

A newborn girl is examined in the delivery room immediately after induced vaginal delivery for intrauterine growth restriction (IUGR). She was born to a 40-year-old woman with no significant medical history. In addition to IUGR, prenatal ultrasound demonstrates a ventricular septal defect. Her parents decline additional prenatal testing. The patient's weight and head circumference are <5th percentile. Examination shows hypertonia and closed fists, with the second digit overlapping the third and the fifth overlapping the fourth. Based on this patient's presumed chromosomal abnormality, which of the following additional physical findings is most likely to be present on examination?

- ☐ A. Cat-like cry
- ☐ B. Cutis aplasia
- ☐ C. Micrognathia
- ☐ D. Microphthalmia
- ☐ E. Transverse palmar crease
- ☐ F. Webbed neck

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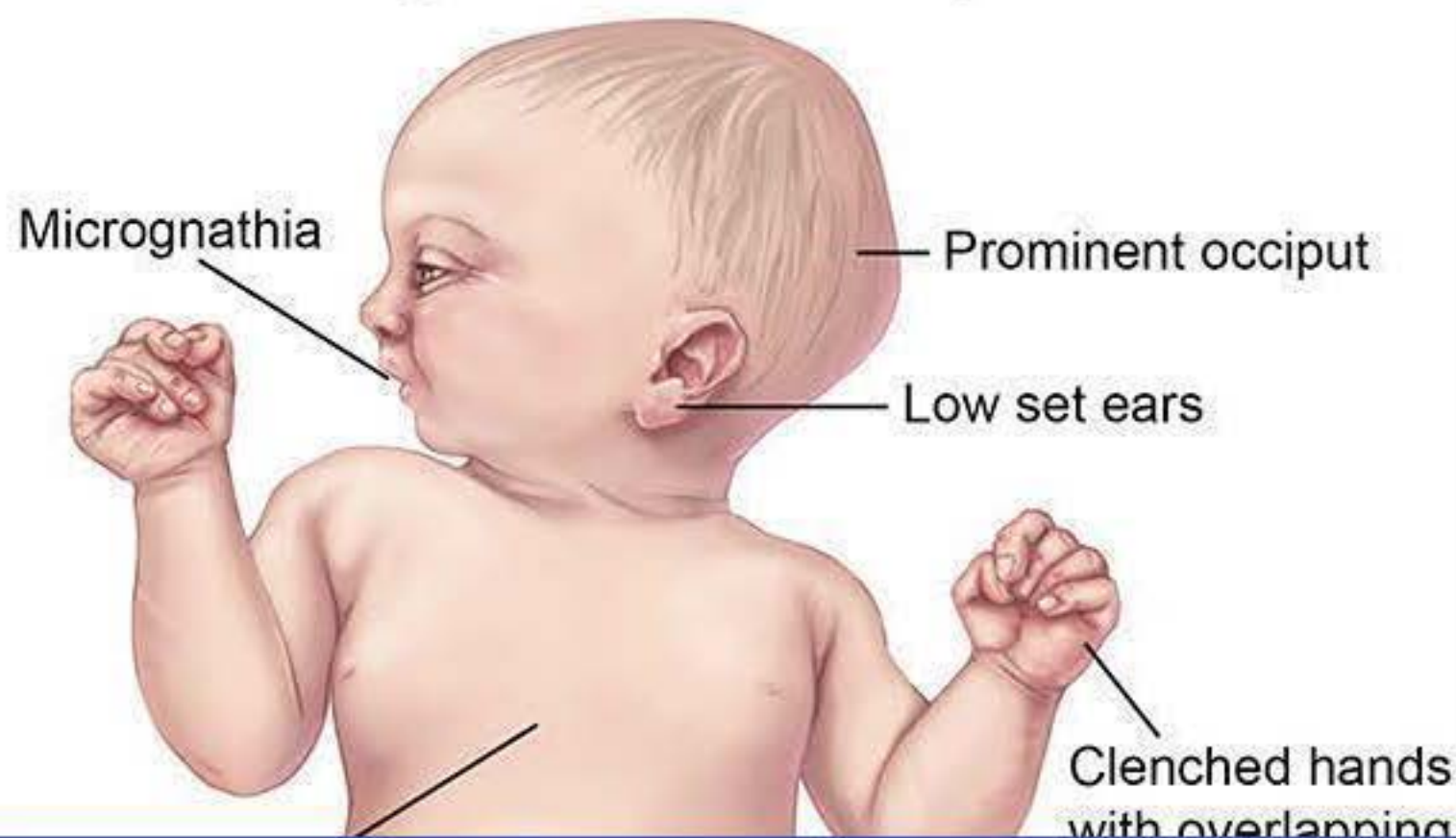
- ☐ A. Cat-like cry [9%]
- ☐ B. Cutis aplasia [7%]
- ☒ C. **Micrognathia** [65%]
- ☐ D. Microphthalmia [8%]
- ☐ E. Transverse palmar crease [10%]
- ☐ F. Webbed neck [1%]

[Proceed to Next Item](#)

Explanation:

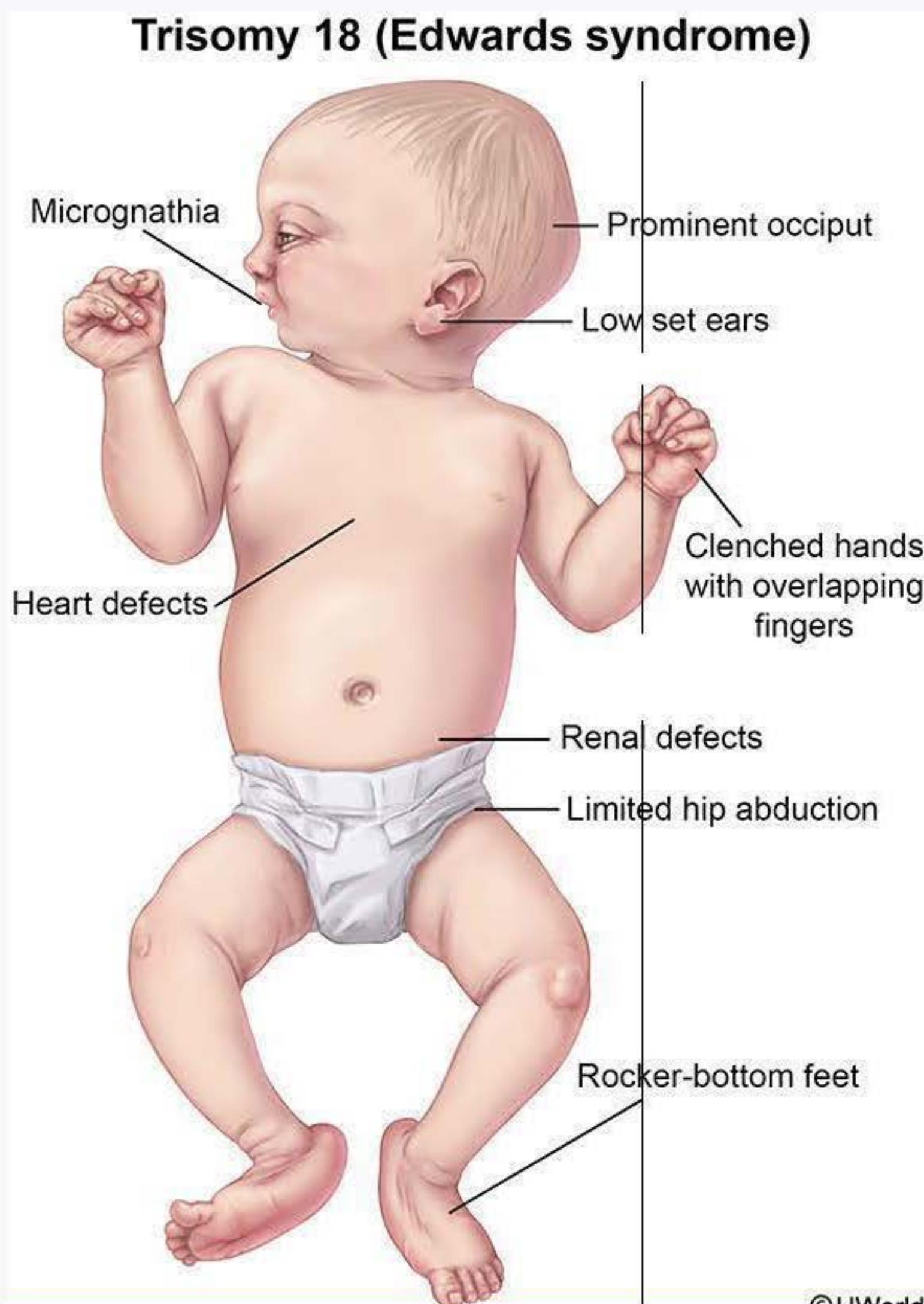
User Id: [REDACTED]

Trisomy 18 (Edwards syndrome)



Explanation:

User Id: [REDACTED]



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This newborn's low birth weight due to **intrauterine growth restriction**, microcephaly, ventricular septal defect, and **closed fists with overlapping fingers** are all classic physical examination features seen in **trisomy 18 (Edwards syndrome)**. Other findings include **micrognathia**, **prominent occiput**, **rocker-bottom feet**, and severe intellectual disability.

After trisomy 21, trisomy 18 is the second most common autosomal trisomy observed in live births. The risk of trisomies increases with maternal age due to meiotic nondisjunction within maternal oocytes. Diagnosis is suspected based on prenatal ultrasonography, and karyotype (prenatal or postnatal) confirms the diagnosis. Approximately 95% of trisomy 18 patients die during their first year of life, most commonly due to cardiac failure from congenital heart disease or respiratory failure from hypoventilation or aspiration.

(Choice A) A cat-like cry is seen in cri-du-chat (5p deletion) syndrome. Infants with this syndrome may also have microcephaly; however, a characteristic protruding metopic suture is present. Other manifestations are hypotonia, short stature, hypertelorism, wide and flat nasal bridge, and intellectual disability.

(Choices B and D) Cutis aplasia (absence of epidermis over the skull) and microphthalmia are both classically seen in **trisomy 13 (Patau syndrome)**. This condition also associated with other midline defects, including holoprosencephaly and omphalocele. Closed fists with overlapping fingers are not seen.

(Choice E) A transverse palmar crease can be a normal variant but is also strongly associated with **trisomy 21 (Down syndrome)**. Infants with Down syndrome also have hypotonia, upward and slanted palpebral fissures, epicanthal folds, Brushfield spots, cardiac malformations, and intestinal atresia.

(Choice F) A webbed neck is a classic feature of **Turner syndrome (45,XO)**. Other features include a low hairline, broad chest with widely spaced nipples, cubitus valgus, and short stature.

Educational objective:

Infants with Edwards syndrome (trisomy 18) commonly have microcephaly, prominent occiput, intrauterine growth restriction, and micrognathia, as well as closed fists with overlapping digits and rocker-bottom feet. The mortality rate in the first year of life approaches 95%.

References:

1. **Trisomy 18: review of the clinical, etiologic, prognostic, and ethical**

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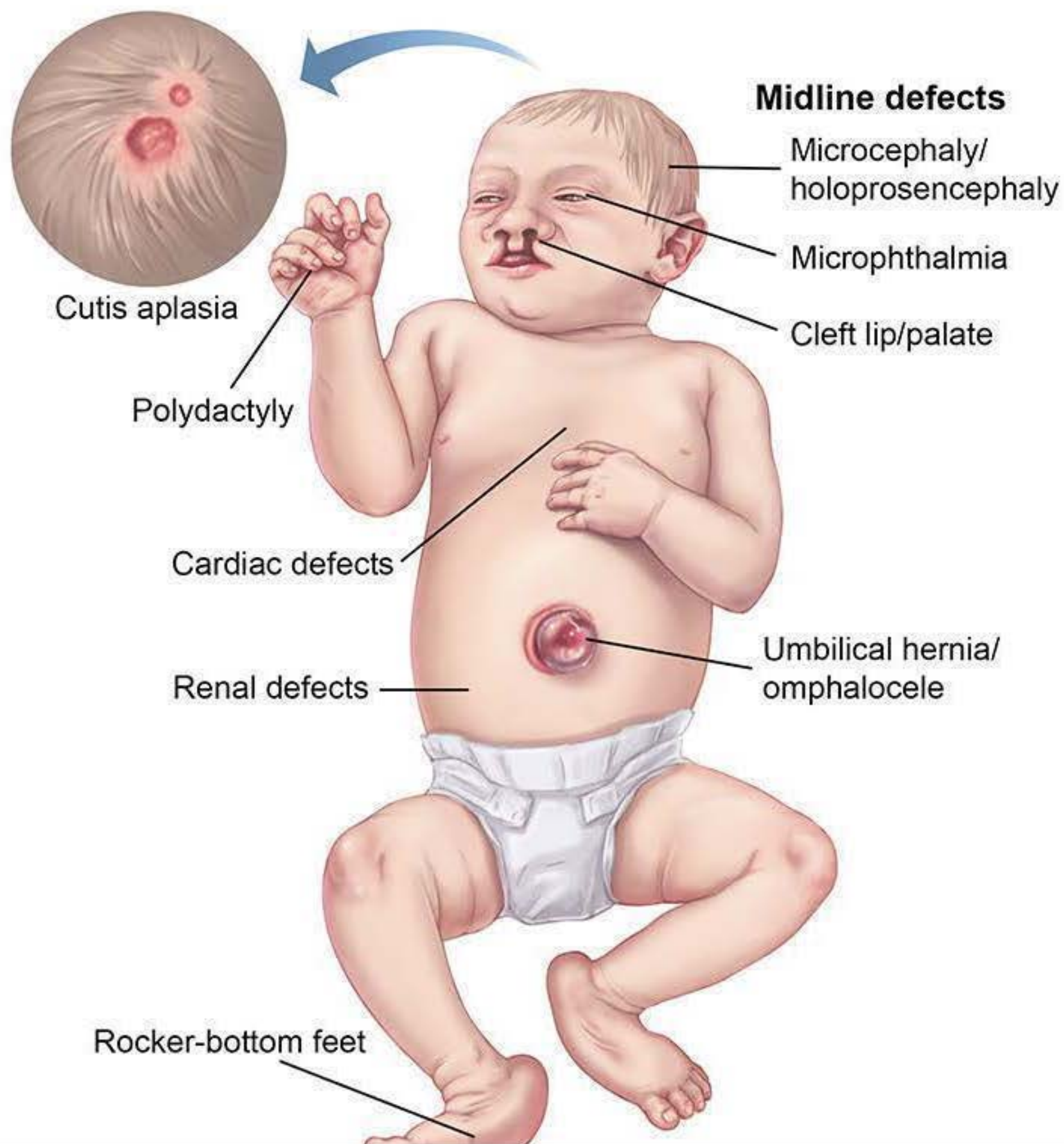
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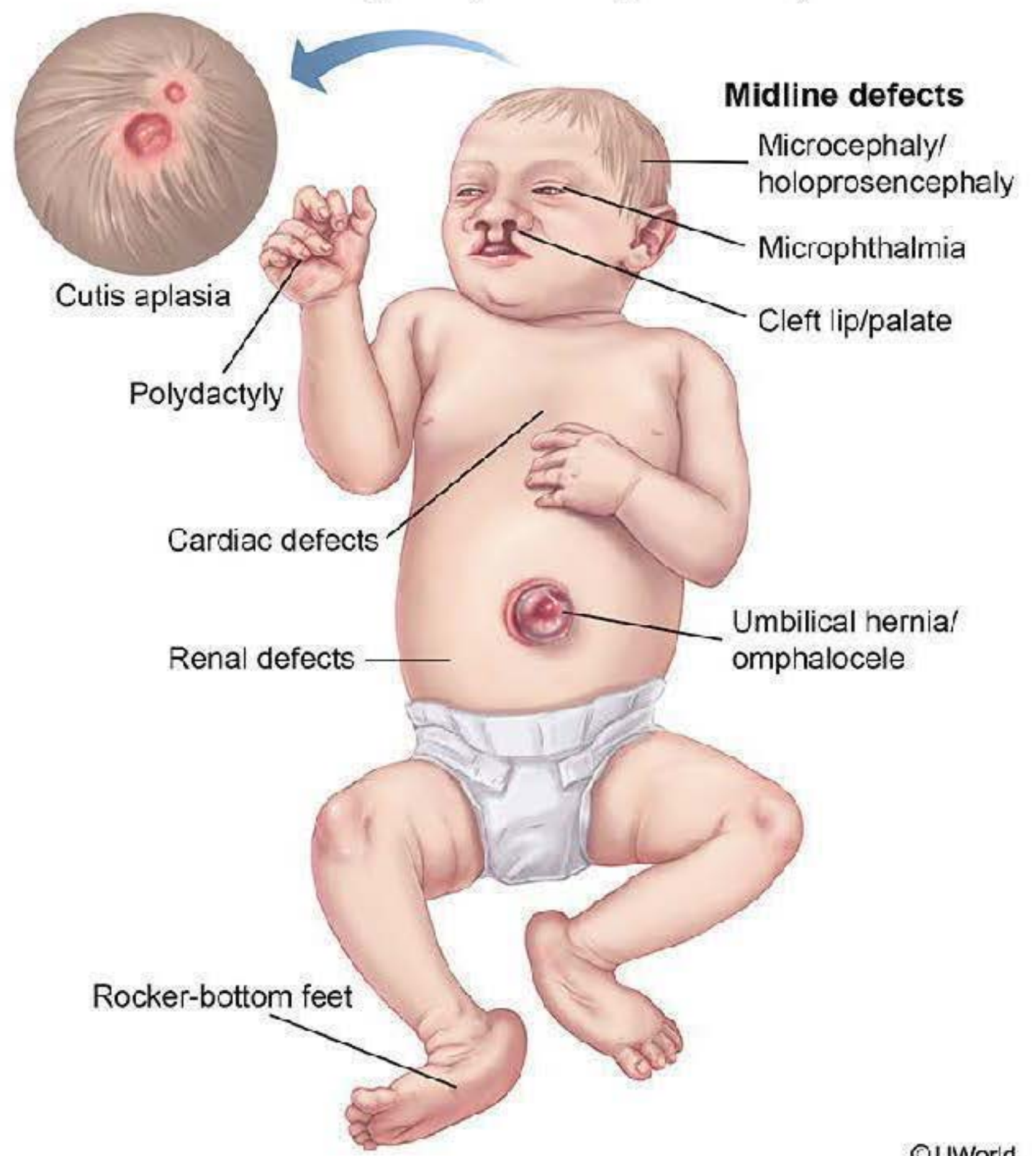
References:

1. **Trisomy 18: review of the clinical, etiologic, prognostic, and ethical aspects.**
2. **Anatomy of trisomy 18.**

Trisomy 13 (Patau syndrome)



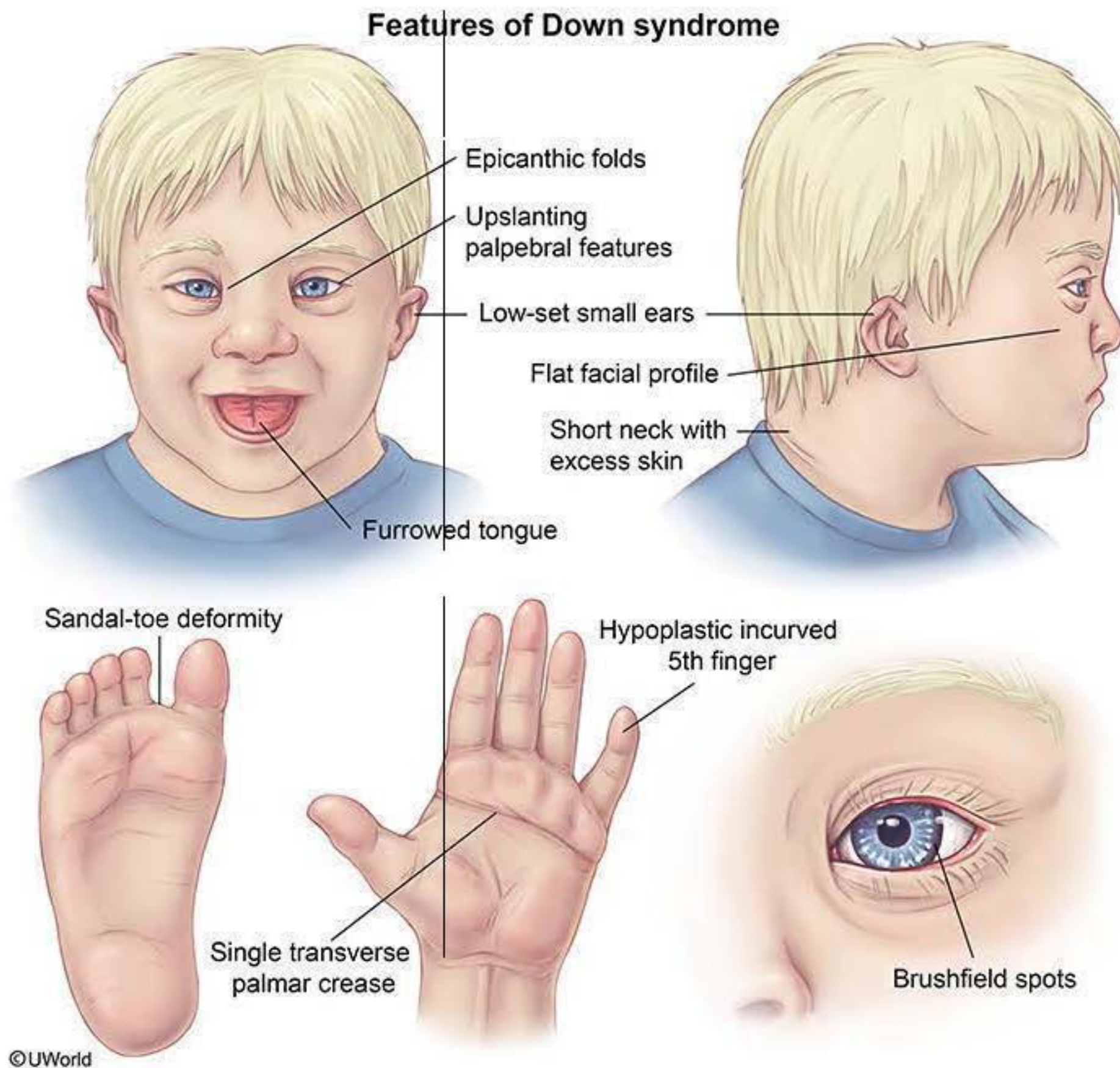
Trisomy 13 (Patau syndrome)



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Media Exhibit

phic features of down syndrome



Media Exhibit

syndrome

