

The Dysmorphology Exam

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Disclosures

Jennifer

- No disclosures

Carolyn

- No disclosures



Objectives

- Describe a basic dysmorphology exam
- Describe phenotypic abnormalities using standardized HPO vocabulary
- Identify clinician resources for dysmorphology exams and HPO terms
- Discuss challenges and solutions to implementing genomic medicine in a NICU setting



Importance of Dysmorphology

- An accurate dysmorphology exam can have a profound impact in clinical diagnostic evaluation
- Dysmorphic features that the provider identifies are used in the analysis of broad genomic sequencing and can be helpful in identifying an underlying etiology for a child
- The goal is to identify patterns of dysmorphology that can provide an understanding of a potential underlying genetic cause



Relevance to Nurses and Providers

- APPs, MDs, RNs in the NICU are the first line for neonates
- Should be able to recognize findings on physical exam and medical history that suggest the presence of a genetic syndrome
- Prompt recognition results in improved outcomes



Birth Defects



Birth Defects

- Rare
- 3% of all children born in any hospital or in any country or in any year will have a significant congenital anomaly
- 15% of newborns have one minor anomaly
 - 0.8% have two minor anomalies
 - 0.5% have three minor anomalies
- Probability of a major anomaly?
 - With two minor anomalies?
 - With three minor anomalies?



Aneuploidies

- Most common aneuploidies include Trisomy 21, 18, and 13
- Typically, if an infant has multiple dysmorphisms and multiple congenital anomalies, we think of chromosomal differences
- If an infant has multiple dysmorphisms, doing additional phenotype (renal US, ECHO) to look for additional congenital anomalies is important
- If an aneuploidy is highly suspected, a karyotype should be completed which should have a fast turnaround time, far faster than more comprehensive genomic testing



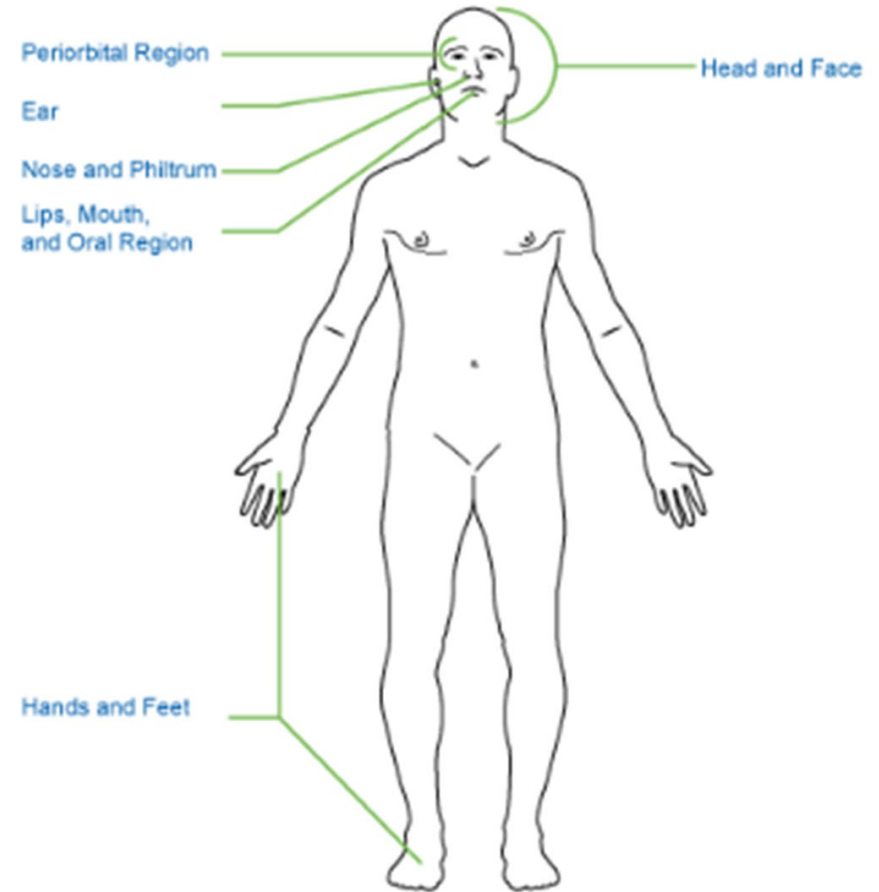
Physical Exam

How to approach Dysmorphology



Elements of Morphology

- NIH website contains 6 key areas
- Goal: develop accurate and clear definitions of terms for craniofacies
 - Head and Face
 - Periorbital Region
 - Ear
 - Nose and Philtrum
 - Lips, Mouth, and Oral Region
 - Hands and Feet



General Assessment

- Birthweight
- Length
- Head circumference
- Muscle tone
- Movements
- Postural abnormalities
- Body proportions



Head and Face Overview



PLATO's Golden Ratio

Golden proportions

- Width of face (two-thirds its length)
- Nose, no longer than the distance between the eyes
- Length divided into perfect $1/3^{\text{rds}}$
- Nose = size of ears
- Pupils drop to corners of mouth



Hair and Head Exam

- Examine child from the front, the back, and from above
 - Hair lines
 - Hair whorls
 - Cowlicks
- Scalp hair distribution
- Cranial shape





Double hair whorl

Aplasia cutis congenita

Abnormal hair whorl position





- Brachycephaly-short head

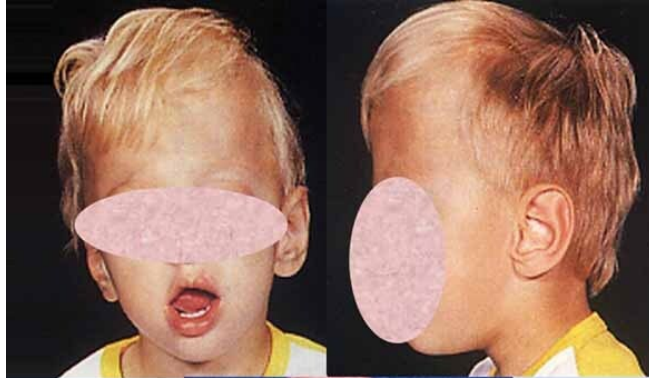


- Dolichocephaly- long head





- Trigonocephaly-Triangular shaped head

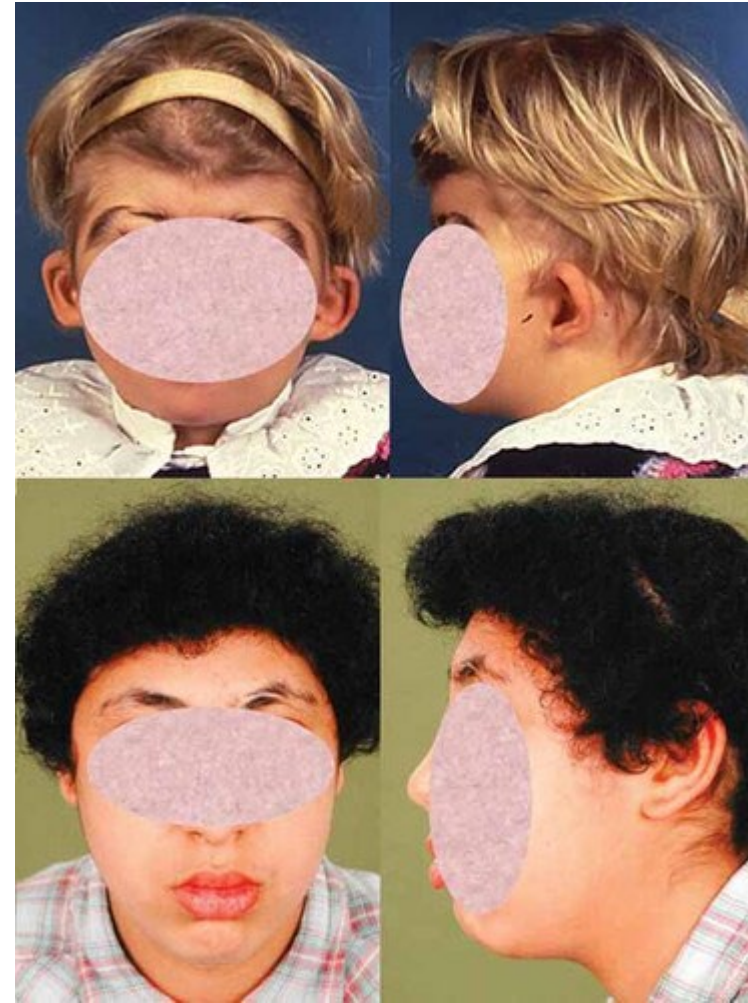


- Turriccephaly-Tower shaped head





High anterior hairline



Low anterior hairline



Forehead



Broad forehead,
frontal bossing



Narrow forehead/
Bitemporal narrowing



Face Variation

- Structure:
 - Asymmetric
 - Flat face
 - Malar flattening
 - Small/recessed mandible
- Size:
 - Small face
 - Thin face
 - Long face
 - Coarse features

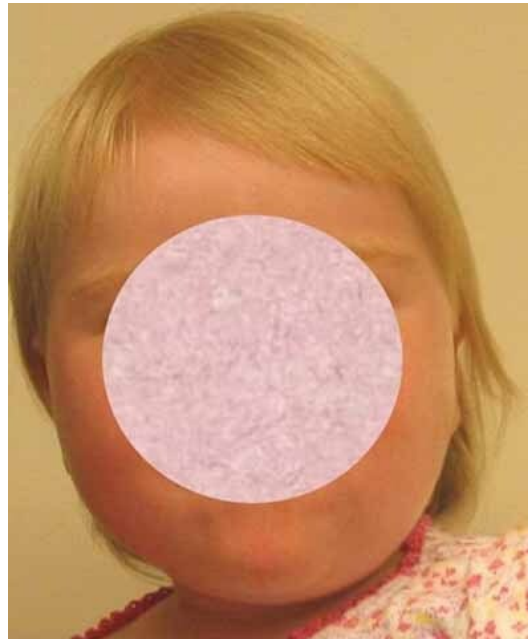




Square-shaped



Triangular



Round





Coarse facies

Flat face

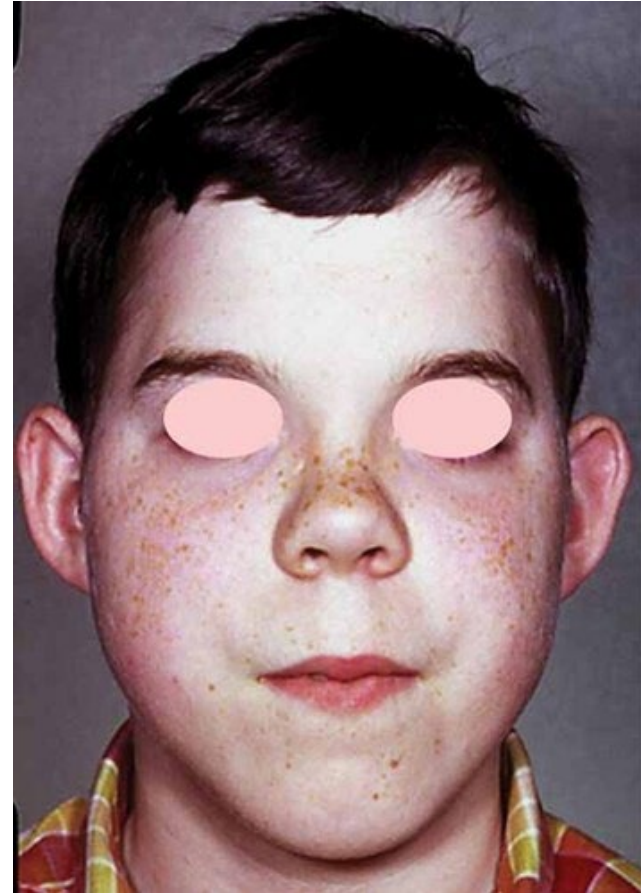


Full cheeks



Midface retrusion

Midface prominence





Cheekbone
Underdevelopment



Malar Flattening



Periorbital Region

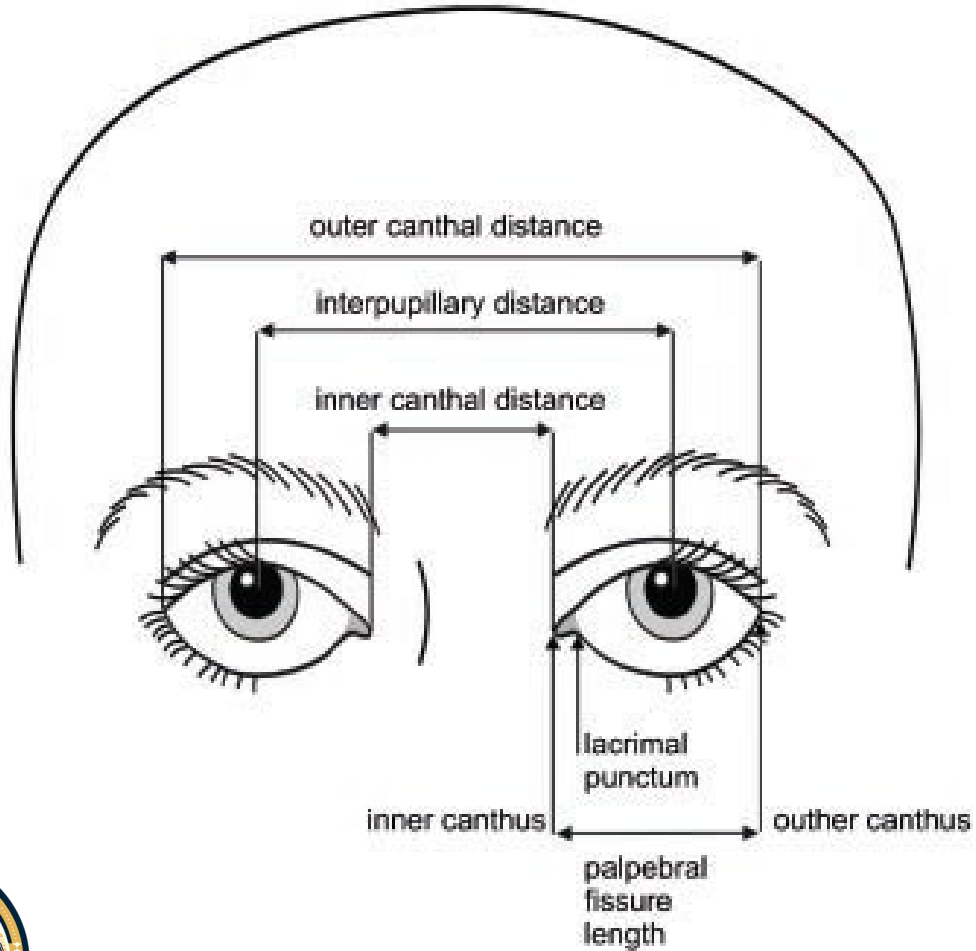
Periorbital Assessment

- Palpebral fissure length (short/long)
- Palpebral fissure slant (up/down)
- Epicanthic folds - a fold of skin which arcs from below the eye into the upper lid
- Eye spacing (use a rough guide of 1:1:1 for the ratio of left palpebral fissure length: inner canthal distance: right palpebral fissure length)
- Palpebral fissure shape
- Red reflex
- Iris color
- Pupil shape
- Retina
- Globe position (assessed from lateral view: protuberant vs deep set globes)



Orbital Spacing

b



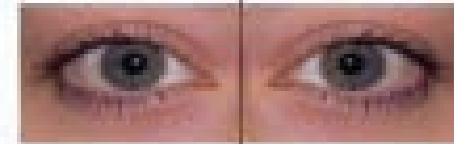
cyclopia



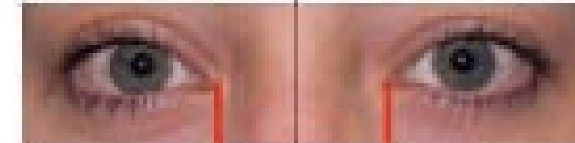
hypotelorism



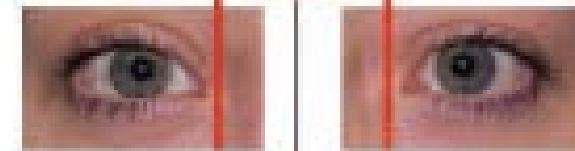
normal



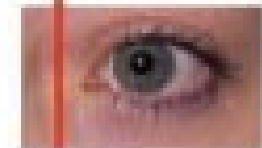
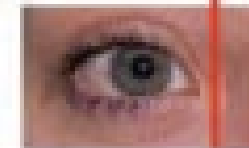
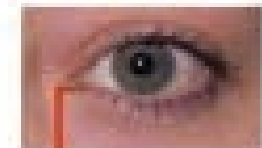
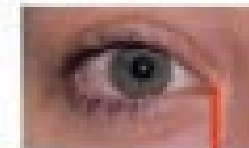
telecanthi



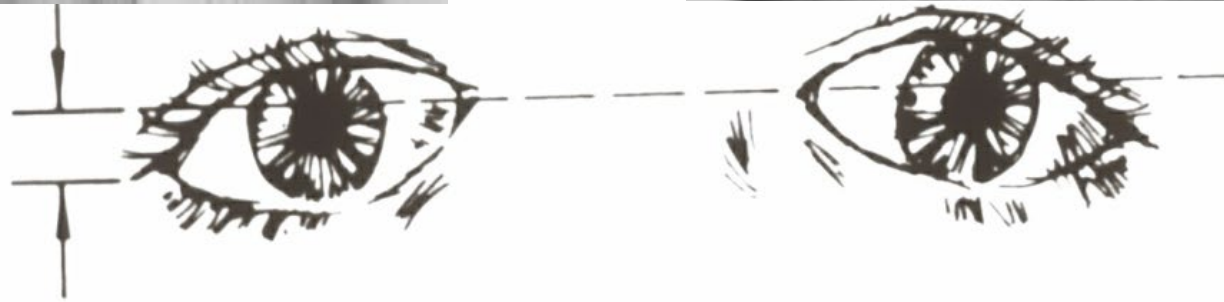
hypertelorism



hypertelorism + telecanthi



Palpebral Fissure



Examples of Periorbital Variation



Epicanthal folds



Deep set



Ankyloblepharon



Synophrys



Blepharophimosis



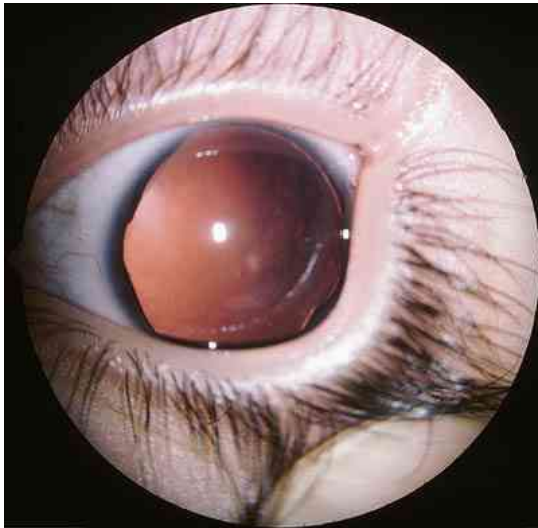
Hypertelorism



Examples of Orbital Variation



Coloboma



Aniridia



Nasal Region

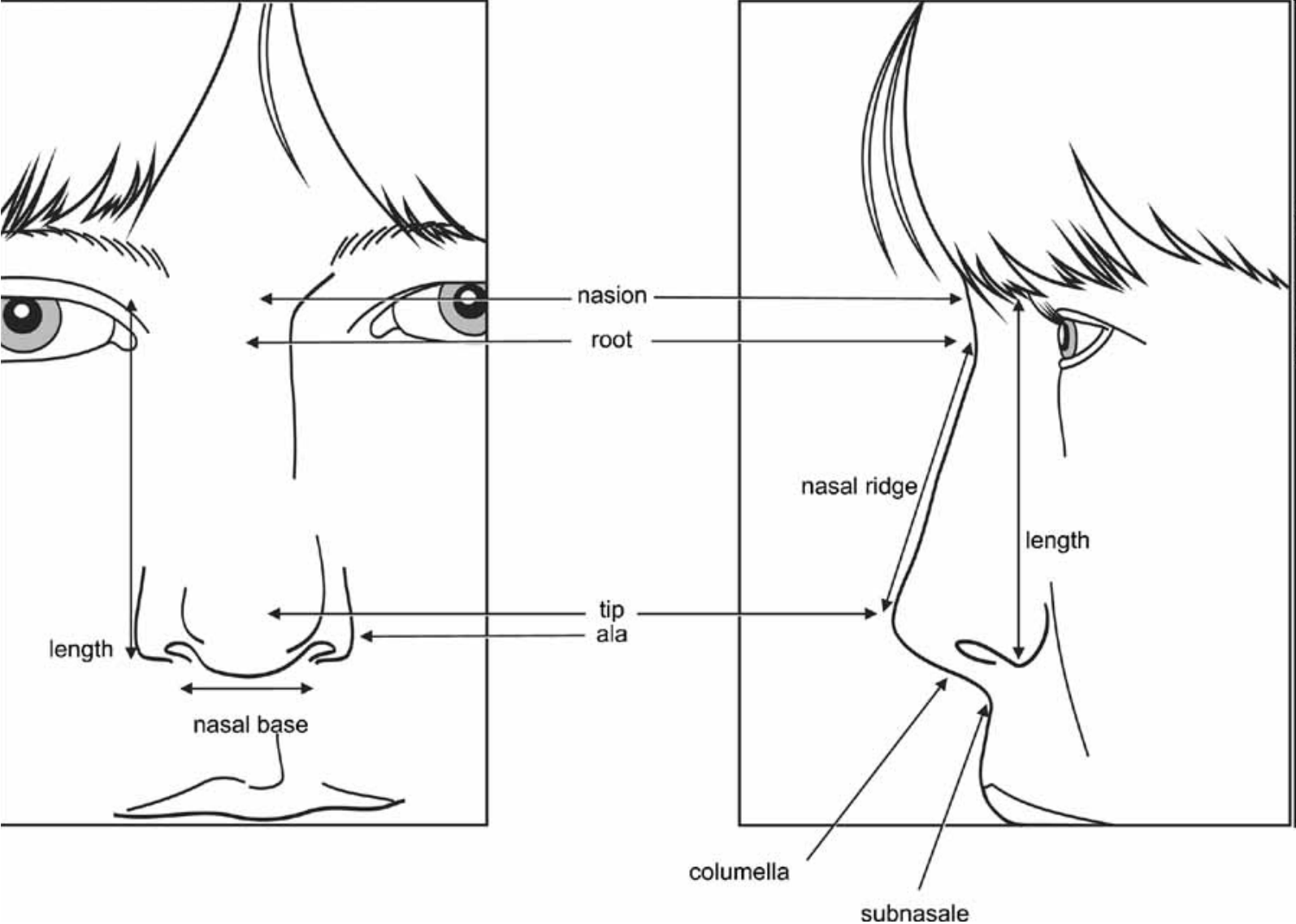


Nasal examination

- Examine the front and side to assess the length and width
- Divide the nose into three sections from the lateral view from superior to inferior into the nasal root, bridge and tip:
 - root
 - bridge (depressed/prominent/broad)
 - tip (broad/narrow,/bulbous)
 - columella (overhanging/low-insertion/short)
- nostrils - patency, position



Nasal Anatomy



Examples of Nasal Variation



Depressed
Nasal
Bridge

Depressed Nasal Ridge



Prominent
Nasal
Bridge

Broad Nasal Bridge



Examples of Nasal Tip Variation



Oral Region

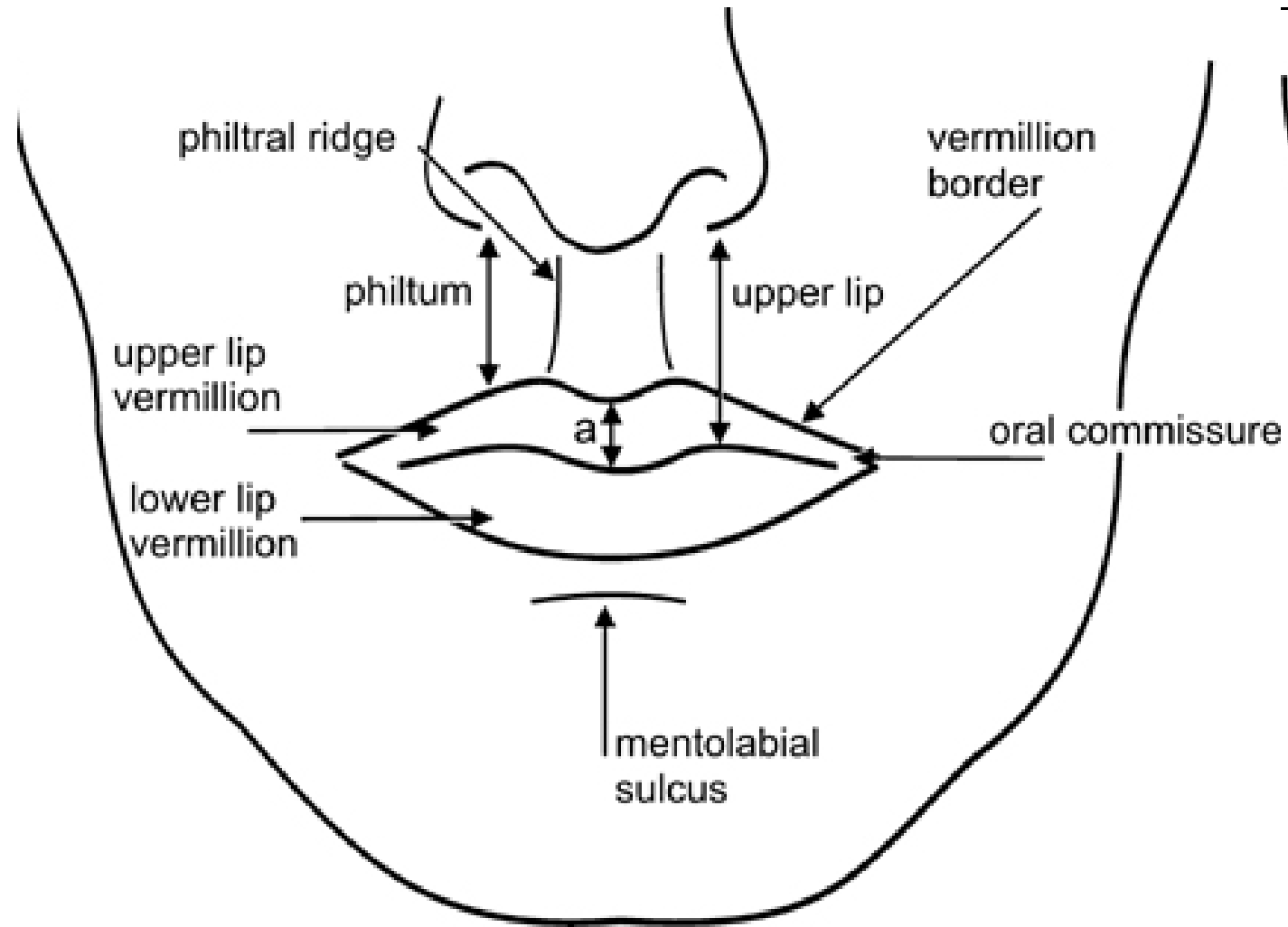


Oral Examination

- Examine size, symmetry, and shape
- Lip shape, thickness
- Philtrum definition and length
- Jaw position (prognathia/micrognathia)
- Oral cavity—palate, natal teeth, tongue size



Philtrum and Lip anatomy



Philtrum Variations

- Smooth/flat philtrum
- Long philtrum
- Short philtrum
- Prominent with deep groove



Flat philtrum, thin upper



Examples of Mouth Variation



Cupid's bow



Tented or "carp shape"



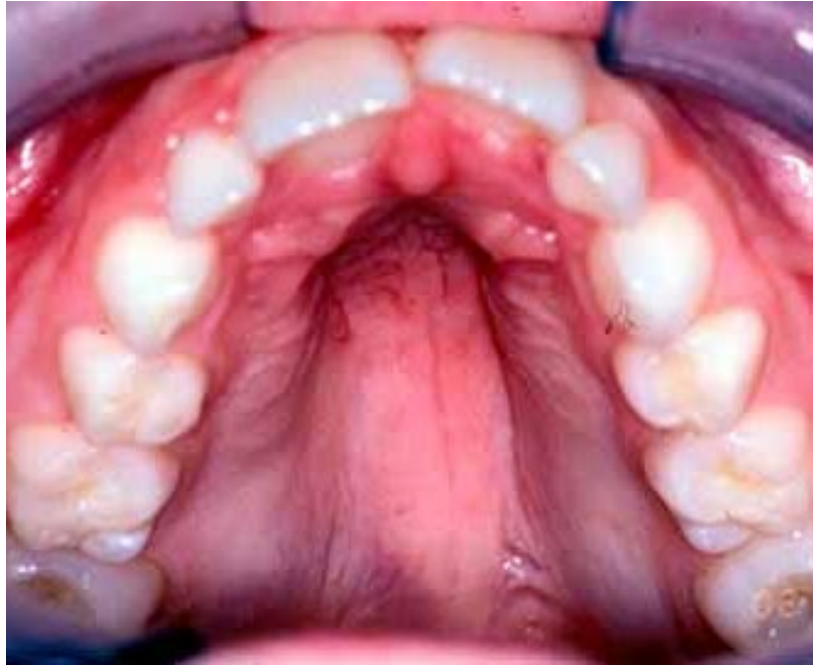
Thin vermilion border
"thin upper lip"



Macro or Microstomia



Examples of Oral Variation



High arched
palate



Bifid Uvula



More Oral Variation



Bifid tongue



Large tongue

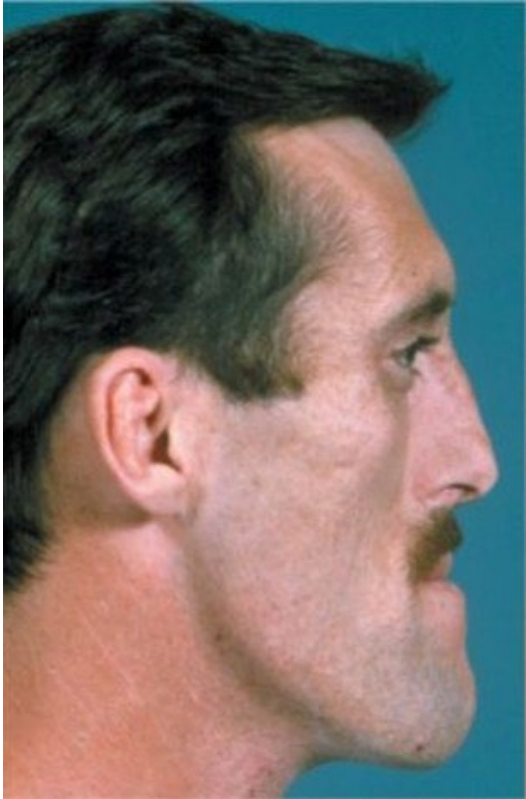


Lobulated tongue



Natal teeth

Examples of Chin Variation



Prognathism



Micrognathia



Retrognathia



Ear

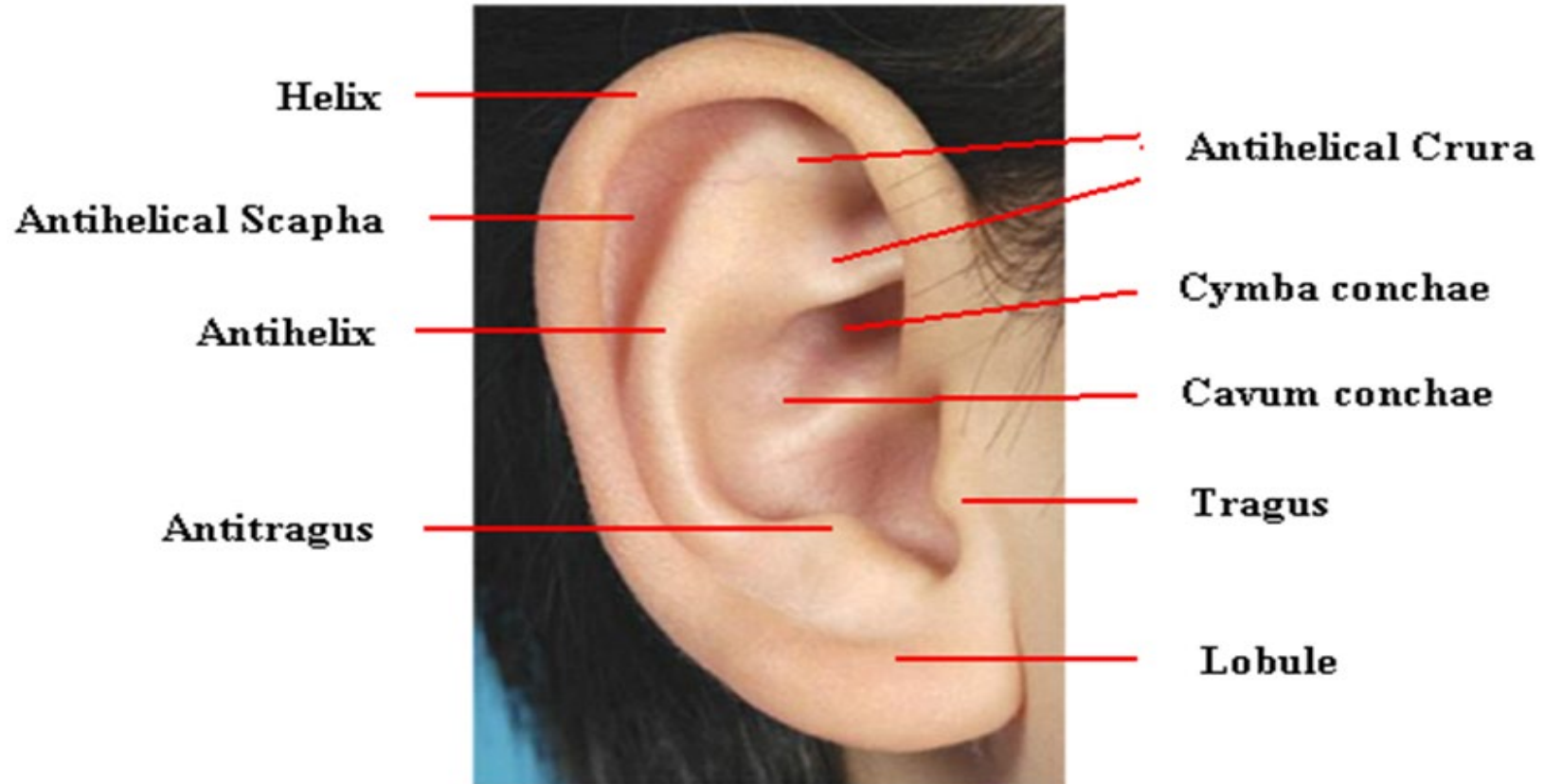


Ear exam

- Includes assessment of:
 - Size
 - Placement including position and rotation
 - Structure
 - Look for abnormalities of the preauricular region including tags and pits.
 - Evaluate the helix, antihelix, tragus, lobe, and external auditory meatus.
 - Both ears should look alike

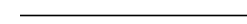
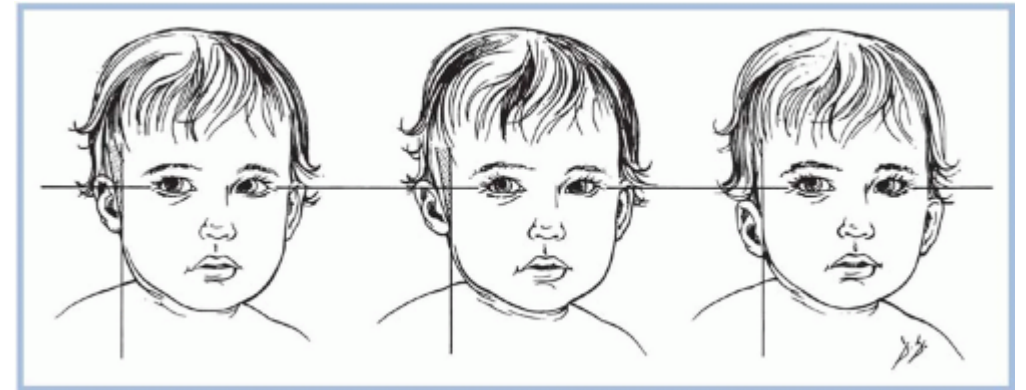
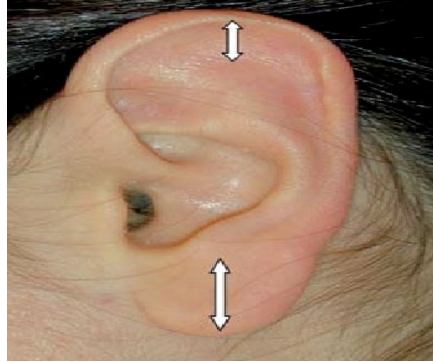


Anatomy of the Ear



Abnormalities

- Variation in size
 - Macrotia, Microtia, Anotia.
- Variation in position
 - Low-set ears, posterior angulation of the ear
- Variations of the individual anatomical part



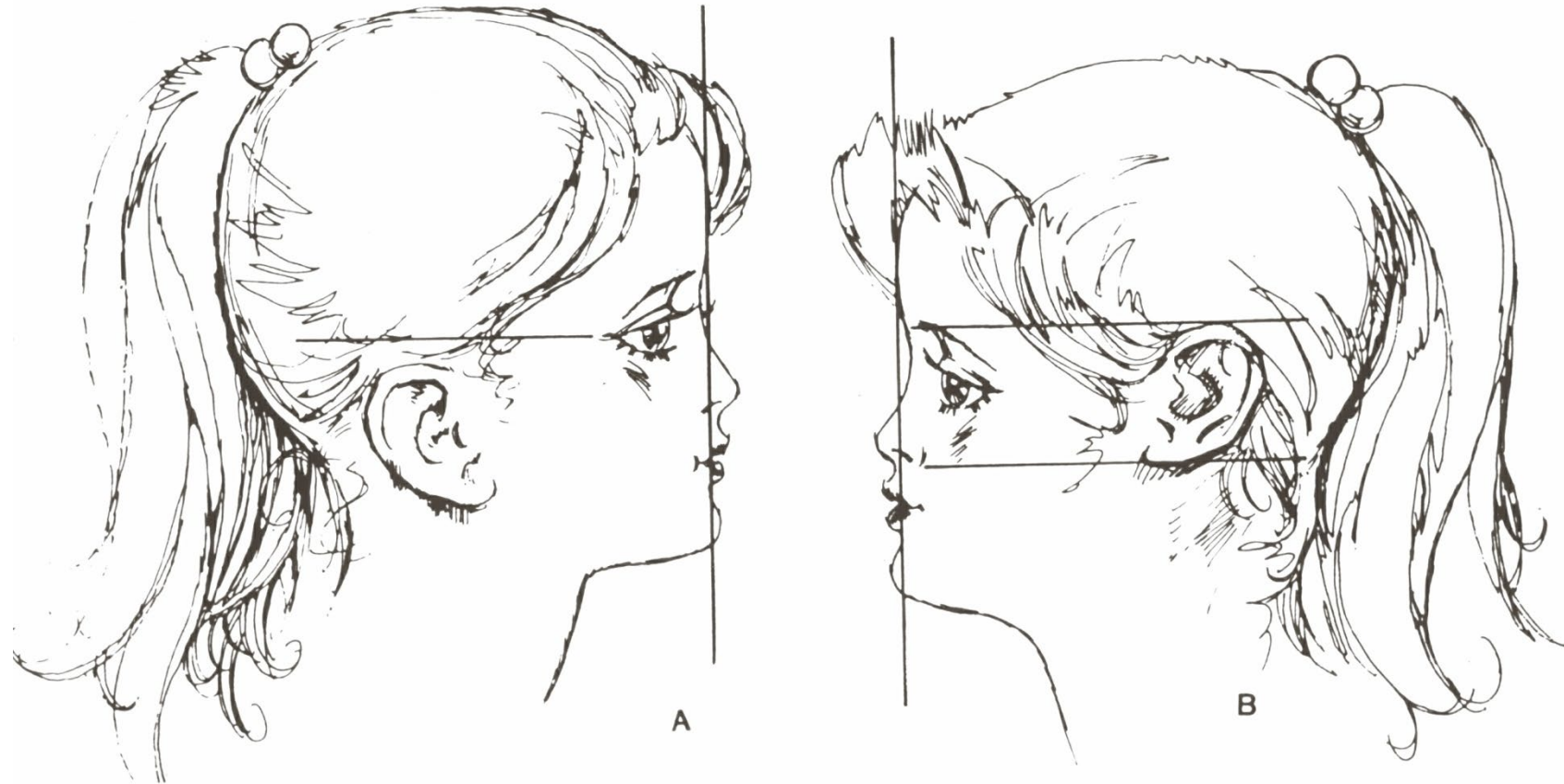
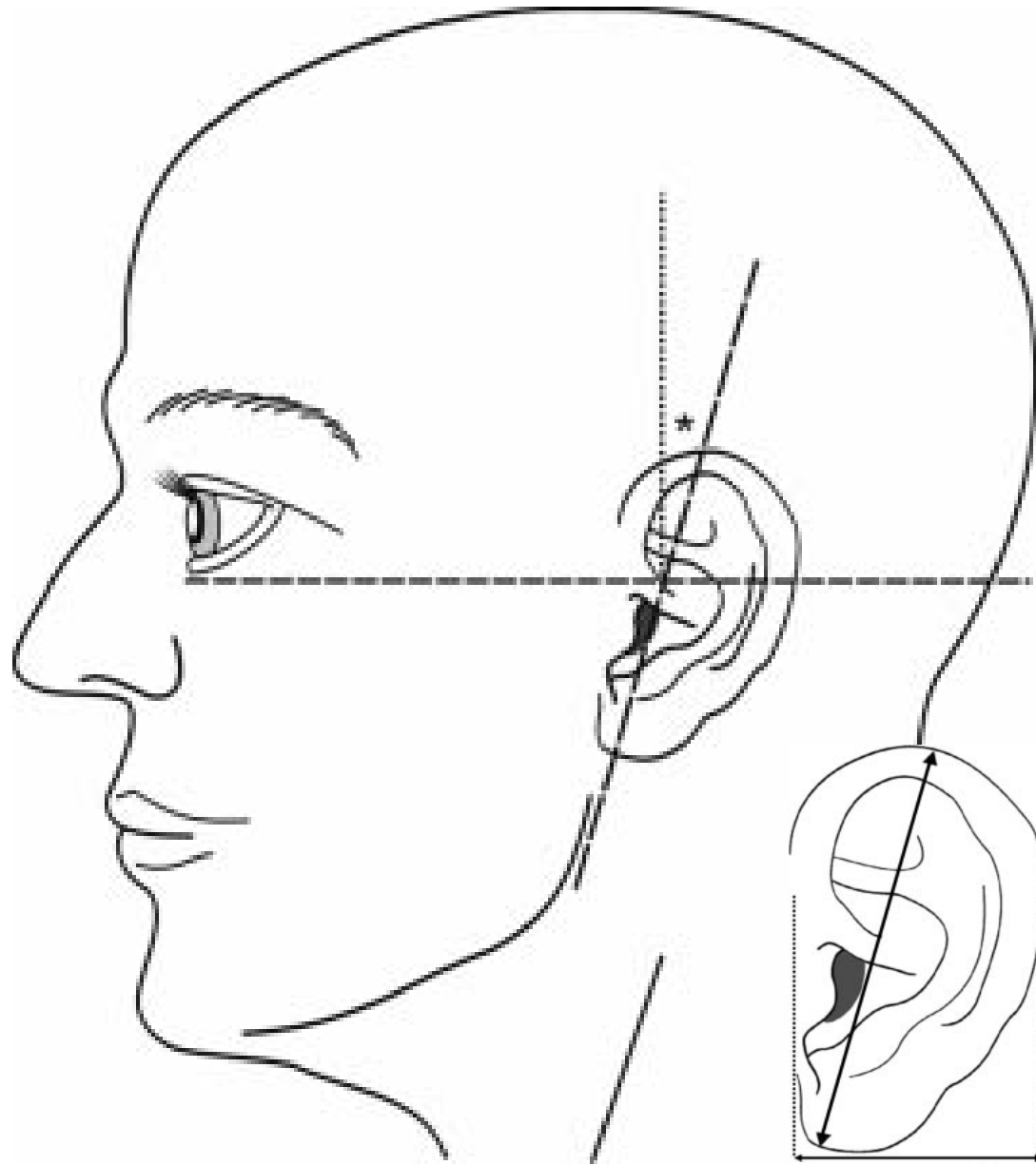


Figure 4.3.3. Methods for clinical assessment of ear position and size. (A) A line perpendicular to the facial plane, passing backward at the level of the outer canthus of the eye, should intersect the root of the helix where it joins the skin of the lateral scalp. This ear is low set and posteriorly rotated. (B) The upper and lower limits of the pinna normally lie at the level of the eyebrow and the base of the alae nasi, respectively.





Variation in Lobe Development



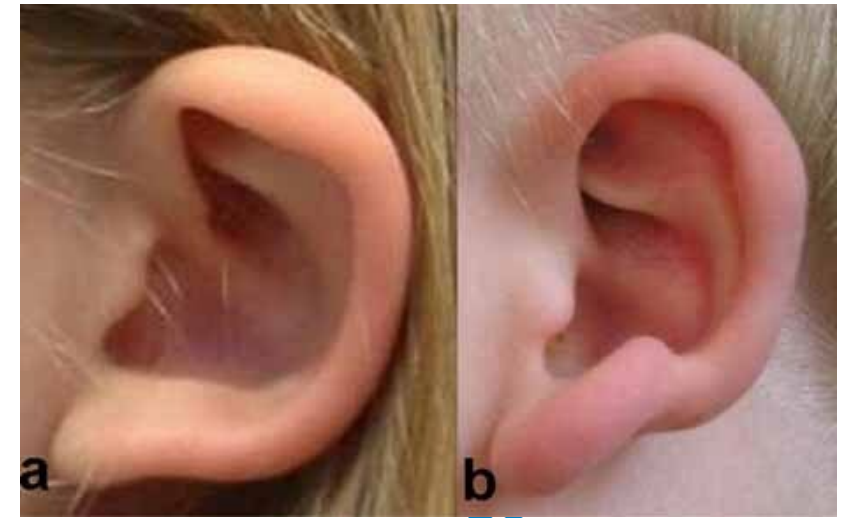
Small



Absent



Anterior creases



Uplifted



Helical Variation



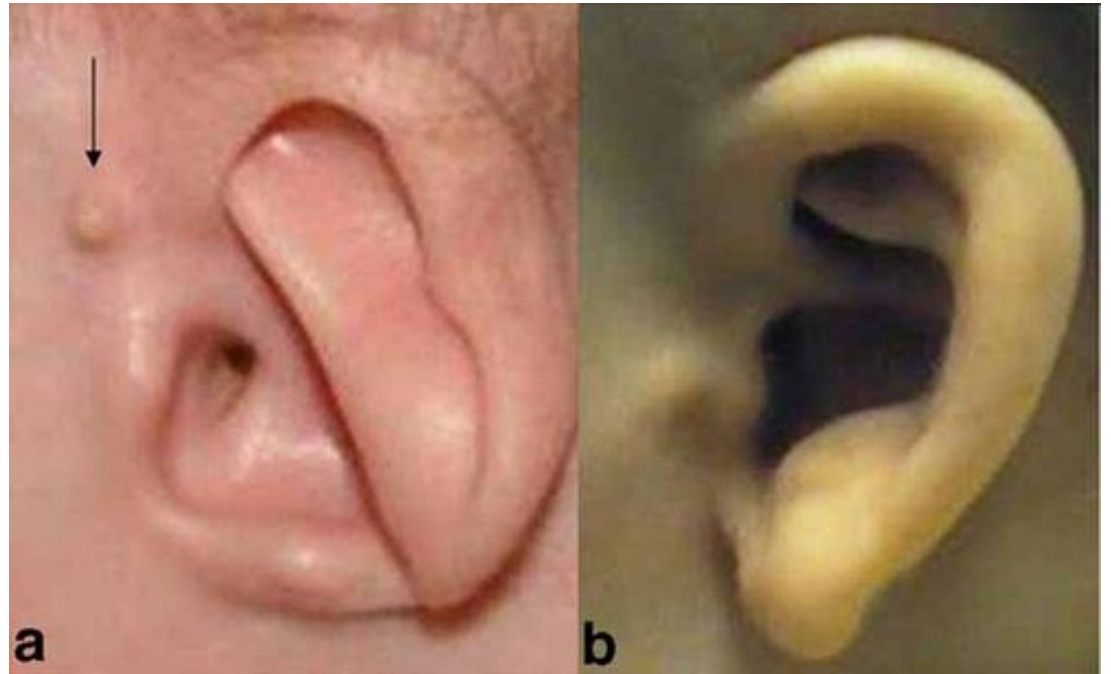
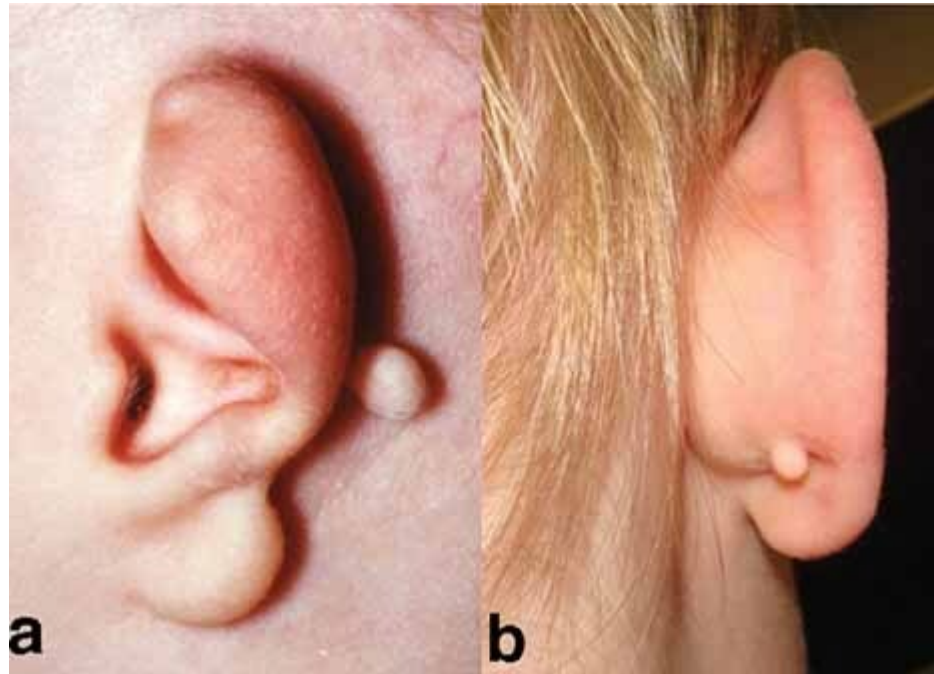
Lop ear



Satyr ear



Ear Tags



Ear Pits



Auricular pit



preauricular pit



Helical pit

More Significant Involvement



Hand, Fingers, and Feet



Assessment

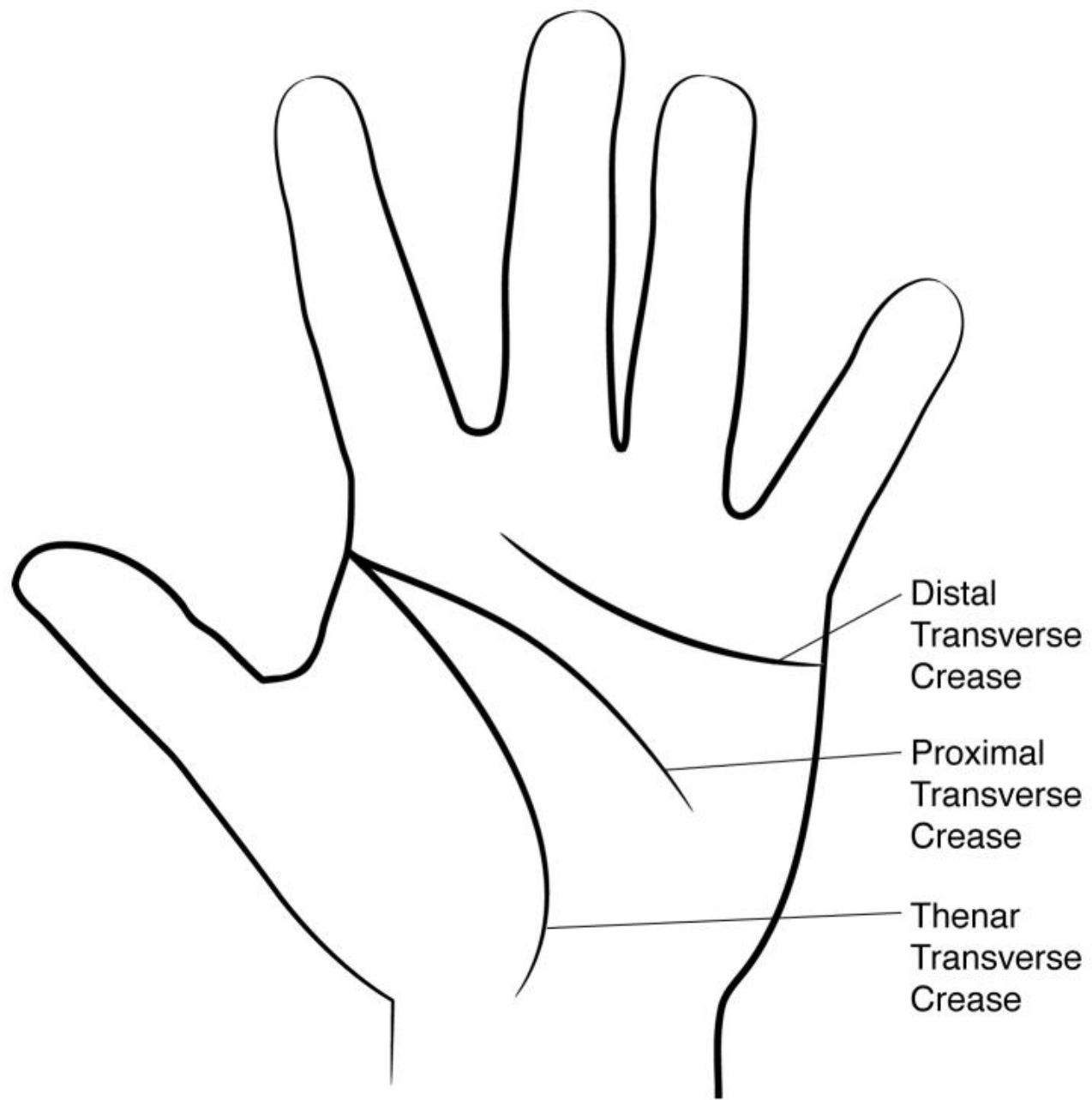
- Inspect the fingers from the palmar and dorsal surfaces
- Inspect for polydactyly & syndactyly
- Looking down on a fistled hand allows easy visualization of the metacarpal lengths

- Inspect finger shape



- Finger lengths should be symmetrical on both hands





Hand Variation

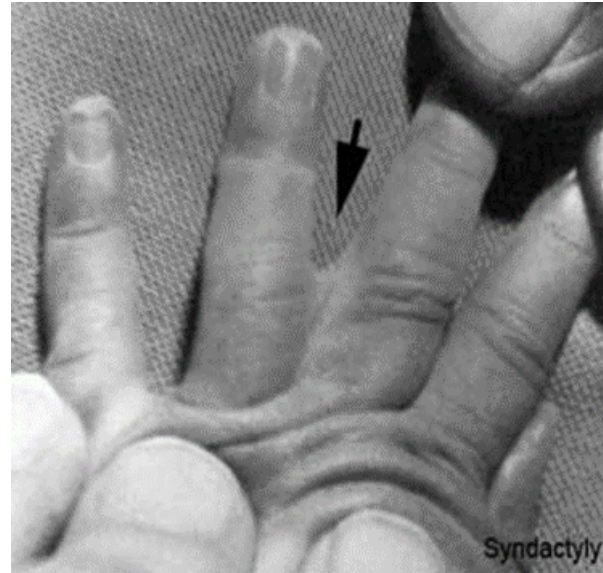


Clinodactyly

Syndactyly



Partial cutaneous



Complete cutaneous



Polydactyly



Preaxial



Mirror Image



Mesoaxial



Postaxial



Finger Variation



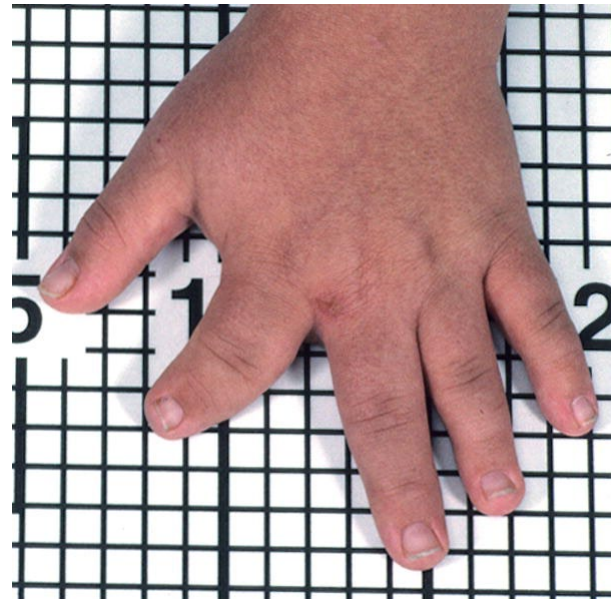
Overlapping fingers



Tapered fingers



Short fingers



Radial deviation



Foot Variation



Sandal Gap



Metatarsus adductus



Rocker bottom



Hypoplastic 5th nail



Significant Variation



Joints and skeleton

- Contractures
- Limb shortening
- Joint range of movement
- Soft tissue webbing across joints (pterygium)
- Sternum length and shape (pectus carinatum / pectus excavatum)
- Shape of thoracic cage
- Spine length, straight/curved
- Neck length, webbing

Examples of Neck Variation



Short neck



Redundant nuchal skin



Examples of Thoracic Variation



Short



Long



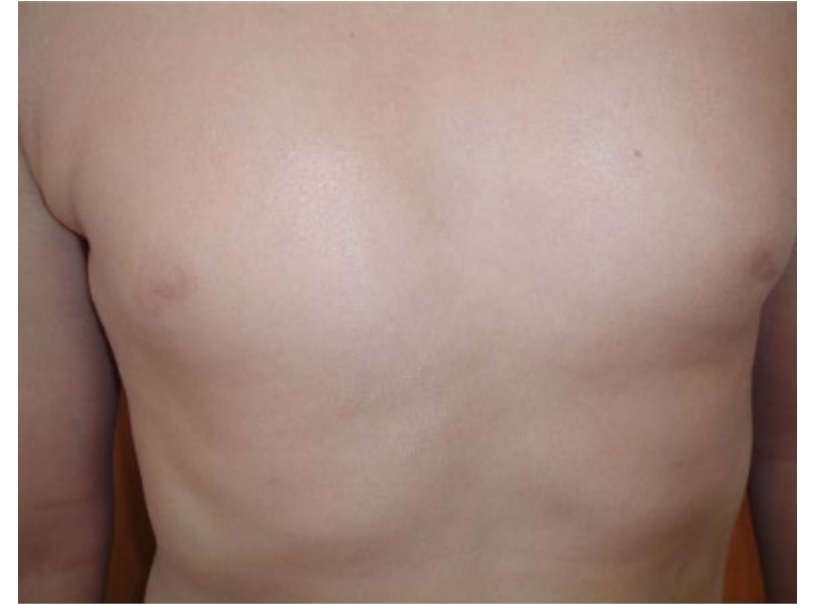
Broad



Examples of Chest Variation



Supernumerary nipple



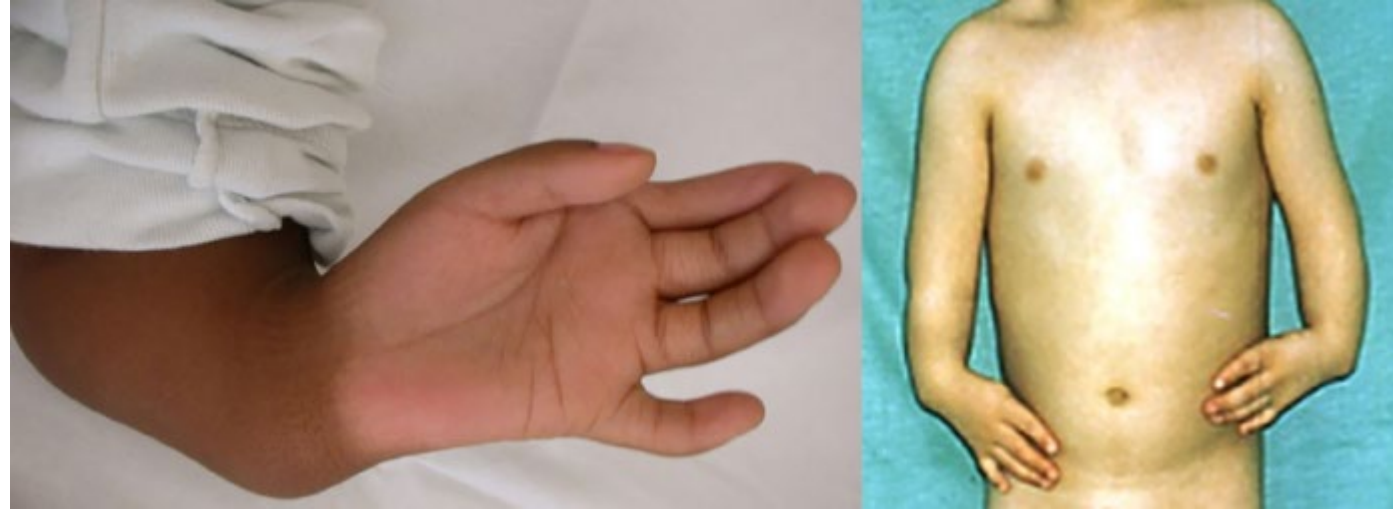
Widely spaced nipples



Examples of Limb Variation



Proportionately short



Mesomelic



Rhizomelic



Genitalia and Anus

- Phallus size, morphology
- Development of scrotum and palpation of testes
- Development of labia
- Position of anus relative to genitalia
- Patency of anus
- Note any genital ambiguity



Examine Family Members

- Examination of other family members may help to determine whether features are dysmorphic or familial.



Sources

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9. <https://hpo.jax.org/>



Dysmorphology Tools for NICU Providers

- Dysmorphology exam checklist: <https://www.safercare.vic.gov.au/best-practice-improvement/clinical-guidance/neonatal/dysmorphology-assessment-in-neonates>
- HPO website for appropriate word usage: <https://hpo.jax.org/>
- Elements of Morphology: Standard terminology for the: (search in PubMed, includes terms and pictures!)
 - Head and face
 - Periorbital region
 - Ear
 - Nose and philtrum
 - Lips, mouth, and oral region
 - Trunk and limbs
 - Hands and feet
 - External genitalia



Questions

