Physical Observations

Eyes
- size - usually bilateral
- position
- structure
- color
- orientation
- function

Eye: small
- diagnostic; indicative of possible developmental errors of underlying brain development
- small size a feature of many syndromes (FAS)
- small eye - microphthalmia
- absent eye - anophthalmia
- measure palpebral fissure

Eye: large
- not syndrome associated
- differs from bulging eyes: associated with syndrome
- bulging eye - exorbitism or exophthalmus
- eye extends out from socket of orbit
- secondary to shallow orbit, maxillary hypoplasia, forehead slope, growths behind eyes

Exorbitism
- also seen in Crouzon's and Aperts Syndromes

Eye: position
- determined by
  - distance between eyes,
  - symmetry,
  - vertical and horizontal placement

Hypertelorism
- increased separation of entire orbit
- common finding in syndromes

Hypotelorism
- decreased space between eyes
- more ominous-associated with underlying brain anomaly
Eye measures
- inner canthal distance
- outer canthal distance
- interpupillary distance
- caution i.e.: small head confound

Eye Orientation
- angle of palpebral fissure
- normal = horizontal
- upslanting assoc. with microcephaly
- downslanting - common in craniofacial syndromes

Eye Structure
- among most common of malformations
- strong association with syndromes
- anomalies of eye, lid, lashes, iris and pupil

Types of Problems
- ocular coloboma - notch like defect in eyelid
- microphthalmia - small eye
- anophthalmia - missing eye
- aniridia - absent iris
- congenital cataracts
- dislocated lens
- absent lashes

Eye Color
- anomalies not common
- heterochromia - two different colors
  - associated with some syndromes but mostly benign
- other
  - unusual iris color, abnormal color of sclera

Eye Function
- relates to abnormalities of eye muscle
- strabismus - impairment of eye convergence
  - exotropia - one eye turns out
  - esotropia - one eye turns in
- nystagmus - movement abnormalities - rapid side to side
- assoc. with over 50 syndromes

Ears - Structure
- microtia
  - grade I - small but normal structures
  - grade II - external ear malformed, canal is stenotic, ossicles malformed
or fused
• grade III - external ear severely malformed or completely absent, middle ear space absent, severely malformed or absent ossicles

17 Other
• Malformation of individual ear components
  • helix or lobule
  • velo-cardio-facial syndrome
• abnormal morphogenesis: grade II, III microtias
• adherence to temporal bone
• protruding ears (Down, V-c-f)

18 Velo-Cardio-Facial Syndrome
• Shprintzen Syndrome
• IQ - 70-90
• cleft palate
• septal defect
• lots of hair
• slender fingers

19 Ear Position
• normal - comparable with eye
• imaginary line - connect palpebral fissures, extend back--line should intersect at least one third of ear
• if below imaginary line - “low set” (benign)