The ultrasound detection of chromosomal anomalies[1]

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Trisomy 21

Synonyms: Down syndrome, Mongolism (do not use!).
Definition: Multiple malformation syndromes with a trisomy for all or a large part of the number 21 chromosome.[4]
Prevalence: 13:10,000 live births with a maternal age effect.
Etiology: Presence of an extra chromosome 21 or a long arm including the q22.1 band in all or some (mosaic) cells.[1]

- 93% are free trisomies due to meiotic non-disjunction in one of the parents
  - 60% are maternal during a 1st meiotic division
  - 20% are paternal non-disjunction during a 1st or second meiotic division
- 5% are Roberstsonian translocations (14/21, 21/21, or 21/22) either de novo or parental
- 2% are mosaic (postzygotic event) and have a variable phenotype

Pathogenesis: Unknown; increased with maternal age.
Differential diagnosis: Multiple malformation syndromes, which include severe growth retardation, polyhydramnios, and congenital heart disease such as trisomy 18, trisomy 13, Smith-Lemli-Opitz or individual anomalies.
Associated anomalies: See table
Prognosis: Approximately 2/3 of the fetuses with trisomy 21 die before delivery. A 1/3 if the survivor die during the 1st year, 50% before the age of 4. The life expectancy of the remainder is shortened. Mental retardation (moderate to severe) is the rule but in some mosaic case it can almost be normal.
Decreased muscle tone.
Some anomalies will need surgery for correction as heart defects or duodenal atresia.
Recurrence risk: For free trisomy in young mothers: 1-2%, for older mother: depend on maternal age. For Robertsonian translocation 21/21: all offspring are either mono- or trisomic, for 14/21 or 21/22 from the mother: 16% from the father: 5%. For the proband's child: 50%.
Management: When ultrasound findings are consistent with trisomy 21, 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.
MESH Trisomy 21 BDE 0171

Associated anomalies

Central nervous system anomalies

- Mild ventriculomegaly (<15mm)

Face, neck and skull anomalies

- Flattened face 90%
- "Nuchal fold"
- Nuchal translucency

- Oblique palpebral fissure 80
- Flat occiput 78
- Brachycephaly 75
- High-arch palate 70
- Low nasal bridge 60
- Ear anomalies 50
- Epicanthal fold 40
- Cataract 3
- Low set ears
- Macroglossia
- Small nose
- Mild microcephaly

Cardio-vascular anomalies 40-60%

- Endocardial cushion defect
- VSD
- ASD
- Aberrant subclavian artery
- Tetralogy of Fallot
- Pericardial effusion

Gastrointestinal anomalies

- Duodenal atresia 30%
- Tracheoesophageal fistula
- Omphalocele
- Pyloric stenosis
- Annular pancreas
- Hirschprung's disease
- imperforate anus
- “Bright bowel”

Urinary tract anomalies

- Mild pyelectasis

Reproductive tract anomalies

- Small penis

Skeletal system anomalies

- Short limbs 70%
- Short finger 70
- Abnormal iliac wing angle 67
- Brachymesophalangia 62
- Clinodactyly 50
- Simian crease 50
- “Sandal gap” 45

- Plantar crease between 1st and 2nd toe 28
- Single flexion crease of 5th phalangeal joint 20
- 11 pairs of ribs
- Instability of atlas-axis joint
- Double ossification of the manubrium
- Funnel or pigeon breast
- Hip anomalies

Other findings & anomalies

- Hypotonia 20-80%
- Goiter
- Leukemia 1

Trisomy 18

Synonyms: Edwards syndrome, Trisomy E.
Definition: Multiple malformation syndromes with a trisomy for all or a large part of the number 18 chromosome.
Prevalence: 3:10,000 live births with a maternal age effect.
Etiology: Parental non disjunction, rarely parental translocation. About 80 % of the parents have a straight trisomy. Another 10 % are mosaics whereas the rest either are double trisomies for another chromosome or have a translocation. Pericentric inversion in chromosome has been described to recombine during meiosis and cause unbalanced offspring phenotypically similar to those fetus with a trisomy 18.
Pathogenesis: results from a faulty chromosomal distribution, which is mostly likely to occur in the older gravida age. Increase maternal age is a risk factor, and the parental origin of extra chromosome is maternal age in 96 % of cases in whom chromosomal origin could be determined.
Differential diagnosis: Multiple malformation syndromes, which include severe growth retardation, polyhydramnios, and congenital heart disease such as, trisomy 13,triploidy, Pena- Shokeir syndrome, should also be .include in the differential diagnosis.
Prognosis: Although trisomy 18 is less common than trisomy 21 it is more lethal. 96 % of live born trisomy 18 infants die in the first month, 50 % within two months, and only 10 % survive the first year and are profoundly mentally retarded. Approximately 68 % of the fetuses with an in utero diagnosis of trisomy 18 die before delivery.
Recurrence risk: For full trisomy 18, the recurrence risk is lower than the 1 % for full trisomy 21 syndrome. A carrier of pericentric inversion in chromosome 18 may produce affected offspring in 6 % of pregnancies and carrier offspring in 53 % of such pregnancies.
Management: When ultrasound findings are consistent with trisomy 18, 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
MESH Trisomy 18 BEE 0160 MIM 257300 POS 3094 ICD 758.2 CDC 758 0000

Associated anomalies:

Central nervous system anomalies

- Meningomyelocele 16 %
- Arnold-Chiari Malformation 13
- Abnormal Gyration 13

- Heterotopics 13
- Abnormal olivary nuclei 10
- Arachnoid cyst 3
- Hypoplastic cerebellar vermis 3
- Alobar Holoprosencephaly 3

Face, neck and skull anomalies

- Low set ear 58 %
- Micrognathia 48
- Small face 35
- Abnormal ears 35
- Small eyes 26
- Small mouth 16
- Microcephaly 16
- Large anterior fontanelle 13
- Hypertelorism 6
- Choanal atresia 6
- Preauricular tag 6
- High arcade palate 6
- Cleft palate 3
- Small anterior fontanelle 3
- Third fontanelle 3
- Low hair line 3

Cardio-vascular anomalies

- VSD 81%
- Polyvalvular dysplasia 65
- Bicuspid aortic valve 45
- Bicuspid pulmonary valve 42
- Coarctation of the aorta 35
- Atrial septal defect 10
- Endocardial cushion defect 10
- Mitral atresia 6
- Double right ventricle 6
- Dextrocardia 6
- Transposition of great vessels 6
- Retroesophageal subclavian vein 6
- Tetralogy of Fallot 3
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- Hypolastic left ventricle 3
- Common atrium 3
- Anomalous pulmonary venous return 6
- Single coronary ostium 3

Gastrointestinal anomalies

- Omphalocele 29
- Meckel's diverticulum 26
- Malrotation of intestine 23
- Diaphragmatic hernia 19
- Ectopic pancreas 16
- Tracheoesophageal fistula 10
- Ectopic gastric tissue 6

- Ileal atresia 3
- Imperforate anus 3
- Absent gallbladder 3
- Absent appendix 3
- Inguinal hernia 3
- Anomalies of the pancreas 6
- Accessory spleen 3

Urinary tract anomalies

- Horseshoe kidney 23
- Hydroureter 16
- Duplicated ureter 13
- Renal microcysts 6
- Renal cystic dysplasia 6
- Bladder diverticulum 3
- Bladder outlet obstruction 3

Reproductive tract anomalies

- Cryptorchidism 26
- Dysplastic ovaries 16
- Bicornuate uterus 10
- Hypospadias 3
- Septate uterus 3
- Abnormal external genitalia 3

Skeletal system anomalies

- Overlapping fingers 71
- Rocker bottom feet 39
- Clubfeet 32
- Single palmar 23
- Hypoplastic nails 19
- Short sternum 13
- Clinodactyly 13
- Syndactyly 10
- Abnormal ribs 10
- Hip dislocation 6
- Deviation of hands 6
- Small pelvis 6
- Hemivertebrae 3
- Redundant skin 3
- Cleft in hand 3
- Small great toe 3

Other Findings and anomalies

Body

- Growth retardation 87
- Thin body habitus 13
- Hydrops 10
- Cystic hygroma 3
- Redundant skin 3

Respiratory System

- Pulmonary hypoplasia 58

Other findings
- Extramedullary hematopoiesis 23
- Adrenal hipoplasia 23

Placental and cord
- Two vessel cord 29
- Polyhydramnios 29
- Villitis 13
- Chorioamnionitis 6
- Trophoblastic inclusions 3

Trisomy 13

Synonyms: Patau Syndrome. D trisomy.
Definition: Multiple malformation syndromes with a trisomy for all or a large part of the number 13chromosome.
Etiology: Presence of an extra chromosome 13 or part of the long arm (proximal segment 13 pter→q 14 or distal segment 13q14→qter).
The majority is free trisomy due to meiotic non-disjunction in one of the parents.
A few are mosaic (postzygotic event) and have a variable phenotype.
Rare Robertsonian translocations (13/14, 13/15) either de novo or parental.
Pathogenesis: Unknown; increased with maternal age.
Differential diagnosis: Multiple malformation syndromes, which include severe growth retardation, polyhydramnios, and congenital heart disease such as trisomy 18, or individual anomalies.
Prognosis: A majority with trisomy 13 dies before delivery or is stillborn. 80 % of the survivor die during the first month, 95 % before 6 months.
Recurrence risk: For the Trisomy: not increased. For Robertsonian translocation 2%.
Management: When ultrasound findings are consistent with trisomy 13, 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

- Holoprosencephaly
- Deafness
- Dysgenesis of the corpus callosum
- Hydrocephaly
- Cerebellar hypoplasia
- Meningomyelocele

Face, neck and skull anomalies

- Cleft lip/ palate 60-80%
- Hypo/Hypertelorism
- Anophthalmia
- Microphthalmia
- Retinal Dysplasia
- Cataract
- Corneal opacities

- Intraocular cartilage
- Microcephaly
- Wide sutures and fontanelles
- Abnormal ears
- Nuchal fold
- Cleft tongue
- Absence of the philtrum
- Micrognathia

Cardio-vascular anomalies 80%

- VSD 50-60 %
- ASD 40-50
- Dextroposition 20-50
- Coarctation
- Anomalous pulmonary venous return
- Overriding aorta
- Pulmonary stenosis
- Hipoplasia aorta
- Mitral atresia
- Aortic Atresia Bicuspid aortic valve

Gastrointestinal anomalies 50-80 %

- Umbilical hernia
- Omphalocele
- Heterotopic pancreas
- Malrotation 20-30 %
- Diaphragmatic hernia
- Elongated gallbladder
- Accessory spleen

Urinary tract anomalies

- Mild pyelectasis

Reproductive tract anomalies 80 %

- Cystic kidneys 40-50 %
- Hydronephrosis 10-20
- Horseshoe kidney
- Multiple renal arteries
- Duplication of the renal pelvis 10-20

Reproductive tract anomalies 50-100 %

- Cryptorchidism
- Hypospadias
- Abnormal scrotum
- Bicornuate uterus 50-80 %

Skeletal System anomalies

- Simian crease
- Clenched fist
- Camptodactyly
- Syndactyly

- Polydactyly
- Club hand with ulnar deviation
- Radial aplasia
- Sandal gap
- Club feet
- Elevation of the big toe 10-50%
- 11 pairs of ribs
- Abnormal iliac wings

Other finding and anomalies

- 2 vessel cord
- Situs inversus
- Growth restriction

References

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