Supplementary Material. Demographic and genetics characteristics of the cohort.

Patient ID	Age*	Sex	Indication	Sanger Sequencing (NM_000500.5)/ACMG classification		MLPA	Final Molecular Diagnosis
P1	17	F	Puberty Problems	c.518T>A p.(Ile173Asn)/ P(PM3,PS3,PM1,PM2,PP2,PP3)	c.1447C>T p.(Pro483Ser)/LP(PM1,PM2,PP2,PP5)	No Reportable Variant	Compound Heterozygous
P2	16	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P3	12	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P4	17	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P5	18	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P6	17	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P7	16	F	Puberty Problems	c.844G>T p.(Val282Leu)/ LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P8	5	F	Disorder of Sexual Development	c.92C>T p.(Pro31Leu)/ P(PM3,PS3,PM1,PM2,PP2,PM5)		No Reportable Variant	Heterozygous
P9	9		Family History	Exon 6-8 micro conversion		Exon 6-8 micro conversion	Heterozygous
P10	0	F	Adrenal Insufficiency	No Reportable Variant		No Reportable Variant	-
P11	15	F	Puberty Problems	c.844G>T p.(Val282Leu)/LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P12	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P13	7	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P14	5	F	Clinical Diagnosis	c.293-13A/C>G /P(PM2,PM3,PS3)		No Reportable Variant	Homozygous
P15	8	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P16	15	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P17	12	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P18	10	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		Exon 1-3 deletion	Compound Heterozygous
P19	0	M	Disorder of Sexual Development	c.518T>A p.(Ile173Asn) /P(PM3,PS3,PM1,PM2,PP2,PP3)		No Reportable Variant	Homozygous
P20	11	F	Puberty Problems	c.293-13 A/C>G /P(PM2,PM3,PS3)		No Reportable Variant	Heterozygous
P21	10	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P22	7	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P23	13	F	Clinical Diagnosis	c.961G>A p.(Glu321Lys) /LP(PM1,PM2,PP2,PP3)	c.293-13 A/C>G /P(PM2,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P24	10	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.1360C>T p.(Pro454Ser) /LP(PM1,PM2,PP2,PP5)	No Reportable Variant	Compound Heterozygous
P25	17	F	Puberty Problems	c.844G>T p.(Val282Leu) / LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P26	17	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P27	9	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P28	7	F	Clinical Diagnosis	c.293-13A/C>G /P(PM2,PM3,PS3)		No Reportable Variant	Homozygous
P29	0	F	Clinical Diagnosis	c. [955C>T; c.1069C>T] p. [Gln319*;p.Arg357Trp]/ P(PM3,PS3,PM5,PP1,PP2,PP3) (heterozygous)	Exon 4-8 micro conversion	Exon 4-8 micro conversion	Compound Heterozygous
P30	7	M	Clinical Diagnosis	c.[1019G>A;1360C>T] p.[Pro454Ser;Arg340His] /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P31	0	F	Disorder of Sexual Development	No PCR products		CAH-X(homozygous large deletion)	Homozygous
P32	0	F	Disorder of Sexual Development	No Reportable Variant		No Reportable Variant	-
P33	0	M	Disorder of Sexual Development	No Reportable Variant		No Reportable Variant	-
P34	0	M	Disorder of Sexual Development	No Reportable Variant		No Reportable Variant	-
P35	0	M	Disorder of Sexual Development	No Reportable Variant		No Reportable Variant	-
P36	3	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.1447C>T p.(Pro483Ser) / LP(PM1,PM2,PP2,PP5)	No Reportable Variant	Compound Heterozygous
P37	16	F	Clinical Diagnosis	c.293-13A/C>G /P(PM2,PM3,PS3)		Exon 1-3 deletion	Compound Heterozygous
P38	10	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P39	12	F	Puberty Problems	c.392G>A p.(Gly131Asp)/VUS(PM2,PP2)		No Reportable Variant	Homozygous
P40	0	M	Clinical Diagnosis	c.293-13A/C>G /P(PM2,PM3,PS3)	Exon 4-9 micro conversion	Exon 4-9 micro conversion	Compound Heterozygous
P41	0	M	Disorder of Sexual Development	No Reportable Variant		No Reportable Variant	-
P42	14	M	Clinical Diagnosis	c.293-13A/C>G /P(PM2,PM3,PS3)		No Reportable Variant	Homozygous
P43	0	M	Disorder of Sexual Development	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P44	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P45	15	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P46	3	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P47	8	F	Clinical Diagnosis	No Reportable Variant		No Reportable Variant	-
P48	15	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P49	0	F	Clinical Diagnosis	c.92C>T p.(Pro31Leu) /P(PM3,PS3,PM1,PM2,PP2,PM5)		No Reportable Variant	Homozygous
P50	13	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P51	0	F	Abnormal Screening Test	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P52	0	M	Abnormal Screening Test	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P53	10	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P54	9	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P55	0	M	Disorder of Sexual Development	No Reportable Variant		No Reportable Variant	-
P56	15	M	Puberty Problems	c.1447C>T p.(Pro483Ser)/LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P57	5	F	Clinical Diagnosis	c.1360C>T p.(Pro454Ser) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P58	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P59	8	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		Exon 7 deletion	Compound Heterozygous
P60	10	M	Puberty Problems	c.518T>A p.(Ile173Asn) /P(PM3,PS3,PM1,PM2,PP2,PP3)	c.1069C>T p.(Arg357Trp) /P(PM3,PS3,PM5,PP1, PP2,PP3)	No Reportable Variant	Compound Heterozygous
P61	9	M	Adrenal Insufficiency	No Reportable Variant		No Reportable Variant	-
P62	6	F	Adrenal Insufficiency	No Reportable Variant		No Reportable Variant	-
P63	0	M	Clinical Diagnosis	c.955C>T p.(Gln319*)/P(PVS1,PM3,PS3)		No Reportable Variant	Homozygous
P64	13	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.293-13A/C>G /P(PM2,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P65	15	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P66	0	F	Clinical Diagnosis	Exon 4-9 micro conversion		Exon 4-9 micro conversion	Homozygous
P67	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P68	1	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P69	9	M	Puberty Problems	c.1447C>T p.(Pro483Ser)/LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P70	8	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.269C>T p.(Ala90Val)/ VUS(PM2,PP2)	No Reportable Variant	Compound Heterozygous
P71	7	M	Puberty Problems	c.844G>T p.(Val282Leu) / LP(PM1,PM2,PP2,PP5)		Total gene deletion	Compound Heterozygous
P72	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P73	10	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P74	0	F	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.293-13A/C>G /P(PM2,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P75	0	F	Disorder of Sexual Development	No Reportable Variant		No Reportable Variant	-
P76	5	F	Family History	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.955C>T p.(Gln319*)/ P(PVS1,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P77	15	M	Puberty Problems	c.1304C>T p.(Ala435Val)		No Reportable Variant	Heterozygous
P78	11	F	Family History	c.392G>A p.(Gly131Asp)/ VUS(PM2,PP2)	c.844G>T p.(Val282Leu) / LP(PM1,PM2,PP2,PP5)	No Reportable Variant	Compound Heterozygous
P79	14	M	Family History	c.392G>A p.(Gly131Asp)/VUS(PM2,PP2)		No Reportable Variant	Heterozygous
P80	12	F	Family History, Disorder of Sexual Development	c.392G>A p.(Gly131Asp)/VUS(PM2,PP2)		No Reportable Variant	Homozygous
P81	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P82	5	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P83	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P84	12	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P85	10	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P86	12	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P87	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P88	16	F	Puberty Problems	c.844G>T p.(Val282Leu) / LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P89	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P90	0	M	Abnormal Screening Test	c.850A>G p.(Met284Val)/ P(PM1,PM3,PP2,PM2)		No Reportable Variant	Heterozygous
P91	4	M	Family History	c.293-13A/C>G /P(PM2,PM3,PS3)	c.397C>T p.(Arg133Cys) /LP(PS3,PM3,PM2,PP2)	No Reportable Variant	Compound Heterozygous
P92	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P93	9	M	Puberty Problems	c.844G>T p.(Val282Leu) / LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P94	15	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P95	12	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P96	15	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	Exon 4 micro conversion	Exon 4 micro conversion	Compound Heterozygous
P97	15	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P98	0	F	Disorder of Sexual Development	c.844G>T p.(Val282Leu) / LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P99	11	M	Clinical Diagnosis	c.92C>T p.(Pro31Leu) /P(PM3,PS3,PM1,PM2,PP2,PM5)	c.844G>T p.(Val282Leu) / LP(PM1,PM2,PP2,PP5)	No Reportable Variant	Compound Heterozygous
P100	0	F	Abnormal Screening Test	c.865C>T p.(Leu289Phe)/ VUS(PM1,PM2,PP2)		No Reportable Variant	Homozygous
P101	0	M	Abnormal Screening Test	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P102	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P103	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P104	16	M	Clinical Diagnosis	c.1069C>T p.(Arg357Trp) /P(PM3,PS3,PM5,PP1,PP2,PP3)	c.1360C>T p.(Pro454Ser) / LP(PM1,PM2,PP2,PP5)	No Reportable Variant	Compound Heterozygous
P105	7	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P106	12	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P107	9	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P108	8	F	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.1352C>T p.(Thr451Met)/ VUS(PM1,PM2,PP2,)	No Reportable Variant	Compound Heterozygous
P109	1	F	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.1069C>T p.(Arg357Trp) /P(PM3,PS3,PM5,PP1, PP2,PP3)	No Reportable Variant	Compound Heterozygous
P110	6	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P111	3	M	Puberty Problems	Exon 4 micro conversion		Exon 4 micro conversion	Homozygous
P112	15	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P113	16	F	Puberty Problems	c.392G>A p.(Gly131Asp)/VUS(PM2,PP2)		No Reportable Variant	Homozygous
P114	1	F	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P115	6	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P116	9	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P117	8	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P118	15	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P119	13	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P120	7	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P121	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P122	3	M	Clinical Diagnosis	c.86A>C p.(His29Pro)		Exon 1 deletion	Compound Heterozygous
P123	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P124	9	M	Adrenal Insufficiency	No Reportable Variant		No Reportable Variant	-
P125	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P126	9	M	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P127	8	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P128	15	F	Puberty Problems	c. 1174G>A p.(Ala392Thr)		No Reportable Variant	Heterozygous
P129	0	F	Puberty Problems	c.293-13A/C>G /P(PM2,PM3,PS3)		No Reportable Variant	Heterozygous
P130	10	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P131	6	F	Puberty Problems	c.188A>T p.(His63Leu)		No Reportable Variant	Heterozygous
P132	15	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P133	7	F	Puberty Problems	c.1360C>T p.(Pro454Ser) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P134	8	F	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P135	9	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P136	15	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P137	5	F	Incidental ACTH Elevation	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P138	10	F	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P139	14	F	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P140	15	F	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.293-13A/C>G /P(PM2,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P141	17	F	Puberty Problems	c.1360C>T p.(Pro454Ser) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P142	15	F	Clinical Diagnosis	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.293-13A/C>G /P(PM2,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P143	7	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P144	11	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P145	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P146	8	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P147	16	F	Puberty Problems	Exon 4 micro conversion		Exon 4 micro conversion	Heterozygous
P148	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P149	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P150	18	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P151	18	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P152	7	F	Clinical Diagnosis	c.961G>A p.(Glu321Lys) /LP(PM1,PM2,PP2,PP3)	c.293-13A/C>G /P(PM2,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P153	12	F	Puberty Problems	c.1360C>T p.(Pro454Ser) /LP(PM1,PM2,PP2,PP5)	c.1019G>A p.(Arg340His) / LP(PM1,PM2,PP2,PM3)	No Reportable Variant	Compound Heterozygous
P154	16	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P155	12	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P156	10	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P157	1	M	Disorder Of Sexual Development	Exon 4 micro conversion		Exon 4 micro conversion	Homozygous
P158	6	F	Puberty Problems	c.1360C>T p.(Pro454Ser) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P159	0	F	Disorder Of Sexual Development	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)	c.955C>T p.(Gln319*)/ P(PVS1,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P160	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P161	3	M	Clinical Diagnosis	c.293-13A/C>G /P(PM2,PM3,PS3)		No Reportable Variant	Homozygous
P162	3	M	Clinical Diagnosis	c.955 C>T p.(Gln319*)/P(PVS1,PM3,PS3)		No Reportable Variant	Homozygous
P163	7	M	Disorder Of Sexual Development	c.1360C>T p.(Pro454Ser) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P164	9	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P165	5	F	Puberty Problems	Exon 4 micro conversion		Exon 4 micro conversion	Homozygous
P166	13	M	Puberty Problems	c.850A>G p.(Met284Val)/ P(PM1,PM3,PP2,PM2)		Total gene deletion	Compound Heterozygous
P167	12	F	Puberty Problems	Exon 4 micro conversion		Exon 4 micro conversion	Homozygous
P168	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P169	11	F	Puberty Problems	c.1360C>T p.(Pro454Ser) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P170	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P171	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P172	8	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P173	0	F	Disorder Of Sexual Development	No Reportable Variant		No Reportable Variant	-
P174	15	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P175	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P176	11	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P177	1	F	Disorder Of Sexual Development	c.955C>T p.(Gln319*)/ P(PVS1,PM3,PS3)	c.293-13A/C>G /P(PM2,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P178	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P179	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P180	0	F	Abnormal Screening Test	c.865C>T p.(Leu289Phe) /VUS(PM1,PM2,PP2)		No Reportable Variant	Heterozygous
P181	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P182	8	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P183	14	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P184	0	F	Clinical Diagnosis	c.518 T>A p.(Ile173Asn) /P(PM3,PS3,PM1,PM2,PP2,PP3)	c.955C>T p.(Gln319*)/ P(PVS1,PM3,PS3)	No Reportable Variant	Compound Heterozygous
P185	12	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P186	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P187	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P188	16	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P189	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P190	14	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P191	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P192	8	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P193	16	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P194	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P195	0	M	Disorder Of Sexual Development	No Reportable Variant		No Reportable Variant	-
P196	15	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P197	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P198	14	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-

P199	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P200	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P201	0	M	Disorder Of Sexual Development	No Reportable Variant		No Reportable Variant	-
P202	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P203	17	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P204	15	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P205	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P206	17	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P207	7	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P208	0	F	Clinical Diagnosis	c.955C>T p.(Gln319*)/P(PVS1,PM3,PS3)		No Reportable Variant	Homozygous
P209	0	F	Disorder Of Sexual Development	No Reportable Variant		No Reportable Variant	
P210	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P211	0	M	Disorder Of Sexual Development	No Reportable Variant		No Reportable Variant	-
P212	0	M	Abnormal Screening Test	c.1360C>T p.(Pro454Ser) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P213	16	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P214	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P215	8	M	Puberty Problems	Exon 4 micro conversion		Exon 4 micro conversion	Heterozygous
P216	9	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P217	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P218	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P219	0	M	Abnormal Screening Test	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P220	7	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P221	17	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P222	11	F	Disorder Of Sexual Development	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P223	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P224	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P225	0	F	Clinical Diagnosis	c.293-13A/C>G /P(PM2,PM3,PS3)	Exon 4 micro conversion	Exon 4 micro conversion	Compound Heterozygous
P226	9	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P227	6	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P228	0	F	Disorder Of Sexual Development	No Reportable Variant		No Reportable Variant	-
P229	15	M	Puberty Problems	c.1069C>T p.(Arg357Trp) /P(PM3,PS3,PM5,PP1,PP2,PP3)		No Reportable Variant	Heterozygous
P230	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P231	0	M	Abnormal Screening Test	c.293-13A/C>G /P(PM2,PM3,PS3)		No Reportable Variant	Heterozygous
P232	15	M	Disorder Of Sexual Development	No Reportable Variant		No Reportable Variant	-
P233	0	M	Disorder Of Sexual Development	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Heterozygous
P234	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P235	15	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P236	5	F	Puberty Problems	c.293-13A/C>G /P(PM2,PM3,PS3)		No Reportable Variant	Heterozygous
P237	15	F	Puberty Problems	c.293-13A/C>G /P(PM2,PM3,PS3)		No Reportable Variant	Heterozygous
P238	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P239	0	M	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P240	0	M	Clinical Diagnosis	Exon 1-3 micro conversion	Total gene conversion	Exon 1-3 micro conversion, Total gene conversion	Compound Heterozygous
P241	9	M	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P242	8	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P243	7	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P244	8	F	Puberty Problems	c.844G>T p.(Val282Leu) /LP(PM1,PM2,PP2,PP5)		No Reportable Variant	Homozygous
P245	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P246	0	F	Abnormal Screening Test	No Reportable Variant		No Reportable Variant	-
P247	7	F	Clinical Diagnosis	c.293-13A/C>G /P(PM2,PM3,PS3)		Exon 1-3 deletion	Compound Heterozygous
P248	13	M	Adrenal Insufficiency	No Reportable Variant		No Reportable Variant	-
P249	17	M	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P250	15	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P251	9	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-
P252	8	F	Puberty Problems	No Reportable Variant		No Reportable Variant	-

*: Designates ages of the probands when tested P:Pathogenic, LP: Likely pathogenic, VUS: Variant of unknown significance F: Female, M: Male.