

- 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
 - 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
 - craniostenosis
 - 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
 - saggital stenosis
- Brachycephaly
 - blateral 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; vermillion = transitional “red” zone
- length: upper - 22mm;
- opening - no greater than 3 mm
- upper lip slightly in front of lower

20 Lip Features

- 1
 - Oral frenulum = midline mucosal attachment
 - mucocutaneous junction = white line
 - cupid’s bow = slope from commissure and peaks from philtrum
- 2
 - Philtral ridge - upward from peaks
 - philtrum - inbetween ridges
 - center fullness = vermillion 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 hp 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 - adenoidectomy
 - contraindicated in cleft population
- 28 **Hypoplasia**
- underdevelopment of a structure (fewer cells make up structure)
 - 2ndary 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
 - micrognathia - shortening of ramus
 - retrognathia - posterior place
 - prognathia

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
- Craniodiaphyseal 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

