

### Down syndrome (trisomy 21)

Incidence 1:800 (US), the most common chromosomal abnormality

#### Causes

95% trisomy, 4% Robertsonian transloc, 1% mosaicism

#### Neonatal

Severe hypotonia, abnormal facial features

#### Limbs

Short/curved 5th finger, single palmar crease

#### Cardiovascular

ASD (Ost. prim.), VSD, PDA, AV canal defects

#### GIT

Duodenal and anal atresia, Hirschsprung D

#### Other

↓ stature, ↓ cognitive ability, strabismus, Alzheimer's (late)

### Turner syndrome (45,X)

Liveborn incidence 1-2:10,000

Caused by absent Barr body

#### Neonatal

1. Low post. hairline
2. ↑ Elbow carry angle
3. Short 4th metacarpals
4. Widely spaced nipples
5. Aortic coarctation

#### Later complications

1. ↓ Stature
2. Infertility (*SHOX* gene rel.)

USG 2nd trim: Hydrops fetalis, thickened nuchal pad

### Patau & Edwards syndromes (trisomies 13, 18)

Incidence 1:5000 (each), ↑ w/ maternal age

10% from mosaicism+Robertsonian transloc

Most term. when discovered, or stillbirth

Most liveborn die within days-weeks

#### Late complications

1. Cardiac defects (90%)
2. Learning difficulties

### Klinefelter syndrome (47,XXY)

1:1000 live births

#### Common features

1. Tall
2. ↓ Verbal IQ 10-20pts
3. Gynecomastia
4. Azoospermia⇒infert. (ALL patients)
5. Small, soft testes

Rarely: >2 X chromosomes (XXXY, XXXXY)⇒ ↓ ↓ cognitive ability, other features more pronounced

### Triploidy (69,XXX, 69,XXY, 69,XYY)

Common in stillbirth

Rare in live births, rarely live beyond neonatal period

#### Hypomelanosis of Ito

Mosaicism w/ triploid cell line

#### Presentation

1. Pigmented+unpigmented streaks on skin
2. Learning difficulties
3. Hard-to-treat convulsions

