Appendix 3. Association of Copy Number Variants With Specific Ultrasonographically Detected Fetal Anomalies

Deletion	Gestational Age (Weeks)	ISCN Nomenclature	US Anomalies
Duplication	11	arr 1q21.1(144,696,283-146,374,546)x3 mat	Nuchal translucency - 3.5 mm; Bladder, Dilated tense; Two vessel cord
Duplication	27	arr 1q21.1(144,083,908-144,643,814)x3 mat	AV canal; Oligohydramnios
Duplication	20	arr 1q21.1(144,083,907-144,723,881)x3 mat	Overlapping fingers - Bilateral; Elbow, Fixed flexed - Bilateral; Knee, Fixed extended - Bilateral; Thorax, other
Duplication	20	arr 2q13(111,085,100-112,782,390)x3 pat	VSD; Bilateral Ventriculomegaly - Present Uncoded; Echogenic bowel
Deletion	23	arr 3p14.1p13(67,917,266-72,754,608)x1 dn	Oligohydramnios; Absent cerebellar vermis; Posterior fossa cyst; Bilateral Ventriculomegaly - Mild; Absent/hypoplastic nose bone; Femur, Short - Bilateral
Duplication	20	arr 3p26.3(832,458-1,403,386)x3 mat	Bilateral Ventriculomegaly - Moderate
Duplication	18	arr 4q13.2q21.1(66,871,838-77,744,787)x3 pat	Micrognathia; Head, other; Echogenic bowel; Two vessel cord
Deletion	19	arr 5q35.1q35.2(171,483,988-176,409,787)x1 dn	Cleft Lip - Unilateral; Cleft Palate
Duplication	19	arr 5p13.2(36,984,535-37,151,099)x3 mat	VSD; Congenital diaphragmatic hernia
Deletion	12	arr 6q23.3q24.1(136,521,187-139,410,871)x1 dn	Cystic hygroma; Nuchal translucency
Deletion	20	arr 7q11.23(72,404,248-73,976,981)x1 dn	Fetal growth restriction - < 10th percentile; Echogenic bowel
Duplication	23	arr 8p22p21.3(17,905,977-19,482,435)x3 mat	Oligohydramnios; Echogenic kidney - Bilateral

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Deletion	Gestational Age (Weeks)	ISCN Nomenclature	US Anomalies
Duplication	18	arr 8q11.1(46,966,687-47,976,420)x3 pat	Hypoplastic right heart
Deletion	21	arr 8p23.1(11,578,132-11,789,207)x1 dn	Abnormal outflow tracts
Deletion	19	arr 8q12.1(61,104,853-62,025,350)x1 dn	Abnormal outflow tracts
Duplication	19	arr 8p23.2(3,674,351-5,938,253)x3 pat	Nuchal fold more or equal to 6mm; Echogenic intracardiac focus; Echogenic bowel
Duplication	12	arr 8p22(13,400,615-14,711,717)x3 mat	Abdominal cyst; Cystic hygroma; Nuchal translucency
Deletion	32	arr 10q21.1(53,697,770-59,847,798)x1 pat	Bilateral Ventriculomegaly - Moderate
Deletion	22	arr 10q21.1(54,376,822-56,475,582)x1 mat	Heart, other; Omphalocele; Chiari malformation; Bilateral Ventriculomegaly - Present Uncoded; Fetal growth restriction - < 5th percentile; Kyphosis; Thorax, other
Duplication	20	arr 10q26.13q26.2(125,730,437-129,696,364)x3 dn	Lissencephaly; Microcephaly
Deletion	23	arr 11q24.2q25(124,104,318-134,373,618)x1 dn	AV canal; Fetal growth restriction - < 10th percentile; Profile Flat; Talipes - Bilateral
Deletion	25	arr 12q24.33(131,718,039-132,287,718)x1 mat	Heart, other; Single effusion - Skin Edema; Fetal growth restriction - < 5th percentile
Deletion	22	arr 14q22.1(50,309,556-51,739,266)x1 mat	Cleft Lip - Unilateral; Cleft Palate; Two vessel cord
Deletion	27	arr 14q32.2q32.31(99,465,564-100,574,047)x1 dn	Fetal growth restriction - < 10th percentile
Duplication	11	arr 15q13.3(29,817,814-30,226,375)x3 mat	Nuchal translucency - 4.2 mm
Duplication	20	arr 15q13.3(29,817,814-30,297,358)x3 pat	Hydrops, Ascites; Hydrops, Pericardial effusion; Hydrops, Pleural effusion; Hydrops, Skin edema

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Deletion	Gestational Age (Weeks)	ISCN Nomenclature	US Anomalies
Deletion	34	arr 16p13.11p12.3(15,311,952-18,539,483)x1 dn	Abnormal outflow tracts; Hypoplastic left heart; Situs inversus
Deletion	19	arr 16p12.1(21,744,792-22,315,572)x1 pat	Hypoplastic left heart; Chiari malformation; Spina bifida; Bilateral Ventriculomegaly - Mild
Duplication	19	arr 16p13.11p12.3(15,311,952-18,048,552)x3 pat	Hypoplastic left heart; Congenital diaphragmatic hernia
Duplication	21	arr 16p13.11(14,805,289-16,203,401)x3 pat	Cerebellar hypoplasia; Ventriculomegaly/Hydrocephaly; Single effusion - Skin Edema; Profile Flat; Stomach bubble - Small; Strawberry sign; Nuchal fold more or equal to 6mm; Overlapping fingers - Bilateral; Elbow, Fixed extended - Bilateral; Knee, Fixed extended - Bilateral
Deletion	19	arr 16p13.11p12.3(15,389,422-18,634,733)x1 dn	Fetal growth restriction - < 10th percentile; Talipes - Bilateral; Fibula, Absent - Bilateral
Deletion	17	arr 16p13.11(14,817,705-16,679,871)x1 mat	VSD
Deletion	13	arr 16p11.2(29,238,063-30,098,209)x1 dn	Nuchal translucency - 4.6 mm
Deletion	20	arr 16p13.11(14,796,083-16,535,551)x1 dn	Ureterocele; Hydronephrosis - Moderate to Severe (AP > 10 mm); Echogenic kidney - Unilateral
Deletion	36	arr 17q12(31,474,734-33,323,031)x1 pat	Polyhydramnios; Femur, Short - Bilateral; Humerus, Short - Bilateral; Congenital diaphragmatic hernia
Deletion	33	arr 17p13.3(513-827,455)x1 mat	Polyhydramnios
Deletion	34	arr 17p13.3p13.2(48,338-4,855,684)x1 dn // 21q22.3(43,364,146-46,912,090)x3 dn	Polyhydramnios; Bilateral Ventriculomegaly - Moderate
Deletion	12	arr 17q12(31,890,368-33,318,471)x1 dn	Nuchal translucency - 3.7 mm
Duplication	20	arr 17q12(32,041,983-33,135,410)x3 pat	VSD; Talipes - Bilateral; Echogenic intracardiac focus

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Deletion	Gestational Age (Weeks)	ISCN Nomenclature	US Anomalies
Deletion	20	arr 17q12(31,930,167-33,323,031)x1 dn	Hydronephrosis - Mild (AP 5-10 mm); Echogenic kidney - Bilateral; Large kidney - Bilateral; Renal, other
Deletion	22	arr 17q12(31,501,499-33,678,831)x1 dn	Echogenic kidney - Bilateral; Large kidney - Bilateral
Deletion	14	arr 17q12(31,929,967-33,323,171)x1 dn	Nuchal translucency - 3.5 mm
Deletion	32	arr 19q13.32(52,174,243-52,869,532)x1 dn	Coarctation; Agenesis of corpus collosum; Bilateral Ventriculomegaly - Moderate; Fetal growth restriction - < 5th percentile
Duplication	23	arr 19q13.42q13.43(59,640,794-61,566,588)x3 dn	Coarctation
Duplication	21	arr 21q21.3(27,186,402-28,309,393)x3 mat	Fetal growth restriction - < 5th percentile; Femur, Short - Bilateral; Humerus, Short - Bilateral; Echogenic bowel
Deletion	11	arr 22q13.33(49,472,416-49,525,855)x1 dn	CNS, other; Cystic hygroma
Deletion	20	arr 22q11.21(17,041,723-19,835,418)x1 dn	Tetralogy of Fallot; Heart, other; Hydronephrosis - Mild (AP 5-10 mm)
Deletion	19	arr 22q11.21(17,041,723-19,835,418)x1 dn	Tetralogy of Fallot
Duplication	12	arr 22q11.2(17,041,523-19,891,669)x3 mat // 15q13.3(29,817,814-30,297,358)x3 mat	Nuchal translucency - 5.9 mm
Deletion	26	arr 22q11.21(19,084,421-19,835,417)x1 dn	Cystic lung lesion
Deletion	19	arr 22q11.21(17,299,741-19,835,557)x1 dn	Truncus Arteriosus; Micrognathia; Anal atresia; Multicystic kidney - Unilateral
Deletion	21	arr 22q11.21(19,411,059-19,835,557)x1 pat	Tetralogy of Fallot; VSD

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Deletion	Gestational Age (Weeks)	ISCN Nomenclature	US Anomalies
Deletion	21	arr 22q11.21(17,256,416-19,795,836)x1 dn	Tetralogy of Fallot; Single effusion - Pleural; Choroid plexus cyst
Deletion	20	arr 22q11.21(17,085,800-19,891,669)x1 dn	Hypoplastic right heart; Bilateral Ventriculomegaly - Severe; Femur, Short - Bilateral; Fibula, Short - Bilateral; Humerus, Short - Bilateral; Radius, Short - Bilateral; Tibia, Short - Bilateral; Ulna, Short - Bilateral
Deletion	22	arr 22q11.21(17,299,741-19,835,557)x1 dn	Fetal growth restriction - < 5th percentile
Deletion	19	arr 22q11.21(17,299,941-19,835,417)x1 dn	Hypoplastic left heart; Absent/hypoplastic nose bone; Multicystic kidney - Unilateral
Deletion	20	arr 22q11.21(17,299,941-19,835,417)x1 dn	Tetralogy of Fallot; Two vessel cord
Duplication	21	arr Xp11.1q11.1(57,307,607-61,734,924)x2 mat	Bilateral Ventriculomegaly - Mild; Nuchal fold more or equal to 6mm; Neck, other; Sandal gap - Bilateral
Duplication	16	arr Xp22.13p22.12(18,498,513-19,414,777)x2 mat	Micrognathia; Choroid plexus cyst
Duplication	20	arr Yq11.223(22,650,981-23,984,067)x2 pat	Bilateral Ventriculomegaly - Mild; Cleft Lip - Midline; Cleft Palate; Hypospadias; Rocker bottom foot - Bilateral; Clinodactyly - Bilateral; Talipes - Bilateral
Deletion	20	arr Yp11.2(6,652,867-8,894,823)x0 pat	Talipes - Bilateral

Complete list of all 61 copy number variants included in this analysis with their associated ultrasound findings. Cases listed have at least one all anomaly, normal karyotype, and copy number variants by microarray.

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