Supplementary Table of Zanolli et al., "Genetic testing for inherited ocular conditions in a developing country", *Ophthalmic Genetics*, 2020.

AREA	Clinical Diagnosis	Gene	Mutation
Adult Retinal Dystrophy	Basal Laminar Drunen	(-)	
Adult Retinal Dystrophy	Bietti Crystalline Dystrophy	*CYP4V2*	*c.1090+1G>T; c.1480C>T*
Adult Retinal Dystrophy	Coats plus Syndrome	*CTC1*[+]	c.2959C>T; c.248_251dupGCCA
Adult Retinal Dystrophy	COD	*ABCA4**	c.1289C>A; c.1289C>A
Adult Retinal Dystrophy	COD	*GUCA1A*	c.429_430insACA; c.428T>C
Adult Retinal Dystrophy	COD	(-)	
Adult Retinal Dystrophy	Congenital Fibrosis of Extraocular Muscles Type 2	(-)	
Adult Retinal Dystrophy	CORD	*ABCA4*	c.3113C>T; c.3113C>T
Adult Retinal Dystrophy	CORD	*ABCA4*	c.5882G>A; c.5461-10T>C
Adult Retinal Dystrophy	CORD	*ABCA4**	c.5929G>A; c.*55G>A
Adult Retinal Dystrophy	CORD	*RPGR*	c.2236_2237delGA
Adult Retinal Dystrophy	Familial Drusen	(-)	
Adult Retinal Dystrophy	Fundus Albipunctatus + COD	*RDH5*	c.712G>T
Adult Retinal Dystrophy	High myopia + retinal detachment + short stature	*COL2A1*[+]	c.2141G>T
Adult Retinal Dystrophy	Maculopathy	*SPATA7*[+]	c.1183C>T

Adult Retinal Dystrophy	Pattern Dystrophy	(-)	
Adult Retinal Dystrophy	Recessive bestrophinopathy	*BEST1**	c.388C>A; c.-37+5G>A
Adult Retinal Dystrophy	RP	*CDHR1*	c.1298delA; c.2108delG
Adult Retinal Dystrophy	RP	*EYS*	c.5834_delA; c.6794_delC
Adult Retinal Dystrophy	RP	*ROM1*	c.713T>C
Adult Retinal Dystrophy	RP	*RP1*	c.2098G>T ; c.2953A>T
Adult Retinal Dystrophy	RP	*RPGR*	c.2452G>T
Adult Retinal Dystrophy	RP	(-)	
Adult Retinal Dystrophy	RP	(-)	
Adult Retinal Dystrophy	RP	(-)	
Adult Retinal Dystrophy	Stargardt disease	*ABCA4*	c.1025_1038del14 ; c.5332A>G
Adult Retinal Dystrophy	Stargardt disease	*ABCA4*	c.4918c>t; 3386G>T
Adult Retinal Dystrophy	Stargardt disease	*ABCA4*	3386G>T; ; c.5332A>G
Adult Retinal Dystrophy	Usher Syndrome	*USH2A*	c.4377A>G; c.1550+16T>C
Adult Retinal Dystrophy	Usher Syndrome	*USH2A**	c.2299delG; c.13436T>G
Adult Retinal Dystrophy	Usher Syndrome	*USH2A**	c.2299delG; c.13436T>G
Childhood Retinal Disease	Achromatopsia	*CNGA3*	c.847C>T; c.1981C>A
Childhood Retinal	Achromatopsia	*CNGA3*	c.847C>T; c.1981C>A

Disease			
Childhood Retinal Disease	Achromatopsia	Opsin	Opsin Exon 2 gene deletion
Childhood Retinal Disease	Best disease	*BEST1*	c.763C>T
Childhood Retinal Disease	CSNB	*CACNA1F*	c.244C>T
Childhood Retinal Disease	CSNB	*TRPM1*	c.2072T>C; c.2899C>T
Childhood Retinal Disease	CSNB	*TRPM1*	c.428-1G>C
Childhood Retinal Disease	High Myopia + Hypoacusia	$(-)^{+}$	
Childhood Retinal Disease	Incontinentia Pigmenti	*IKBKG*	c.1116delT
Childhood Retinal Disease	Kearns Sayre syndrome	*Mitochondrial deletion*	Mt deletion 4400 BP
Childhood Retinal Disease	Knobloch syndrome	*COL18A1*	c.3513delG; c.3627+9_3627+10delCT
Childhood Retinal Disease	Knobloch syndrome	(-)	
Childhood Retinal Disease	LCA	*CRB1*	c.1666_1667dupCT; c.3110_3143dupTGACCCTTTCCATGACAGACCCACTGTCCCAGAC
Childhood Retinal Disease	LCA	*CRB1*	c.2843G>A; c.2843G>A
Childhood Retinal Disease	LCA	*CRB1*	c.2843G>A; c.2843G>A
Childhood Retinal Disease	LCA	*CRB1*	c.2843G>A; c.2843G>A
Childhood Retinal Disease	LCA	*CRB1*	c.2843G>A; c.2264T>C
Childhood Retinal Disease	LCA	*CRB1*	c.3110_3143dup34; c.2843G>A
Childhood Retinal Disease	LCA	*CRB1*	c.3988delG; c.2501G>A

Childhood Retinal Disease	LCA	*CRB1*	c.2843G>A; c.2843G>A
Childhood Retinal Disease	LCA	*CRB1*	c.2843G>A; c.2843G>A
Childhood Retinal Disease	LCA	*CRB1*	2843G>A; 2843G>A
Childhood Retinal Disease	LCA	*CRB1*	2843G>A; 2843G>A
Childhood Retinal Disease	LCA	*GUCY2D**	c.1762C>T; c.2190T>A
Childhood Retinal Disease	LCA	*LCA5*	c.1243G>T; c.1243G>T
Childhood Retinal Disease	LCA	*LCA5*	c.1243G>T; c.1243G>T
Childhood Retinal Disease	LCA	*LCA5*	c.1243G>T; c.1243G>T
Childhood Retinal Disease	LCA	*LCA5*	c.1243G>T; c.1243G>T
Childhood Retinal Disease	LCA	*RDH12*	c.295C>A; c.295C>A
Childhood Retinal Disease	LCA	*RDH12*	c.295C>A; c.716G>T
Childhood Retinal Disease	LCA	*RP1L1*$^{+}$	c.1138G>A
Childhood Retinal Disease	LCA	(-)	
Childhood Retinal Disease	LCA	(-)	
Childhood Retinal Disease	Norrie disease	*NDP*	Deletion of exon 2
Childhood Retinal Disease	Recessive bestrophinopathy	*BEST1**	c.172G>C; c.599delC
Childhood Retinal Disease	Recessive bestrophinopathy	*BEST1*	c.888C>A; c.584C>T
Childhood Retinal	Recessive bestrophinopathy	*BEST1*	c.920+1G>A; c.920+1G>A

Disease			
Childhood Retinal Disease	Retinopathy associated with *NR2E3* gene	*NR2E3*	c.767C>T; c.311G>A
Childhood Retinal Disease	RP	*RPGR*	c.2426_2427delAG
Childhood Retinal Disease	RP	*RPGR*	c.2426_2427delAG
Childhood Retinal Disease	RP	*RPGR**	c.2431G>T
Childhood Retinal Disease	Stargardt disease	*ABCA4*	3386G>T; 3386G>T
Childhood Retinal Disease	Stargardt disease	*ABCA4*	c.1025_1038del14; c.3113C>T
Childhood Retinal Disease	Stargardt disease	*ABCA4**	c.1289C>A;c.768G>C
Childhood Retinal Disease	Stargardt disease	*ABCA4*	c.388C>A; c.388C>A
Childhood Retinal Disease	Stargardt disease	*ABCA4**	c.1289C>A; c.1289C>A
Childhood Retinal Disease	Stargardt disease	*ABCA4*	c.1025_1038del14; c.3113C>T
Childhood Retinal Disease	Stargardt disease	*ABCA4*	c.1622T>C; c.3113C>T
Childhood Retinal Disease	Stargardt disease	*ABCA4*	c.1025_1038del14; c.3113C>T
Childhood Retinal Disease	Stargardt disease	*ABCA4*	c.5692C>T; c.1937+1G>A
Childhood Retinal Disease	Stargardt disease	*ABCA4**	c.5929G>A; c.388C>A
Childhood Retinal Disease	Stargardt disease	*ABCA4*	c.634C>T;c.768G>C
Childhood Retinal Disease	Usher syndrome	*CDH23*	c.6393delC; c.7979_7986delACTGGGAG
MAC spectrum	Gorlin Goltz syndrome	*PORCN*	c.178G>A

MAC spectrum	MAC spectrum	*PUF60*[+]	c.850dup
MAC spectrum	MAC spectrum		arr[hg19] 14q22.2q23.2(54331256-63789238)x1 (including *SIX1, OTX2*)
MAC spectrum	MAC spectrum	(-)	
MAC spectrum	MAC spectrum	(-)	
MAC spectrum	MAC spectrum	(-)	
MAC spectrum	MAC spectrum	(-)	
MAC spectrum	MAC spectrum	(-)	
MAC spectrum	MAC spectrum	(-)	
MAC spectrum	MAC spectrum	(-)	
Other	Aniridia	*PAX6*	c.408_409dupGA
Other	Autosomal Dominant Optic Atrophy	*OPA1*	c.3G>A
Other	Autosomal Dominant Optic Atrophy	(-)	
Other	Congenital Glaucoma	(-)	
Other	FEVR	*NDP*	c.162G>C
Other	FEVR	(-)	
Other	FEVR	(-)	
Other	Leber Hereditary Optic Neuropathy	*MT-MD1*	m.3460G>A
Other	Sotos syndrome	*NSD1*	Deletion of entire sequencing region
Other	Wolfram Syndrome	*WFS1*	c.409_424dup16; c.409_424dup16

MAC: Microphthalmia Anophthalmia Coloboma; COD: Cone Dystrophy; CORD: Cone and Rod Dystrophy; CSNB: Congenital Stationary Night Blindness; LCA: Leber Congenital Amaurosis; FEVR: Familial exudative vitreoretinopathy; RP: Retinitis pigmentosa; [+] Diagnosed with Whole Exome Sequencing.
*Eleven patients with variances of unknown significance were re-classified into probably pathogenic or pathogenic variant (10,6%).