

Appendix 2. Association of Copy Number Variants With Specific Ultrasonographically Detected Fetal Anomalies

Ultrasound Anomaly Details

- Abdominal wall
 - Bladder exstrophy
 - Body-stalk anomaly
 - Cloacal exstrophy
 - Gastroschisis
 - Omphalocele
 - Other:

- CNS
 - Absent cerebellar vermis
 - Agenesis of corpus callosum
 - Anencephaly
 - Arachnoid cyst
 - Cerebellar hypoplasia
 - Chiari malformation
 - Dandy-Walker malformation
 - Encephalocele
 - Anterior
 - Posterior
 - Holoprosencephaly
 - Hydranencephaly
 - Iniencephaly
 - Lissencephaly
 - Parenchymal defect
 - Posterior fossa cyst
 - Spina bifida
 - Vascular anomaly
 - Ventriculomegaly/Hydrocephaly
 - Unilateral
 - Mild (10-12mm)
 - Moderate (13-15mm)
 - Severe (>15mm)
 - Bilateral
 - Mild (10-12mm)
 - Moderate (13-15mm)
 - Severe (>15mm)
 - Other:

- Ear
 - Outer ear malformation
 - Unilateral
 - Bilateral
 - Other:

- Effusion
 - Hydrops
 - Single effusion only
 - Ascites
 - Pericardial effusion
 - Pleural effusion
 - Skin edema

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The authors provided this information as a supplement to their article.



- Other:
- Fac
 - Eye anomalies
 - Cyclopia
 - Hypertelorism
 - Hypotelorism
 - Microphthalmia
 - Other:
 - Facial tumor
 - Lip - Cleft
 - Unilateral
 - Midline
 - Bilateral
 - Nose
 - Absent / hypoplastic nose bone
 - Depressed nasal bridge
 - Palate – Cleft
 - Profile
 - Frontal bossing
 - Micrognathia
 - Profile Flat
 - Other:
- Fetal growth restriction
 - Expected fetal weight
 - < 3rd percentile
 - < 5th percentile
 - < 10th percentile

- Gastro-intestinal tract
 - Abdominal cyst
 - Anal atresia
 - Bowel
 - Large obstruction
 - Meconium ileus
 - Small obstruction
 - Duodenal atresia
 - Liver nodules
 - Situs inversus
 - Stomach bubble
 - Enlarged
 - Small/absent
 - Other:

- Genitalia -
 - Ambiguous genitalia
 - Hydrocele
 - Hydrometrocolpus
 - Hypospadias
 - Micropenis
 - Other:

- Head shape -
 - Cloverleaf skull
 - Craniosynostosis

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- Macrocephaly
- Microcephaly
- Strawberry sign
- Other:

Heart -

- Abnormal 4-chamber view
- Abnormal outflow tracts
- Anomalous pulmonary venous return
- ASD
- AV canal
- Coarctation
- Dextrocardia
- Dilatation
- Ebsteins anomaly
- Heart tumor
- Hypoplastic left heart
- Hypoplastic right heart
- Interrupted aortic arch
- Pulmonary stenosis
- Tetralogy of Fallot
- Transposition
- Truncus Arteriosus
- VSD
- Other:

Neck -

- Cystic hygroma
- Nuchal fold $\geq 6\text{mm}$
- Nuchal translucency (in mm)
- Teratoma
- Other:

Renal tract -

- Abnormal adrenal glands
 - Hemorrhage
 - Tumor
- Bladder
 - Bladder Collapsed
 - Dilated tense
 - Dilated floppy
 - Ureterocele
- Duplex system
 - Unilateral
 - Bilateral
- Hydronephrosis
 - Mild (AP 5-10mm)
 - Moderate to severe (AP > 10mm)
- Kidney
 - Echogenic kidney
 - Unilateral
 - Bilateral
 - Horseshoe kidney
 - Large kidney
 - Unilateral
 - Bilateral
 - Multicystic kidney

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- Unilateral
- Bilateral
- Pelvic kidney
- Polycystic kidney
 - Unilateral
 - Bilateral
- Small kidney
 - Unilateral
 - Bilateral
- Renal agenesis
 - Unilateral
 - Bilateral
- Urethra
 - Absent
 - Dilated/Valves
- Other:

Skeletal -

- Skeletal dysplasia
(if yes, check individual components below)

Foot

- Absent Left Right Both
- Oligodactyly Left Right Both
- Polydactyly Left Right Both
- Rocker bottom foot Left Right Both
- Sandal gap Left Right Both
- Split foot Left Right Both
- Syndactyly Left Right Both
- Other:

Hand

- Absent Left Right Both
- Brachydactyly Left Right Both
- Clinodactyly Left Right Both
- Oligodactyly Left Right Both
- Overlapping fingers Left Right Both
- Polydactyly Left Right Both
- Split hand Left Right Both
- Syndactyly Left Right Both
- Other:

Joints

- Ankle
 - Fixed extended Left Right Both
 - Fixed flexed Left Right Both
 - Talipes Left Right Both
- Elbow
 - Fixed extended Left Right Both
 - Fixed flexed Left Right Both
- Knee
 - Fixed extended Left Right Both
 - Fixed flexed Left Right Both
- Wrist
 - Fixed extended Left Right Both
 - Fixed flexed Left Right Both
- Other:

Long bones

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- Femur
 - Absent Left Right Both
 - Bowed Left Right Both
 - Short Left Right Both
- Fibula
 - Absent Left Right Both
 - Bowed Left Right Both
 - Short Left Right Both
- Humerus
 - Absent Left Right Both
 - Bowed Left Right Both
 - Short Left Right Both
- Radius
 - Absent Left Right Both
 - Bowed Left Right Both
 - Short Left Right Both
- Tibia
 - Absent Left Right Both
 - Bowed Left Right Both
 - Short Left Right Both
- Ulna
 - Absent Left Right Both
 - Bowed Left Right Both
 - Short Left Right Both
- Other:

Spine -

- Hemivertebrae
- Kyphosis
- Sacral agenesis
- Sacrococcygeal teratoma
- Scoliosis
- Sirenomelia
- Other:

Thorax -

- Congenital diaphragmatic hernia
- Cystic lung lesion
- Hydrothorax
- Hypoplastic thorax
- Short ribs
- Other:

Soft markers

- Choroid plexus cyst(s)
- Echogenic bowel
- Echogenic intracardiac focus
- Nuchal Translucency of 3.0-3.4 mm
- Other:

Amniotic fluid anomaly -


- Oligohydramnios
- Polyhydramnios

Placental anomaly -

- Edema

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- Tumor
 - Umbilical cord anomaly -
 - 2 vessel cord
 - Velamentous cord insertion

Full list of ultrasound detected anomalies broken down by category and subcategory as entered into online database.

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