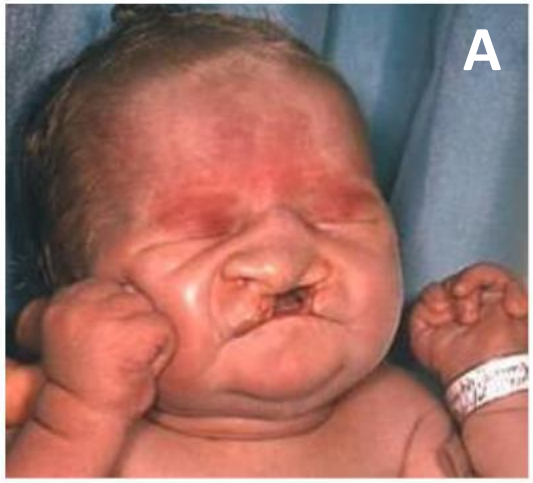
Part 2: Describe the Dysmorphism

1. Describe the dysmorphism in all cases
2. In case 13-17 state who is affected.
3. You don't have to suggest a diagnosis unless you really want to ☺

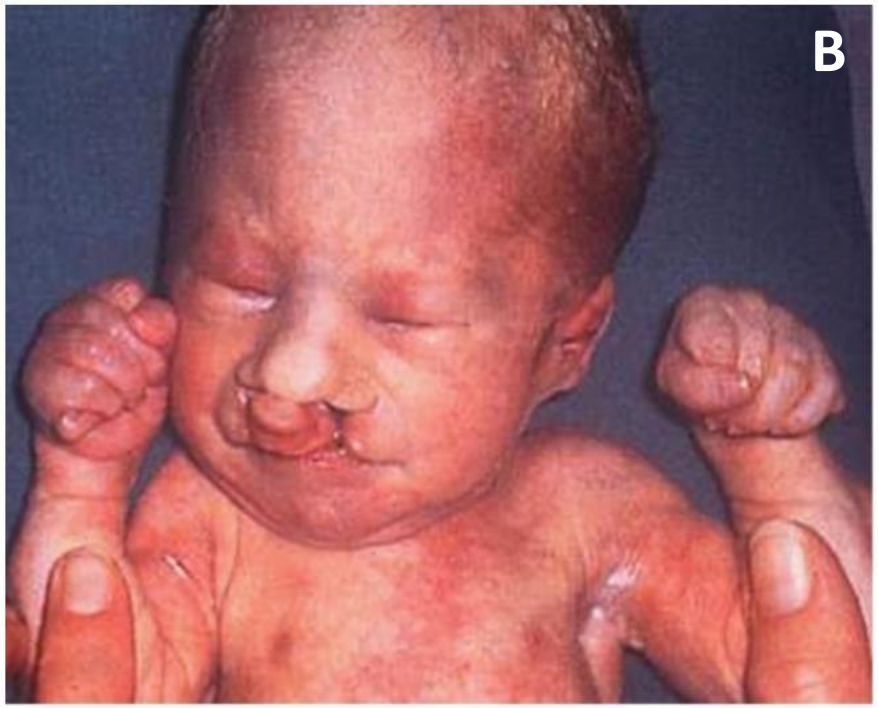
40 minutes

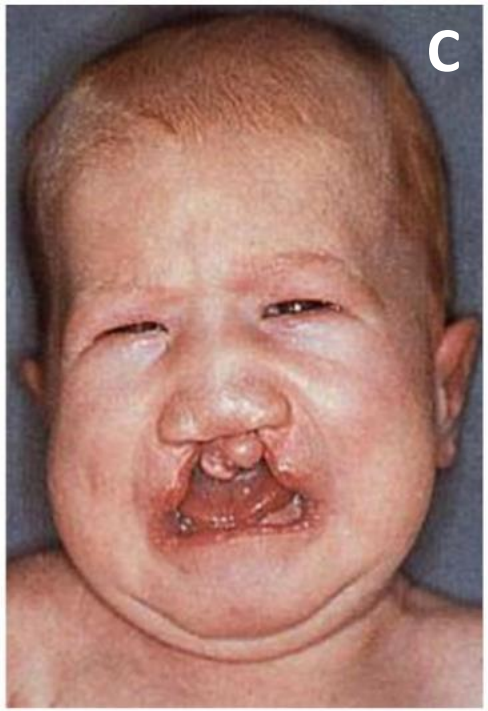
Snir Boniel, MD

Case 1

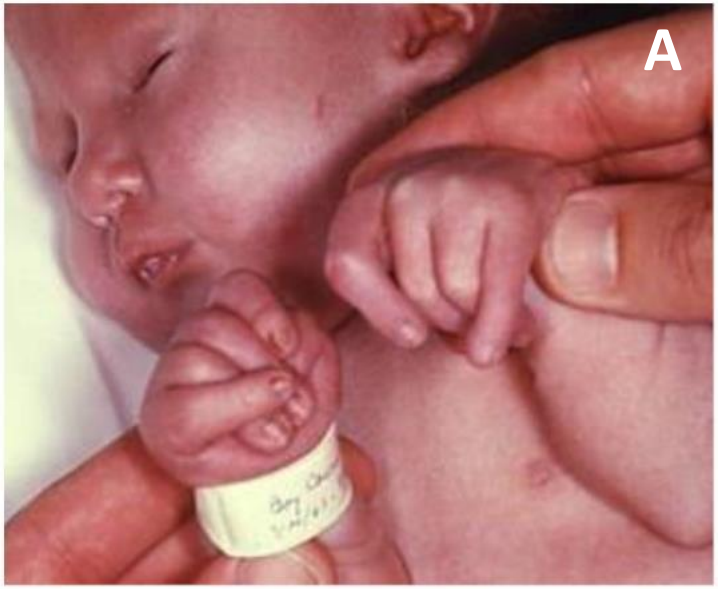


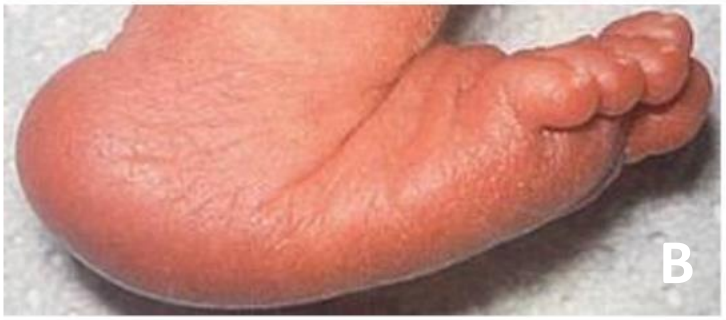
- Cleft palate (unilateral or bilateral?)
- Nasal anomaly
- Polydactyly / hexadactyly.
- *Patau syndrome (Trisomy 13)*





Case 2

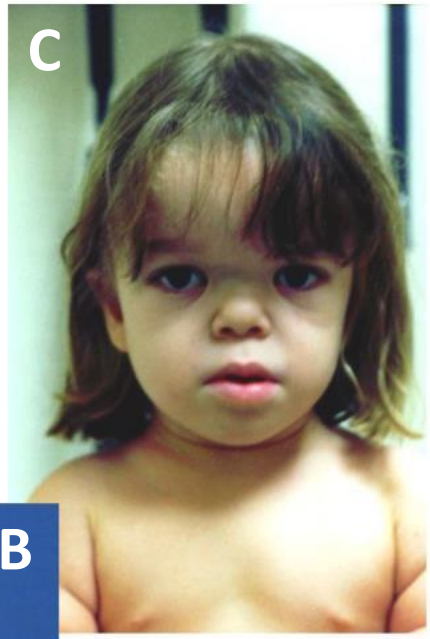


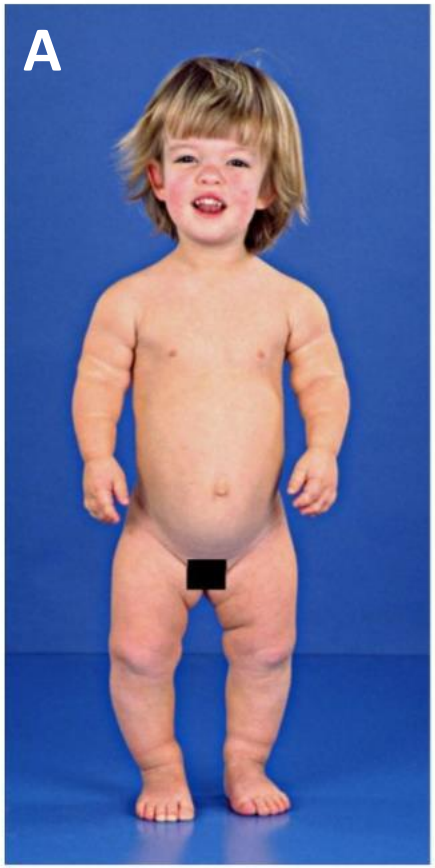


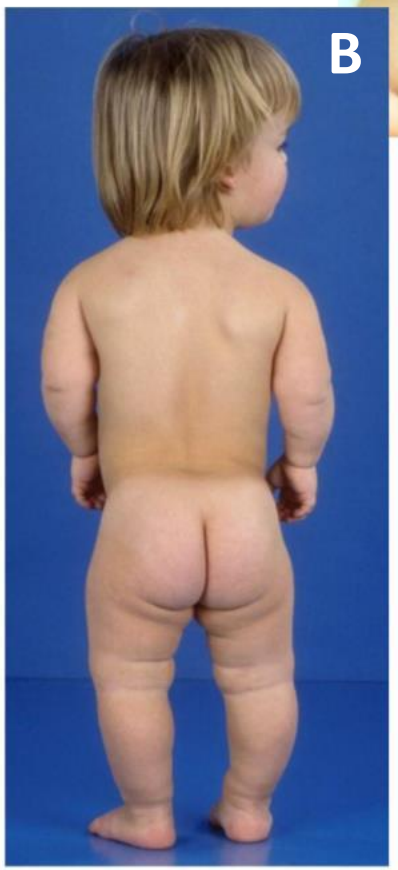
- Club foot?
- No…the opposite. Simply foot dorsiflexion.
- Arthrogryposis – fingers 2&5 cross over 3&4.
- Palpebral fissure deformity
- *Edwards Syndrome (Trisomy 18)*

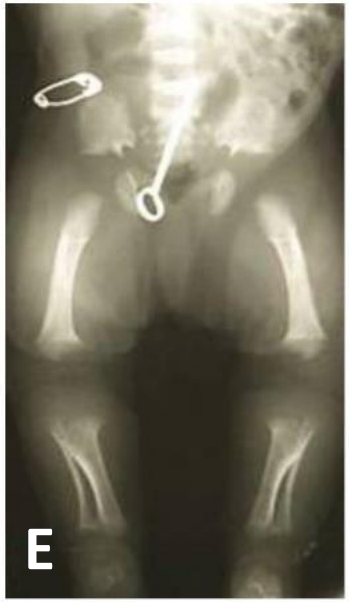
Case 3

- Disproportionate short stature. Skin folds.
- Proximally short bones = genu varum
- Large protruding forehead? Bangs?
- *Achondroplasia*



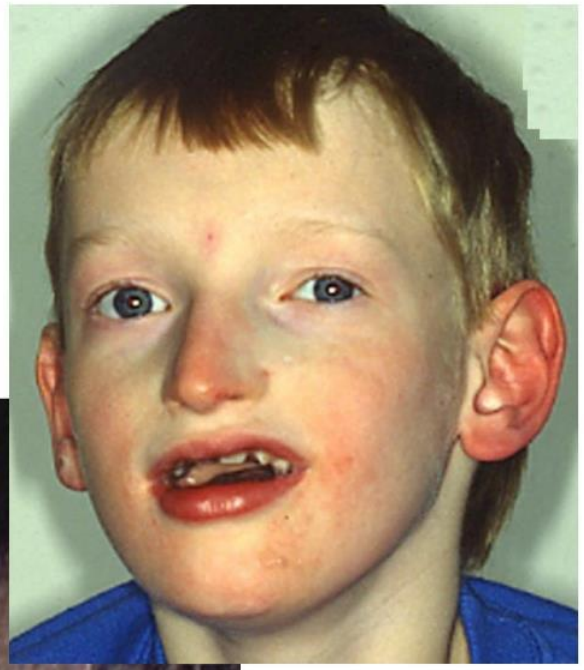


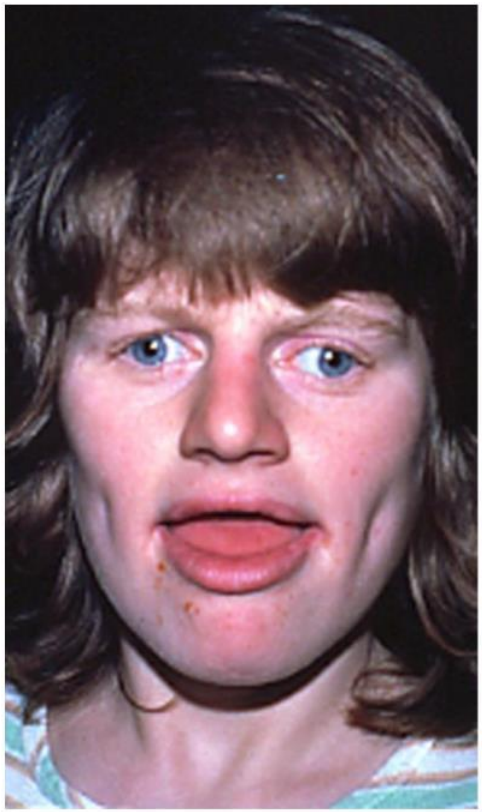


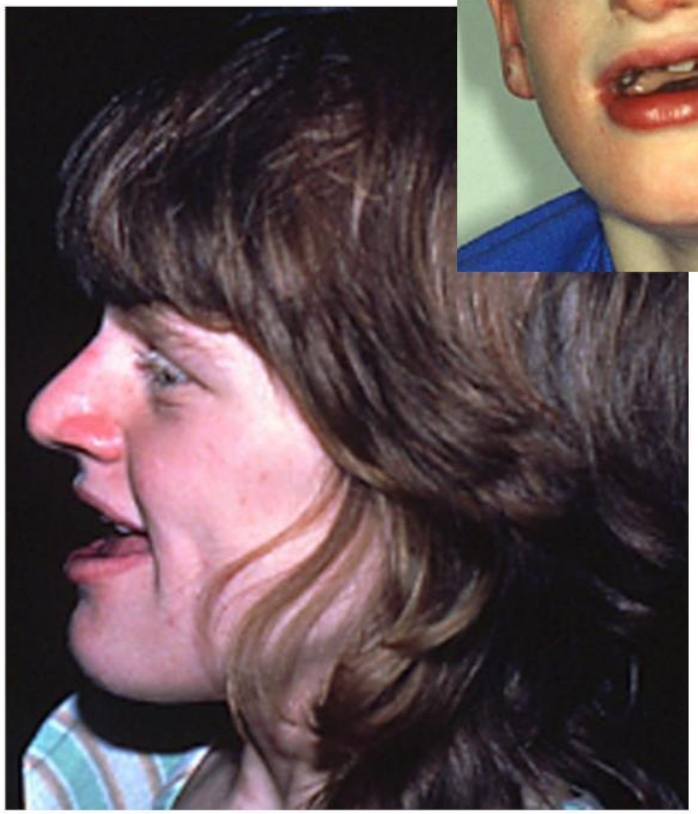


Case 4

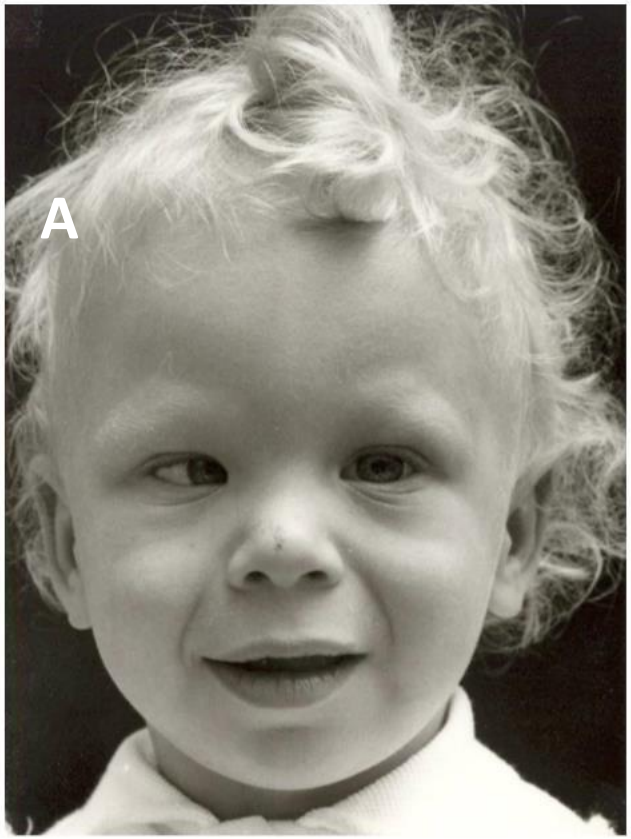
- Prominent nasal ridge
- Large lower jaw, open mouth.
- Prominet dimples – facial hypotonia
- *Angelman syndrome* –
    - behavioral phenotype, no consistent dysmorphism.

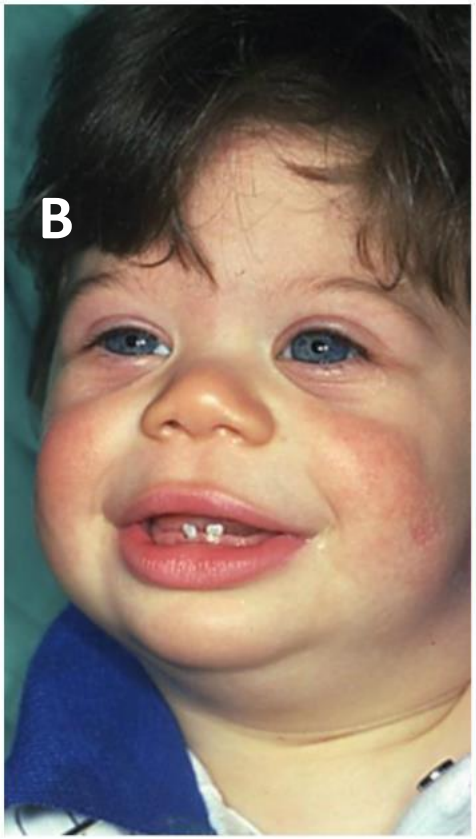


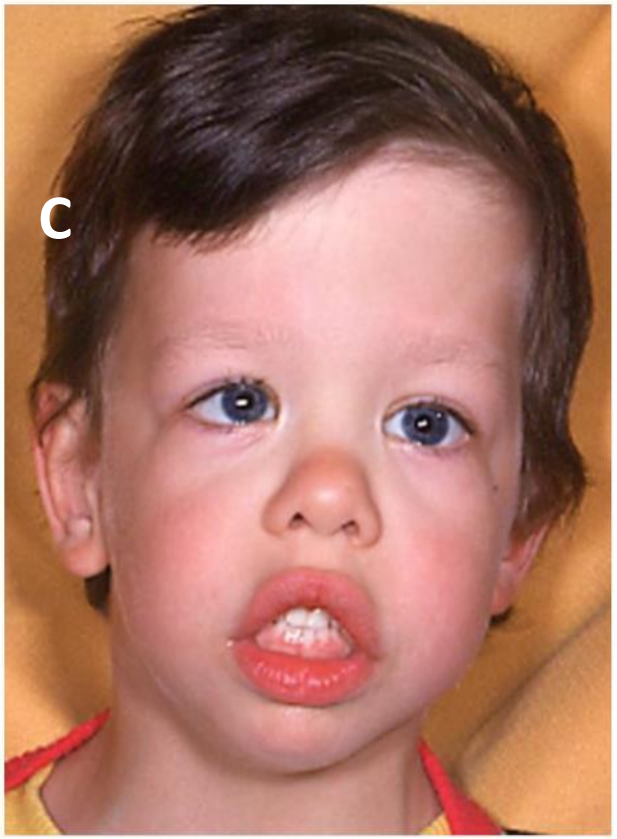




Case 5





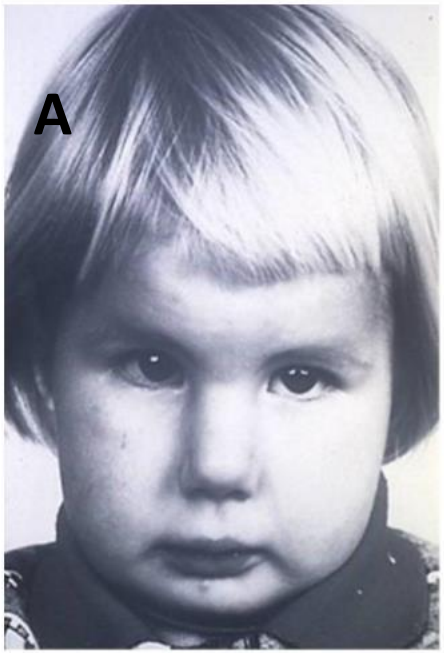


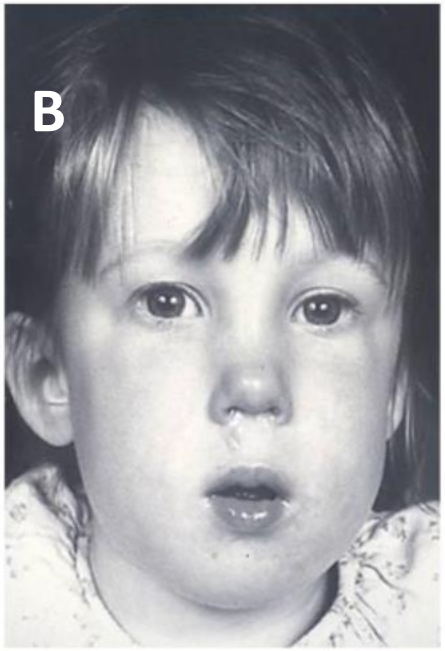
- Common features between A, B and C? Round face – excess subcutaneous tissue.
- External supraorbital area looks the same
- Thick lips
- Facial feature hypomimia
- Triangular nose
- Elfin face
- Long/misshapen palpebral features
- *Williams syndrome* – connective tissue problems (aorta).

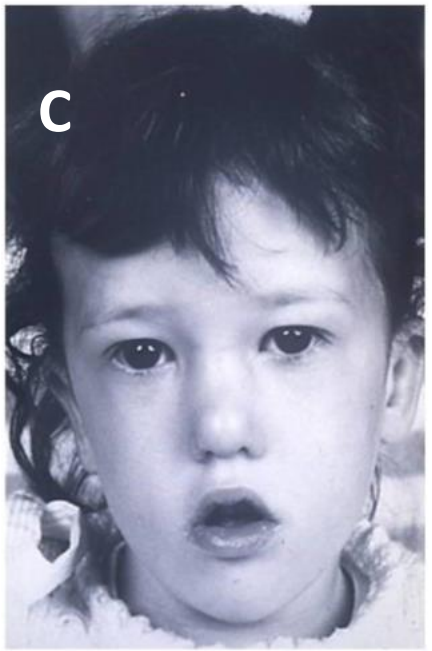
Case 6

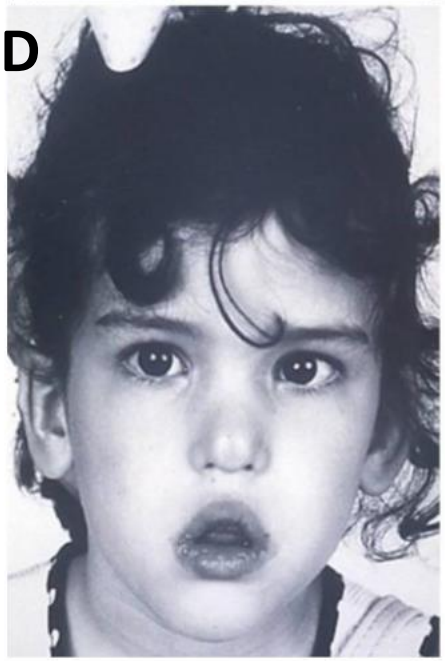
Small mouth
Tubular nose
Lots of neck tissue
Hypotonia
DiGeorge Syndrome

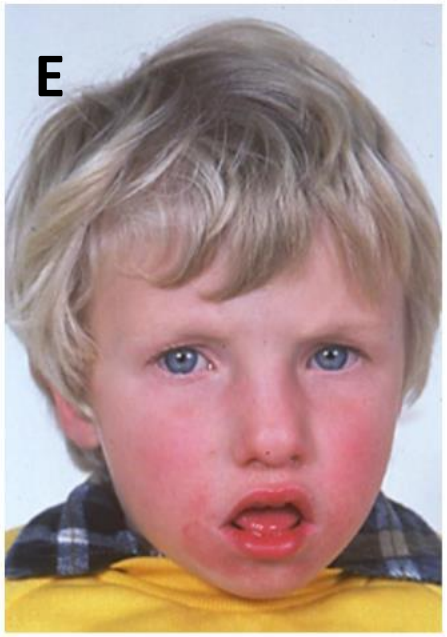
- **CATCH-22**
- **C**ardiac anomaly:
  Interrupted aortic arch, truncus arteriosus, ToF
- **A**bnormal facies
- **T**hymic aplasia
- **C**left palate
- **H**ypocalcemia
- **22**q11del

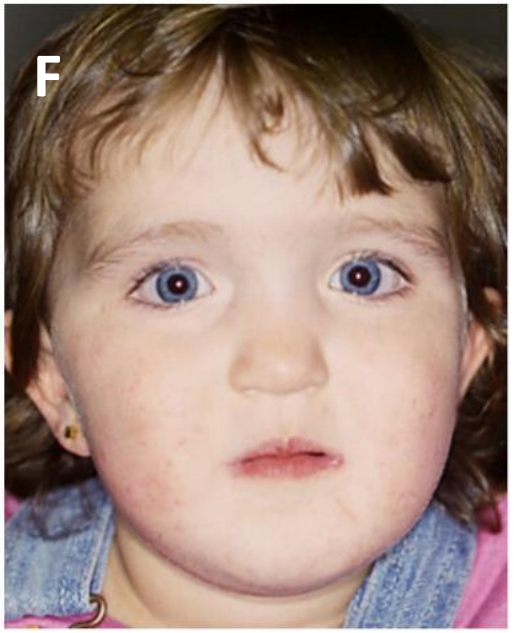




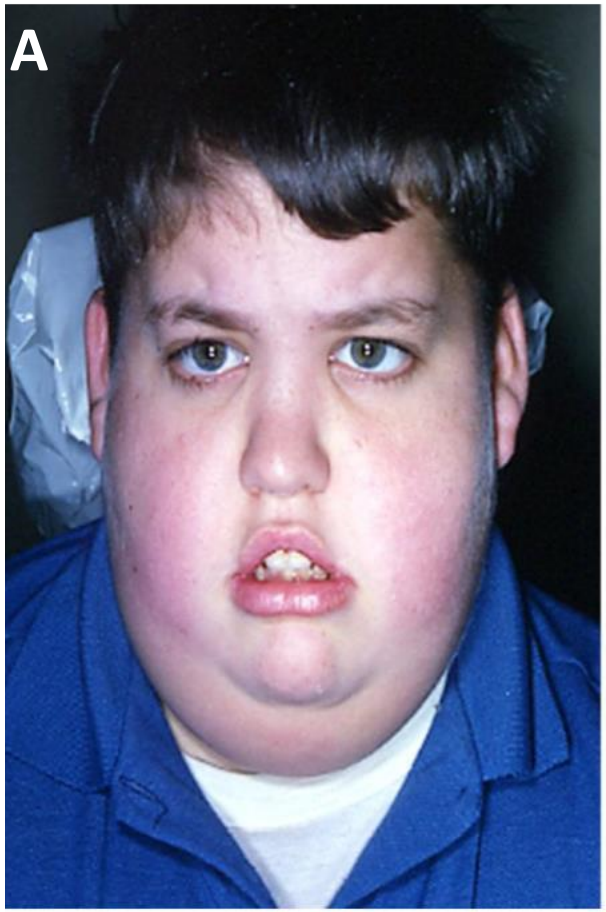


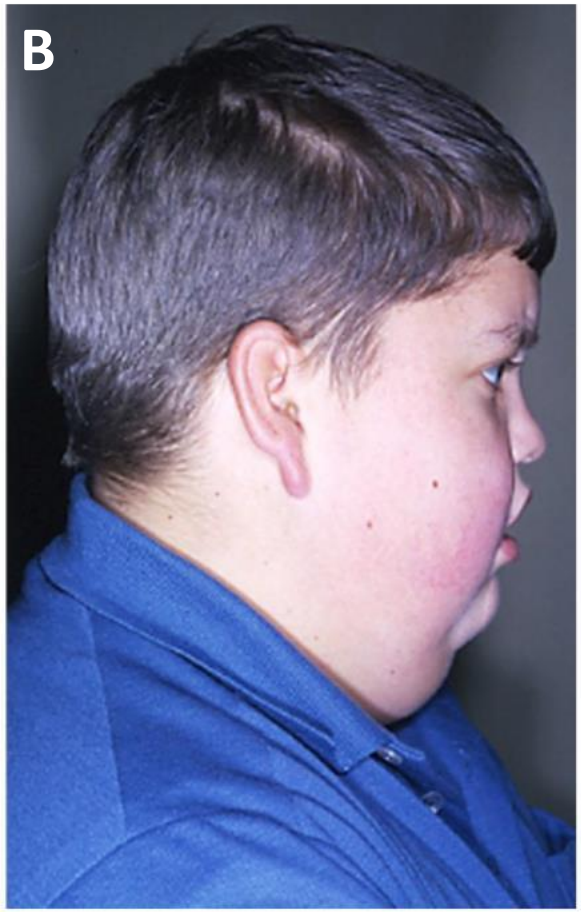


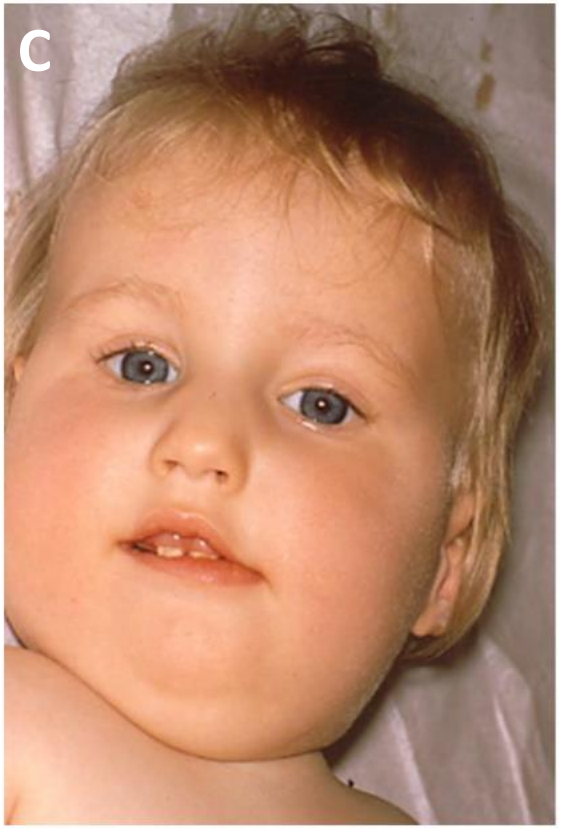




Case 7

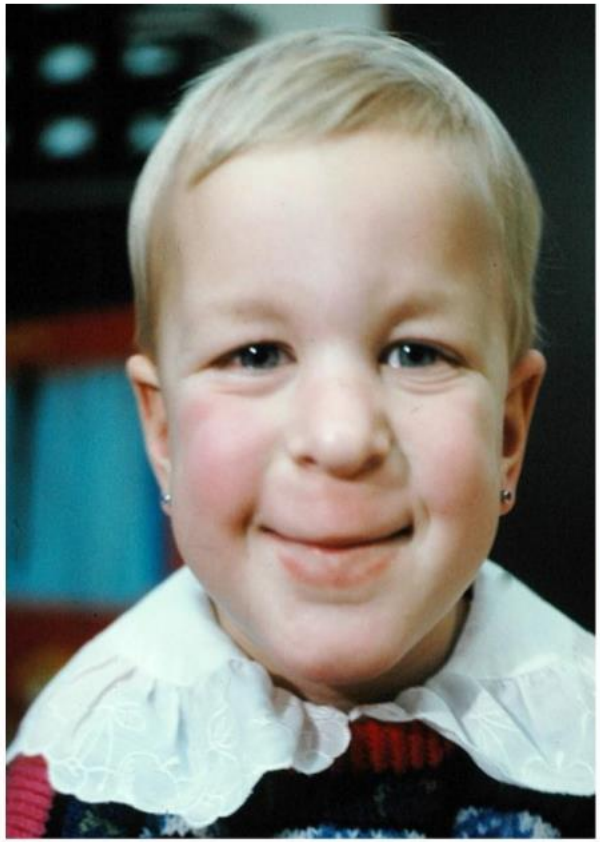
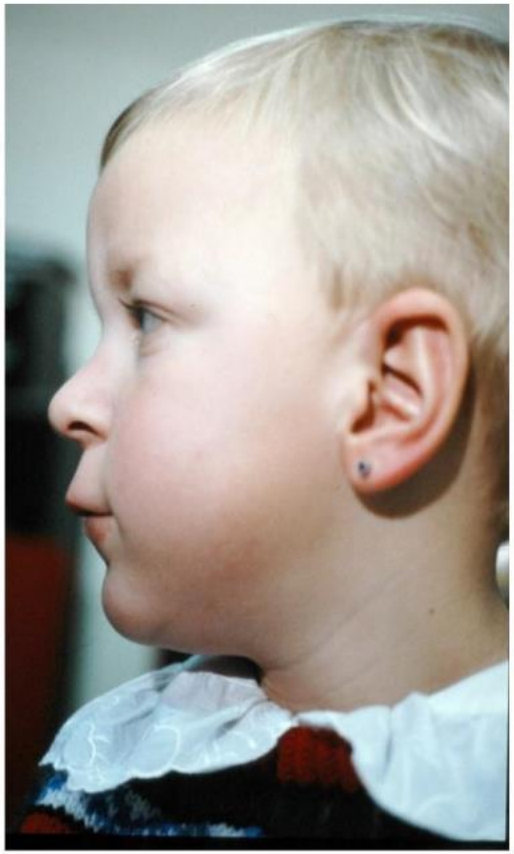
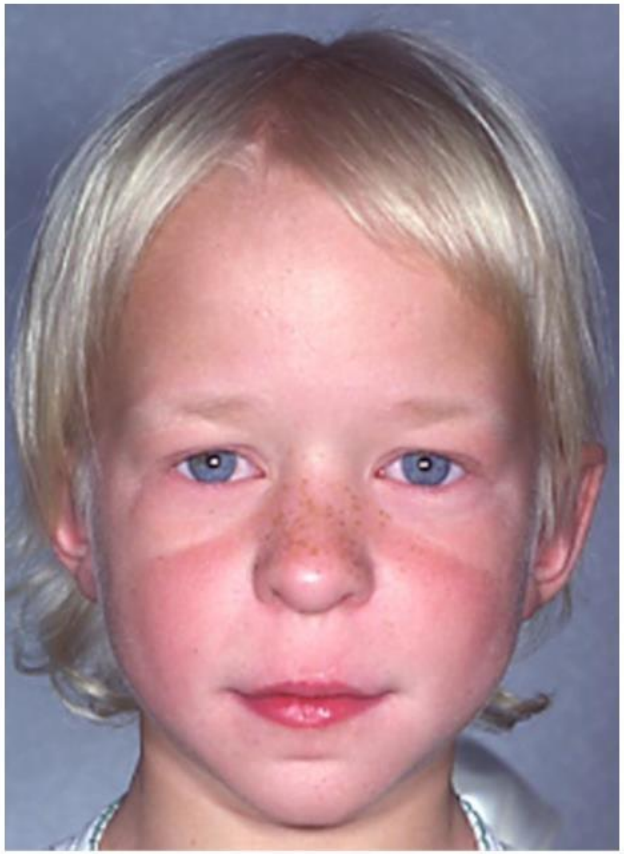






- Obesity
- Hypomimia/ generalized hypotonia
- Open mouth – dry saliva
- *Prader-Willi Syndrome*

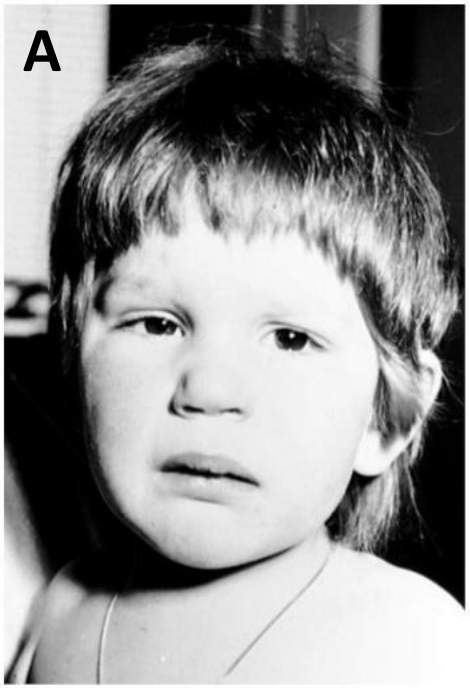
Case 8

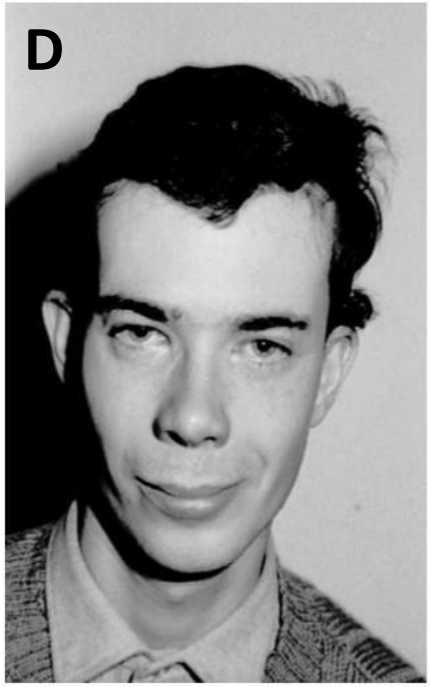




- How old does she look? Why does she look old?
- Because she has a high hairline. No teeth, thin lips, long face.
- Temporal balding
- Lack of teeth. Maxilla and mandible too close together. Chin folds.
- *Trichorhinophalangeal syndrome*

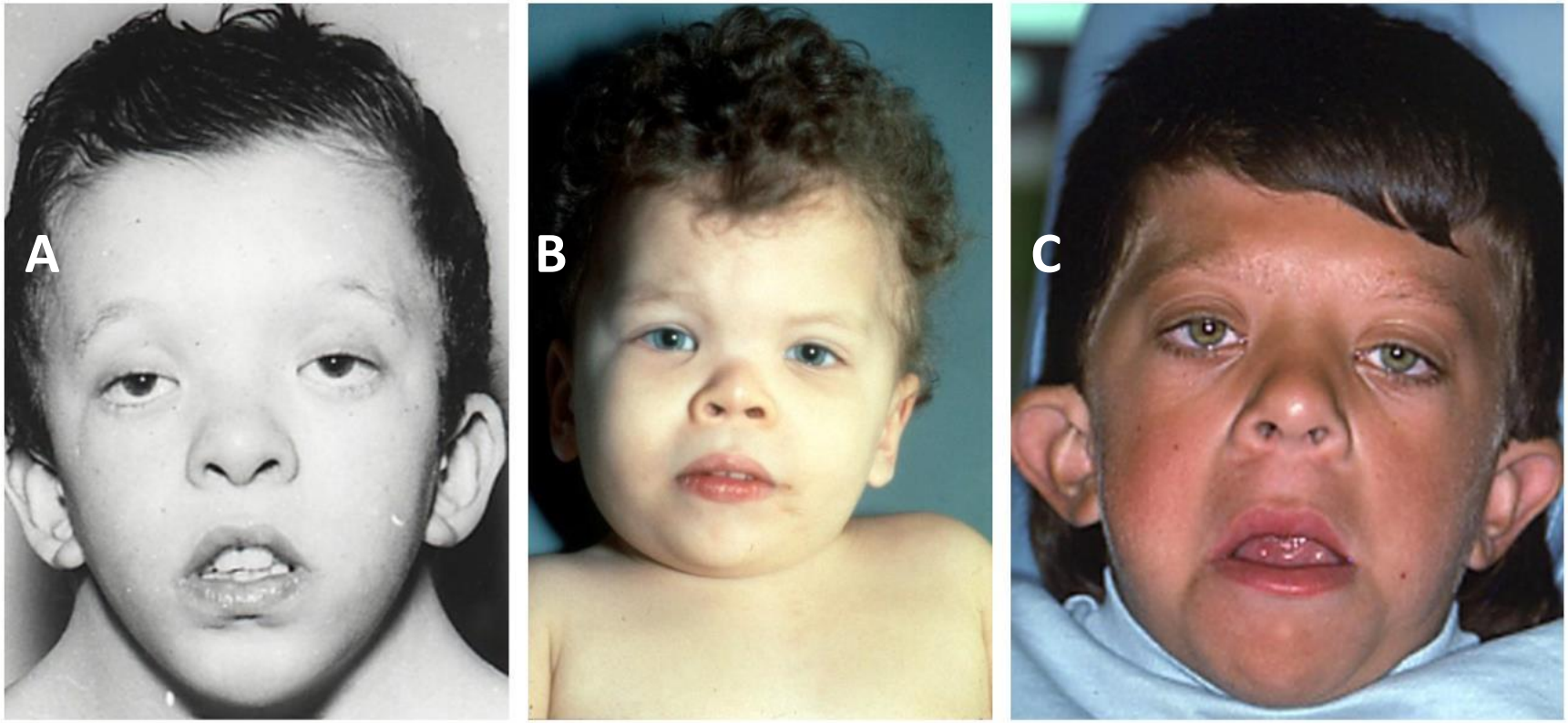
Case 9

- Triangle face
- Large protruding ears
- Microcephaly? normocephaly or macrocephaly?
- Large testicles
- Severe developmental delay
- “Hand flapping” behavioral phenotype
- *Fragile X syndrome*



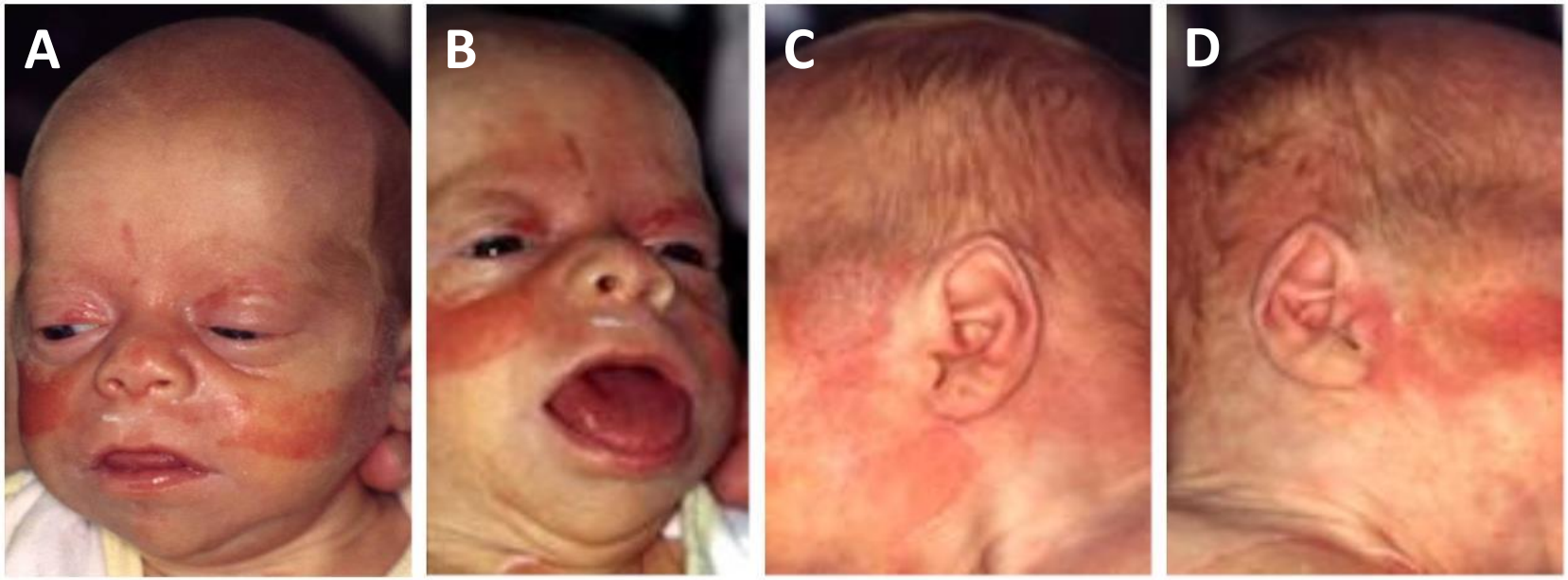


Case 10



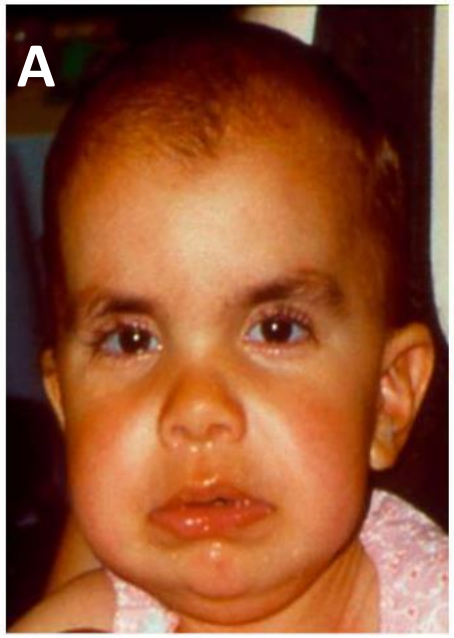
- Downslanting palpebral fissures (old terminology: antimongoloid).
- Low-set ears
- Low nasolabial folds
- Drooping mouth corners
- Ptosis
- Webbed neck
- As if you pull the face down. Why does it look like that? What happened prenatally?
- *Noonan syndrome.*

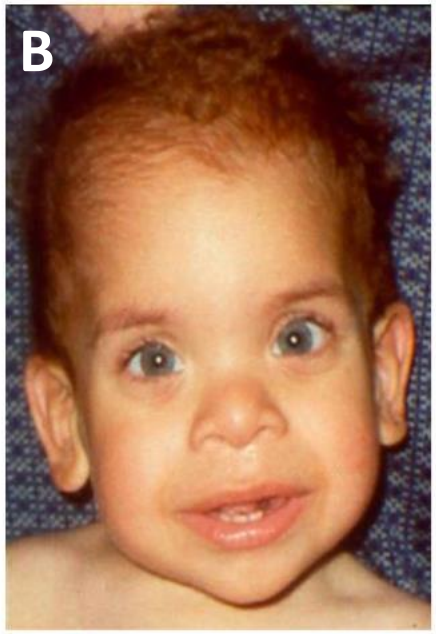
Case 11

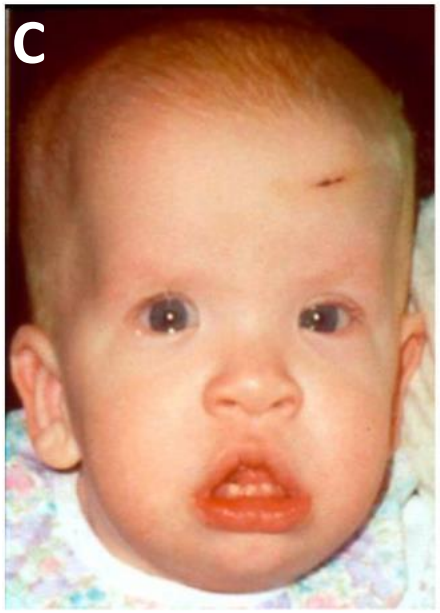


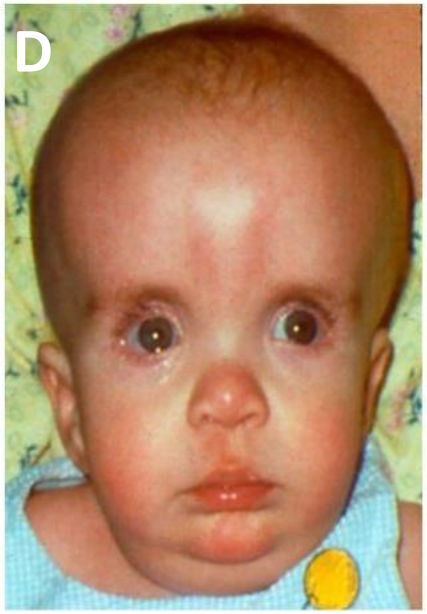
- Large tongue
- Crumpled/dysmorphic ears
- Skin lesions? No clinical significance
- *Beckwith-Widemann / Overgrowth Syndrome*

Case 12





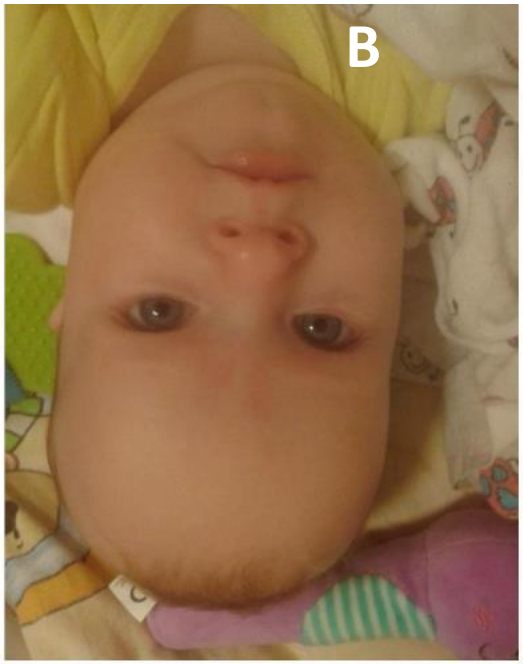


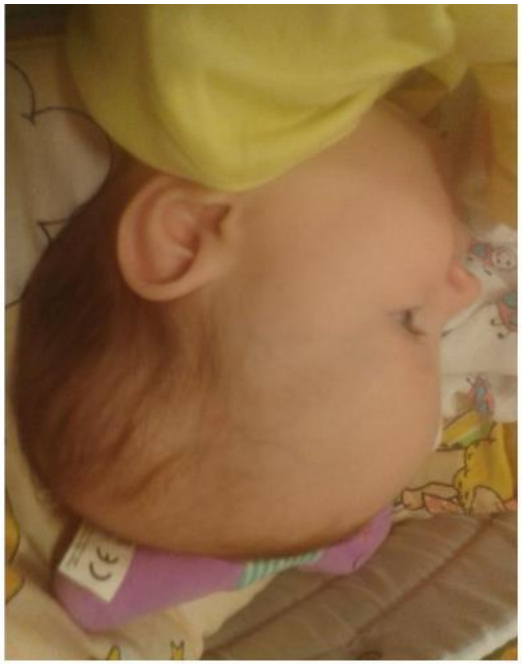


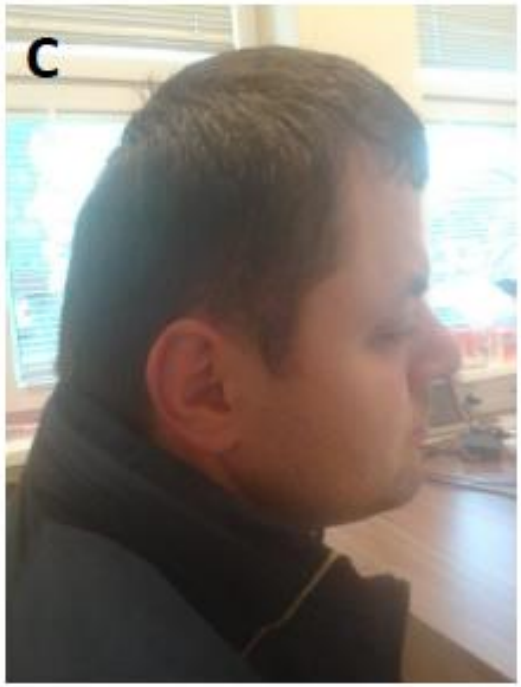
- Wide forehead.
- Narrow temples
- Macrocephaly – ”overturned pear”
- Developmental delay
- Short nose
- *Sotos Syndrome*

Case 13

- Father's affected for sure.
- Flat occiput.
- Microcephaly?
- No – normocephaly, just dysmorphic shape.
- Why does the head grow? What if it doesn't?
- Premature fontanelle closure. Wide forehead. Child
- Craniosynostosis. What are we afraid of? Craniostenosis.
- How about the child?
- *Cruzon syndrome / Craniosynostosis*

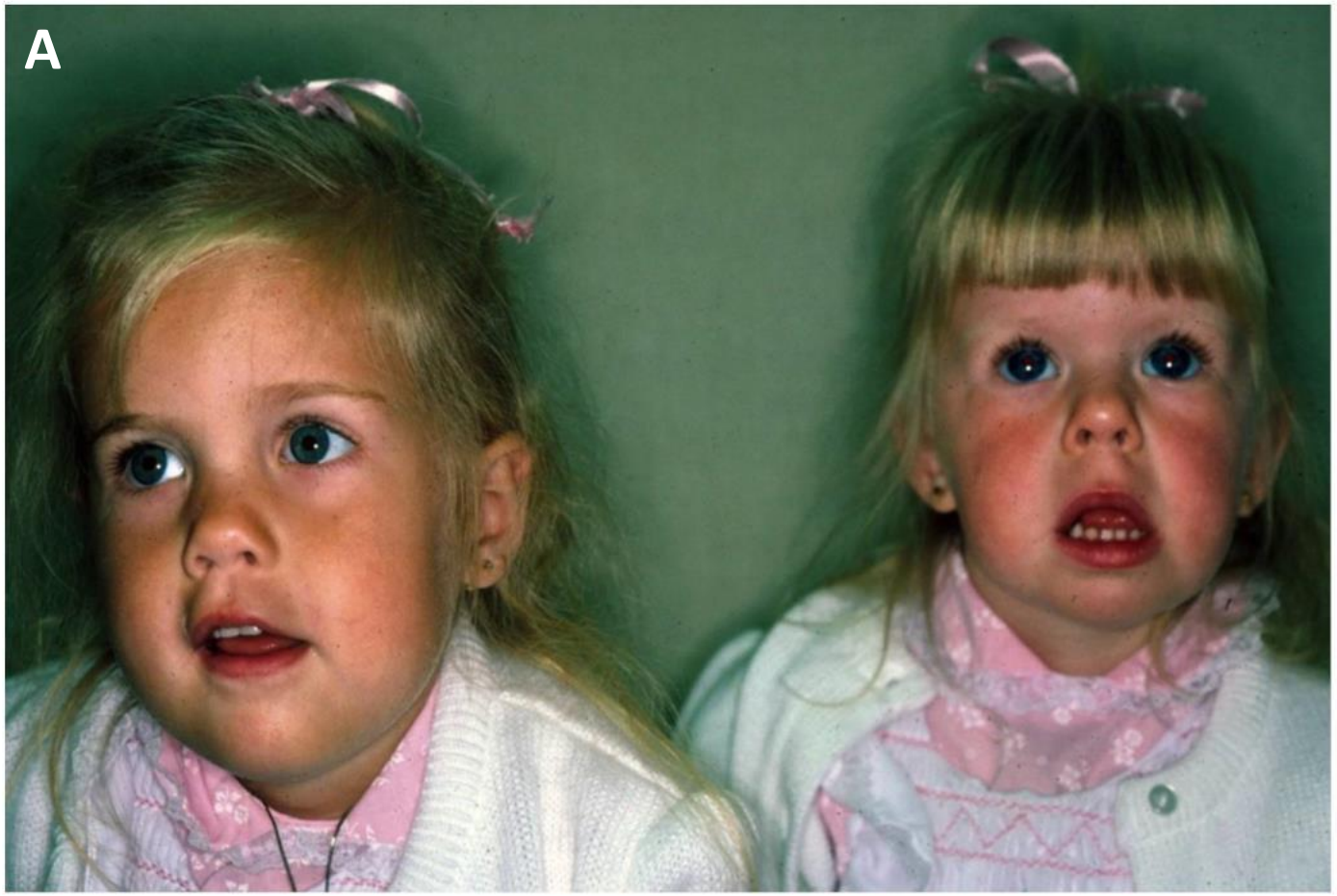






Father

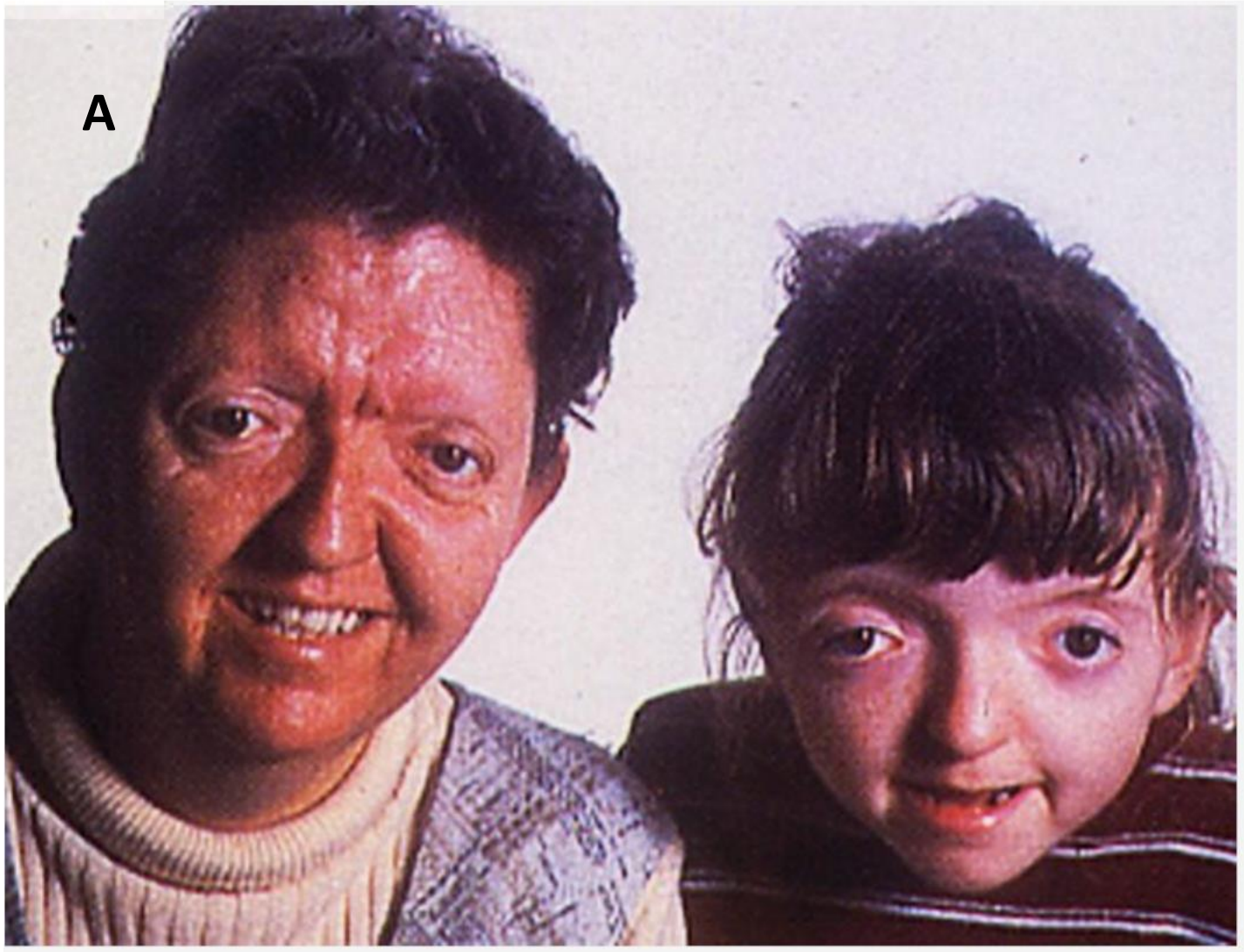
Case 14



Twin 1 Twin 2

Only the second twin is affected. Face ”pulled low” – facial hypotonia.
Williams? / Microdeletion?

Case 15



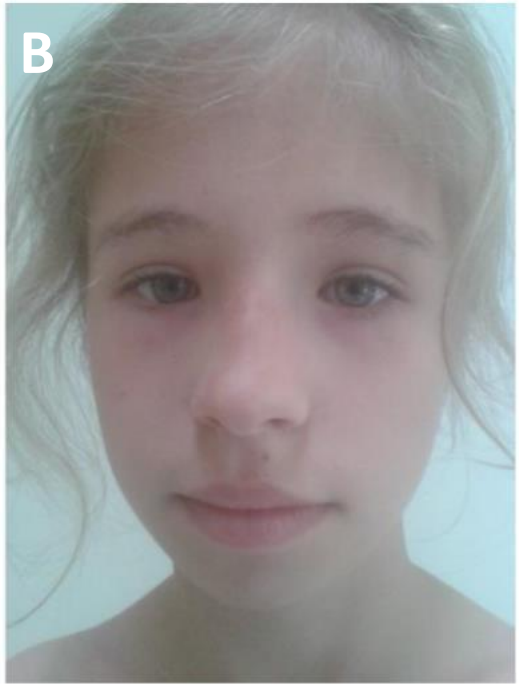
Patient 1 Patient 2

Increased forehead size. Bangs? Hypertelorism. *Cruzon / craniosynostosis*. Both affected.

Case 16

- Both sons are affected.
- Hair is thin and fragile
- Thin lips
- Thin and fragile nails
- Dental pathology
- They like being in cold places cuz they're always sweating.
- ECTODERMA! *Ectodermal dysplasia.*
- *10-20% of general population are carriers of X-linked recessive disorder. May be asymptomatic or have very mild signs / symptoms (i.e. lab test deviations like high CPK).*

Case 17



Sister

Brother

- Both pts:
- Microtia (small ears)
- Micrognathia(small jaw)
- Feeding difficulties in early childhood.
- *Pierre-Robin sequence disease*