

Reference	Number of observation	Karyotype	FISH	aCGH or MLPA			Main outcome	origin	Duplicated breakpoint
				Genome	Initial breakpoint	Final breakpoint			
Xu et al., 2024	U	47,XY,+mar	-	GRCh37/hg19	16,888,900	18,644,241	Dysmorphic with severe unilateral hearing loss at 2 years	dn	←A
Jiang et al., 2024a	2	47,XN,+mar	-	GRCh37/hg19	16,888,900	18,644,241	Interrupted pregnancy	mat	←A
	17	47,XN,+mar	-	GRCh37/hg19	16,888,899	18,649,190	Interrupted pregnancy	-	←A
	26	47,XN,+mar	-	GRCh37/hg19	16,888,899	18,649,190	Interrupted pregnancy	dn	←A
Mansur et al., 2023	U	-	-	GRCh37/hg19	16,888,899	17,950,504	Unique Liver and dermatological presentation	-	Before A
Lu et al., 2023	Father	47,XY,+mar	ish mar (RP11-958H20++ or enh)	GRC38/hg38	16,500,000	18,200,000	CES	-	←A
	Daughter	47,XX,+mar	ish mar (RP11-958H20++ or enh)	GRC38/hg38	16,500,000	18,200,000	CES	pat	←A
Wang et al., 2023	Mother	-	-	GRCh37/hg19	7100001	18,650,000	Eye and heart defects	-	←A
	Daughter	47,XX, + mar	-	GRCh37/hg19	7100001	18,650,000	Eye defect	mat	←A
Gaspar et al., 2022	1	47, XY,+idic(22)(q11.21)	-	-	17,068,189	18,894,894	CFM/CES	-	←A
Serra et al., 2022	U	47,XX,+mar	-	GRCh37/hg19	16,054,691	19,010,508	CES	dn	←A
Wu et al., 2022	13	47,XY,+psu idic(22)(q11.2)	Two 22q11.2 signals (probe?)	GRCh37/hg19	16,888, 899	18,649,190	Died 10 days after birth	-	←A
Xue et al., 2021	1	47,XY,+mar	RP11-958H20x4	GRCh37/hg19	16,900,884	18,400,884	CES	dn	Before A
Li et al., 2021	U	47,XY,+mar.ish idic(22)(q11.2)	?	GRCh37/hg19	16,888, 899	18,644, 241	Dysmorphic with heart defect. Recurrent respiratory infections	dn	←A
Alsat et al., 2018	U	47,XX,+mar.ish idic(22)(11.2)	(wcp22+, TUPLE1-)	MLPA	IL17RA	USP18	Died on his second day of life	mat	←A
AlSubaihini et al., 2018	U	47,XX,+idic(22)(q11.21)	-	GRCh37/hg19	16,888,899	18,648,856	Mullerian Agenesis	-	←A
Jedraszak et al., 2015a	1	47,XY,+der(22)(q11.1q11.21)	ish(RP11-112D4)×4[12/15].nucish(RP11-112D4)×4[85/100]	NCBI36/hg18	15,443,579	17,021,468	Growth hormone deficiency and pituitary malformation	dn	Before A
	2	47,XY,+der(22)(q11.1q11.21)	ish(RP11-112D4)×4[14/20].nucish(RP11-112D4)×4[70/100]	NCBI36/hg18	15,443,579	17,021,468		dn	Before A
Tsai et al., 2015	U	47,XY,+mar(22)[10]/46,XY[10]	-	-	15,777, 528	17,145,109	Type B Interrupted Aortic Arch and Hydrocephalus	dn	Before A
Jedraszak et al., 2015b	U	47,XY,+idic(22)(q11.21)	ish22q11.21(RP11-112D4)x4[15]	GRCh37/hg19	16,053,473	18,641,468	Severe Psychomotor Delay	dn	←A
Haltrich et al., 2014	U	47,XY,+mar[60]/46,XY[40]	ish idic r(22)(q11.1.q11.21)×4	GRCh37/hg19	17,435, 645	18,656,678	Hyperinsulinaemia, severe immunodeficiency and grave cognitive deficits	dn	←A
Chen et al., 2013	U	47,XY,+mar[15]/46,XY[6]	-	GRCh37/hg19	17,397,498	18,661,773	Interrupted pregnancy	-	←A
Chellapandian & Schneider, 2013	U	47 XY, isodic(22)(q11.2)	-	-	14, 435, 171	17, 035, 860	Anatomical Asplenia	-	←A
Quintero-Rivera, 2013	U	47,XX,+mar.	ish idic(22)(q11.?) (SKY+, wcp22+, D22Z1++, TUPLE1-)	-	17,381,569	18,451,769	CFM/CES	dn	Before A
Córdova-Fletes et al., 2012	U	47,XX,+psu dic(22)(q11.21)	-	NCBI36/hg18	15,250,000	17,035,860	Dysmorphic with psychomotor development delay	dn	←A

Reference	Number of observation	Karyotype	FISH	aCGH or MLPA			Main outcome	origin	Duplicated breakpoint
				Genome	Initial breakpoint	Final breakpoint			
Belangero et al., 2012	1 *			MLPA	IL17RA	USP18	Preauricular tags/pits and heart defect	dn	←A
	2			MLPA	IL17RA	USP18	Dysmorphic whith development delay and heart defect	dn	←A
	3	XX,+idic(22)(pter→q11.21:q11.21→pter)	ish idic(22)(RP11- 81B3+,RP11-479G10-)	MLPA	IL17RA	USP18	Dysmorphic whith development delay and heart defect	dn	←A
	4			NCBI36/hg18	14,435,171	17,190,083	Dysmorphic whith development delay and heart defect	dn	←A
	5			MLPA	IL17RA	USP18	Dysmorphic	mat	←A
	6			MLPA	IL17RA	USP18	Dysmorphic whith development delay	mat	←A
	7			MLPA	IL17RA	USP18	Dysmorphic	dn	←A
Kvarnung et al., 2012	II:2	47,XY,+mar[4]/46,XY[260]	inv dup(22)(RP11-423L23; RP11-419G2)x4 Blood: 2.8% [787]; Bucal mucsa: 5.4% [165]; Spermatozoa: 49.6% [889]	-		CESCR	Discrete downslanting of the palpebral fissures and preauricular pits	dn	←A
	III:1	-	inv dup(22)(RP11-423L23; RP11-419G2)x4 Blood: 20% [530]; Bucal mucsa: 32% [78]	-		CESCR	Healthy	pat	←A
	III:2	47,XX,+mar(28%)/46,XY	inv dup(22)(RP11-423L23; RP11-419G2)x4 Blood: 29% [525]; Bucal mucsa: 63% [52]	-		CESCR	CES	pat	←A
	III:3	47,XY,+mar(50%)/46,XY	inv dup(22)(RP11-423L23; RP11-419G2)x4 Blood: 27% [550]; Bucal mucsa: 47% [96]	NCBI36/hg18	15275597	17,086,045	CES	pat	←A
Knijnenburg et al., 2012	I	-	-	MLPA	SLC25A18	MICAL3		-	←A
	II-1	-	-	MLPA	SLC25A18	MICAL3		mat	←A
	III	46,XX	ish 22q11.1q11.21(RP11-958H20, RP11-155N18)x2[10].nuc ish 22q11.1(RP11-155N18x2)[29],22q11.21(RP11-958H20x3)[12/30]	NCBI36/hg18	16,179,398	16,777,897	Anorectal, renal and preauricular anomalies	mat	Before A
	II-2	-	-	MLPA	SLC25A18	MICAL3	Preauricular anomalies, renal anomalies and idiopathic growth hormone deficiency	mat	←A
Ko et al., 2010	U	47,XY,+mar.ish idic(22)(q11.1)(D22S43+)	ish idic(22)(q11.1)(D22S43+)	-	15,500,000	15,900,000	CES	dn	Before A
Belangero et al., 2009	U *	XX,+idic(22)(pter→q11.21:q11.21→pter)	ish idic(22)(RP11- 81B3+,RP11-479G10-)	MLPA	IL17RA	USP18	Interrupted Aortic Arch Type B	dn	←A
Bélien et al., 2008	U	47,XX,+mar	(RP11-154H4+ , TBX1-)	-	.	17,385,163	CES	dn	←A or before
Gomez-Lado et al., 2006	U	47 XX,/mar.	WCP22x3;(D22S553; D22S609; D22S942-TUPLE1; ARSA)x2	-	-	-	Duane's syndrome, CES	dn	←A or before

Reference	Number of observation	Karyotype	FISH	aCGH or MLPA			Main outcome	origin	Duplicated breakpoint
				Genome	Initial breakpoint	Final breakpoint			
Win et al., 2006	U	47,XX,+mar.ish psudic(22;22)(q11.2;q11.2)	(D14Z1/D22Z1++, D22Z4++,D22S75-)	-	-	-	Duane's syndrome, CES		←A
Gentile et al., 2005	U	-	(CTA154H4+, CTA433F6+, RP11-155F20+, RP11-47L18-,RP11-165F18-)	-	-	-	CES	dn	←B
	2	48,XY,+marx2[12]/47,XY,+mar[9]	ish dic(22)(q11.1)(D14Z1/D22Z1++,cos121/Z00042-)	-	-	-	Liveborn, normal at 6 years	mat	-
	7A	47,XY,+mar	ish i(22)(p10)(D22Z2++,D14Z1/D22Z1 dim)	-	-	-	TOP, anhydramnios, spina bifida aperta, and dysplastic kidneys on US, no postmortem	pat	←A
	18	47,XY,+mar	ish i(22)(p10)(D22Z2+or++,D14Z1/D22Z1-)	-	-	-	Liveborn, normal at 2.5 years	pat	←A
Bartsch et al., 2005	20	47,XY,+mar	ish i(22)(p10)(D22Z2++,D14Z1/D22Z1+,cos121/Z00042-)	-	-	-	Liveborn, normal at 2.5 years	pat	-
	26	47,XY,+mar	ish der(22)t(11;22)(q23.3q11.2)(CTA-919E7/D22S942+,MLL+)	-	-	-	Liveborn, multiple malformations, died 1 day old	pat	-
	28	47,XX,+mar	ish i(22)(p10)(D22Z2++,D14Z1/D22Z1 dim)	-	-	-	Liveborn, aside from adducted feet normal at the age of 4 months	pat	←A
Denavit et al., 2004	U	47, XX, +idic(22)(pter-->q11.2 ::q11.2-->pter)	-	-	-	-	Died at age one month of cardiac failure	dn	←B

* Same patient; U = Unic patient reporter; mar = marker chromosome; ish = FISH; mat = maternal inheritance; pat = paternal inheritance; dn = de novo.; A = LCR22A; B = LCR22B