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 convusions