Trisomy ١٨ (Edward syndrome)

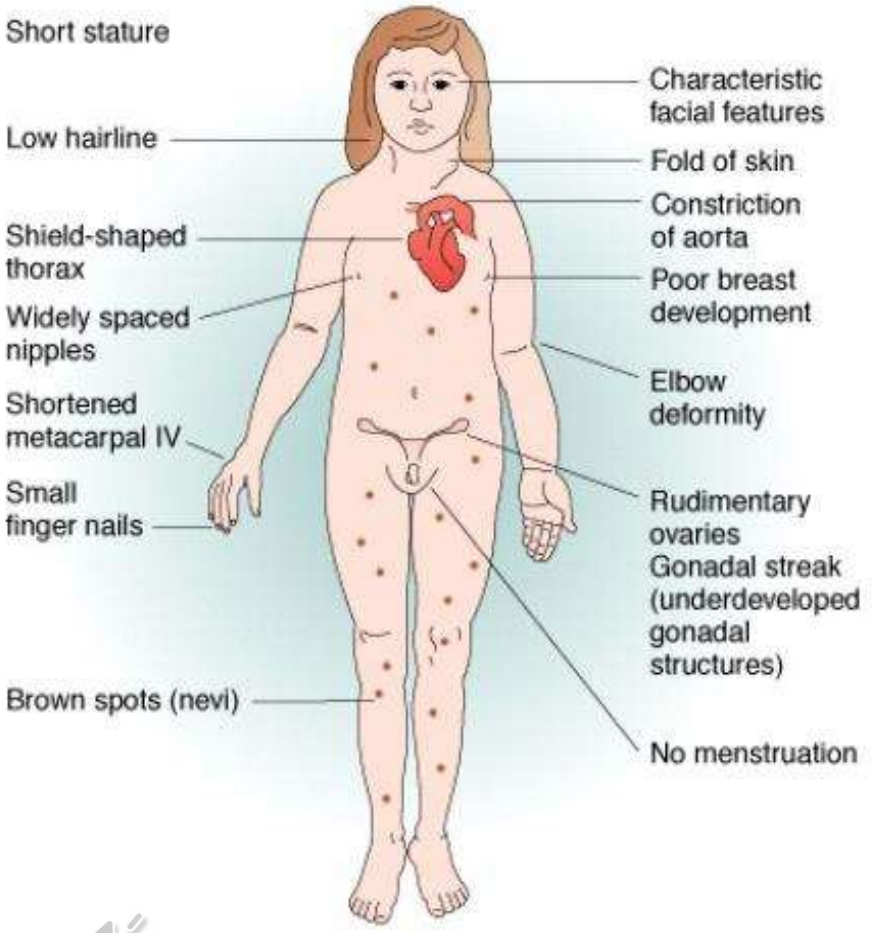
- Fewer than ١٠% reach her first birthday
- Small for gestational age
- Hypertonia
- Rocker bottom feet
- Prominent occiput

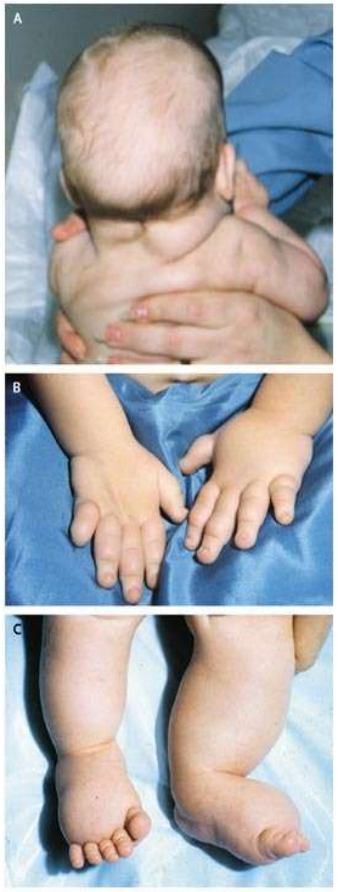
Trisomy ١٣ (Patau syndrome)

- Usually fatal before the age of ١
- Small for gestational age
- Aplasia cutis of scalp
- Microcephaly
- Cleft lip and palate

Turner syndrome

- ۴۵XO, female with normal intelligence and life expectancy
- Relatively mild phenotype
- **Short stature** is a cardinal feature
- ۴۵% **cardiac involvement** (coarctation of the aorta, bicuspid aortic valve, aortic aneurysm)
- ۵۰% **renal anomalies** (horseshoe kidney)
- ۵ fold risk of **acquired hypothyroidism**
- **Gonadal dysgenesis** and amenorrhea

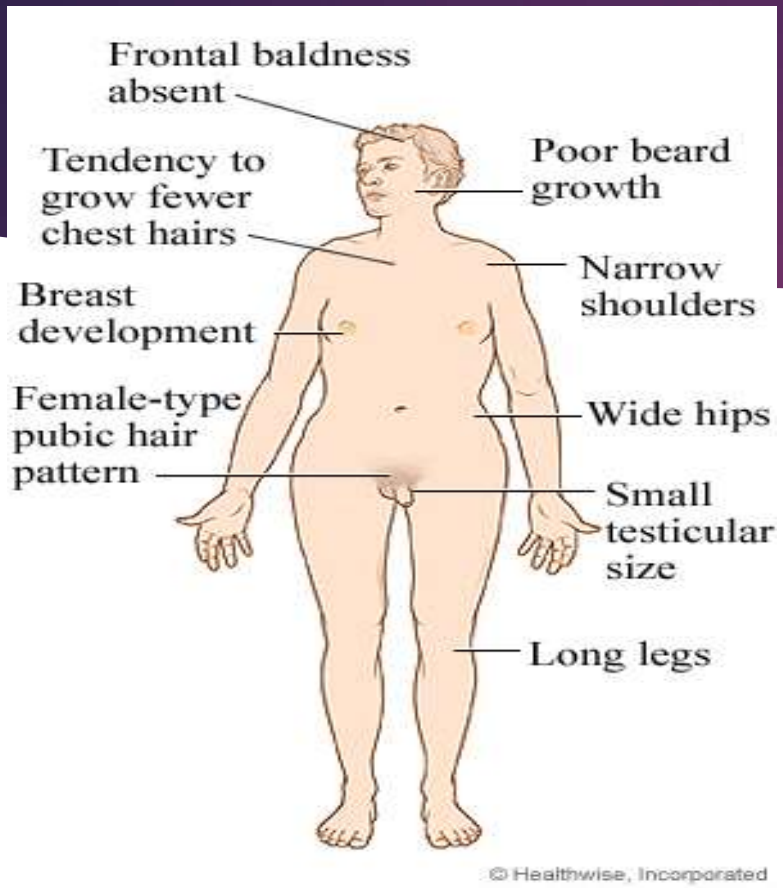

Short stature
Low hairline
Shield-shaped thorax
Widely spaced nipples
Shortened metacarpal IV
Small finger nails
Brown spots (nevi)
Characteristic facial features
Fold of skin
Constriction of aorta
Poor breast development
Elbow deformity
Rudimentary ovaries
Gonadal streak (underdeveloped gonadal structures)
No menstruation


A
B
C


Though she be but
LITTLE
she is
FIERCE

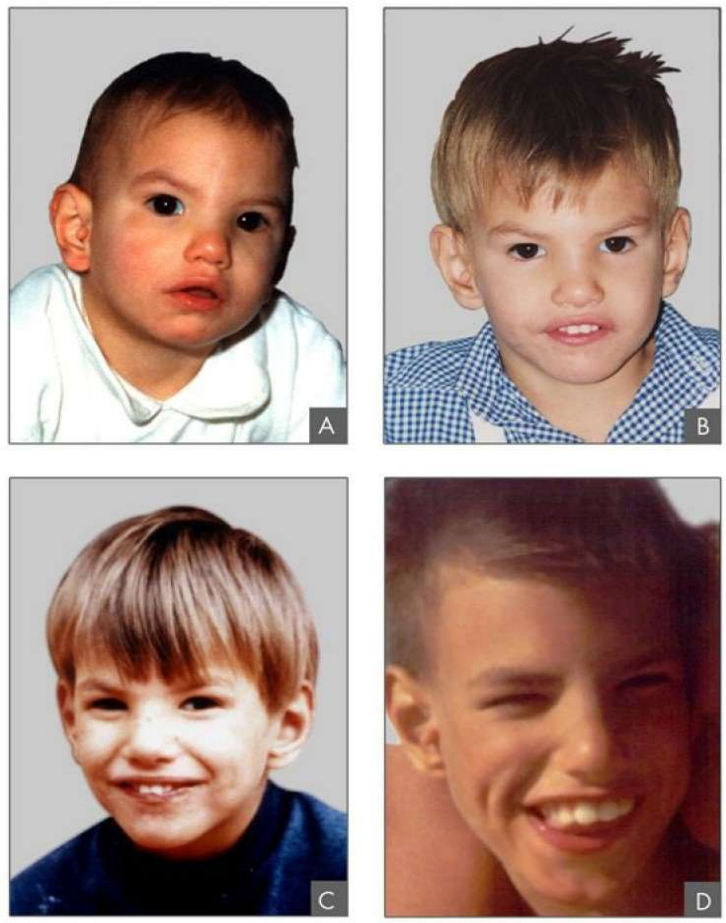
Klinefelter syndrome

- The most common cause of **infertility in men**
- ۴۷XXY, diagnosis at the age of adolescence
- Tall stature, long limbs, infantile testes, gynecomastia
- Failure of secondary sexual characteristics
- Osteopenia and osteoporosis


Frontal baldness absent
Tendency to grow fewer chest hairs
Breast development
Female-type pubic hair pattern
Poor beard growth
Narrow shoulders
Wide hips
Small testicular size
Long legs
© Healthwise, Incorporated

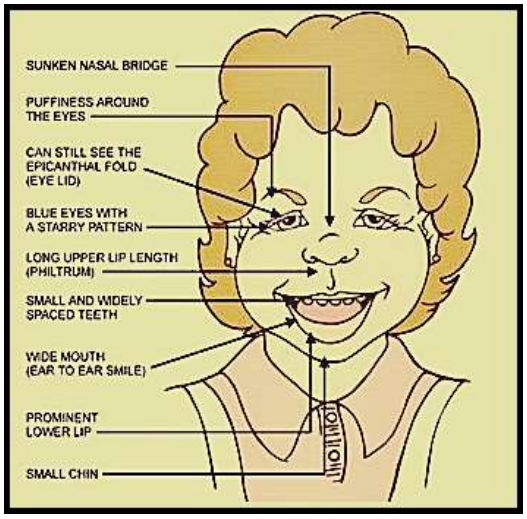
Cri du chat syndrome

- Deletion of short arm of chromosome ٥
- Catlike cry in infancy
- Low birth weight
- Failure to thrive
- Microcephaly
- Developmental delay
- Cardiac involvement



William syndrome

- Deletion of long arm of chromosome ٧
- ٨٠% **cardiac involvement** (supravalvular aortic and pulmonic stenosis)
- ١٠% **autistic**, moderate mental retardation
- Striking personality
- Growth delay and short stature
- ٢٠% **hypercalcemia**

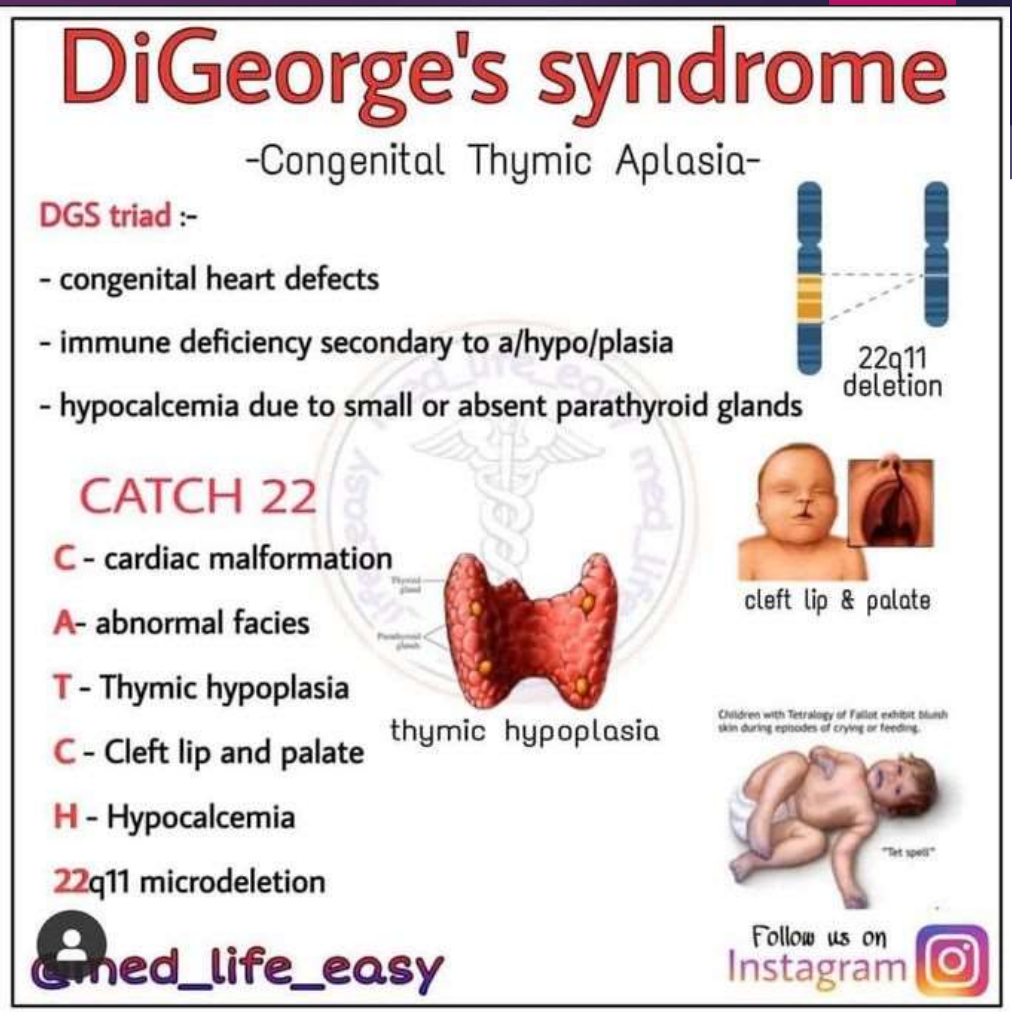


Aniridia Wilms tumor association

- Deletion of short arm of chromosome ١١
- WAGR syndrome (Wilms tumor, Aniridia, Genitourinary anomalies, Mental Retardation)
- ٥٠% microcephaly
- Cryptorchidism and hypospadias
- Short stature
- ٥٠% **Wilms tumor**

DiGeorge syndrome

- Deletion of long arm of chromosome ٢٢
- **Cardiac involvement** (TOF, VSD, truncus arteriosus, right sided aortic arch)
- Mild mental retardation



Approach to the dysmorphic child

- Detailed **history taking** and **physical examination**
- Request **some imaging** and **genetic tests**
- Refer to clinical **genetic specialist**
- Offer an **explanation to the family** why they child have a congenital anomaly, **reduce their guilt feeling**
- **Anticipation of medical problems** associated with a particular syndrome
- Identify the **risk of another pregnancy** and plan tests

Reference

- Nelson essentials of Pediatrics, Eight edition, Section ٩, Human genetics and dysmorphology