

**Supplementary Table 3. Pathogenicity classification of all variants in this study.**

Patient ID	Transcript ID	Genetic variants	Zygoty	Hereditate mode	Classification	Evidence level			gnomAD frequency	Popmax	Reference
						Strong	Moderate	Supporting			
1880	NM_003140.2	<i>SRY</i> c.397C>T, p.Arg133Trp	hemi	YL	P	PS1+PS2	PM2	PP3+PP4	/	/	9150734, 8353496
1886	NM_000348.3	<i>SRD5A2</i> c.419_421delinsATTC	het	AR	P	PVS1	PM2	PP4	/	/	/
1909	NM_000233.3	<i>LHCGR</i> c.1541G>C, p.Cys514Ser	homo	AR	VUS	/	PM2	PP3+PP4	/	/	/
2036	NM_000044.3	<i>AR</i> c.142G>T, p.Ala48Ser	hemi	XR	VUS	/	PM2	PP3+PP4	0.000009492	0	/
2061	NM_000216.2	<i>ANOS1</i> del Exon 4-5	hemi	XR	LP	PVS1	PM2	/	/	/	16882753
2356	NM_000406.2	<i>GNRHR</i> c.806C>T, p.Thr269Met	homo	AR	LP	/	PM2+PM5	PP1+PP2+PP3	0.00001194	0.00000293 (European non-Finnish)	22031817, 26708526, 27544332
2367	NM_000102.3	<i>CYP17A1</i> c.985_987delinsAA, p.Tyr329Kfs*90	homo	AR	P	PVS1	PM2	PP4	0.00004599	0.0006317 (East Asian)	/
2420	NM_000348.3	<i>SRD5A2</i> c.680G>A, p.Arg227Gln	homo	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
2439	NM_004959.4	<i>NR5A1</i> c.721C>T, p.Arg241Trp	het	AD	LP	PS2	PM1+PM2	PP3	/	/	29095814
2473	NM_032551.4	<i>KISS1R</i> c.631G>A, p.Ala211Thr	het	AR	VUS	/	PM1+PM2	PP3	0.0001812	0.001506 (East Asian)	/
2540	NM_000044.3	<i>AR</i> c.1787G>A, p.Cys596Tyr	hemi	XR	P	PS2	PM1+PM2	PP3+PP4	/	/	/
2692	NM_144773.2	<i>PROKR2</i> c.533G>C, p.Trp178Ser	het	AD	VUS	PS3+ BS1	/	PP3	0.0002227	0.00231 (East Asian)	17054399
2752	NM_000044.3	<i>AR</i> c.2528T>C, p.Ile843Thr	hemi	XR	LP	/	PM2+PM5	PP3+PP4	/	/	8723113
2782	NM_000348.3	<i>SRD5A2</i> c.680G>A, p.Arg227Gln	het	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
2786	NM_000348.3	<i>SRD5A2</i> c.281G>A, p.Arg94Lys	het	AR	LP	/	PM2+PM3	PP3+PP4	/	/	/
		<i>SRD5A2</i> c.702C>G, p.Phe234Leu	het	AR	VUS	/	PM2	PP3	0.0002037	0.004319 (East Asian)	15266301
2892	NM_000348.3	<i>SRD5A2</i> c.607G>A, p.Gly203Ser	het	AR	P	PS1	PM1+PM2	PP3+PP4	0.00002629	0.0001567 (East Asian)	9135696, 15266301, 32713132
		<i>SRD5A2</i> c.737G>A, p.Arg246Gln	het	AR	LP	/	PM1+PM2+PM5	PP3+PP4	0.000126	0.0004087 (South)	1522235, 26446026
2955	NM_000216.2	<i>ANOS1</i> c.1712G>A, p.Trp571*	hemi	XR	P	PVS1+PS2	PM2	/	/	/	/
		<i>LHCGR</i> c.681-1Gly>Ala	het	AR	P	PVS1	PM2	PP3+PP4	/	/	29305568
3167	NM_000233.3	<i>LHCGR</i> c.1582_1585delATAT, p.Ile528*	het	AR	P	PVS1	PM2+PM3	PP4	/	/	29305568
3828	NM_000348.3	<i>SRD5A2</i> c.457T>C, p.Phe153Leu	het	AR	VUS	/	PM2	PP3	/	/	/
3874	NM_021044.2	<i>DHH</i> c.968G>A, p.Gly323Asp	het	AR	VUS	/	PM2	PP3	/	/	/
3879	NM_000216.2	<i>ANOS1</i> c.984C>G, p.Tyr328*	hemi	XR	LP	PVS1	PM2	/	/	/	8989261
3973	NM_005634.2	<i>SOX3</i> c.424C>A, p.Pro142Thr	hemi	XL	LP	PS3	PM2	PP4	/	/	30125608
4004	NM_000044.3	<i>AR</i> c.1175C>G, p.Pro392Arg	hemi	XR	LP	/	PM1+PM2	PP3+PP4	0.00001776	0	10571951
4119	NM_003309.3	<i>TSPYL1</i> c.67C>T, p.Gln23*	het	AR	LP	PVS1	PM2	/	0.0001441	0.001463 (East Asian)	/
4136	NM_001059.2	<i>TACR3</i> c.196dupG, p.Ala66fs*84	het	AR	LP	PVS1	PM2	/	/	/	/
4208	NM_000044.3	<i>AR</i> c.2256G>A, p.Trp752*	hemi	XR	P	PVS1	PM2	PP4	/	/	19463997, 10458483, 27284311
4274	NM_000145.3	<i>FSHR</i> c.227A>C, p.Glu76Ala	het	AR	VUS	/	PM1+PM2	PP3	/	/	/
4431	NM_000941.2	<i>POR</i> c.1196_1204delCCTCGGAGC, p.Pro399_Glu401del	het	AR	VUS	/	PM2+PM4	/	/	/	21843508
4500	NM_144773.2	<i>PROKR2</i> c.892C>T, p.Arg298Cys	het	AD	VUS	/	PM2	PP3	/	/	/
		<i>SRD5A2</i> c.269A>C, p.His90Pro	het	AR	LP	/	PM2+PM3	PP3	/	/	/
4622	NM_000348.3	<i>SRD5A2</i> c.680G>A, p.Arg227Gln	het	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
4940	NM_000102.3	<i>CYP17A1</i> c.785T>G, p.Met262Arg	het	AR	VUS	/	PM2	PP3+PP4	/	/	/
		<i>CYP17A1</i> c.1193C>T, p.Ala398Val	het	AR	VUS	/	PM2	PP3+PP4	/	/	14552333
4986	NM_004959.4	<i>NR5A1</i> c.251G>A, p.Arg84His	het	AD	P	PS3	PM1+PM2+PM5	PP3	/	/	17694559, 27899157, 24434652
		<i>AMH</i> c.585C>A, p.Tyr195*	het	AR	LP	PVS1	PM2	/	/	/	/
5015	NM_000479.3	<i>AMH</i> c.1165G>T, p.Glu389*	het	AR	LP	PVS1	PM2	/	1.97884E-05	/	31277073, 32172781
5175	NM_005270.4	<i>GLI2</i> c.3463_3464delGA, p.Asp1155Arg fs*39	het	AD	P	PVS1+PS2	PM2	/	/	/	/
5337	NM_000044.3	<i>AR</i> c.626G>A, p.Gly209Glu	hemi	XR	VUS	/	PM2	PP3	/	/	/
5421	NM_005921.1	<i>MAP3K1</i> c.2062C>G, p.Leu688Val	het	AD	VUS	/	PM1+PM2	PP3	/	/	/
5589	NM_005270.4	<i>GLI2</i> c.3137delG, p.Gly1046Ala fs*84	het	AD	LP	PVS1	PM2	/	/	/	/
6500	NM_000163.4	<i>GHR</i> c.136+1Gly>Ala	homo	AD/AR	VUS	/	PM2	PP3	/	/	/
6606	NM_005270.4	<i>GLI2</i> c.3640C>T, p.Gln1214*	het	AD	LP	PVS1	PM2	/	/	/	/
6791	NM_000348.3	<i>SRD5A2</i> c.680G>A, p.Arg227Gln	homo	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	/	8784107
6928	NM_000197.1	<i>HSD17B3</i> c.466T>A, p.Cys156Ser	het	AR	VUS	/	PM1+PM2	PP3	/	/	/
		<i>SRD5A2</i> c.656delT, p.Phe219Ser fs*60	het	AR	P	PVS1	PM2+PM3	PP4	0.00000657	/	20736251
7847	NM_000348.3	<i>SRD5A2</i> c.680G>A, p.Arg227Gln	het	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
7929	NM_004959.4	<i>NR5A1</i> c.848G>T, p.Cys283Phe	het	AD	LP	/	PM1+PM2+PM5	PP3	/	/	/
8065	NM_000044.3	<i>AR</i> del Exon5-8	hemi	XR	P	PVS1	PM2	PP4	/	/	8339746
8585	NM_144773.2	<i>PROKR2</i> c.1004C>T, p.Thr335Met	het	AD	VUS	/	PM2	PP3	0.00000701	0.00000701 (European non-finnish)	/
8803	NM_000044.4	<i>AR</i> c.2270A>G, p.Asn757Ser	hemi	XR	LP	/	PM1+PM2	PP3+PP4	0.000008892	0	8723113
8945	NM_000475.4	<i>NR0B1</i> c.1169-2Ala>Gly	hemi	XL	P	PVS1+PS2	PM2	PP3	/	/	/
9052	NM_000044.4	<i>AR</i> c.2495G>A, p.Arg832Gln	hemi	XR	LP	/	PM1+PM2+PM5	PP3+PP4	0.000008987	0	2082179, 26778393, 30815925
9171	NM_023110.2	<i>FGFR1</i> c.1961A>G, p.Tyr654Cys	het	AD	VUS	/	PM1+PM2	PP3	/	/	/
		<i>SRD5A2</i> c.650C>A, p.Ala217Glu	het	AR	LP	/	PM1+PM2+PM3	PP3+PP4	/	/	/
9242	NM_000348.3	<i>SRD5A2</i> c.680G>A, p.Arg227Gln	het	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107

9431	NM_000348.3	<i>SRD5A2</i> c.607G>A,p.Gly203Ser	het	AR	P	PS1	PM1+PM2+PM3	PP3+PP4	0.00002629	0.0001567 (East Asian)	9135696
		<i>SRD5A2</i> c.680G>A,p.Arg227Gln	het	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
9471	NM_000044.4	<i>AR</i> c.4G>A, p.Glu2Lys	hemi	XR	LP	PS3	PM2	PP3+PP4	/	/	8823308
9482	NM_000348.3	<i>SRD5A2</i> c.680G>A,p.Arg227Gln	homo	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
10066	NM_000044.4	<i>AR</i> c.899G>C, p.Gly300Ala	hemi	XR	LP	/	PM1+PM2	PP3+PP4	0.00002657	0.00001494 (European)	/
10239	/	7q33-q35Del	het	-	P	PVS1+PS2	PM2	PP3	/	/	/
10482	NM_002934.4	<i>PTPN11</i> c.1472C>A,p.Pro491His	het	AD	P	PS2	PM1+PM2	PP2+PP3+PP4	/	/	17020470
10724	NM_000348.3	<i>SRD5A2</i> c.408C>A,p.Tyr136*	het	AR	VUS	/	PM2	PP3+PP4	/	/	/
10800	/	2q33.1Del	het	-	LP	PS2	PM2	PP3	/	/	/
10892	NM_002934.4	<i>PTPN11</i> c.922A>G,p.Asn308Asp	het	AD	P	PS2	PM1+PM2	PP2+PP3+PP4	0.00001193	0	11704759
10908	NM_000044.4	<i>AR</i> c.1737delC, p.Cys580Alafs*46	hemi	XR	P	PVS1	PM2	PP3+PP4	/	/	19463997
11072	NM_000044.4	<i>AR</i> c.2113C>T, p.Leu705Phe	hemi	XR	LP	/	PM1+PM2	PP3+PP4	/	/	25613104 1406794, 1944596, 12576851
11757	<i>NM_000348.3</i>	<i>SRD5A2</i> c.16C>T,p.Gln6*	het	AR	P	PVS1	PM2+PM3	PP3+PP4	/	/	/
		<i>SRD5A2</i> c.680G>A,p.Arg227Gln	het	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
11867	NM_000348.3	<i>SRD5A2</i> c.680G>A,p.Arg227Gln	homo	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
12343	NM_017780.3	<i>CHD7</i> c.5211-2A>G	het	AD	P	PVS1+PS2	PM2	PP3+PP4	/	/	/
12480	/	18p11.31-p11.21del	het	-	P	PVS1+PS2	PM2	PP3	/	/	/
12888	/	16p11.2del	het	-	P	PVS1+PS2	PM2	PP3	/	/	/
13048	/	17p11.2del	het	-	P	PVS1+PS2	PM2	PP3	/	/	/
13317	NM_012082.3	<i>ZFPM2</i> c.1124_1127del,p.Gln375Argfs*40	het	AD	VUS*	PS3	PM2	PP3	/	/	/
13421	NM_000044.4	<i>AR</i> c.2020T>C, p.Phe674Leu	hemi	XR	LP	/	PM1+PM2	PP3+PP4	/	/	/
14250	NM_000348.3	<i>SRD5A2</i> c.607G>A,p.Gly203Ser	het	AR	P	PS1	PM1+PM2+PM3	PP3+PP4	0.00002629	0.0001567 (East Asian)	9135696, 15266301, 32713132
		<i>SRD5A2</i> c.680G>A,p.Arg227Gln	het	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
14317	NM_003106.3	<i>SOX2</i> c.259A>G,p.Lys87Glu	het	AD	LP	PS2	PM2	PP3+PP4	/	/	/
14376	NM_000216.2	<i>ANOS1</i> c.515G>A,p.Cys172Tyr	hemi	XR	LP	PS1	PM2	PP3+PP4	/	/	11297579
15071	NM_006941.3	<i>SOX10</i> c.506C>T,p.Pro169Leu	het	AD	LP	PS2	PM2	PP3+PP4	/	/	25256313
15128	NM_005996.3	<i>TBX3</i> c.932dupA,p.Glu312Glyfs*6	het	AD	P	PVS1+PS2	PM2	PP4	/	/	/
15362	NM_000479.3	<i>AMH</i> c.259-270del,p.Val187-Ala90del	het	AR	LP	/	PM2+PM3	PP3+PP4	/	/	/
15562	NM_006941.3	<i>AMH</i> c.1432C>T,p.Leu478Phe	het	AR	LP	/	PM2+PM3	PP3+PP4	/	/	/
		<i>SOX10</i> c.506C>T,p.Pro169Leu	het	AD	LP	PS2	PM2	PP3+PP4	/	/	25256313
16213	NM_000348.3	<i>SRD5A2</i> c.680G>A,p.Arg227Gln	homo	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
16255	NM_023110.2	<i>FGFR1</i> c.2173T>C,p.Cys725Arg	het	AD	LP	PS2	PM2	PP3+PP4	/	/	/
16746	NM_000348.3	<i>SRD5A2</i> c.680G>A,p.Arg227Gln	homo	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
17641	NM_000435.2	<i>NOTCH3</i> c.2129A>G, p.Tyr710Cys	het	AD	VUS	/	PM2	PP4	/	/	24000151
18385	NM_144773.2	<i>PROKR2</i> c.533G>C,p.Trp178Ser	het	AD	VUS	PS3	/	PP3	0.0002227	0.00231 (East Asian)	17054399
18423	NM_006912.6	<i>RIT1</i> c.170C>G,p.Ala57Gly	het	AD	P	PS1+PS2	PM2	PP4	/	/	23791108
18553	NM_000348.3	<i>SRD5A2</i> c.680G>A,p.Arg227Gln	homo	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
		<i>SRD5A2</i> c.467G>A, p.Gly156Glu	het	AR	LP	/	PM2+PM3	PP3+PP4	0.00006677	0	/
19697	NM_000348.3	<i>SRD5A2</i> c.680G>A,p.Arg227Gln	het	AR	P	PS3	PM1+PM2	PP3+PP4	0.0002235	0.004817 (East Asian)	8784107
		<i>AR</i> c.2521C>T, p.Arg841Cys	hemi	XR	LP	/	PM1+PM2	PP3+PP4	/	/	11788673
20472	NM_000044.6	<i>AR</i> c.1175C>G, p.Pro392Arg	hemi	XR	LP	/	PM1+PM2	PP3+PP4	0.00001776	0	10571951
20685	NM_000044.6	<i>AR</i> c.1159A>T, p.Ile387Phe	hemi	XR	VUS	/	PM2	PP3+PP4	/	/	/
21257	NM_144773.2	<i>PROKR2</i> c.533G>C,p.Trp178Ser	het	AD	VUS	PS3	/	PP3	0.0002227	0.00231 (East Asian)	17054399
21288	NM_017934.7	<i>PHIP</i> c.600+1G>C	het	AD	LP	PS2	PM2	PP3	/	/	/

Abbreviations: PVS, pathogenic very strong; PS, pathogenic strong; PM, pathogenic moderate; PP, pathogenic supporting; BS, benign strong; P, pathogenic; LP, likely pathogenic; VUS, uncertain significance.

\*: This variant on *ZFPM2* was inherited from the subject's mother who had no similar DSD phenotype, thus considering VUS.