Table S3. Clinical findings in 35 patients with 46, XY DSD.

ID	sex#	DSD subcategory	clinical diagnosis	age at referral	cause of referral	EMS score	Tanner stages	associated conditions
1	M	DGD	TRS	1y11m	empty scrotum	9	prepubertal	complex obstetric history (1 miscarriage, death of 2nd child in 1st day of life due to combined congenital defects)
2	M	DGD	TRS	2y8m	empty scrotum	9	prepubertal	
3	M	DGD	TRS	5y7m	empty scrotum	9	prepubertal	
4	M	DGD	TRS	7y4m	empty scrotum	9	prepubertal	SGA, gestational hypertension
5	M	DGD	TRS	8y10m	empty scrotum	9	prepubertal	
6	F	DGD	CGD	13y	abdominal mass, delayed puberty	9	B1P3	yolk sac tumor, tall stature
7	F	DGD	CGD	14y6m	recurrent abdominal pain, delayed puberty	0	B1P4	no hormonal work-up prior to surgical treatment, bilateral gonadoblastoma, bilateral immature teratoma
8	F	DGD	CGD	16y2m	abdominal mass, primary amenorrhea	0	B3P3	c-section due to placenta praevia, prematurity, mixed germ cell tumor secreting E2, tall stature
9	F	DGD	CGD	17y8m	primary amenorrhea	0	B1P4	bilateral gonadoblastoma secreting T, tall stature
10	M	DAS	5αRD	10m	atypical genitalia	9	prepubertal	
11	M	DAS	5αRD	12m	atypical genitalia	9	prepubertal	bilateral sandal gap (limb anomalies)
12	M	DAS	5αRD	2y1m	atypical genitalia	9	prepubertal	prematurity

13	F	DAS	DAS	21m	atypical genitalia	3	prepubertal	right side inguinal hernia, gonadectomy at the age of 2y10m, testes on histopathology
14	M	DAA	NSDUM	11d	atypical genitalia	9	prepubertal	SGA, normal urinary system on US, glanular hypospadias in father
15	M	DAA	NSDUM	22d	atypical genitalia	5	prepubertal	positive family history for AIS
16	M	DAA	NSDUM	3m	atypical genitalia	6	prepubertal	
17	M	DAA	NSDUM	3m	atypical genitalia	?	prepubertal	SGA, c-section due to fetal distress, umbilical hernia
18	M	DAA	NSDUM	6m	atypical genitalia	6	prepubertal	c-section due to breech position
19	M	DAA	NSDUM	3y3m	atypical genitalia	7.5	prepubertal	SGA, c-section due to fetal distress, prematurity, underweight
20	F	DAA	CAIS	10d	atypical genitalia	3	prepubertal	bilateral inguinal hernias, positive family history for AIS
21	F	DAA	CAIS	5y10m	atypical genitalia	1	prepubertal	asthma
22	F	DAA	CAIS	165m	primary amenorrhea	0	B5P2	tall stature, obesity
23	F	DAA	CAIS	17y4m	primary amenorrhea	0	B5P2	pectus excavatum
24	F	DAA	CAIS	17y9m	primary amenorrhea	2	B5P2	c-section due to fetal distress, bilateral inguinal hernias, tall stature, overweight
25	F	DAA	CAIS	17y9m	primary amenorrhea	1	B5P2	tall stature, unilateral inguinal hernia
26	F	DAA	CAIS	14y6m	primary amenorrhea	2	B5P2	c-section due to placental abruption, bilateral inguinal hernias, hyposmia
27	F	DAA	PAIS	6m	atypical genitalia	3	prepubertal	SGA, c-section due to fetal distress, oligohydramnios, prematurity
28	M	sDSD	sDSD	2m	atypical genitalia	6	prepubertal	atrial septal defect type II, left kidney ectopy, with time idiopathic hypertransaminasemia developed

29 †	M	sDSD	sDSD	2m	atypical genitalia	8	prepubertal	SGA, c-section due to breech position, congenital heart defect, left kidney ectopy, nonautoimmunological primary hypothyroidism, with time hypergonadotropic hypogonadism developed, older brother of patient ID 30
30 †	M	sDSD	sDSD	6m	atypical genitalia	6	prepubertal	facial dysmorphia, multiple pituitary hormone deficiency (TSH, GH, ACTH), pituitary posterior lobe ectopy, younger brother of patient ID 29
31	M	sDSD	sDSD	18m	atypical genitalia	4	prepubertal	SGA, complex obstetric history (4th pregnancy, 2nd delivery, 2 miscarriages), facial dysmorphia, psychomotor delay, hypotonia, left side hypoacusis, patent foramen ovale, hip dysplasia, asthma, dilatation of the ventricular system and the frontal lobes subarachnoid spaces on MRI
32	M	sDSD	sDSD	2y2m	atypical genitalia with adrenal insufficiency	8	prepubertal	primary adrenal insufficiency, epilepsy, mild developmental delay, with time hypergonadotropic hypogonadism developed
33 ‡	M	sDSD	sDSD	6y10m	atypical genitalia	3	prepubertal	SGA, epilepsy, diaphragmatic hernia, younger brother of patient ID 34
34 ‡	M	sDSD	sDSD	12y10m	atypical genitalia	3	G2P2	obesity, short stature (rhGH treatment), hypermetropia, older brother of patient ID 33
35	F	sDSD	sDSD	7d	atypical genitalia	0	prepubertal	c-section due to fetal distress, facial dysmorphia, microcephaly, syndactyly of 2nd and 3rd toes, clinodactyly of 2nd and 3rd fingers of the left hand, psychomotor delay, short stature (rhGH treatment), dilatation of subarachnoid spaces on MRI

#sex of rearing; †first pair of siblings; ‡second pair of siblings

Abbreviations: DSD, disorder of sex development; EMS, external masculinisation score; M, male; DGD, disorder of gonadal development; TRS, testicular regression syndrome; y, years; m, months; SGA, small for gestational age; F, female; CGD, complete gonadal dysgenesis; E2, estradiol; T, testosterone; DAS, disorder of androgen synthesis; 5αRD, 5α-reductase deficiency; NSDUM, non-specific disorder of

undermasculinisation; d, days; US, ultrasound; AIS, androgen insensitivity syndrome; CAIS, complete androgen insensitivity syndrome; sDSD, syndromic DSD; rhGH, recombinant human growth hormone; TSH, thyroid stimulating hormone; GH, growth hormone; ACTH, adrenocorticotropic hormone