Supplementary Data 3: Pathogenic, likely pathogenic and variants of unknown significance found in a cohort of 95 individuals with Peters' anomaly.

Caption: D: deleterious, LB: Likely Benign, LP: Likely Pathogenic, MAF: minor allele frequency, NA: not applicable, P: Pathogenic, pD: possibly damaging, PD: probably damaging, T: tolerated, VUS: Variant of unknown significance. ^ain the following order: SIFT, MutTaster, Polyphen-2 ^baccording to Richards et al. 2015,[1]

A: Pathogenic and likely pathogenic SNVs

Gene	Individuals	Nucleotide	Protein alteration	gnomAD	Prediction	Functional	ClinVar	Literature	ACMG classification ^b
	D15			IVIAF	LOOIS	domain	Variation ID	Web at al	
NM_194318.4	P15	C.459+1G>A	p.r		NA		P (449340)	2014,[2]	P (PVS1, PIVIZ, PP3, PP5)
	P3, P56, P71, P87, P93	c.660+1G>A	p.?		NA		P (1264)	Weh <i>et al.</i> 2014,[2]	P (PVS1, PM2, PP3, PP5)
	Р3	c.1178G>A	p.(Gly393Glu)		D, D, PD	fringe-like	P (1268)	Weh <i>et al.</i> 2014,[2]	LP (PM2, PM3, PM5, PP3)
	P87	c.1184+3A>G	p.?	0.0007%	NA				LP (PS3, PM2, PM3)
<i>PAX6</i> NM_000280.4	P7	c.38G>C	p.(Gly13Ala)		D, D, PD	paired domain			P (PS2, PM1, PM2, PP3)
	P33	c.52G>C	p.(Gly18Arg)		D, NA, PD	paired domain	P (289849)	Hever et al. 2006,[3]	P (PM1, PM2, PM5, PP3, PP5)
	P46	c.622C>T	p.(Arg208Trp)	0.0004%	D, D, PD	homeodomain	P (372441)	Lim et al. 2012,[4]	P (PM1, PM2, PP3, PP5)
	P74	c.786G>C	p.(Arg262Ser)		D, D, PD	homeodomain			P (PM1, PM2, PM5, PP1, PP3)
SOX2	P45	c.22G>T	p.(Glu8*)		NA				LP (PVS1, PM2)
NM_003106.4	P91	c.70_89del	p.(Asp24Argfs*65)		NA		P (94104)	Suzuki <i>et al.</i> 2014,[5]	P (PVS1, PM6, PM2, PP5)
	P95	c.70_89dup	p.(Gly31Thrfs*22)		NA				P (PVS1, PM6, PM2)
	P94	c.103A>T	p.(Lys35*)		NA			Shima <i>et al.</i> 2017,[6]	P (PVS1, PM2, PP5)
<i>PITX3</i> NM_005029.4	P14	c.38G>A	p.(Ser13Asn)	0.0004%	D, D, PD		P (6938)	Semina <i>et al</i> . 1998,[7]	P (PM2, PP1, PP3, PP5)
	P41, P50	c.640_656dup	p.(Gly220Profs*95)		NA		P (468353)	Summers <i>et al.</i> 2008,[8]	P (PVS1, PM2, PP5)
CYP1B1 NM_000104.4	P36	c.171G>A	p.(Trp57*)	0.018%	NA		P (7737)	Ma et al. 2020,[9]	P (PVS1, PM2, PM3, PP5)
	P5	c.317C>A	p.(Ala106Asp)	0.002%	D, D, PD			Medina-Trillo et al. 2016,[10]	P (PS3, PM2, PP2, PP3)
	P36	c.868dup	p.(Arg290Profs*37)	0.003%	NA		P (68468)	Zhou <i>et al.</i> 2017,[11]	P (PVS1, PM2, PM3, PP5)
	P41	c.1064_1076del	p.(Arg355Hisfs*69)	0.022%	NA		P (282564)	García-Antón et al. 2017,[12]	P (PVS1, PM2, PP3, PP5)
	P5	c.1200_1209dup	p.(Thr404Serfs*30)	0.026%	NA		P (68466)	García-Antón <i>et</i> <i>al.</i> 2017,[12]	P (PVS1, PM2, PP3, PP5)
	P38	c.1310C>T	p.(Pro437Leu)	0.0008%	D, D, PD			Medina-Trillo et al. 2016,[10]	P (PS3, PM2, PP2, PP3)
<i>FOXE3</i> NM_012186.3	P64, P90	c.232G>A	p.(Ala78Thr)	0.004%	D, D, PD	forkhead	P (427852)	Plaisancié <i>et al.</i> 2018 (same patient,P2),[13]	LP (PM2, PP1, PP2, PP3, PP5)
	P90	c.253dup	p.(Ala85Glyfs*200)		NA				LP (PVS1, PM2)
	P70	c.960A>C	p.(*320Cys*72)		NA			Plaisancié <i>et al</i> . 2018,[13]	LP (PM2, PM4, PP5)
PITX2 NM_153426.2	P52	c.311T>C	p.(Phe104Ser)		D, D, PD	homeobox			LP (PM1, PM2, PP2, PP3)
RARB NM_000965.5	P65	c.1151G>A	p.(Gly384Asp)		D, D, PD				LP (PM1, PM2, PP2, PP3)
COL4A1 NM_001845.6	P27	c.615+1G>A	p.?		NA				P (PVS1, PM2, PP1)
HCCS NM_005333.5	P6	c.589C>T	p.(Arg197*)		NA		P (11670)	Van Rahden <i>et</i> <i>al.</i> 2014 (same patient :P2),[14]	P (PVS1, PM2, PM6, PP3, PP5)

B: Pathogenic CNVs

Of the 95 patients, three had a CNV in an eye development gene (bold) and four patients had pathogenic CNVs that did not encompass ocular development genes.

Individuals	CNVs	Size	OMIM morbid genes	Literature
P2	arr[GRCh38]11p13(31141793_32226400)x1	1,1Mb	РАХ6	Lima Cunha <i>et al.</i> 2019[15]
P59	arr[GRCh38]6p25.3(1386760_1822396)x3	435 kb	FOXC1, FOXF2, GMDS	D'haene <i>et al.</i> 2011[16]
P58	rsa[GRCh38] 13q12.3(<i>B3GLCT</i>)x0	NA	B3GLCT (exons 1-2)	
P4		2 4146	TOV1 17TD1 SIC2EA1 COMT	Poir at a/ 2015[17]
P83	an[Grch59]22411.2(22.13022273_21030120)x1	2,41010	IBAT, LETAT, SLOZSAT, CONT	Keis et ul. 2013[17]
P72	arr[GRCh38]8q21.11q21.2(75652023_85480932)x1	10Mb	PEX2, ZFHX4, IL7, MRPS28, PMP2, IMPA1, CA2	Happ <i>et al.</i> 2016[18]
P86	arr[GRCh38]13q31.1q33.2(82528272_106198042)x1	27,7Mb	CLDN10, DAOA, DNAKC3, ERCC5, FRF14, GPC6, MIR17HG, NALCN, PCCA, SLC10A2, SLITRK1, SLITRK6, TGDS, ZIC2	Quélin <i>et al</i> . 2009[18]

C: Variants of unknown significance (VUSs)

We identified a total of 62 of unknown significance variants (60 SNVs and 2 CNV) among the 95 patients of our cohort. For some of these VUSs, further investigations, including segregation analyses, are needed to conclude on the benign or pathogenic status of these variants.

SNVs

All of these variants were found at heterozygous state in individuals, with the exception of the *PXDN* (NM_012293.2):c.2569T>C found at homozygous state in patient P18. None of the detected intronic and synonymous variants is predicted to significantly alter the splicing according to *in silico* software (SSF-like, MaxEnt, NNSPLICE, GeneSplicer and SpliceAI).

In P18, who had isolated bilateral PA, we identified a novel homozygous variant in *PXDN*: c.2569T>C p.(Cys857Arg), predicted to be deleterious by the in silico prediction tools. Although classified as a VUS (segregation analysis not performed due to the absence of parental DNA), it remains a good candidate as *PXDN* is a gene involved in microphthalmia and ASD previously reported in patients with PA.[19]

Two previously reported patients (P16 and P27) each had a VUS in *PTCH1* according to ACMG criteria, originally described as pathogenic after *in vivo* modelling studies.[20] Although we did not find a causative variant in the patient P16, the P27 patient, however, has a pathogenic variant in *COL4A1*.

Gene transcript	Individuals	Nucleotide alteration	Protein alteration	gnomAD MAF	Prediction tools ^a	Domain	ClinVar variation ID	Literature	ACMG classification ^b
ABCB6 NM_005689.4	P79	c.2314G>T	p.(Val772Phe)	0.014%	D, D, PD	ABC transporter			VUS (PM1, PP3, BS2)
ADAMTS18 NM_199355.3	P49	c.779-4G>A	p.?	0.0092%	NA, NA, NA	intron	LB (1124887)		VUS (PM2, BP4)
AHR NM_001621.4	P91	c.2525T>A	p.(Leu842*)		NA, NA, NA				VUS (PM2, BP5)
ALDH1A3 NM_000693.3	P68	c82C>G		0.049%	NA, NA, NA	5' UTR			VUS (PM2)
	P19	c.275A>G	p.(Asp92Gly)	0.00040%	D, D, PD	aldehyde dehydrogenase			VUS (PM2, PP3)
	P51	c.852G>T	p.(Lys284Asn)		D, D, pD	aldehyde dehydrogenase			VUS (PM2, PP3)
COL4A1 NM_001845.5	P69	c.3876+14T>C	p.?		NA, NA, NA	intron			VUS (PM2, BP4)
	P23	c.4755+24A>G	p.?		NA, NA, NA	intron			VUS (PM2, BP4)
CPAMD8 NM_015692.5	P69	c.434-28T>C	p.?	0.021%	NA, NA, NA	intron			VUS (PM2, BP4)
	P80	c.671A>G	p.(Tyr224Cys)	0,002%	D, B, PD				VUS (PM2, PP3)
	P81	c.700+19G>A	p.?	0,032%	NA, NA, NA	intron			VUS (PM2, BP4)
CREBBP NM_004380.2	P93	c.914A>T	p.(Asn305Ile)		D, B, PD				VUS (PM2, PP3, BP5)
	P77	c.2283+16C>G	p.?		NA, NA, NA	intron			VUS (PM2, BP4)
CYP1B1 NM_000104.3	P12	c.101A>T	p.(His34Leu)		B, B, pD				VUS (PM2, PP2)
	P30	c.1044-11A>C	p.?	0.0090%	NA, NA, NA	intron			VUS (PM2, BP4)
	P44	c.1204G>A	p.(Ala402Thr)		D, B, pD				VUS (PM2, PP3)
EP300 NM_001429.3	P13	c.4448A>G	p.(Tyr1483Cys)		D, D, PD	HAT			VUS (PM2, PP3)
FAT1 NM_005245.3	P17	c.155A>T	p.(Tyr52Phe)	0.00080%	D, B, PD	cadherin			VUS (PM2, PP3)
	P66	c.4323+26T>C	p.?		NA, NA, NA	intron			VUS (PM2, BP4)
	P93	c.5563C>T	p.(Pro1855Ser)	0.0020%	D, B, PD	cadherin			VUS (PM2, PP3, BP5)

Gene transcript	Individuals	Nucleotide alteration	Protein alteration	gnomAD MAF	Prediction tools ^a	Domain	ClinVar variation ID	Literature	ACMG classification ^b
	P17	c.6796G>A	p.(Glu2266Lys)		D, B, pD	cadherin			VUS (PM2, PP3)
FRAS1 NM_025074.6	P62	c.2345A>T	p.(His782Leu)	0.019%	D, D, B	Furin-like repeat	VUS (906237)		VUS (PM2, BP4)
	P76	c.3058C>T	p.(Arg1020Cys)	0.079%	D, D, PD		VUS (290048)		VUS (PM2, PP3)
	P78	c.4579C>T	p.(Arg1527Trp)	0.2707%	D, B, pD		LB/VUS (196928)	Kohl <i>et al.</i> 2014, [21]	VUS (PP5)
	P76	c.7769G>A	p.(Arg2590His)	0.0096%	В, В, В				VUS (PM2, BP4)
	P78	c.9364C>T	p.(Arg3122Trp)	0.030%	D, D, PD		LB/VUS (198324)		VUS (PM2, PP3)
FREM1 NM_144966.5	P78	c.1493G>A	p.(Arg498Gln)	0.039%	D, D, PD		VUS (30767)		VUS (PM2, PP3)
FZD4 NM_012193.3	P26	c.1468A>T	p.(Thr490Ser)		D, B, PD	trans- membrane			VUS (PM2, PP3)
FZD5 NM_003468.3	P54	c25T>A	p.?		NA, NA, NA	5' UTR			VUS (PM2, BP4)
GDF3 NM_020634.1	P89	c.796C>T	p.(Arg266Cys)	0.20%	D, D, PD	disulfide bond	P/LP/LB (30591)		VUS (PM1, PP5, BS1)
HCCS NM_005333.4	P1	c.608+5G>A	p.?		NA, NA, NA	intron			VUS (PM2, BP4)
HMX1 NM_018942.2	P39	c.250C>G	p.(Arg84Gly)	0.0071%	B, B, pD		VUS (939776)		VUS (PM2, BP4)
ITPR1 NM_001099952.2	P32	c.4222C>T	p.(Arg1408Cys)		D, D, PD				VUS (PM2, PP3)
LTBP2 NM_000428.2	P55	c.494+37G>A	p.?		NA, NA, NA	intron			VUS (PM2, BP4)
	P90	c.1612C>T	p.(Arg538Trp)	0.022%	D, B, PD		VUS (888083)		VUS (PM2, PP3, BP5)
	P92	c.1987+20G>A	p.?	0.012%	NA, NA, NA	intron			VUS (PM2, BP4)
MFRP NM_031433.3	P53	c.*1C>G	p.?	0.0015%	NA, NA, NA	3' UTR			VUS (PM2)
NOTCH1 NM_017617.4	P63	c.3989G>T	p.(Arg1330Leu)		D, B, pD	EGF-like			VUS (PM2)
PAX6 NM_000280.4	P69	c129+273G>T	p.?		NA, NA, NA	5' UTR			VUS (PM2, BP4)
	P69	c128-1601A>G	p.?		NA, NA, NA	5' UTR			VUS (PM2, BP4)
	P82	c128-1096C>T	p.?		NA, NA, NA	intron			VUS (PM2, BP4)
	P63	c.358-124C>T	p.?		NA, NA, NA	intron			VUS (PM2, BP4)
PRSS56 NM_001195129.1	P18	c.1400G>T	p.Arg467Leu	0.051%	D, NA, PD		LP (191153)		VUS (PM2, PP5)
<i>PTCH1</i> NM_000264.5	P69	c.2561-656C>T	p.?	0.0032%	NA, NA, NA	intron			VUS (PM2, BP4)
	P16	c.2695A>G	p.(Ile899Val)	0.0016%	B, D, B	extracellular	LP/VUS (221968)	Chassaing <i>et al.</i> 2016 (same	VUS (BS2, PS3, PM2)
	P35	c.3715C>T	p.(Arg1239Trp)	0.0018%	D, B, pD		VUS (569497)	patient:CC10),[20]	VUS (PM2)
	P27	c.3947A>G	p.(Tyr1316Cys)	0.05%	T, D, PD		LP/VUS/LB/B (136471)	Chassaing et al. 2016 (same	VUS (PS3, BS1)
PXDN NM 012293.2	P83	c.1992G>C	p.(Pro664=)	0.0032%	NA, NA, NA			patient:P20),[20]	VUS (PM2, BP5)
	P18	c.2569T>C	p.(Cys857Arg)		D, D, PD	disulfide bond domain			VUS (PM1, PP2, PP3)
	P48	c.3189C>T	p.(Asn1063=)	0.0082%	NA, NA, NA	haem peroxydase			VUS (PM2, BP4)
RAB3GAP2 NM_012414.3	P10	c.2578-3T>C	p.?		NA, NA, NA	intron			VUS (PM2, BP4)
RAX NM 013435.2	P53	c.197G>C	p.(Arg66Thr)	0.21%	D, D, B		VUS (496932)		VUS (PM2)
RERE NM_012102.3	P61	c.263G>A	p.(Arg88Lys)	0.00080%	D, B, pD				VUS (PM2, PP3)

Gene transcript	Individuals	Nucleotide alteration	Protein alteration	gnomAD MAF	Prediction tools ^a	Domain	ClinVar variation ID	Literature	ACMG classification ^b
SALL1 NM_002968.2	P63	c.466_477del	p.(Ser156_Ser159del)		NA, NA, NA				VUS (PM2)
SALL2 NM_005407.2	P66	c.947C>T	p.(Ser316Leu)		D, NA, pD				VUS (PM2, PP3)
	P82	c.2855G>A	p.(Arg952Gln)	0.0024%	B, NA, PD	Zinc finger C2H2			VUS (PM2)
SRD5A3 NM_024592.4	P76	c.951_955del	p.(Phe318Ser)	0.053%	D, NA, PD	C-Terminal	VUS (632442)		VUS (PM2, PP3)
TENM3 NM_001080477.3	P55	c.788T>C	p.(Leu263Pro)		D, NA, PD	teneurin N- terminal			VUS (PM2, PP3)
	P63	c.4124G>T	p.(Arg1375Leu)		B, NA, PD				VUS (PM2)
VSX2 NM_182894.2	P35	c.539T>C	p.(Met180Thr)	0.012%	D, D, B	homeobox	VUS (967386)		VUS (PM2, PP3)

CNVs

In two patients we found a CNV of unknown significance.

First, patient P67, investigated for right PA, intellectual disability, GR, brachydactyly, polycystic ovary and Hirschsprung's disease, who has the recurrent 16p11.2 deletion (arr[GRCh38] 16p11.2(29662633_30187279)x1). This deletion has previously been described as a risk factor for neurodevelopmental disorders and ocular malformations have occasionally been described (one patient with colobomatous microphthalmia and one patient with coloboma).[22] We did not find enough criteria to consider this CNV responsible for her ocular anomaly.

In the other patient (P29), we found a 436kb microduplication in 22q11.23 (arr[GRCh38] 22q11.23(23261243_23698049)x3). This rare duplication (8/21692 in gnomAD SVs v2.1 for a similar duplication: DUP_22_50935) has not been described in any other patient. It only contains three OMIM genes that have not been associated with ocular anomalies: *BCR*, *IGLL1* and *RLG4*. One patient with a larger (1.3Mb) and more distal 22q11.23 duplication has been reported to have an optic nerve coloboma but no sequencing of OD genes was performed to rule out another causative variant.[23] It was therefore considered as of uncertain significance.

Individuals	CNVs	Size	OMIM morbid genes	Literature
P67	arr[GRCh38] 16p11.2(29662633_30187279)x1	525 kb	PRRT2, TBX6, KIF22, TLCD3B, ALDOA, CORO1A	Stingl <i>et al.</i> 2020[22]
P59	arr[GRCh38] 6p25.3(1386995_1822630)x3	435 kb	BCR, IGLL1	D'haene <i>et al.</i> 2011[16]

Web resources:

Polyphen2: <u>http://genetics.bwh.harvard.edu/pph2/</u> Mutation Taster: <u>http://www.mutationtaster.org</u> SIFT: <u>https://sift.bii.a-star.edu.sg</u> SSF-like (Splice Site Finder-like): <u>https://extranet.interactive-biosoftware.com/AlamutVisual-</u> <u>2.15 Documentation/splicing.html</u> MaxEnt: <u>http://hollywood.mit.edu/burgelab/maxent/Xmaxent.html</u> NN SPLICE: <u>https://www.fruitfly.org/seq_tools/splice.html</u> GeneSplicer: <u>https://ccb.jhu.edu/software/genesplicer/</u> SpliceAI: <u>https://github.com/Illumina/SpliceAI</u> gnomAD: https://gnomad.broadinstitute.org

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