Edward's syndrome

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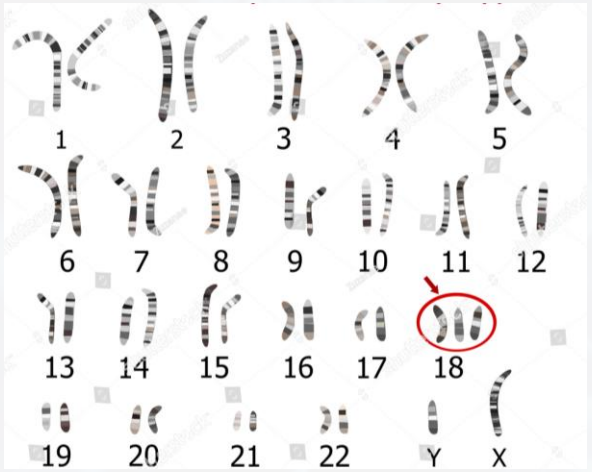
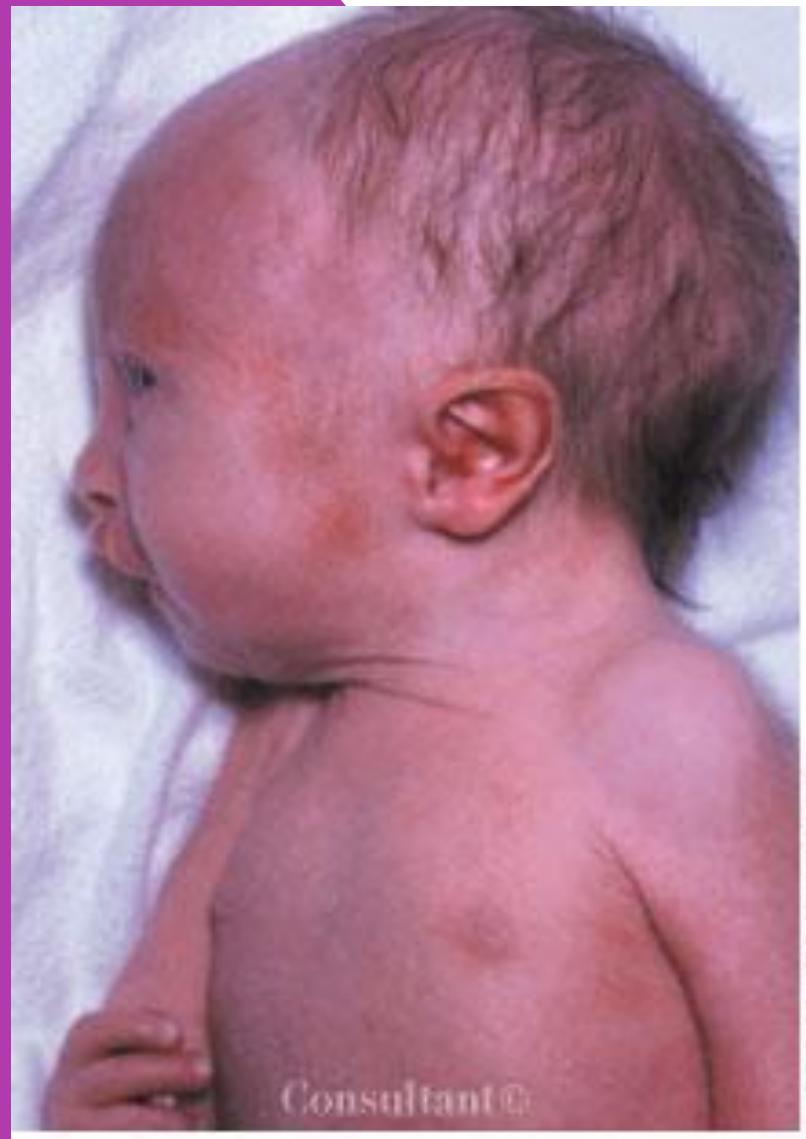


Generally:

- Trisomy 18 (Edwards syndrome)
- chromosomal condition, affects multiple parts of the body
- intrauterine growth retardation & low birth weight
- heart defects and abnormalities of other organs
- small, abnormally shaped head with a small jaw and mouth
- clenched fists with overlapping fingers
- Lifespan rarely lasts 1 month after birth (only 5-10 percent of affected children live past their first year while often having severe intellectual disability)



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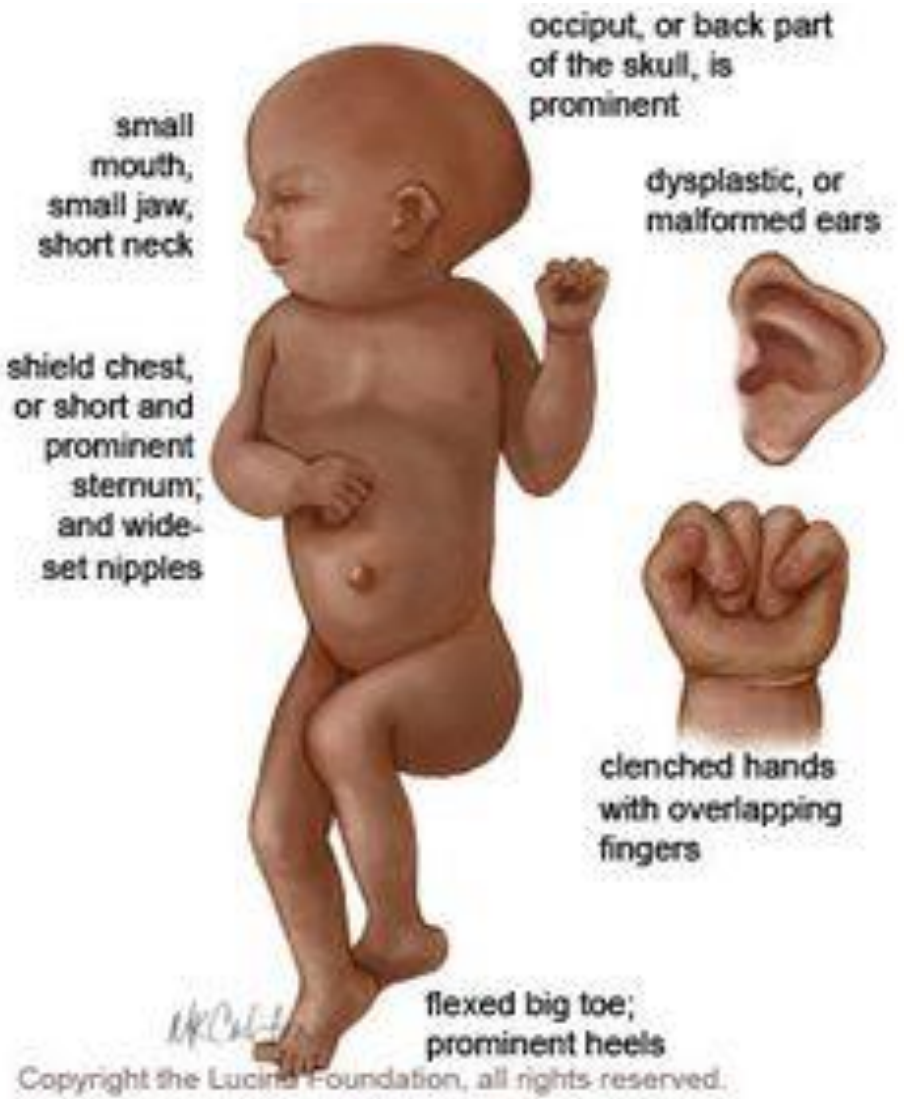

Incidence

- Occurs in about 1 in 5,000 live-born infants
- Around 80% of those affected are female
- Many affected fetuses do not survive to term
- Women of all ages can have a child with trisomy 18
- As the woman gets older, the possibility of having a child with this condition increases

Types

- 95% of children with Edward's syndrome have full-trisomies
- 2% are due to trans-locations → only a portion of an extra chromosome is present (partial Edward's syndrome)
  - The extent of impairment of the baby depends on which part of chromosome 18 is present in their cells
- 3% of children with Edward's syndrome have 'mosaic trisomies' (extra chromosome is not in every one of the child's cells)
  - Usually milder expression, depending on amount and type of cells affected
  - With this type of Edward's syndrome, the life span can even reach up to adulthood in some cases

Symptoms

The majority of children appear fragile and weak; many are underweight

Their heads are unusually small, while the backs of their heads are prominent. Their ears are low-set and malformed, and their mouths and jaws are small, a condition referred to as, 'micrognathia.' Babies with the syndrome may experience a cleft palate or lip. Their hands are often clenched into fists, with their index finger overlapping their other fingers. Babies with Edward's syndrome can have clubfeet, as well as toes that may be fused or webbed.

Children with the syndrome can experience problems with their lungs and diaphragm, and blood vessels which are malformed. They may present with a number of types of congenital heart disease, to include atrial septic defect, ventricular septal defect, or patent ductus arteriosus. Children with the syndrome might have an inguinal or umbilical hernia, abnormalities of their urogenital

Screening for Edward's syndrome

- Women are offered screening for Edwards' syndrome between 10 & 14 weeks of pregnancy
- This screening test is a combined test and it works out the chance of a baby having Edwards' syndrome, Down's syndrome and Patau's syndrome (it includes a blood test and an ultrasound scan to assess nuchal translucency)
- Identification of the type of Edward's syndrome is not possible here

Diagnosing of Trisomy 18 during pregnancy

- If the combined test indicates a relatively higher chance of the baby having Edward's syndrome, more diagnostic (and invasive) tests are conducted to check for an extra copy of chromsome 18:
  - Chorionic villus sampling (sample from placenta)
  - Amniocentesis (sample of amniotic fluid)

Treatment of Edward's syndrome

- There is no cure, only management of the symptoms
  - Heart conditions
  - Breathing difficulties (apnea, pulmonary hypertension)
  - Infections (pneumonias, sinusitis,ear/ eye infections, UTIs)
- Surgical interventions might be limited due to the child's cardiac health
- Palliative care mostly
- Feeding difficulties
  - Use of a nasogastric tube may be necessary
  - Constipation due to poor abd ominal muscle tone

Prognosis

- The majority do not live past their first year of life
- Average lifespan for half of the children born with this syndrome is less than two months
- Children who do survive their first year experience severe developmental disabilities, require walking support and their ability to learn is limited
- Their verbal communication abilities are limited although they are able to respond to comforting and have the ability to learn to smile, recognize and interact with caregivers and others
- They can acquire skills such as self-feeding and rolling over

Sources

- https://www.nhs.uk/conditions/edwards-syndrome/
- https://www.youtube.com/watch?v=EFP671hzcil&vl=en
- https://ghr.nlm.nih.gov/condition/trisomy-18
- https://www.disabled-world.com/disability/types/edwards-syndrome.php
- Pathoma
- https://edwardssyndromeinfo.weebly.com/birth-defects-and-characteristics.html

Děkuji za pozornost!