Dysmorphology Practical Class

March 2023

Snir Boniel, MD

Agenda

1. Warm-up
2. Describe the dysmorphism – group exercise
3. Clinical cases – group exercise
4. Quiz!

Warm-up!

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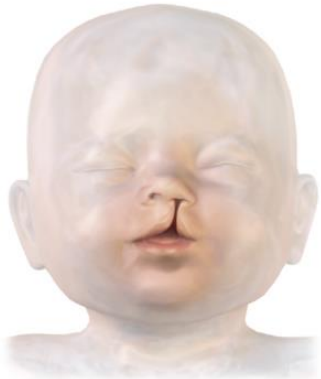
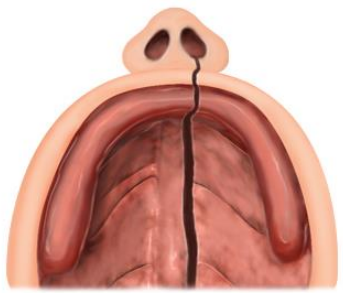
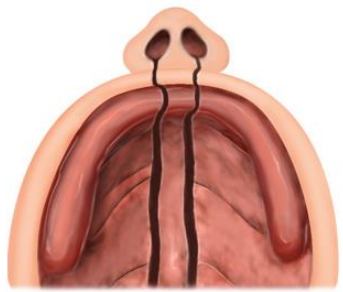


Unilateral Cleft Lip

Bilateral Cleft Lip

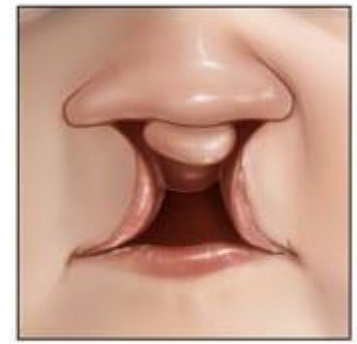


Unilateral Cleft Lip and Palate



Bilateral Cleft Lip and Palate

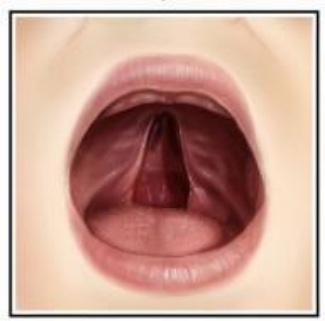
Unilateral cleft lip

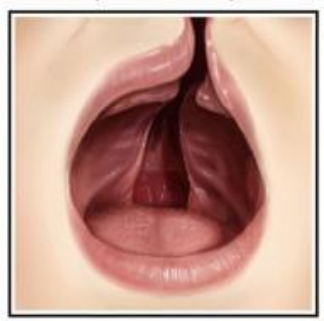


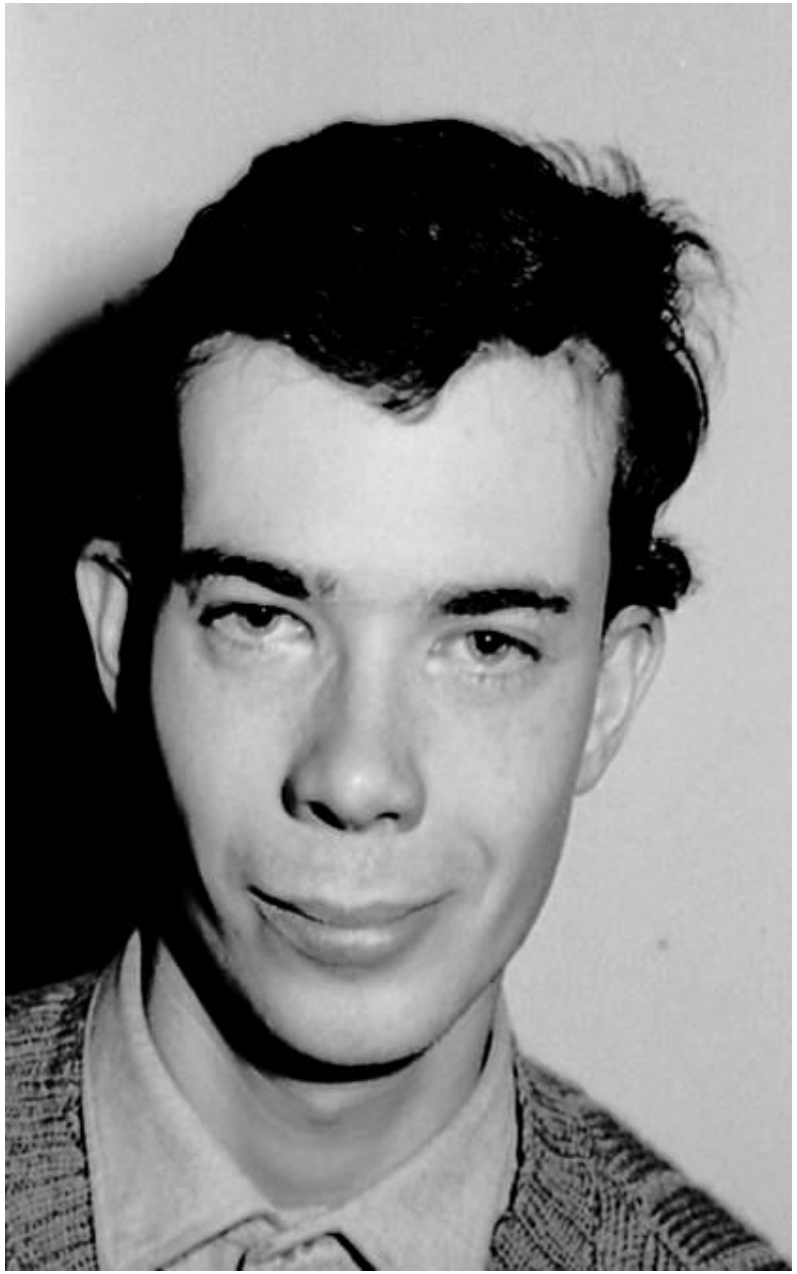
Bilateral cleft lip

Cleft palate

Cleft lip and cleft palate







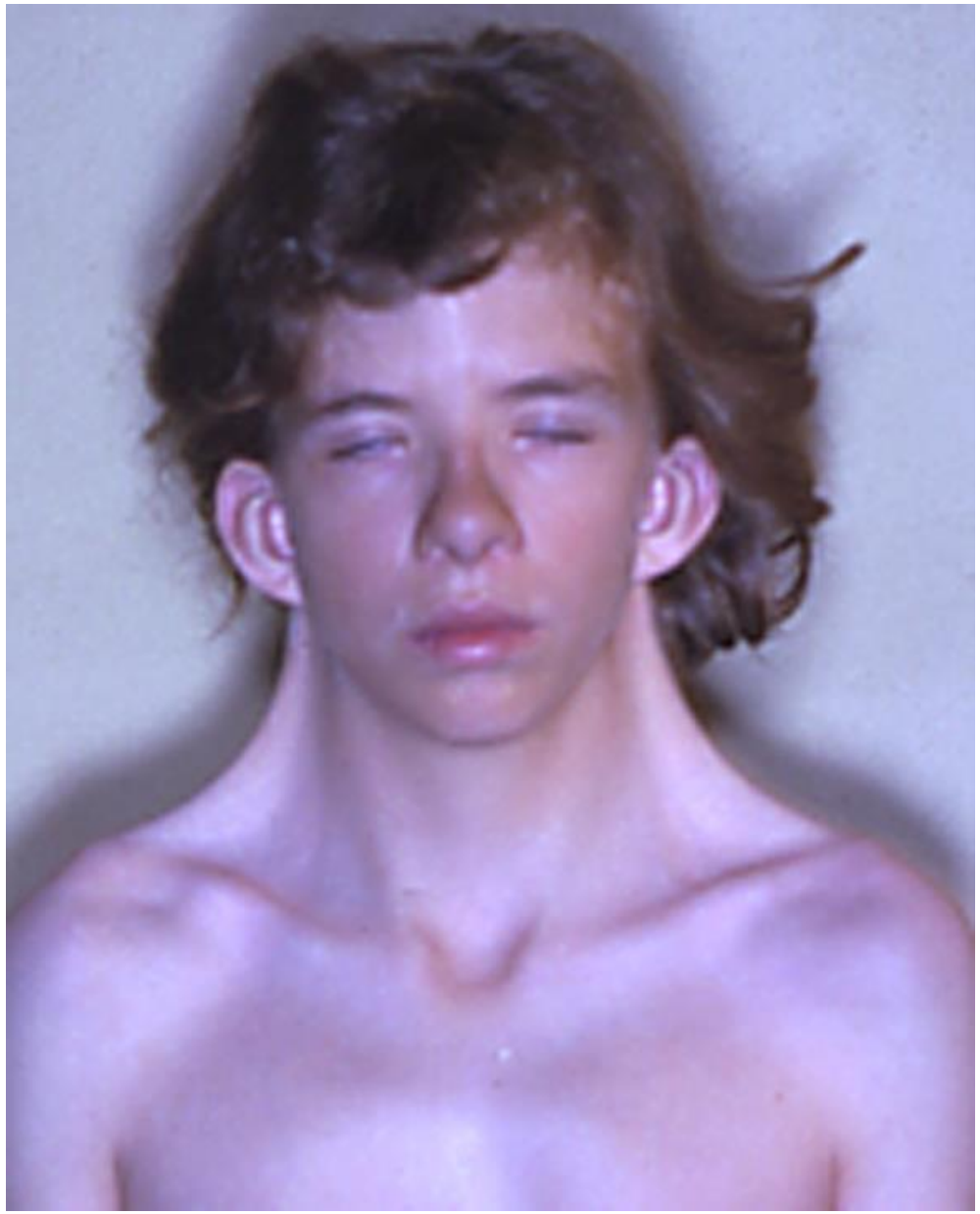
Theoretical question:

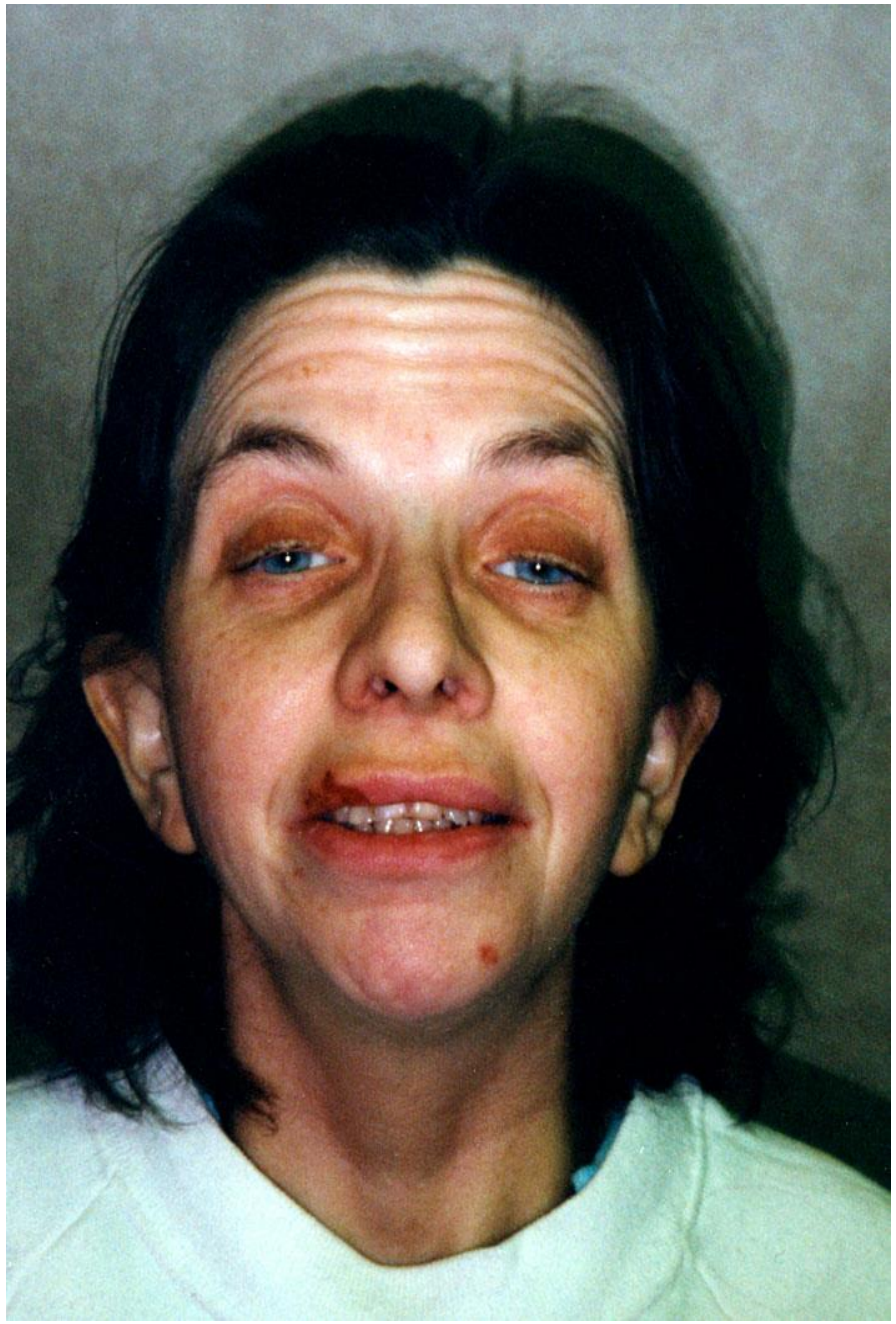
You are waiting at the bus stop. A group of young people with developmental delay are out on a trip, they arrive at the bus stop with their teachers from a special education school.

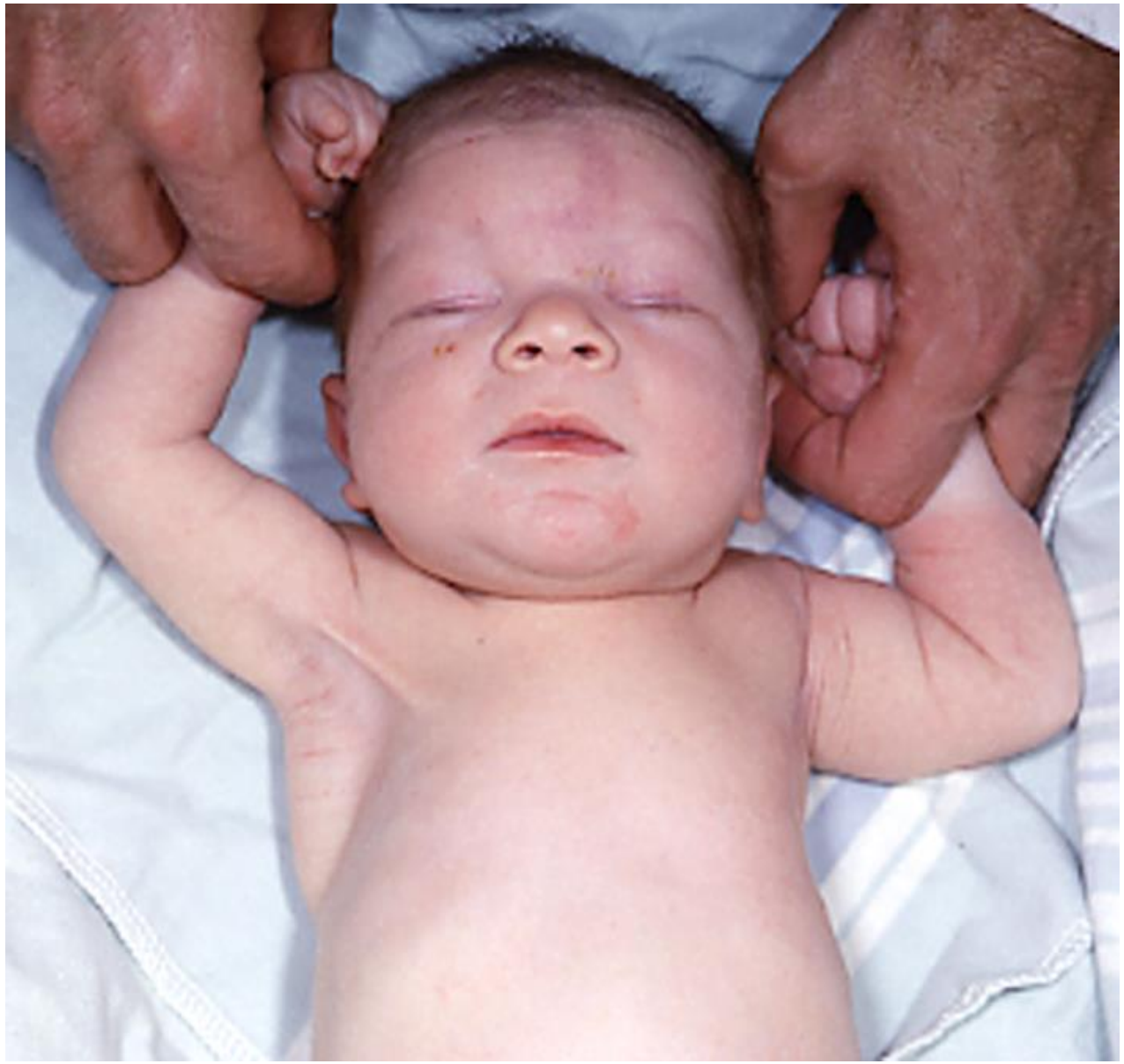
1. Will there be more men or women in this group?
2. What is the most common cause of developmental delay?
3. What is the second most common cause?
4. Will these people present with microcephaly, normocephaly or macrocephaly?w

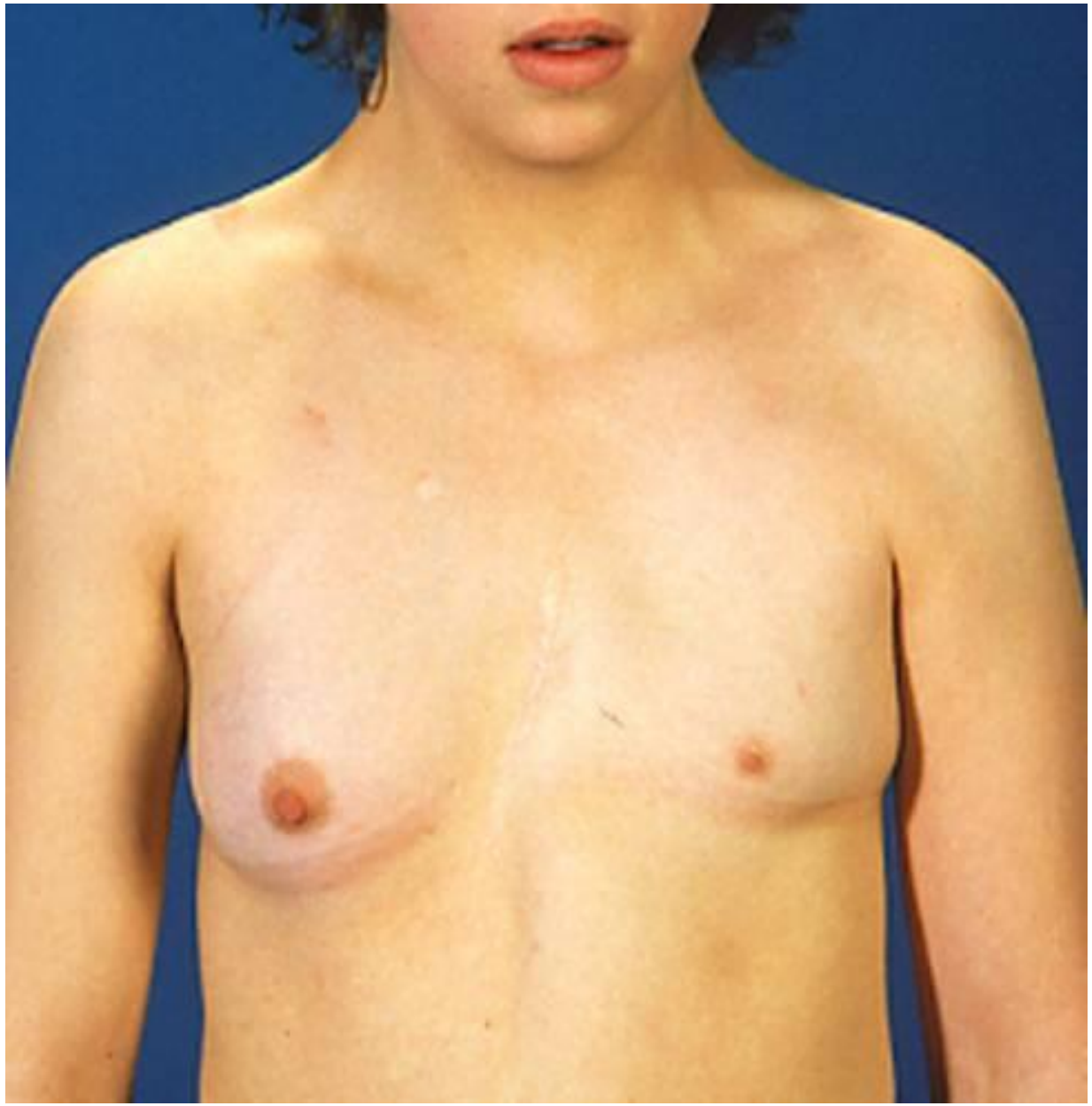
70% microcephaly – associated with developmental delay. But, 30% - normo or macrocephaly. If you're not sure what's inside the brain – perform brain imaging.

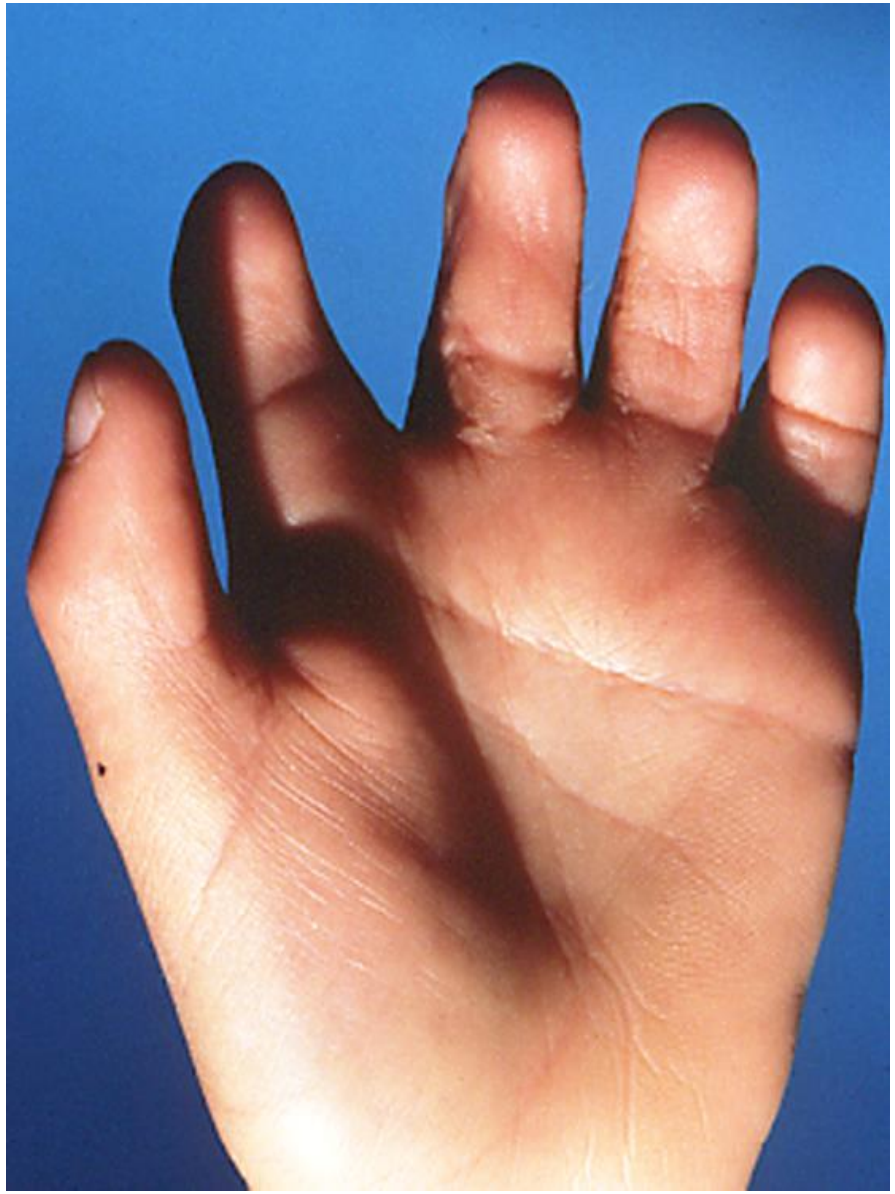
Patients with FraX usually present with either normocephaly or macrocephaly.

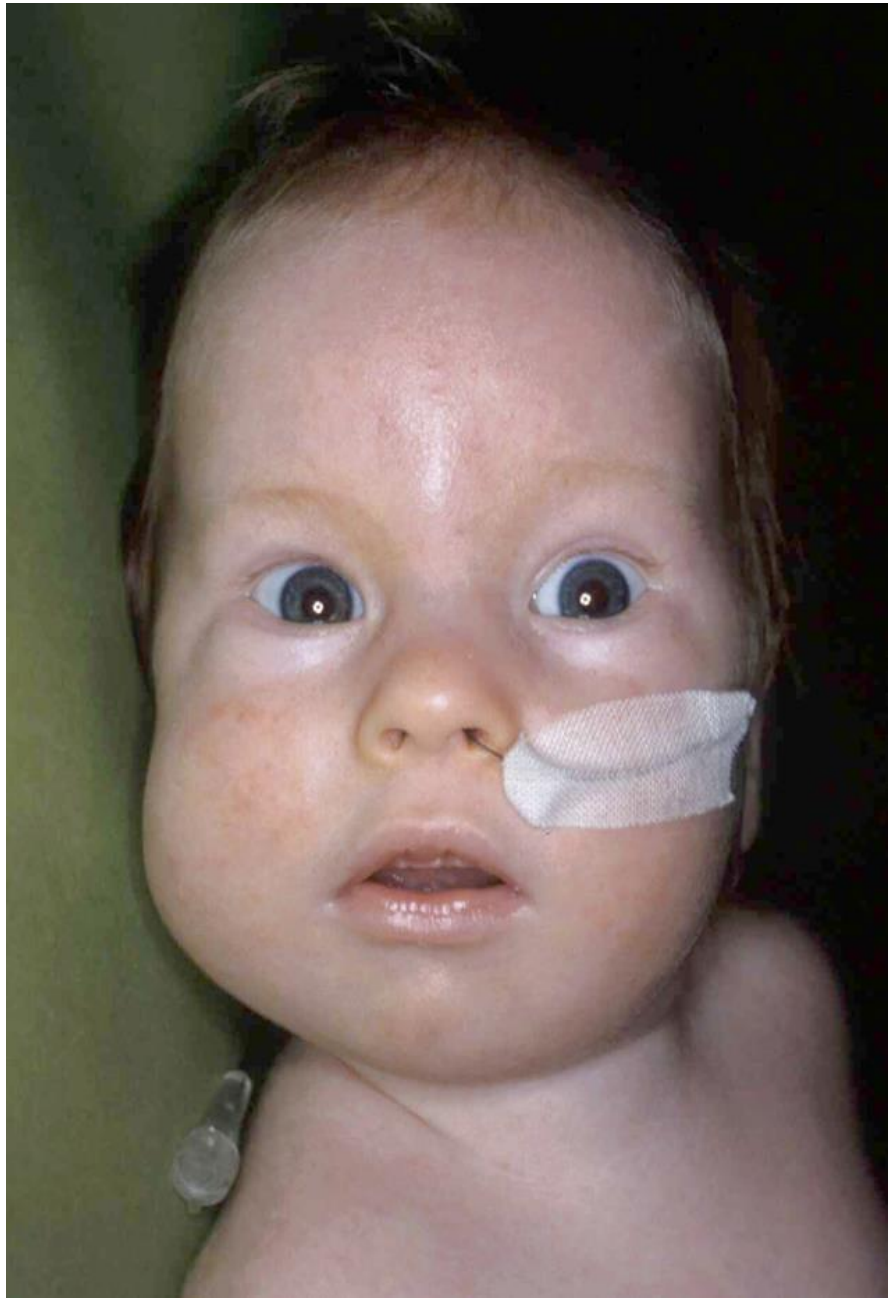


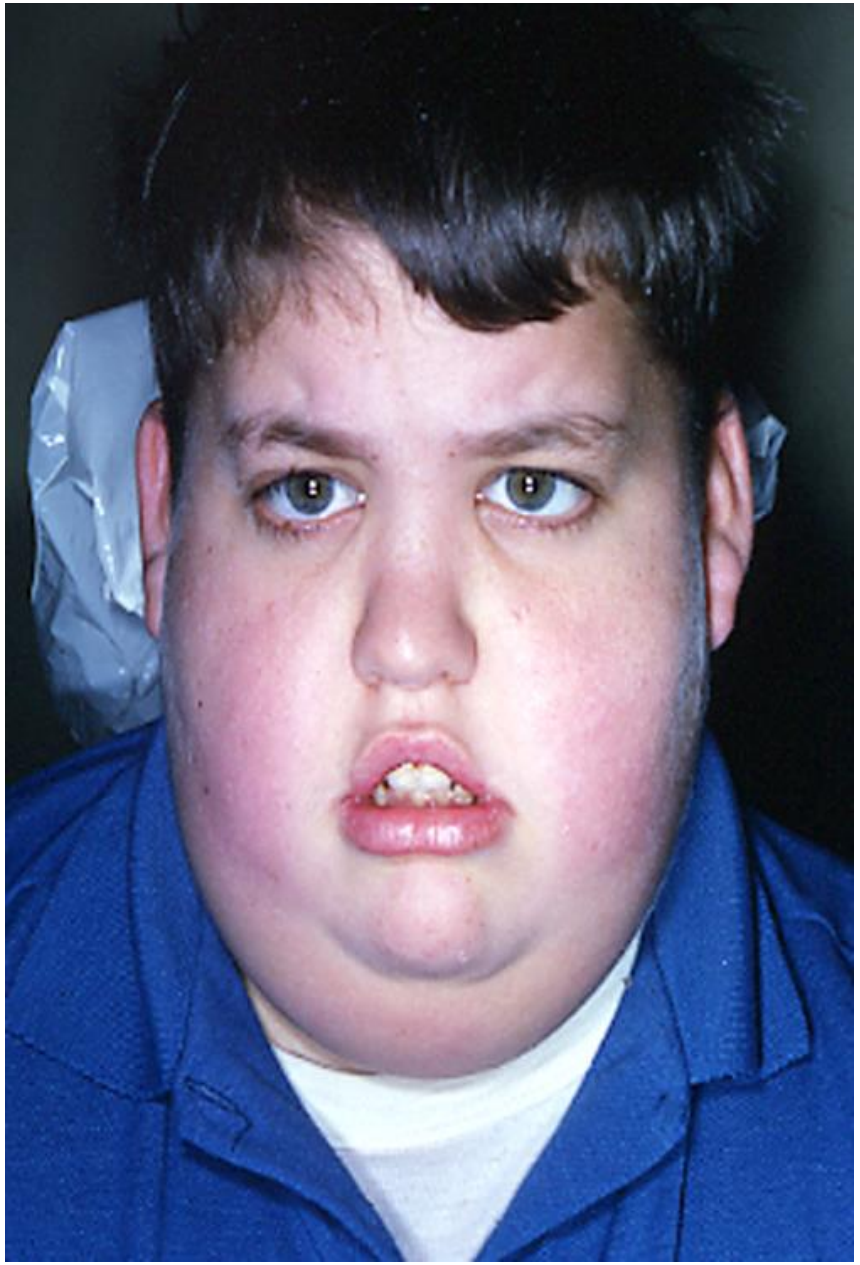






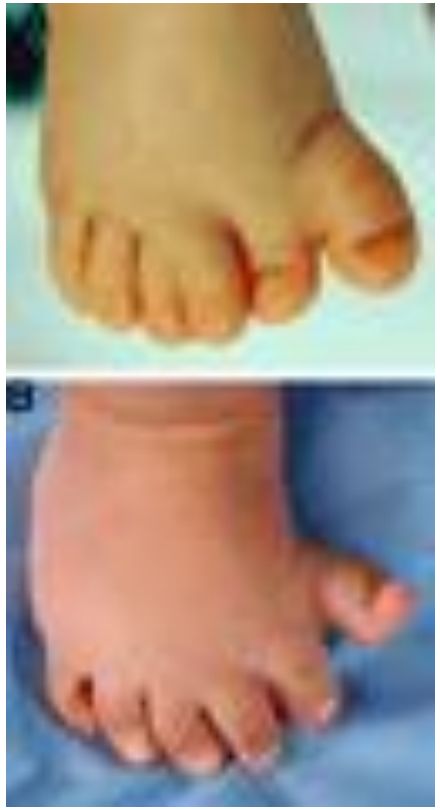




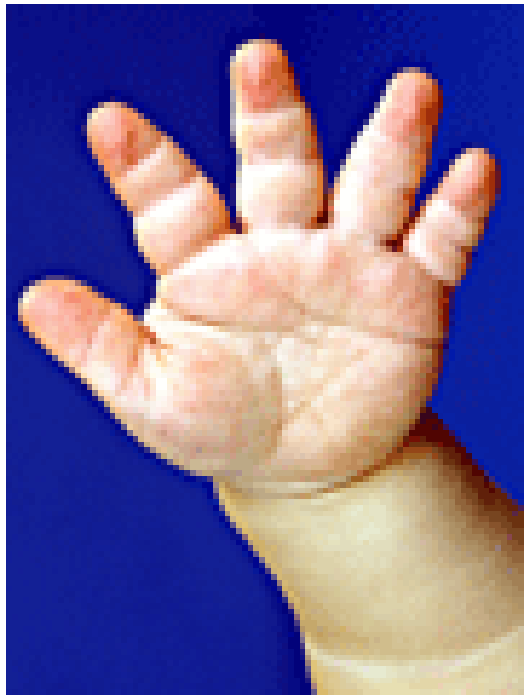


Trigonocephaly

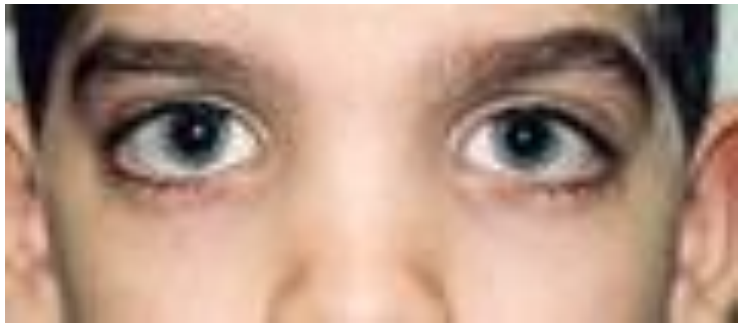
Webbed neck



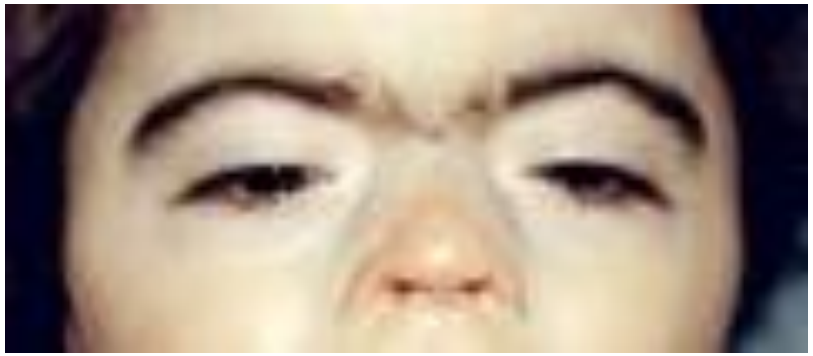
Foot preaxial polydactyly



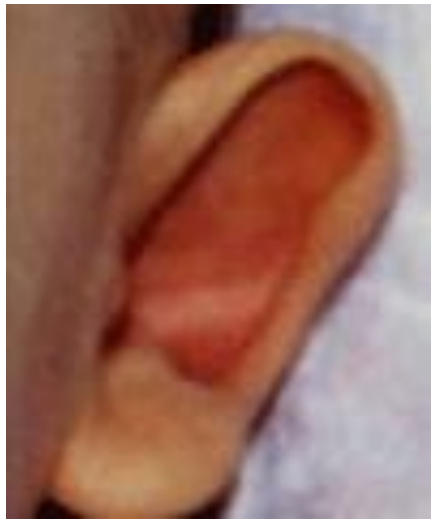
Trident hand



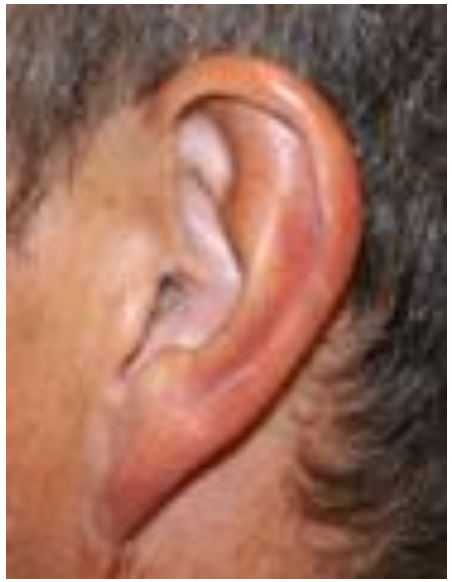
Long palpebral fissures



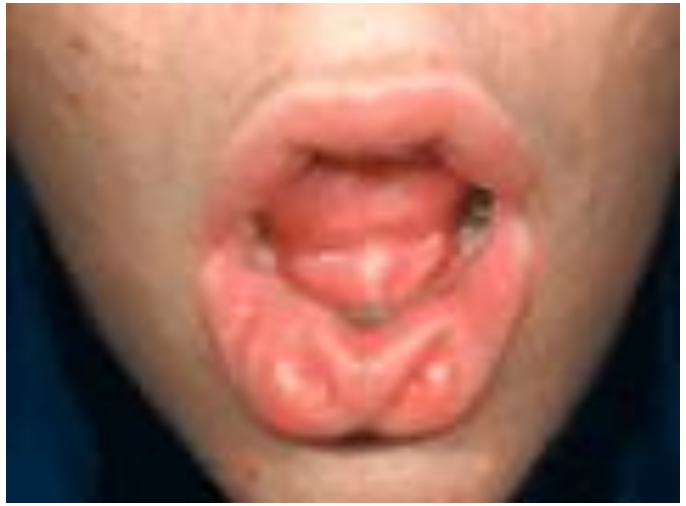
Synophrys



Cupped ear



Attached earlobe



Lip pit

Ankyloglossia

Submucous cleft palate

Lastly, remember

- You **MUST** know these 28 genetic diseases/syndromes inside out:
  - Main symptoms / clinical issues
  - Inheritance method
  - Gene, locus, principal method of diagnosis:

1. Factor V Leiden Thrombophilia
2. Hemophilia A
3. Hemochromatosis
4. 22q11.2del / DiGeorge Syndrome
5. Noonan syndrome
6. Williams syndrome
7. Neurofibromatosis Type 1
8. Tuberous Sclerosis
9. Joubert Syndrome
10. Prader-Willi Syndrome
11. Patau Syndrome
12. Edwards' Syndrome
13. Down Syndrome
14. Wolf-Hirschhorn Syndrome
15. Charcot-Marie-Tooth Disease
16. Duchenne Muscular Dystrophy
17. Spinal Muscular Atrophy
18. Phenylketonuria
19. Autosomal recessive deafness (GJB2, DFNB1)
20. Angelman Syndrome
21. Fragile X Syndrome
22. Huntington's Disease
23. Rett Syndrome
24. Cystic Fibrosis
25. Achondroplasia
26. Marfan Syndrome
27. Klinefelter Syndrome
28. Turner Syndrome