

OTHER ANOMALIES AND SYNDROMES

There is typically 1 syndrome tested each year. With hundreds of syndromes listed in the medical literature, it impossible to commit all of them to memory. The following syndromes have appeared on the Registry in the past.

TERMINOLOGY

Sequence:	A single defect leading to the development of other defects.
Syndrome:	The presence of two or more embryologically unrelated anomalies occurring together with relatively high frequency and having the same etiology. Two or more different organs must be affected.
Association:	Non-random occurrences of two or more defect occurring less frequently than in a syndrome, but more frequently than by chance alone. An example is VACTERL association.

AMNIOTIC BAND SYNDROME

The early rupture of the amnion may result in bands of tissue that insert on the fetal body early in pregnancy. As the fetus grows, the non-stretchable bands restrict tissue growth. Must be differentiated from uterine synechia. Multiple and severe abnormalities may result including:

LIMB DEFECTS

- Amputations
- Clubfeet
- Constriction rings
- Pseudosyndactyly

CRANIOFACIAL DEFECTS

- Anencephaly
- Encephalocele
- Facial clefting
- Severe facial dysmorphism

VISCERAL DEFECTS

- Gastroschisis
- Omphalocele

SONOGRAPHIC FINDINGS:

- Presence of two or more of the defects listed above
- Presence of multiple encephaloceles
- Identification of an echogenic linear structure attached to the fetus\
- Fetal postural deformities

TRISOMY 13 (PÄTAU SYNDROME)

A malformation syndrome with characteristic facies, hexadactyly, psychomotor retardation and multiple anatomic abnormalities. 85% die before 1 year. Occurs 1 in 5000 live births.

SONOGRAPHIC FINDINGS:

- ***holoprosencephaly***
- IUGR
- polyhydramnios
- CNS anomalies including: agenesis of the corpus callosum
- micrognathia, cleft lip/palate
- other congenital abnormalities associated with trisomy 13 may be visualized: omphalocele, VSDs, persistent truncus arteriosus, polycystic kidney disease, other GU abnormalities and holoprosencephaly

TRISOMY 18 (EDWARD'S SYNDROME)

A syndrome of primordial growth deficiency, typical facial dysmorphism, profound psychomotor retardation and other abnormalities. Occurs 1 in 3000 live births.

SONOGRAPHIC FINDINGS:

- ***clenched hand with finger/thumb crossover (arthrogryposis)***
- IUGR
- polyhydramnios (3rd trimester)
- VSD's, ASD's
- micrognathia
- dolichocephaly, prominent occiput
- enlarged cisterna magna
- limb malformations
- other congenital abnormalities associated with trisomy 18 may be visualized: diaphragmatic hernia, omphalocele, hydronephrosis

TRISOMY 21 (DOWNS' SYNDROME)

A malformation syndrome with mental retardation and a characteristic physical appearance. Occurs 1:710 live births.

SONOGRAPHIC FINDINGS:

- ***Nuchal fold measuring > 5mm at 20 weeks***
- There are no specific diagnostic findings for Down's syndrome
- Other congenital anomalies associated with Down's syndrome may be visualized including: cystic hygroma, mild ventriculomegaly, VSD's, ASD's, duodenal atresia, diaphragmatic hernia, renal pyelectasis

TURNER'S SYNDROME

A malformation syndrome attributed to complete or partial absence of the X chromosome in phenotypic females. Prognosis depends on extent of cardiac and renal abnormalities.

Pathology:

- Lymphedema, diffuse lymphangiectasia, cystic hygromqa
- Cardiac anomalies, aortic stenosis
- Horseshoe kidney, unilateral renal agenesis

KLEEBLATTSCHADEL SYNDROME

Pathology:

- Cloverleaf skull
- Normally proportioned trunk and extremities
- Absence of other apparent malformation

HOLT - ORAM SYNDROME

A hereditary syndrome of malformations of the heart and upper extremities.

Pathology:

- Varied hand anomalies
- Cardiac septal defects

FETAL ALCOHOL SYNDROME

Results from excessive maternal alcohol intake. More common in binge drinkers.

Pathology:

- IUGR
- Mental deficiency
- Microcephaly
- Microphthalmos
- Short metacarpals
- Cardiac septal defects
- Hypospadias, other genital anomalies

BECKWITH-WEIDEMAN SYNDROME

A group of congenital disorders characterized by the presence of EMG anomalies, i.e. exomphalos (omphalocele), macroglossia and gigantism.

Associated anomalies include:

Pathology:

- Cardiac malformations
- Placental enlargement
- Placental chorioangiomas
- Renal tumors (Wilm's tumors)

PENTALOGY OF CANTRELL

The association of two major defects: **omphalocele and ectopia cordis**. Three other defects are also present in the lower sternum, the anterior diaphragm and the diaphragmatic pericardium.

Pathology:

- Vertebral anomalies
- Atrial and ventricular septal defects
- Tetralogy of Fallot
- Trisomies 13 and 18
- Turner syndrome
- Craniofacial abnormalities
- Two-vessel cord
- Fetal ascites

MECKEL - GRUBER SYNDROME

Related to autosomal recessive inheritance.

Pathology:

- Encephalocele
- Polycystic kidneys
- Cleft lip
- Polydactyly

While the following syndromes have not been reported to have appeared on the exam, they are more commonly included in standard OB/GYN ultrasound textbooks,

TREACHER COLLINS SYNDROME

(Mandibulofacial dysostosis). An autosomal dominant disorder that affects the ears, mandible and palate. Frequently associated with congenital heart defects.

Pathology:

- Small mandible
- Micrognathia (receding chin)
- Autosomal trisomy anomalies
- Holoprosencephaly

CAUDAL REGRESSION SYNDROME

A severe fetal malformation consisting of agenesis of the sacrum and lumbar spine and hypoplasia of the lower extremities. Frequently associated with obstructive urinary tract pathology.

Pathology:

- Dilated bladder and ureters
- Thickened bladder wall
- Absent sacrum
- Small lower extremities

ELLIS - van CREVELD SYNDROME

Chondrodermal dysplasia. Occur 1:200,000 with a high prevalence among the Amish.

Pathology:

- Polydactyly
- Thoracic dysplasia
- Mild-moderate limb shortening
- 50% have associated congenital heart disease, most commonly atrial septal defects.

VATERS Association

Sometimes referred to as **VACTERL** association.

Pathology:

- Vertebral anomalies
- Anal atresia
- Cardiac anomalies
- Tracheo esophageal atresia
- Renal and radial ray anomalie
- Limb anomalies



HELPFUL HINTS

<i>FETAL TRISOMY SYNDROMES</i>	
Most suggestive sonographic finding:	
Trisomy 13	Holoprosencephaly
Trisomy 18	Clenched hands, overlapping digits
Trisomy 21	Nuchal thickening, endocardial cushion defect