

Table S4. Biochemical and radiological findings in 35 patients with 46, XY DSD.

ID	LH [#] [IU/L]	peak LH after GnRH test [IU/L]	FSH [IU/L]	peak FSH after GnRH test [IU/L]	E2 [pg/mL]	T [ng/mL]	T after hCG test [ng/mL]	AMH [ng/mL]	Inhibin b [pg/mL]	distinctive biochemical trait	abdomen ultrasound
1	0.1	nd	6.5	nd	nd	0.1	nd	0	7.4		no Müllerian structures, hypoplastic left gonad in inguinal canal
2	9.7	nd	91.1	nd	nd	0	0.1	nd	nd		no Müllerian structures, gonads not visible
3	1.5	32.3	38.1	111.2	nd	0.1	0.1	0.1	0.9		no Müllerian structures, gonads not visible
4	<0.5	5.3	7.2	31.1	nd	0.1	0.1	1.2	0.3		no Müllerian structures, gonads not visible
5	<0.5	nd	3.4	nd	nd	0.1	0.1	0.1	0.9		no Müllerian structures, gonads not visible
6	0.7	nd	0.7	nd	<20	nd	nd	nd	nd	AFP 23440 [ng/mL]	infantile uterus, normal vagina's length, tumor arising from left gonad
7	nd	nd	nd	nd	nd	nd	nd	nd	nd		infantile uterus, normal vagina's length, tumors arising from both gonads
8	1.5	nd	4.7	nd	125	nd	nd	nd	nd	AFP 1210 [ng/mL]; βhCG 13658 [mIU/mL]; LDH 1158 [IU/mL]	infantile uterus, normal vagina's length, tumor arising from left gonad
9	27	nd	53.6	nd	10	1.3	nd	nd	nd		infantile uterus, normal vagina's length, visible gonads in typical localization for ovaries
10	<0.5	nd	0.5	nd	nd	0	5.2	211.6	174.6	T/DHT after hCG test 56	no Müllerian structures
11	<0.5	nd	0.8	nd	<20	0.1	3.2	123.2	67.9	T/DHT after hCG test 32	no Müllerian structures
12	0.1	nd	0.4	nd	nd	0.1	4.5	262.3	355.5	T/DHT after hCG test 32	no Müllerian structures
13	0.8	nd	0.9	nd	23	0	nd	nd	nd		no Müllerian structures, blind ended vagina
14	6.5	nd	1	nd	12	1.5	nd	130.4	nd		no Müllerian structures
15	6	nd	1.2	nd	<20	4.9	nd	>440.9	544.9		no Müllerian structures, gonads in inguinal canals
16	4.8	nd	2.3	nd	31	0.6; at 7y [†] 0.1	at 7y 2.8	at 13y 7.8	at 13y 208.9		no Müllerian structures
17	8.3	nd	4.7	nd	nd	1	nd	at 17y 1	at 17y 52.1		no Müllerian structures
18	<0.5	nd	0.8	nd	nd	0.1	1.2	175.8	194.6		no Müllerian structures
19	<0.5	nd	0.8	nd	nd	0.3	1.3	93.7	65.7		no Müllerian structures, left gonad invisible
20	0.4	nd	0.2	nd	11	0.1	5.4	155.7	401.9		no Müllerian structures, blind ended vagina

21	0.3	nd	1.5	nd	11	0.3	2.9	80.7	196.5		no Müllerian structures, blind ended vagina
22	19.2	nd	7	nd	39	8.4	nd	>440.9	298		no Müllerian structures, blind ended vagina
23	24	nd	19	nd	39	10.7	nd	184.3	175.8		no Müllerian structures, blind ended vagina
24	9.7	nd	4	nd	48	5.1	nd	193.6	346.1		no Müllerian structures, blind ended vagina
25	21.9	nd	8.3	nd	38	8	nd	393.8	401.2		no Müllerian structures, blind ended vagina
26	23.4	nd	11	nd	53.8	11.5	nd	nd	nd		no Müllerian structures, blind ended vagina
27	4.7	nd	1.1	nd	<20	0.5	2.7	47.6	299.7		no Müllerian structures
28	15.4	nd	56.3	nd	nd	0.8; at 2y 0.1	at 2y 0.3	nd	nd		no Müllerian structures
29	at 13y8m 4.8	nd	at 13y8m 12.2	nd	nd	6.8	nd	at 13y8m 4.2	at 13y8m 73.3		no Müllerian structures, gonads in inguinal canals
30	0.5	2	1.3	2.9	nd	nd	nd	222.7	165.2	ACTH [8.8 pg/mL]; cortisol [29 ng/mL]	no Müllerian structures
31	0.12	4.8	0.5	1.8	nd	0.1	2.6	206	nd	T/DHT after hCG test 44	no Müllerian structures
32	1.1	nd	1.2	nd	nd	nd	0.1	nd	nd	ACTH [511 pg/mL]; cortisol [8 ng/mL]	no Müllerian structures, gonads in inguinal canals
33	<0.5	5.7	3	124.3	nd	0.1	0.8	46.8	90.6		no Müllerian structures
34	2.2	nd	5.6	nd	nd	0.9	4.3	2.7	122.9		no Müllerian structures
35	nd	nd	nd	nd	46	1.4	nd	nd	nd		no Müllerian structures

Abbreviations: LH, luteinizing hormone; FSH, follicle-stimulating hormone; E2, estradiol; T, testosterone; AMH, anti-Müllerian hormone; nd, no data; AFP, alfa-fetoprotein; βhCG, beta-human chorionic gonadotropin; LDH, lactate dehydrogenase; DHT, dihydrotestosterone; y, years; m, months; ACTH, adrenocorticotrophic hormone

†the specific age is given when assessed at different time than age of referral

age specific reference values: LH: 0.5-12.0; FSH: 1.0-12.0; E2: 11.0-44.0; T median (2.5-97.5 percentile): mini-puberty: 1.6 (0.1-3.6), childhood: 0.1 (0-0.3), Tanner 1: 0.1 (0-0.2), Tanner 2: 0.2 (0-1.6), Tanner 3: 2.3 (0-5.1), Tanner 4: 3.5 (1.2-6.9), Tanner 5: 5 (2.2-7.6); AMH

median (2.5-97.5 percentile): mini-puberty: 198.2 (141.8-365.3), childhood: 142.9 (74.8-252.7), Tanner 1: 131.9 (60.8-230.5), Tanner 2: 47.9 (8.7-212), Tanner 3: 12.5 (4.2-138.9), Tanner 4: 8.1 (2.8-21.2); Tanner 5: 9.3 (4.4-24.2); Inhibin B median (2.5-97.5 percentile): mini-puberty: 270 (210-335), childhood: 80 (50-130), Tanner 1: 78 (35-182), Tanner 2: 195 (62-338), Tanner 3: 163 (78-323), Tanner 4: 188 (67-304), Tanner 5: 187 (95-323)