1. **Multiple anomalies**
   - often noted by clinician observation of physical symptoms
   - two major features affected in majority of multiple anomalies
     - craniofacial complex - head, face, eyes, ears and hair
     - limbs - hands

2. **Multiple Anomaly**
   - Unilateral cleft of the primary palate

3. **Multiple Anomalies**
   - Ear anomaly
   - Eye anomaly

4. **Head**
   - abnormalities of size, shape and symmetry
   - size and shape related to brain size and craniostenosis

5. **Diagnostic concerns**
   - normal size does not indicate normal brain
   - abnormal size - strong correlation with function
   - not possible to eyeball, use anthropometric measures (head circumference)

6. **Head: Cranial size and brain**
   - microcephaly - small brain, small skull
     - small head circumference
     - common in many syndromes
     - prognostic of MR
   - macrocephaly - brain is too large (hydrocephalus), skull large

7. **Cranial size and craniostenosis**
   - craniostenosis - premature fusion of cranial bones which normally remain separated by sutures
   - sutures allow expansion of brain til 2nd decade
   - bones of skull base may also be fused
   - brain growth compromised, bulging eyes (exophthalmos), underdeveloped midface (maxillary hypoplasia), protruding lower jaw (prognathism).

8. **Craniostenosis**
   - 1/2000 live births
   - most commonly limited to one suture
   - feature of over 150 syndromes
   - most inherited

9. **“Bulging Eyes”**
   - Exophthalmos
• associated with shallow orbits

10  Head: cranial shape
• related to
  – underlying brain anomalies
  – craniosenosis
  – anomalies of the skull base
• other physical findings: frontal bossing, prominent occiput, temporal bulging, posteriorly sloping forehead, flattening of occiput

11  Skull shapes & Stenosis
• Shape determined by stenosis
• Scaphocephaly
  – sagittal stenosis
• Brachycephaly
  – bilateral coronal stenosis

12  Cloverleaf
• Cloverleaf skull anomaly
• referred to as kleeblattschadel
• involves multiple sutures
• downslanting eyes

13  Face
• weaker relationship between shape and CNS
• kids with abnormal brains have normal faces and visa versa
• abnormalities described as hypoplasia, hyperplasia, clefts, asymmetries, structural abnormalities or redundancies

14  Face structures
• frontal bone of calvarium (forehead)
• orbits (eyes)
• maxilla (region from base of orbits to mouth)
• nasal bones
• mandible (lower part of face)

15  Facial Proportions
• Approx 3 equal parts: forehead, midface and lower face
• lower face divided into 1/3 and 2/3 above and below oral commissure

16  Facial Proportions
• Distance between inner canthi (corners) of eyes approximately equal to alar nasal base width
• alar = wings

17  
• Mouth should be as wide as distance from right to left medial limbus
  • limbus = border
  • in this case, border between the cornea and sclera of eyeball

18 Facial Features

19 Lips
• composed of muscle and glands
• outer - skin; inner - mucous membrane; vermilion = transitional “red” zone
• length: upper - 22mm;
• opening - no greater than 3 mm
• upper lip slightly in front of lower

20 Lip Features
• Oral frenulum = midline mucosal attachment
• mucocutaneous junction = white line
• cupid’s bow = slope from commissure and peaks from philtrum
• Philtral ridge - upward from peaks
• philtrum - inbetween ridges
• center fullness = vermilion tubercle

21 Nose
• length: 75% of height of lower 1/3rd of face
• upper bony part and lower cartilagenous part
• nares - nostrils

22 Hard Palate: underlying structure
• paired palatine process of maxilla
• posterior 1/4 = palatine bones
• midline suture
• anterior midline = incisive foramen
• posterior midline = posterior nasal spine

23 Hard Palate: Surface features
• keratinized rugae mucosa
• palatine torus - prominent longitudinal ridge
• anterior midline = incisive papilla

24 Maxillary/Alveolar Process
• forms and supports teeth
• alveolar bones envelopes tooth buds
• gingival mucosal covering
• anterior portion = premaxilla
• premaxilla houses incisors
25  Soft Palate
   • marks lower border of nasopharynx
   • fibromuscular structure
   • palatine aponeurosis - fibrous tissue extending 1 cm posterior to border
   • mucosal covering on oral and nasal surface; different characteristics
   • functional musculature: levator, tensor, muscularis uvula

26  Tonsils
   • lymphoid mass: palatine, lingual, pharyngeal
   • Waldeyer’s Ring
   • involute at puberty
   • palatine - between faucial pillars
   • little significance to speech

27  Adenoids
   1  • pharyngeal tonsil
      • posterior corner, nasopharynx
      • not visible intraorally
      • size affects nasopharynx
      • in kids, contact point for velum

   2  • Involute at puberty
      • velar accommodation to change
      • hypertrophic, hyponasal/denasal
      • 50% obstruction - adnoidectomy
      • contraindicated in cleft population

28  Hypoplasia
   • underdevelopment of a structure (fewer cells make up structure)
   • secondary to lack of vascular supply, intrinsic developmental disorder, genetic or environmental etiologies
   • affects various facial components

29  Maxillary hypoplasia
   • underdevelopment middle third of face
   • common anomaly in craniofacial syndromes
   • may be related to craniostenosis as face “hangs from” skull
   • often associated with brain dysfunction, especially with closely placed eyes
   • seen in FAS, holoprosencephaly, other chromosomal deficiencies

30  maxillary hypoplasia
   • Patient with cl & p
   • lower lip everted
   • negative overjet
• difficulty with bilabials

31  Fetal Alcohol Syndrome (p. 78)
• Thin upper lip
• poorly defined philtrum
• short nose
• broad, low nasal bridge
• mild ptosis
• epicanthal folds

32  Other features
1  • small size at birth
• microcephaly
• congenital heart defects
• cleft lip +/- palate
• MR - average IQ=63
• behavior problems (hyperactivity, poor judgement etc
2  • Difficulty with social pragmatics
• distractibility

33  Holoprosencephaly
• Developmental anomaly
• absence of cranial vault
• cerebral hemispheres missing or single mass
• absence of midline structures

34  Holo...cont
• Also called anencephaly
• median cleft
• absent nasal structures
• premaxially agenesis
• bleak outlook

35  Maxillary Hypoplasia As isolated anomaly
• some people have smaller maxillary complex “naturally”
• ethnic differences: negroid = larger and broader than caucasian
• caucasian larger and broader than asian

36  Mandibular hypoplasia
• as separate bone, no implications for CNS
• tends to be familial trait and benign, especially with normal shape
• other: genetic syndromes: Treacher Collins - abnormal size and shape
• other descriptors
  – microagnathia - shortening of ramus
  – retroagnathia - posterior place
  – proagnathia

37  Hyperplasia
• overdevelopment; overgrowth syndrome
• associated with active disease process
• few syndromes with hyperplasia
• example: neurofibromatosis (NF 1, von Recklinghausen disease
• Proteus syndrome - elephant man; large tumors
• Cranjodiaphyseal dysplasia - (Mask) - skeletal overgrowth

38  Facial Clefts
• occur anywhere along lines of embryologic development
• Tessier - classified 14 clefting lines-cl/p only one of 14
• can occur as isolated events
• clefts most often clinical feature of multiple anomaly syndrome

39  Assymetries
• isolated to single part of face or in multiple craniofacial structures
• most common affects multiple structures (eyes, ears, mandible)
• some syndromes identified by asymmetry- Goldenhar

40  Hemifacial Microsomia
1  • Goldenhar
   • oculoauriculovertebral dysplasia
   • 1/3000-5000 births
   • 70% unilateral
   • major feature: unilateral hypoplasia
2  • Malar, maxillary and mandibular hypoplasia
   • ear involvement-anotia (absent external canal)
   • eye anomalies
   • brain anomalies
   • vertebral anomalies

41  Hemifacial microsomia
• Oculo-auriculo-vertebral spectrum
• +/-cl&p
• ocular anomalies
• dermoid cysts
• hearing loss
• facial paresis
• other occasionals

42  Hemifacial microsomia
• Skin tags
• anterior to auricle

43  Others
44 Last One