Gln301* fs*5	-	[60]					f	3	LCA	ar	yes	KSA
124	TULP1	c.901C>T	p.Gln301* fs*5	-	[60]	c.901C>T	p.Gln301* fs*5	-	[60]	RPT1: c.5248G>T	p.Glu1750*	-	[a]	m	6	LCA	ar	possible	KSA

Reference

1. Eisenberger T, Neuhaus C, Khan AO, Decker C, Preising MN, et al. (2013) Increasing the Yield in Targeted Next-Generation Sequencing by Implicating CNV Analysis, Non-Coding Exons and the Overall Variant Load: The Example of Retinal Dystrophies. PLoS ONE 8(11): e78496. doi:10.1371/journal.pone.0078496.