The ultrasound detection of chromosomal anomalies[1]

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Trisomy 8

Synonyms: Warkany syndrome
Definition: Predominantly final malformations syndrome with a trisomy for all or a large part of the number 8 chromosome
Prevalence: less than 100 cases reported.
Etiology: Presence of an extra chromosome 8 or part of the short arm.
Pathogenesis: Unknown
Differential diagnosis: Other aneuploidies.
Prognosis: Better survivals than other trisomies, some reasonably healthy adults exist.
Recurrence risk: Unknown
Management: When ultrasound findings are consistent with trisomy 8, prenatal karyotyping should be undertaken. Pregnancy termination can be offered before viability. No alterations of prenatal care are necessary should the pregnancy be allowed to go to term.

Associated anomalies

Central nervous system anomalies

- Agenesis of the corpus callosum
- Hydrocephalus

Face, neck and skull anomalies

- Everted lips
- Large dysplastic ears
- Prominent fore head
- Broad nose
- Microphtalmia
- Cataract

Cardio-vascular anomalies 40-60%

- VSD
- ASD
- Great vessels anomalies

Gastrointestinal anomalies

- Diaphragmatic hernia
- Esophageal atresia
- Absence of the gallbladder

Urinary tract anomalies

- Hydronephrosis

- Reflux

Reproductive tract anomalies

- Cryptorchidism

Skeletal system anomalies

- Vertebral anomalies (Hemivertebrae, spina bifida, kyphoscoliosis)
- Joint contractures
- Abnormal metacarpals and metatarsals
- Simian crease
- Deep longitudinal plantar crease

Other findings & anomalies

- Associated malignancy
- May be related to old parental age.

Trisomy 9

Synonyms: None
Definition: Trisomy for all or a large part of the number 9 chromosome. Most cases are mosaic with 2-97 % abnormal cells.
Prevalence: Less than 30 cases reported.
Etiology: not associated with parental age.
Pathogenesis: Unknown.
Differential diagnosis: Other aneuploidies.
Prognosis: Very lethal trisomy with most newborn dying and very rare surviving 4 months.
Recurrence risk: Unknown.
Management: When ultrasound findings are consistent with trisomy 9, prenatal karyotyping should be undertaken. Pregnancy termination can be offered before viability. No alterations of prenatal care are necessary should the pregnancy be allowed to go to term. Tocolysis for preterm labor and cesarean section should be avoided

Associated anomalies

Central nervous system anomalies

- Dysgenesis of the corpus callosum.

Face, neck and skull anomalies

- Abnormal and low set ear
- Micrognathia
- Small palpebral fissure
- Microcephaly
- Broad nose
- Wide sutures-Craniosynostosis
- Cleft -high arched palate
- Short neck

Cardio-vascular anomalies

- ASD/VSD

- Persistent left superior vena cava
- Single umbilical artery.

Gastrointestinal anomalies

- Malrotation
- Diaphragmatic hernia
- Omphalocele 29

Reproductive tract anomalies

- Cryptorchidism
- Small penis
- Hypoplastic scrotum
- Various

Skeletal system anomalies

- hypoplastic bones
- Hand anomalies (abnormal fingers, overlapping, hypoplastic phalanges).
- Simian Crease
- Rocker bottom feet
- Deep palmar furrow
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Other findings and anomalies

- IUGR

4p- syndrome

Synonyms: Monosomy 4p. Partial deletion of chromosome 4. Wolf Syndrome.
Definition: Absence of part of the short arm of chromosome 4. 87% are due to de novo deletions (predominantly paternal origin) 13 % are inherited from parents with balanced translocations (predominantly maternal).
Prevalence: Over 100 cases reported.
Etiology: Abnormal chromosome breakage during synapsis and recombination.
Pathogenesis: Unknown.
Differential diagnosis: Other aneuploidies in particular trisomy 13.
Prognosis: Lethal with most newborn dying and very rare surviving 1 year.
Recurrence risk: Unknown.
Management: When ultrasound findings are consistent with partial deletion of chromosome 4, prenatal karyotyping should be undertaken. Pregnancy termination can be offered before viability. No alterations of prenatal care are necessary should the pregnancy be allowed to go to term. Tocolysis for preterm labor and cesarean section should be avoided

Associated anomalies

Central nervous system anomalies

- Absence of the cavum septum pelludidum
- Interventricular cysts
- Unspecified

Cardio-vascular anomalies 80%

- ASD
- Unspecified.

Gastrointestinal anomalies 50-80 %

- Malrotation
- Unspecified.

Urinary tract anomalies

- Hypoplastic kidney
- Reflux
- Various

Reproductive tract anomalies

- Hypospadias

Skeletal System anomalies

- Simian crease
- Scoliosis
- Abnormal ossification of the sternum
- Unspecified others

Other finding and anomalies

- IUGR
- Decreased fetal movements
- Weak cry after birth

Triploidy

Synonyms: Incomplete molar gestation, partial triploid mole.
Definition: Presence of 3 set chromosomes resulting in focal hydropic swelling of chorionic villi with throphoblastic hyperplasia and identifiable embryonic or fetal tissues.
Prevalence: 1- % conceptuses, but 0.1 - 1: 10.000 of all pregnancies.
Etiology: Abnormal fertilization
Pathogenesis: Fertilization of a normal (haploid) ovum by two normal (haploid) sperm or fertilization of a normal ovum by an abnormal (diploid) sperm. All configurations (XXX; XXY; XYY) have been found.
Differential diagnosis: Twin gestation with one fertilized ovum undergoing molar degeneration and hydropic changes in a missed abortion.
Associated anomalies: See table
Prognosis: Most die in utero during first or early second trimester and if born alive they die within a few hours.
Recurrence risk: Unknown, but probably none.
Management: When ultrasound findings are consistent with triploidy, prenatal karyotyping should be undertaken. Pregnancy termination can be offered before viability. No alterations of prenatal care are necessary should the pregnancy be allowed to go to term. Tocolysis for preterm labor and cesarean section should be avoided

Associated anomalies

Central nervous system anomalies

- Relative macrocephaly
- Agenesis of corpus callosum
- Dandy-Walker malformation
- Holoprosencephaly
- Arnold Chiari Malformation
- Spina bifida
- Meningomyelocele
- Hydrocephalus
- Central and cerebellar hipoplasia

Face, neck and skull anomalies

- Cleft lip
- Low set ears
- Micrognathia
- Hypertelorism
- Cystic Hygroma

Cardio-vascular anomalies 40-60%

- VSD
- ASD
- Many others.

Gastrointestinal anomalies

- Omphalocele
- Unspecified

Urinary tract anomalies

- Hydronephrosis
- Dysgenesis of kidneys
- Multicystic kidneys

Reproductive tract anomalies

- Hypospadias
- Ambiguity of external genitalia
- Cryptorchidism

Skeletal system anomalies

- Skeletal dysplasias
- Syndactyly of 3rd and 4th toes and fingers
- Short hallux
- Club foot
- Rocker-bottom foot
- Unspecified other

Other findings & anomalies

- IUGR
- Decreased fetal movement

- Lung hypoplasia
- Head to abdomen discrepancy with much decreased abdominal size.

Monosomy X

Synonyms: Turner Syndrome, 45, X0.
Definition: Absence of one X chromosome, either in all cells or as mosaic (45, X0/ 46, XX).
Absence of short arm results in the Turner phenotype. Absence of the short arm up to Xp11 or the long arm up to Xq21 results in gonadal dysgenesis.
Prevalence: 2:10,000 live births.
Etiology: Complete monosomy (57%), presence of an abnormal X chromosome (ring chromosome (10 %); isochromosome (17%)) or mosaic with 46, XX or 46 XY. Mosaic 45X / 46XY may span the rage between phenothipical Turner syndrome, ambiguous genitalia, hermaphroditic and almost normal male.
Pathogenesis: Meiotic non-disjunction during gametogenesis (complete monosomy) or mitotic error. The missing X is of paternal origin. Not increased with maternal age.
Differential diagnosis: Hydrops Fetalis
Prognosis: 95 % die in utero, those who survive may have a normal life span but infertility (99 %) or early menopause.
Recurrence risk: Not known but probably low.
Management: When ultrasound findings are consistent with Monosomy X, prenatal karyotyping should be undertaken. Pregnancy termination can be offered before viability. No alterations of prenatal care are necessary should the pregnancy be allowed to go to term. Tocolysis for preterm labor and cesarean section should be avoided

Associated anomalies:

Central nervous system anomalies

- Nothing special

Face, neck and skull anomalies

- Cystic Hygroma >80%
- Ear anomalies 80
- Short neck 80
- Narrow palate 80
- Micrognathia >70%
- Increased nuchal thickness 50
- Short metacarpal of 4th digit 50
- Short metatarsal of 4th digit 50
- Hypertelorism

Cardio-vascular anomalies 40-60%

- Bicuspid aorta valve
- Coarctation of the aorta
- Dilatation of the aorta
- Hypoplastic left heart syndrome
- Cardiac anomalies are more likely if there is a nuchal thickening. [Clark]

Gastrointestinal anomalies

- Nothing special.

Urinary tract anomalies

- Horseshoe kidneys
- Duplicated renal pelvis.

Reproductive tract anomalies

- Nothing special prenatally
- Primary or secondary amenorrhea at puberty

Skeletal system anomalies

- Simian crease
- Cubitus valgus (not detected prenatally)
- Short stature (probably not detected prenatally).

Other findings & anomalies

- Broad chest
- Pectus excavatum
- IUGR.

References

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