Additional file 2.

Title: Karyotypes of the patients

Legend: Karyotypes of the patients were systematized according to the main type of chromosome aberration and composition of mosaicism.

TYPES OF SCAs & OGDs (number of patients)				PATIENTS	KARYOTYPES • CNV with pathogenicity according to ACMG recommendation
ALL (137 patients)					
TURNER SY. & VS. (52 patients)					
CLASSIC (39)	*NON-MOSAIC (19)*			P1-19	45,X[100]dn
	MOSAIC (20)	*46,XX (8)*		P20	45,X[5]/46,XX[95]dn
				P21	45,X[6]/46,XX[94]dn
				P22	45,X[7]/46,XX[93]dn
				P23	45,X[10]/46,XX[90]dn
				P24	45,X[25]/46,XX[75]dn
				P25	45,X[52]/46,XX[48]dn
				P26	45,X[78]/46,XX[22]dn
				P27	45,X[97]/46,XX[3]dn
		TRIPLE/POLY X (8)		P28	45,X[3]/47,XX,+X[1]/46,XX[96]dn
				P29	45,X[2]/48,XX,+X,+X[2]/46,XX[96]dn
				P30	45,X[3]/49,XX,+X,+X,+X[1]/46,XX[96]dn
				P31	45,X[5]/47,XX,+X[2]/46,XX[93]dn
				P32	45,X[6]/47,XX,+X[2]/46,XX[92]dn
				P33	45,X[7]/47,XX,+X[1]/46,XX[92]dn
				P34	45,X[6]/47,XX,+X[1]/48,XX,+X,+X[1]/46,XX[92]dn
				P35	45,X[79]/47,XX,+X[21]dn
		STRUCTURAL ABNORMALITY (4)	*RING X (1)*	P36	45,X[70]/46,X,r(X)(p22q28)[30]dn
			ISOCHROMOSOME Xq (3)	P37	45,X[50]/46,X,i(X)(q10)[50]dn
				P38	45,X[50]/46,X,i(X)(q10)[50]dn
				P39	45,X[81]/46,X,i(X)(q10)[19]dn
STRUCTURAL ABNORMALITY (13)	*PARTIAL Xp DELETION (2)*	*NON-MOSAIC (1)*		P40	46,X,del(X)(p11pter)[100]dn
		MOSAIC (1)		P41	46,X,del(X)(p10)[78]/45,X[22]dn
		NON-MOSAIC (3)		P42	46,X,del(X)(q22q23)[100]dn

	PARTIAL Xq DELETION (4)		P43	46,X,del(X)(q22.1q23)[100]dn
			P44	46,X,del(X)(q22qter)[100]dn
		MOSAIC (1)	P45	46,X,del(X)(q26)[53]/45,X[24]/46,XX[23]dn
	RING X, MOSAIC (1)		P46	46,X,r(X)(q10q28)[30]/45,X[20]/46,X,dic r(X;X)(p22q28;p22q28)[5]/46,XX[45]dn
	ISOCHROMOSOME Xq (6)	*NON-MOSAIC (5)*	P47-51	46,X,i(X)(q10)[100]dn
		MOSAIC (1)	P52	46,X,i(X)(q10)[50]/45,X[44]/47,X,i(X)(q10),+i(X)(q10)[6]dn
KLINEFELTER SY. & VS. (29 patients)				
CLASSIC (25)	*NON-MOSAIC (23)*		P53-75	47,XY,+X[100]dn
	MOSAIC WITH 46,XY (2)		P76	47,XY,+X[3]/46,XY[97]dn
			P77	47,XY,+X[4]/46,XY[96]dn
>1 EXTRA X CHROMOSOME (2)	*NON-MOSAIC (1)*		P78	48,XY,+X,+X[100]dn
	MOSAIC (1)		P79	49,XY,+X,+X,+X[90]/48,XY,+X,+X[8]/46,XY[2]dn
STRUCTURAL ABNORMALITY (2)	*ISOCHROMOSOME Xq, MOSAIC (1)*		P80	48,XY,+i(X)(q10),+21[18]/47,XY,+21[82]dn
	PARTIAL Xp DUPLICATION (1)		P81	46,dup(X)(p22.33)mat,Y[100]
TRIPLE & POLY X SYS. & VS. (9 patients)				
TRIPLE X (8)	*CLASSIC (7)*	*NON-MOSAIC (6)*	P82-87	47,XX,+X[100]dn
		MOSAIC (1)	P88	47,XX,+X[7]/45,X[2]/46,XX[91]dn
	PARTIAL, NON-MOSAIC (1)		P89	46,X,dup(X)(p22.33)[100]unknown
POLY X, NON-MOSAIC (1)			P90	48,XX,+X,+X[100]dn
DOUBLE Y SY. & VS. (11 patients)				
NON-MOSAIC (10)			P91-100	47,XY,+Y[100]dn
MOSAIC (1)			P101	47,XY,+Y[8]/46,XY[92]dn
COMBINED PARTIAL DELETION & DUPLICATION OF X (1 patient)				
NON-MOSAIC (1)			P102	46,X,der(X)del(X)(p22.33)dup(X)(p22.33p11.22),dup(7)(p22.3p21.3)[100]dn • del(X)(p22.33)-pathogenic • dup(X)(p22.33p11.22)-likely pathogenic • dup(7)(p22.3p21.3)-pathogenic
PARTIAL Y DELETION (1 patient)				
NON-MOSAIC (1)			P103	46,X,del(Y)(q11.1q11.23)[100]dn • del(Y)(q11.1q11.23)-pathogenic
X-AUTOSOME TRANSLOCATIONS (4 patients)				
BALANCED (3)	*t(X;9) (1)*		P104	46,XX,t(X;9)(q22;p24.2)[100]dn
	t(X;10) (1)		P105	46,XX,t(X;10)(q13.3;q11)[100]dn
	t(X;19) (1)		P106	46,XX,t(X;19)(p11;p13),del(15)(q26.3)[100]dn

			• del(15)(q26.3)-likely pathogenic
UNBALANCED (1)	*t(X;10) (1)*	P107	46,XX,t(X;10)(p21;p12)dn,dup(7)(q31.1)pat,dup(X)(p22.11)mat[100] • dup(7)(q31.1)pat-likely benign • dup(X)(p22.11)mat-likely benign
X-Y TRANSLOCATIONS (9 patients)			
t(Xp;Yp) (5)		P108-112	46,XX.ish,der(X)t(X;Y)(p22.3;p11.3)(*SRY*+)[100]dn
t(Xp;Yq) (4)	*IN FEMALE (3)*	P113	46,X,der(X)del(X)(p22.3pter)add(Y)(q11.21q12)mat[100] • der(X)del(X)(p22.3pter)add(Y)(q11.21q12)mat-pathogenic
		P114	46,X,der(X)del(X)(p22.3pter)add(Y)(q11.21q12)mat[100] • der(X)del(X)(p22.3pter)add(Y)(q11.21q12)mat-pathogenic
		P115	46,X,der(X)del(X)(p22.3pter)add(Y)(q11.21q12)dn*[100] • der(X)del(X)(p22.3pter)add(Y)(q11.21q12)dn*-pathogenic
	IN MALE (1)	P116	46,der(X)del(X)(p22.3pter)add(Y)(q11.21q12)mat,Y[100] • der(X)del(X)(p22.3pter)add(Y)(q11.21q12)mat-pathogenic
OGDs (21 patients)			
COMPLETE/PURE (16)	*46,XX DSD (1)*	P117	46,XX(*SRY*-)[100]dn*
	46,XY DSD (15)	P118-132	46,XY(*SRY+*)[100]dn*
MIXED/MOSAIC (5)	*46,XX/46,XY DSD (1)*	P133	46,XX(*SRY*-)[22]/46,XY(*SRY*+)[78]dn
	45,X/46,XY DSD (3)	P134	45,X(*SRY-)*[17]/46,XY(*SRY*+)[83]dn
		P135	45,X(*SRY*-)[28]/46,XY(*SRY*+)[72]dn
		P136	45,X(*SRY*-)[49]/46,XY(*SRY*+)[51]dn
	45,X/46,XX/46,XY DSD (1)	P137	45,X(*SRY*-)[17]/46,XX(*SRY*-)[78]/46,XY(*SRY*+)[5]dn

Abbreviations: +: positive; -: negative; *: supposedly *de novo* according to the nature of the genetic abnormality (e.g. infertility) or negative family history; ACMG: American College of Medical Genetics and Genomics; add: addition; CNV: copy number variation; del: deletion; der: derivative chromosome; dic: dicentric; dn: *de novo*; DSD: disorders of sex development; dup: duplication; i: isochromodome; mat: maternally inherited; OGD: other gonadal dysgenesis; p: short arm of a chromosome; P: patient; pat: paternally inherited; q: long arm of a chromosome; r: ring; SCA: sex chromosome abnormality; *SRY*: Sex Determining Region Y gene; sy.: syndrome; t: translocation; ter: terminal; unkown: inheritance is unknown; v.: variant; X: X chromosome; Y: Y chromosome