

# Chromosomal aberrations and congenital malformations in the newborn

Věra Hořínová, Michael Doubek

# Learning outcomes

- The student knows clinical manifestations of rare chromosomal aberrations
- The student knows clinical manifestations of congenital malformations in the newborn

# Lecture content

- Videos of patients with chromosomal aberrations
- Videos of patients with congenital malformations

# Chromosomal aberrations

# Down syndrome; 47,XY,+21; 47,XX,+21



# Down syndrome; 47,XY,+21; 47,XX,+21



# Patau syndrome; 47,XY,+13; 47,XX,+13



# Patau syndrome; 47,XY,+13; 47,XX,+13





# Edwards syndrome; 47,XY,+18; 47,XX,+18



# Edwards syndrome; 47,XY,+18; 47,XX,+18



# Cri du chat syndrome; 46,XY,del(5p)



# Wolf-Hirshor syndrome; 46,XX,del(4p)



# Turner syndrome; 45,X0



# Klinefelter syndrome; 47,XXY



# Beckwith–Wiedemann syndrome



# Congenital malformations



# Goldenhar syndrome



# Congenital hydrocephalus, CMV infection



# Fetal alcohol spectrum syndrome



# Amniotic lane syndrome



# Amniotic lane syndrome



# Encephalocele



# Neural tube cleft



# VACTERL syndrome





# VACTERL syndrome



# Hydrops fetalis



# Scrotal hypoplasia



# Holoprosencephaly



# Craniosynostosis, cloverleaf skull



# Take home message

- Chromosomal aberrations and congenital malformations are a wide range of abnormalities
- Many of the chromosomal aberrations are characterized by typical phenotypic changes, which should be an indication for genetic testing

MUNI  
MED