

Chromosomal anomaly	Causative gene(s)	Location	Transmission	Phenotype(s)
46,XX (deletion diagnosed though MLPA)	<i>WNT4</i>	1p36.12	AD	46,XX DSD (Zhu et al., 2021)
46,XY.arr[GRCh37] 2p16.3(48,861,903-48,988,580)x1	<i>LHCGR</i>	2p16.3	AD	46,XY DSD (Abualsaud et al., 2021; Kon and Fukami, 2015) (MIM#238320)
2p16.3 deletion			AD	
46,XY.arr cgh 2q22.2q22.3(RP11-734C21-.RP11-294G19)x1	<i>ZEB2</i>	2q22.3	AD	46,XY DSD (Tannour-Louet et al., 2010) Mowat-Wilson syndrome (MIM#235730)
2q31.1q32.1 deletion	<i>HOXD cluster</i>	2q31.1	AD ?	46,XY DSD (Kon and Fukami, 2015)
46,XY.der(14)t(3;14)(q11.1;p11.2)dmarr[GRCh37]3q11.1q29(93580736_197840339)x3 arr[GRCh37] 3p26.3p26.1(93949_8411977)x1,3q26.1q29(162391365_197840339)x3	<i>TBL1XR1</i>	3q26.32	AD	This study (Dworschak et al., 2017)
46,XY.arr[GRCh37] 3q26.33(180,678,656-181,472,237)x1	<i>SOX2</i>	3q26.33	AD	46,XY DSD (Abualsaud et al., 2021)
46,XY.arr[GRCh37] 4p16.3p15.2(51,413-27,238,111)x3 dn,4q32.3q35.2(165,361,405-190,896,674)x1 dn	<i>NSD2</i>	4p16.3	AD ?	This study (Battaglia et al., 2015)
46,XY.der(5)(qter->q34::p14->qter)	<i>ADCY2</i>	5p15.31	AD	46,XY DSD (Tannour-Louet et al., 2010)
arr[GRCh37]5q11.1q11.2(49430268_56998592)x1	<i>MAP3K1</i>	5q11.2	AD	46,XY DSD (Bayat et al., 2021) MIM#613762
Homozygous gene deletion of various sizes	<i>CYP21A2</i>	6p21.33	AR	46,XX DSD (Tolba et al., 2022) (MIM#201910)
46,XX. arr[GRCh37] 6q27(168845632_170892302)x1	<i>DLL1</i>	6q27	AD	This study, (Lesieur-Sebellin et al., 2022)
46,XY.arr[GRCh37] 4p16.3(51,413-3,872,980)x1,7p22.3p22.1(54,185-6,777,295)x3 dpat	<i>ACTB</i>	7p22.1	AD	This study (Ronconi et al., 2017)
46,XY.del(8)(p23.1p23.1)	<i>GATA4</i>	8p23.1	AD	46,XY DSD (Wagner-Mahler et al., 2019)
46,XY.t(8;20)(q22.3;p13)	<i>ZFPM2</i>	8q23.1	AD	46,XY DSD (Tar et al., 1997) MIM#616067
mos 45,XY,-9[8]/46,XY,r(9)[29]/47,XY,+idic r(9)x 2[1]/46,XY,idic r(9)[1]/46,XY[1]	<i>DMRT1</i>	9p24.3	AD	46,XY complete gonadal dysgenesis (Marsudi et al., 2018) (when deleted)
47,XX,+der(9)dup(9)(q12q33)del(9)(q33q34.3)				46,XX DSD (Wyandt et al., 2000) (when duplicated)
46,XY.arr[GRCh37] 9q33.3(127,251,113-127,487,356)x1	<i>NR5A1</i>	9q33.3	AD	46,XY DSD (MIM#612965) (Harrison et al., 2014) 46,XX DSD (MIM#617480) (Jaillard et al., 2020) POI (MIM#612964)
Deletions ~600 kb upstream of SOX9	<i>SOX9</i>	17q24.3	AD	46,XY complete gonadal dysgenesis (Kim et al., 2015; Morozumi et al., 2018) (MIM#114290)
Duplications ~500 kb upstream of SOX9			AD	46,XX testicular DSD (Croft et al., 2018; Kim et al., 2015)
46,XY.arr[GRCh37] 10q25.3q26.12(118907361-122761687)x1	<i>EMX2</i>	10q26.11	AD	46,XY DSD (Piard et al., 2014) (when deleted)
46,XY.arr[GRCh37] 10p13(12342101-12730020)x3	<i>CAMK1D</i>	10p13	AD	46,XY DSD (White et al., 2011)
46,XY.arr[GRCh37] 10q25.3q26.13(117,012,878-125,217,066)x1 dn	<i>FGFR2</i>	10q26.13	AD	46,XY DSD (Tosur et al., 2015)
46,XY.del(11)(p12p14.1)	<i>WT1</i>	11p13	AD	WAGR syndrome (MIM#194072) (Le Caignec et al., 2007) Denys-Drash syndrome (MIM#194080) Frasier syndrome (MIM#136680)
46,XX.arr[GRCh37] 11q24.2q25(125,101,271-134,927,114)x1 dn	<i>BARX2</i>	11q24.3	AD ?	This study
46,XY.der(11)t(9;11)(q33.3;q24.3)dn.arr[GRCh37]9q33.3q34.3(126027848_141008915)x3,11q24.3q25(129034189_134868407)x1				
47,XY,+i(12)(p10)dn.arr[GRCh37] 12p13.33p11.1(163593_34398316)x4 dn	<i>CHD4</i>	12p13.31	AD	This study
46,XY.arr[GRCh37] 13q24.13q24.21(113,120,652-115,362,584)x1	<i>TBX3</i>	12q24.21	AD	46,XY DSD (Abualsaud et al., 2021) (MIM#181450)
46,XY.arr[GRCh37] 13q12.3(29,245,232-31,511,604)x1	<i>KATNAL1 ?</i>	13q12.3	AD	46,XY DSD (Abualsaud et al., 2021)
46,XX.arr[GRCh37] 13q12.11(22245874-22276184)x3,13q31.1(80909499-80915699)x3	<i>FGF9, SPRY2</i>	13q12.11, 13q31.1	AD	46,XX testicular DSD (Chiang et al., 2013; Ürel Demir et al., 2017)
47,XX,+13			NA	
46,XY.arr[GRCh37] 15q11.2q15.1(22,770,421-42,889,047)x3	<i>MAGEL2, SNRPN</i>	15q11.2	AD, imprinted genes	46,XY DSD (Abualsaud et al., 2021)

46,XY.arr[GRCh37] 16p13.3(chr16:446961-1013822)x3 Upstream of SOX8	SOX8	16p13.3	AD	46,XY DSD (Erickson et al., 2011)
46,XX.arr[GRCh37] 19q13.42q13.43(55784730_57496219)x1	UBE2S, NLRP2, NLRP5	19q13.42q13.43	AD ?	This study
mos 47,XX,+der(22)(pter->q11.21:)[5]/46,XX[25].arr[GRCh37] 22q11.1q11.21(17096855_22055183)x2-3[0.50]	CECR2	22q11.1q11.21	AD ?	This study
	COMT, HIRA	22q11.21		
47,XY,+der(Y),t(X;Y)(p21.1;p11.2) 46,XY,der(22)t(X;22)(p11.4;p13)	NROB1, MAGEB	Xp21.2	XLR	46,XY complete gonadal dysgenesis (McElreavey and Cortes, 2001; Simioni et al., 2016; Zarate et al., 2011) (when duplicated)
46,XY.arr[GRCh37] Xq12(66841607-66885230)x2	AR	Xq12	XLR	46,XY DSD (Sreenivasan et al., 2022) (MIM#300068)
46,XY.arr[GRCh37] Xq13.3q21.1(75583865-77097512)x2	ATRX	Xq21.1	XLR	46,XY DSD (Chen et al., 2017) (MIM#301040)
Duplications of various sizes	SOX3	Xq27.1	XLD	46,XX testicular DSD (Tasic et al., 2019)
Deletion 343kb upstream of SOX3				
Xp-Yp or Xq-Yp translocations	SRY	Yp11.3	NA	46,XX (or 45,X) testicular DSD (McElreavey and Cortes, 2001)

Supplementary Table 1: Review of previously published CNV linked to DSD with the related causative gene(s).

AD: autosomal dominant, AR: autosomal recessive, XLR: X-linked recessive, XLD: X-linked dominant

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