NNP Genetics Education

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Objectives

1. Review genetic testing orders… in EPIC!
2. Discuss the discipline of dysmorphology and how to approach these findings clinically.
3. Introduce appropriate terminology for both typical features and dysmorphic features.

No disclosures.

Genetic Testing Review

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AMA Joy in Medicine Recognized Organization Gold 2022–2026

Best Children's Hospitals U.S. News Ranked in 9 Specialties 2025–2026

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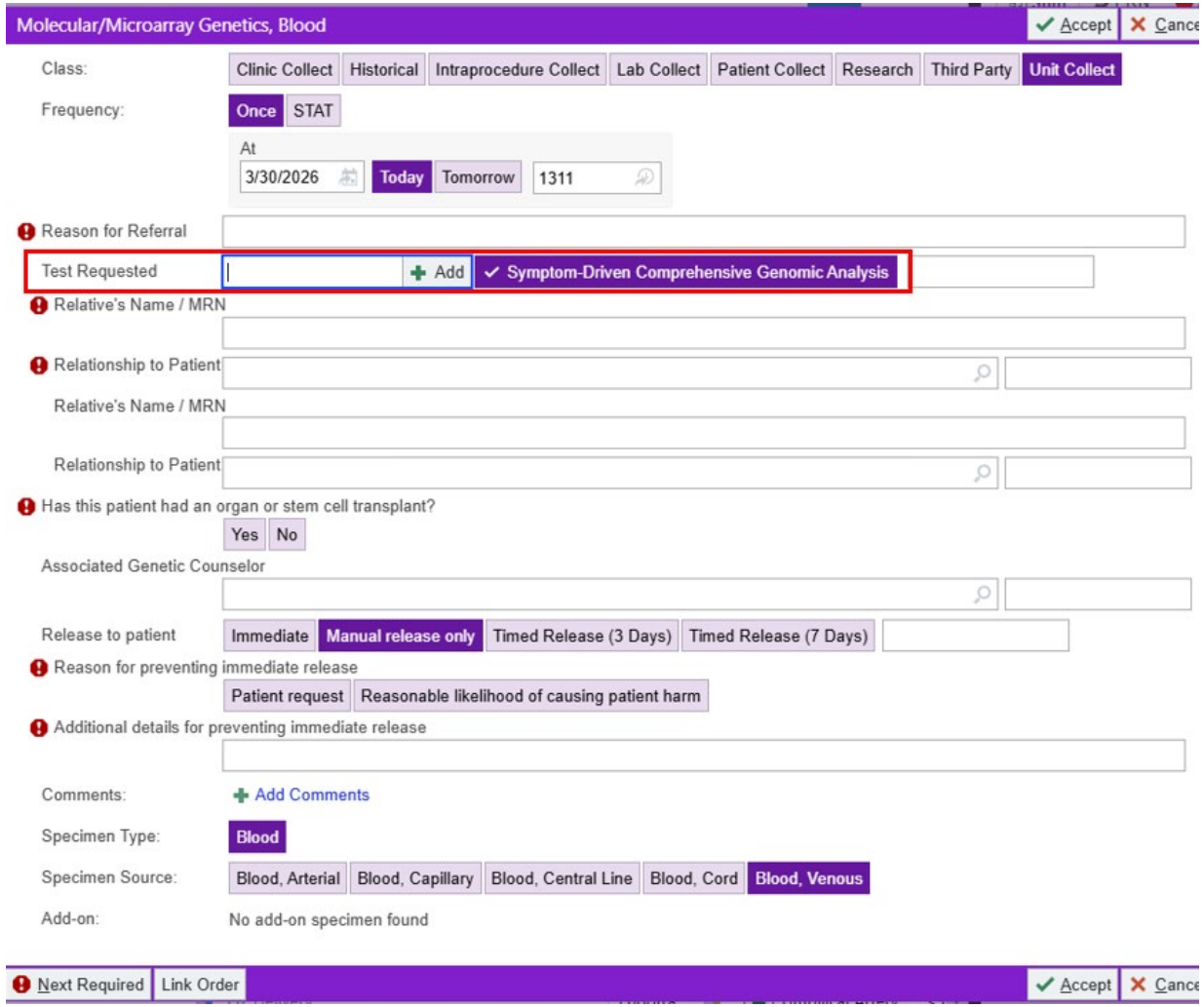
What do we order now?

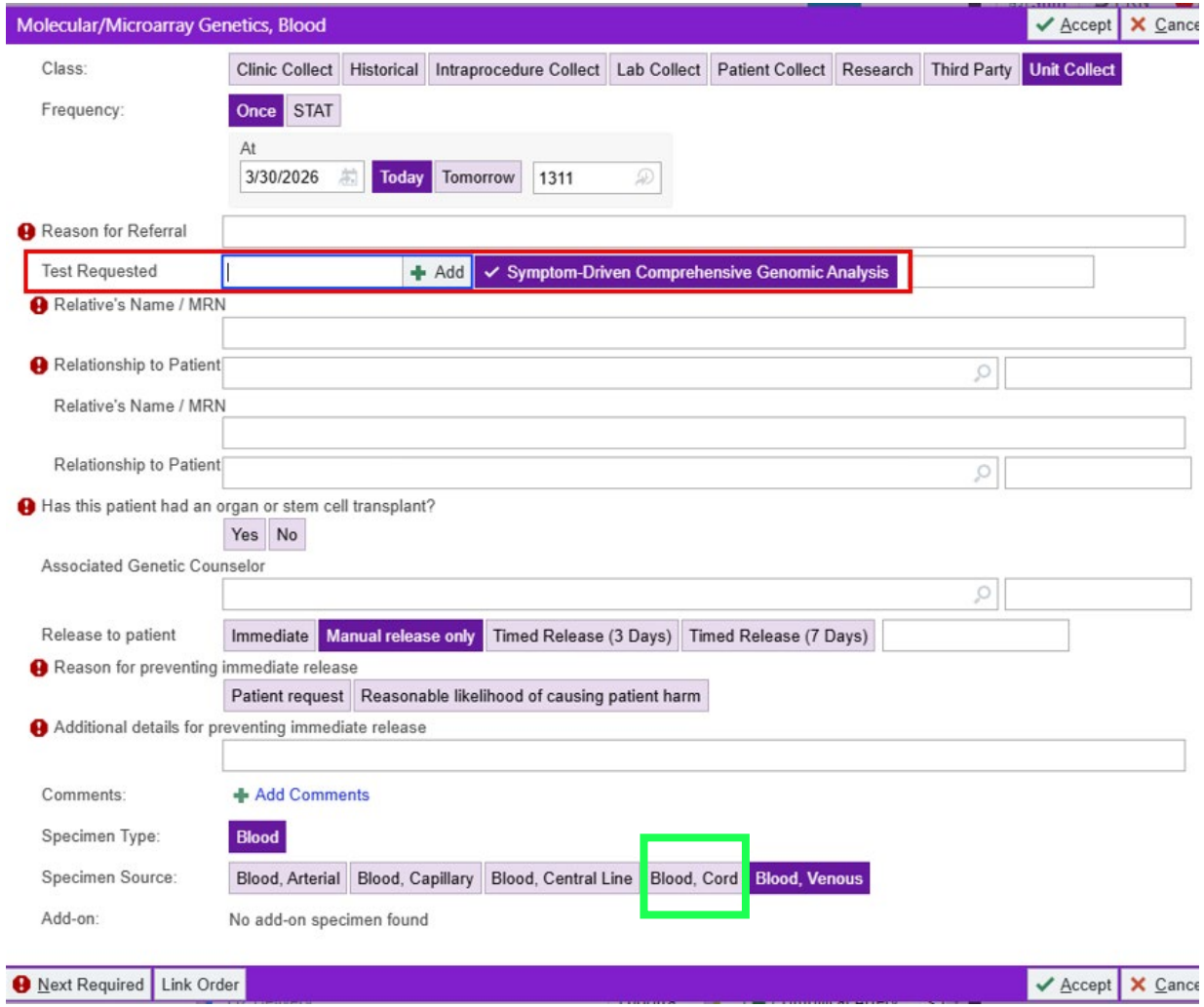


- Most common order in the NICU: Symptom-driven comprehensive genomic analysis
- With this selection, new required fields pop up for parent names and MRNs
- PLEASE do not change the release from “Manual release only”

Molecular/Microarray

- Name of the order because it includes DNA-based tests and the microarray, which is a chromosome test
  - This does not mean that the baby has a microarray pending!
  - These tests are lumped together because the sample should be collected in a lavender top (EDTA) tube
- DNA isolation is still an option
  - Add on ordering from a previously obtained sample is available
- New box to select the Genetic Counselor who is following the patient

A Note about Samples…



- Cord blood can be a great option if the baby is not going to have access or other labs
- Cord blood samples can be affected by maternal cell contamination and may need additional processing

Cytogenetics

- The order is still “Cytogenetics, Blood”
- This testing is used for FISH and karyotypes
- For FISH, type in the chromosome of consideration
  - 13, 18, 21, X, Y
- Different tube is used for sample collection -> Green top, sodium heparin tube

Dysmorphology

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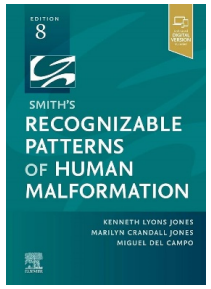
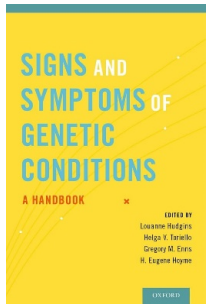
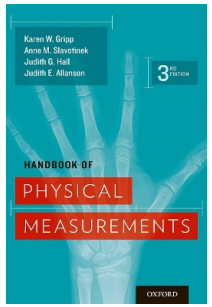
What is Dysmorphology?

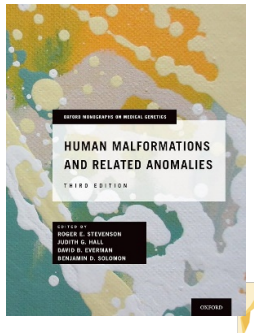
- Term coined in the 1960s by Dr. David W. Smith
- Discipline of clinical genetics that studies and attempts to interpret the patterns of human growth and structural defects
- Historically, the main tool for diagnosis of a genetic disorder

- In practice: explaining abnormal physical features by understanding the abnormal morphogenesis or anatomical development









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Terminology

National Human Genome Research Institute
Advancing human health through genomics research

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Human Malformation Terminology

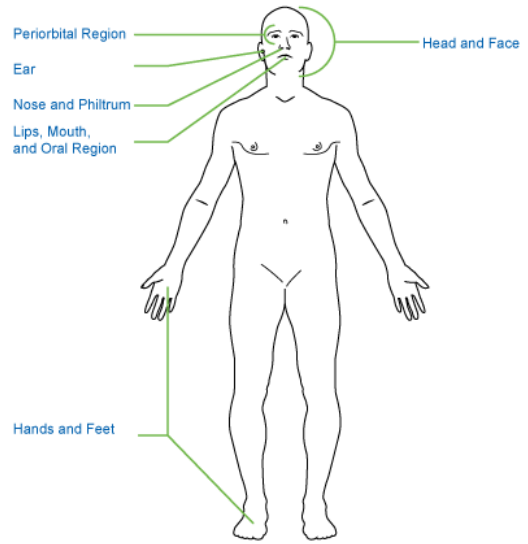
- Human Malformation Terminology Home Page
- Head and Face Terminology
- Periorbital Region Terminology
- Ear Terminology
- Nose and Philtrum Terminology
- Lips, Mouth, and Oral Region Terminology
- Hands and Feet Terminology
- References

Elements of Morphology: Human Malformation Terminology

An international group of clinicians working in the field of dysmorphology has initiated the standardization of terms used to describe human morphology. The goals are to standardize these terms and reach consensus regarding their definitions. In this way, we will increase the utility of descriptions of the human phenotype and facilitate reliable comparisons of findings among patients. Discussions with other workers in dysmorphology and related fields, such as developmental biology and molecular genetics, will become more precise. Here we describe the general background of the project and the various issues we have tried to take into account in defining the terms. Published 2009 Wiley-Liss, Inc.

This Web site contains six articles that describe the initial results of a project intended to develop accurate and clear definitions of terms for the craniofacies in general, the major components of the face, and the hands and feet [Allanson et al., [2009]; Biesecker et al., [2009]; Carey et al., [2009]; Hall et al., [2009]; Hennekam et al., [2009]; Hunter et al., [2009]]. These articles are the result of a significant amount of planning, organization, negotiation, review, and writing, while, at the same time, they are but a start.

Dysmorphology evolved from a small nucleus of clinicians in the 1950s into a recognized and widely practiced discipline, and more recently has incorporated translational research into developmental biology, molecular genetics, and metabolic medicine. The terms that clinicians use to describe a body part have gradually evolved in a haphazard and uncoordinated manner, and have not been critically reviewed. Clinicians and researchers have always made comparisons among patients and syndromes, and in the last decade it has become increasingly possible and necessary to use clinical data for studies of etiology and pathogenesis, epidemiology, the isolation of causative gene mutations, and for evaluation of interventions. Therefore, we need to have uniform and internationally accepted terms to describe the human phenotype.



The Netherlands

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Conversations in Practice

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Striking Features

- Compare to parents
  - Are features familial?
- Other symptoms
  - What is else is going on with the patient?
- Abnormal exam
  - Has the anterior fontanelle closed early?
  - Do you feel a big spleen and/or liver?
  - Is muscle tone abnormal?

What to Say

- Address other features first
  - Delays
  - Growth differences
  - Congenital anomalies
- Concern for a structural issue
  - Head shape concerning for craniosynostosis
  - Micrognathia concerning for feeding difficulties
- Age and cognitive status of the child matter
- Role of Genetics -> Looking for clues on the physical exam
  - That includes facial features too

Helpful Considerations

- If you are concerned about a baby being dysmorphic, do not place a consult only for this indication.
  - Is the baby growth restricted, are congenital anomalies present, is the head shape or anterior fontanelle abnormal, etc.?
- Do not tell the parents that we are consulted only because of dysmorphic features.
- Ethnicity and race greatly affects morphology.
- Facial features do NOT provide prognostic information.
- Please be kind -> we are ALL dysmorphic!
  - No FLK…

Terms

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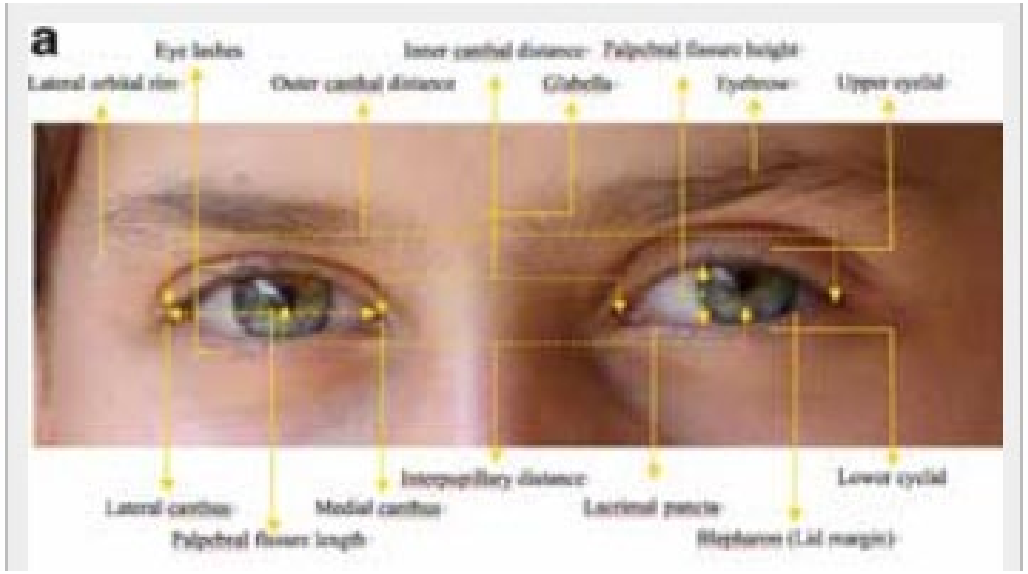
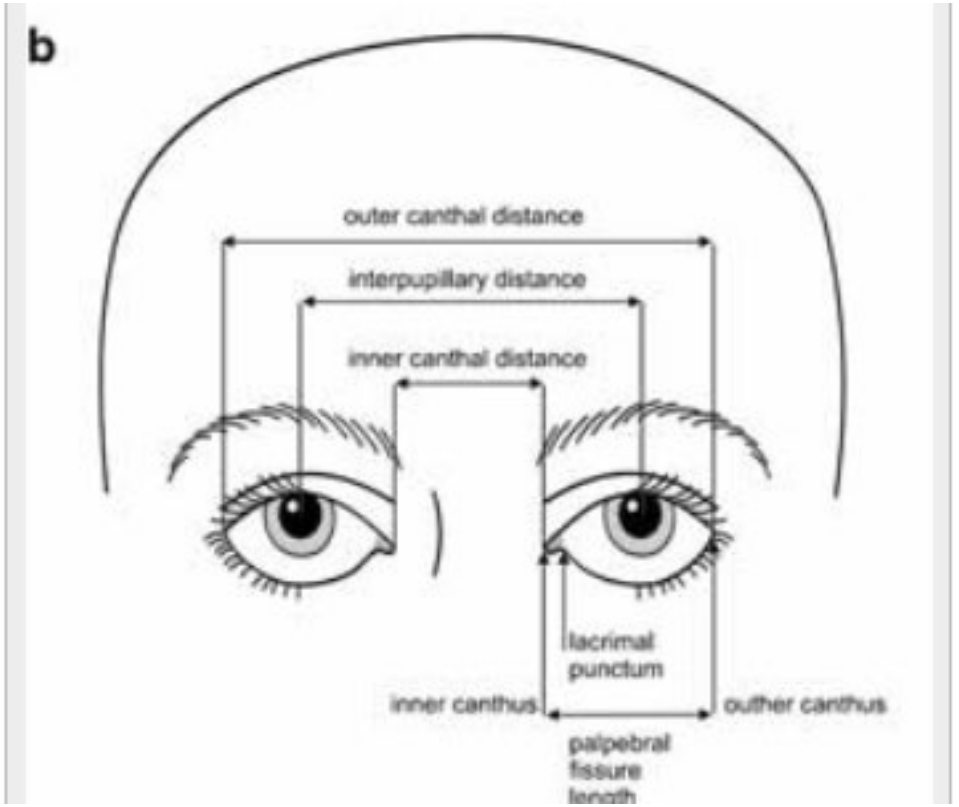
Periorbital Region

Landmarks





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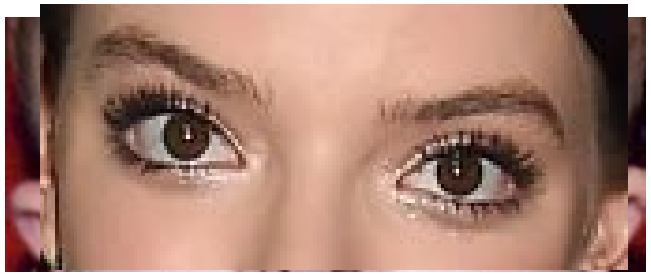
Eyes

Palpebral Fissures

Down-slanting

Up-slanting

Spacing



Hypertelorism

Hypotelorism

Other Findings

Epicanthic Fold

Deeply set

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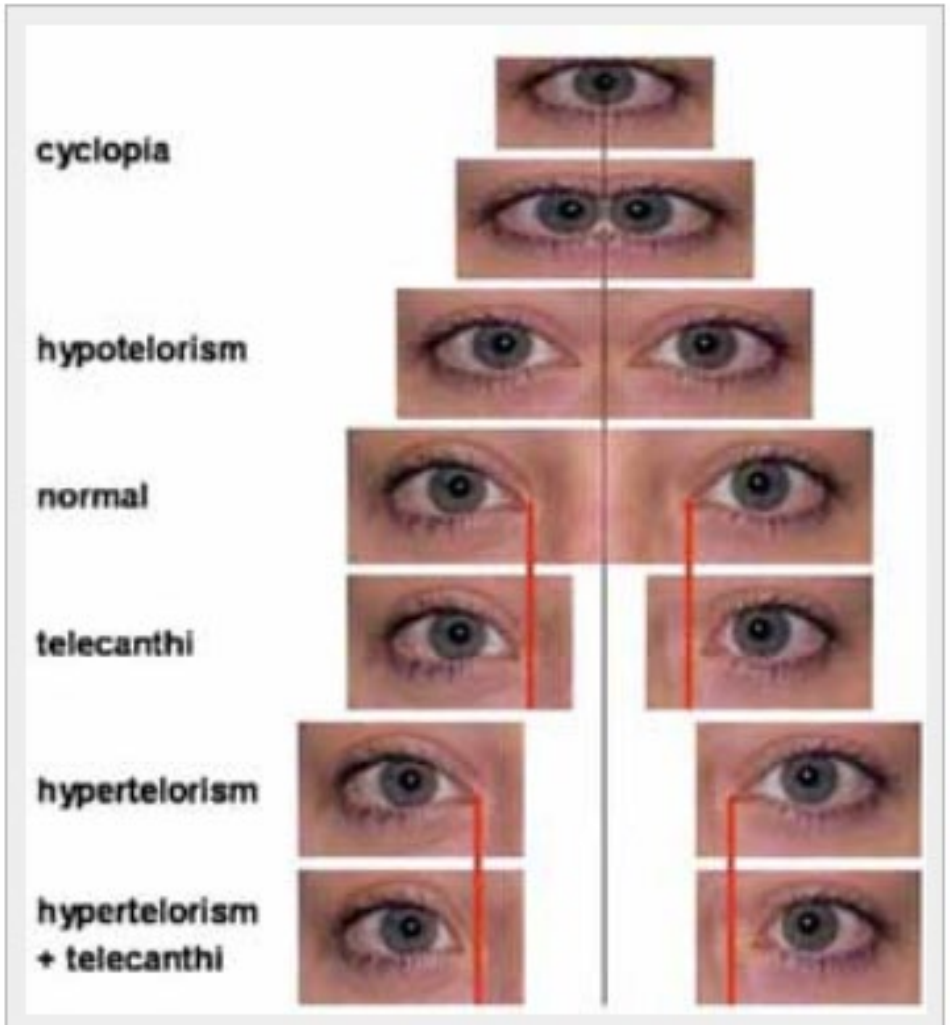
Eye Spacing



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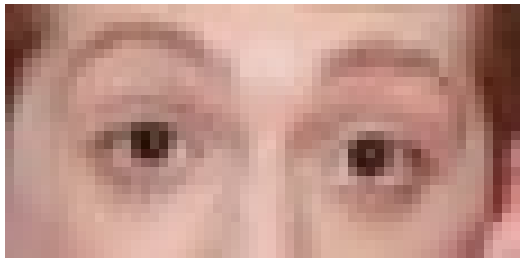
Eyebrows and Eyelashes



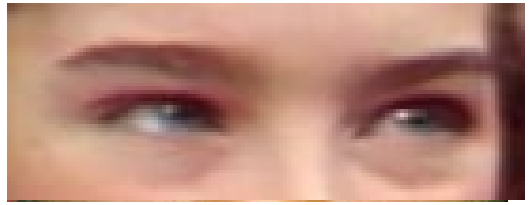
Highly Arched

Synophrys

Prominent Eyelashes

Long Eyelashes

Straight



Thick

Sparse Eyelashes

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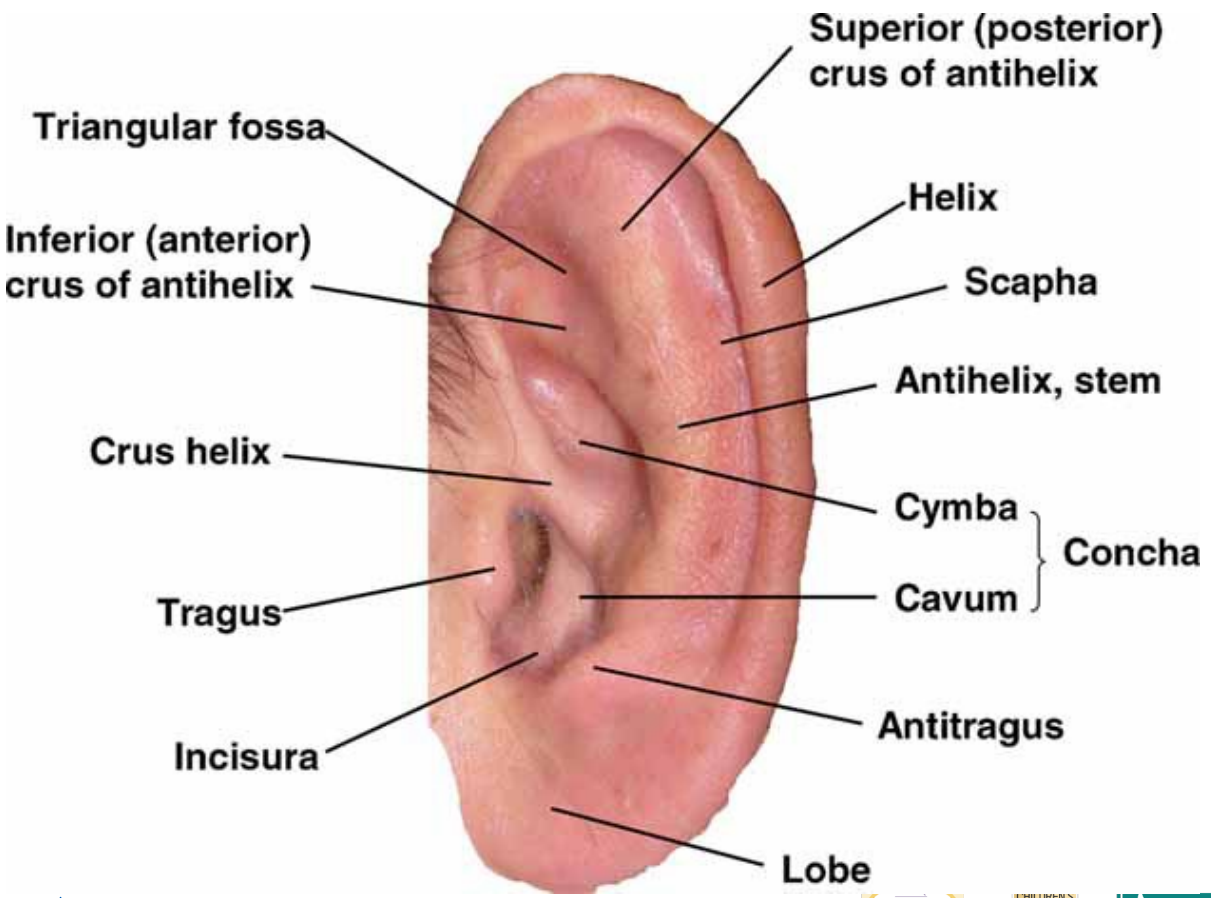
Ears

Landmarks



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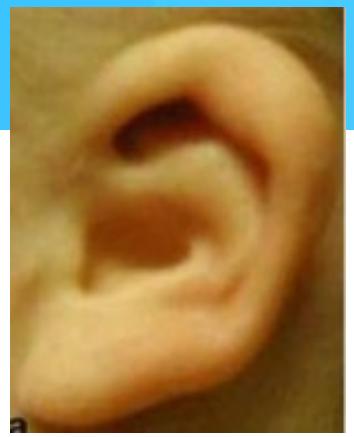
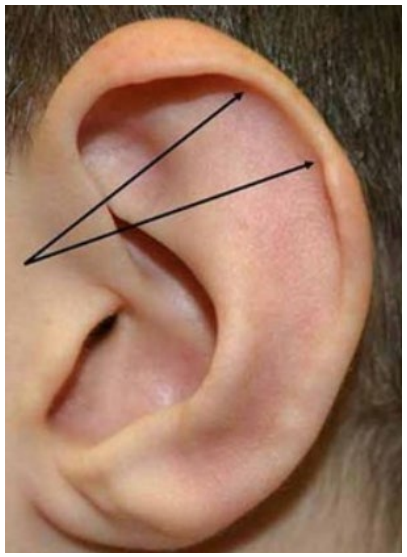
Ears

Darwinian Notch

Overfolded Helix



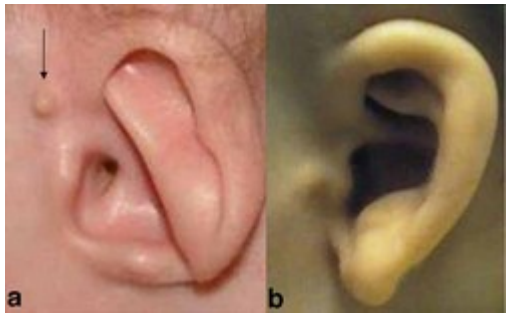
Absent Tragus



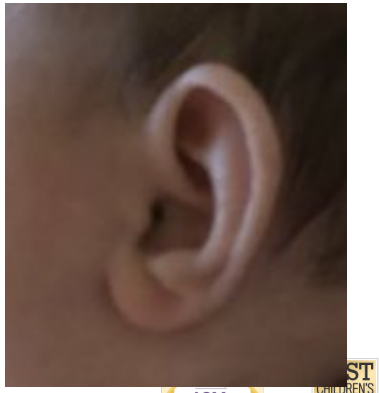
Underfolded Helix

Prominent Tragus

Preauricular Pit



Preauricular Tag



Ear Lobe Crease

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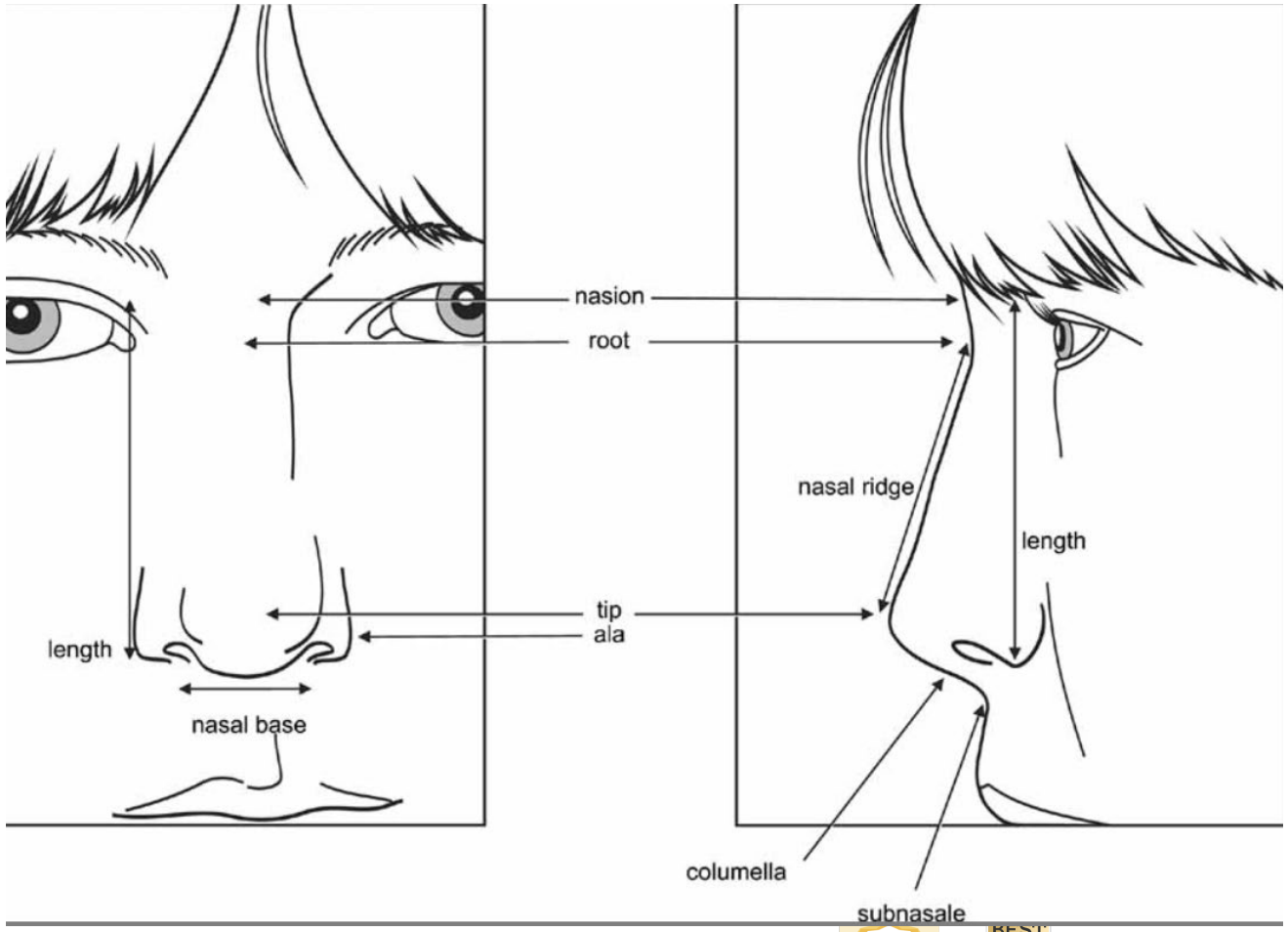
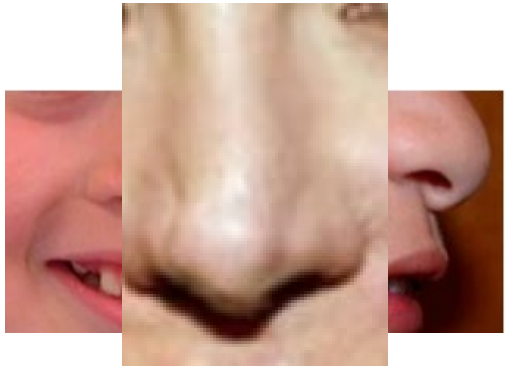
Ear Placement

Nose

Landmarks

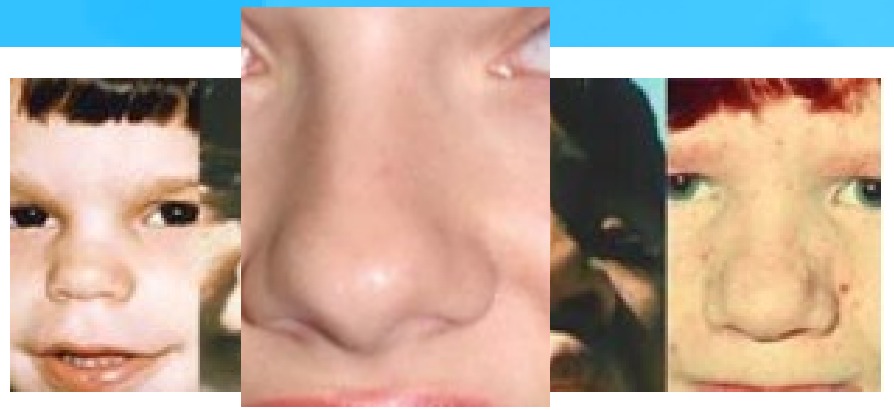

nasion
root
nasal ridge
length
tip
ala
length
nasal base
columella
subnasale

Nose



Low Hanging Columella

Anteverted Nares



Bulbous Nose

Wide Nasal Bridge

Thick Ala Nasi

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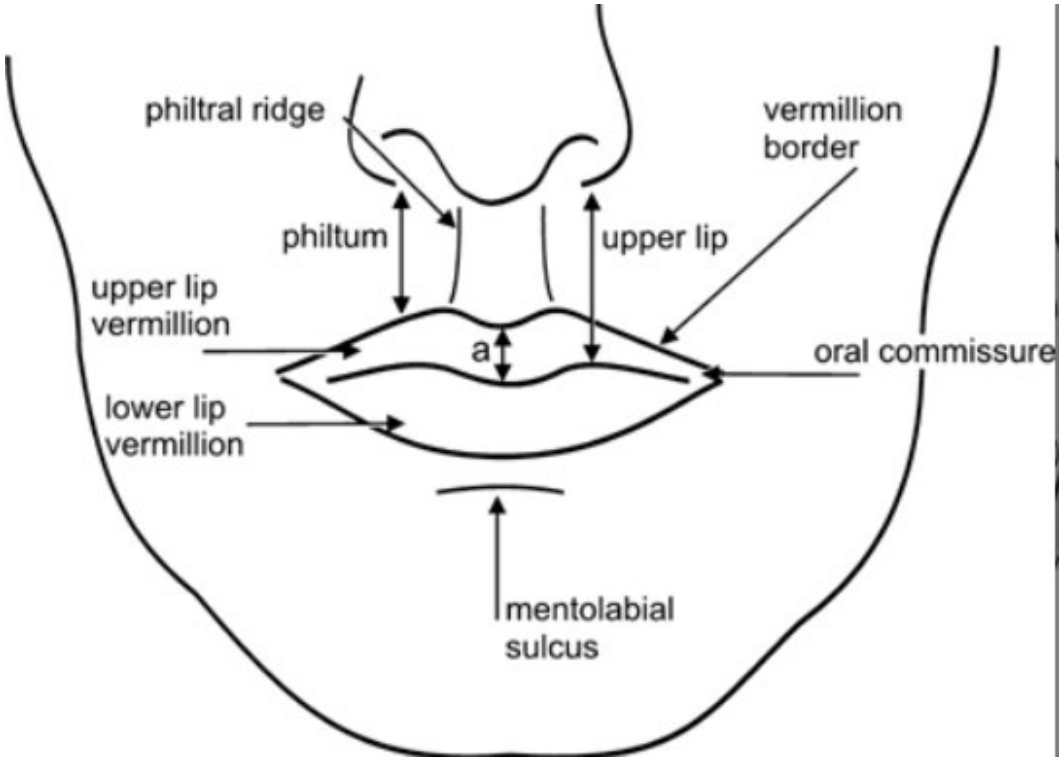
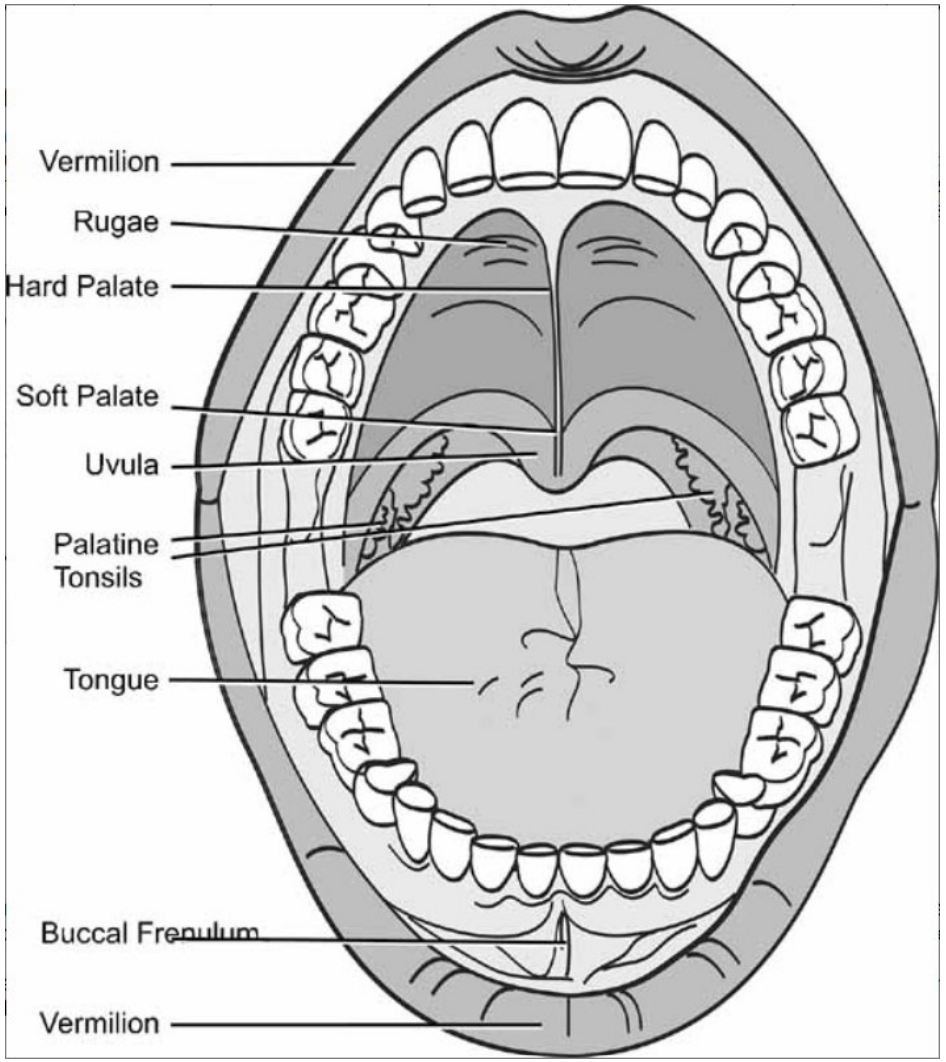
Mouth

Landmarks





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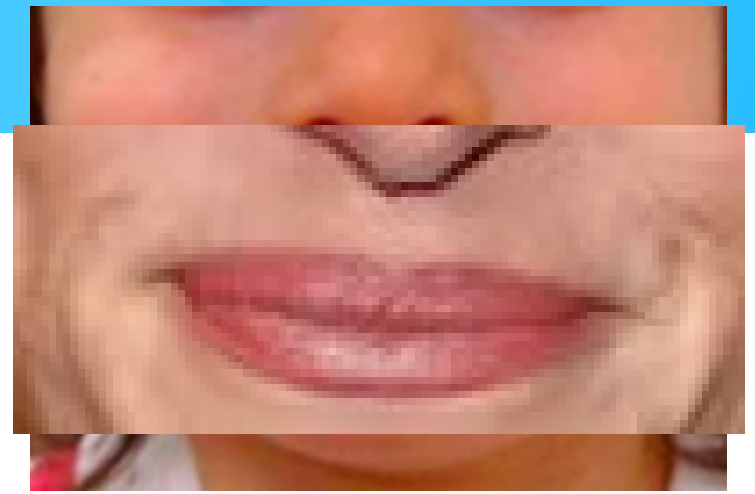
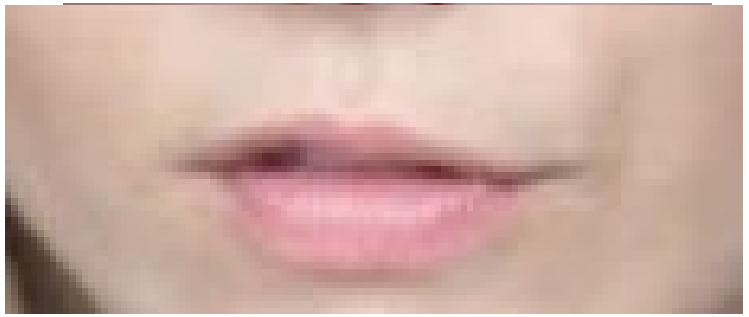
Mouth

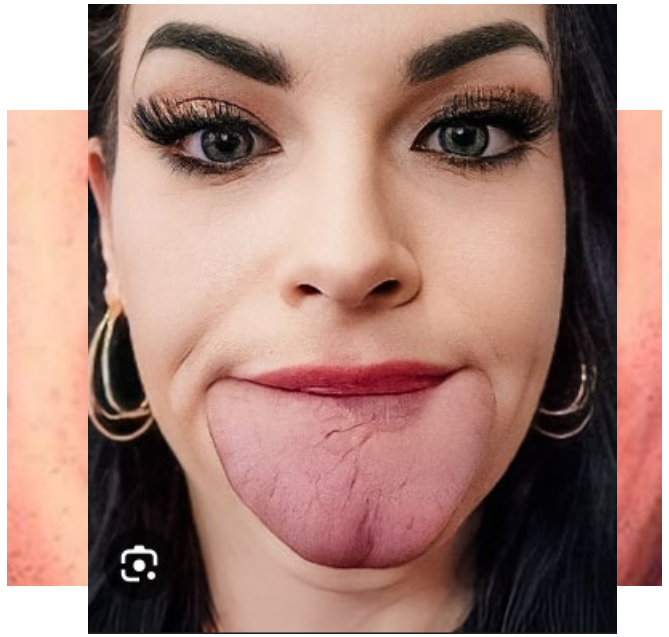


Macrostomia



Thin Upper Llp

Microstomia



Macroglossia

Teeth

Macrodontia

Dental Crowding

Cleft Uvula

Microdontia

High Palate

Absent Uvula

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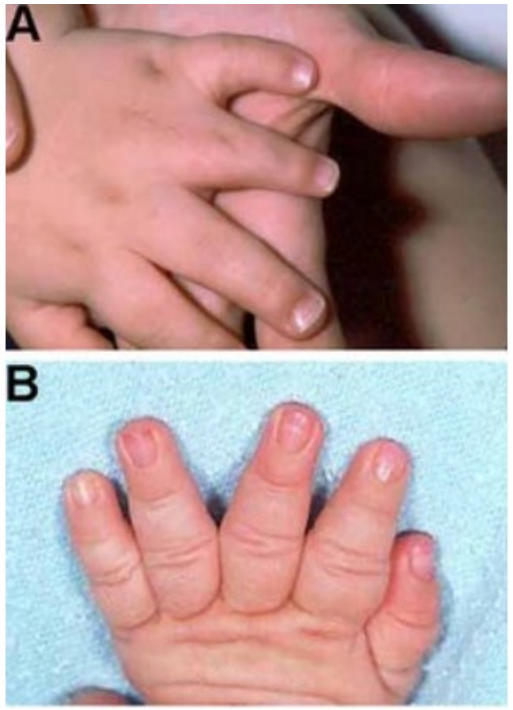
Hands and Feet

Hands

Camptodactyly: The DIPJ and/or PIPJ of the fingers cannot be extended to 180 by either active or passive extension

- Fetal Fingertip Pads: A soft tissue prominence of the ventral aspects of the fingertips or toe tips

- Tapered Fingers: The gradual reduction in girth of the digit from proximal to distal



- Clinodactyly: A digit that is laterally curved in the plane of the palm

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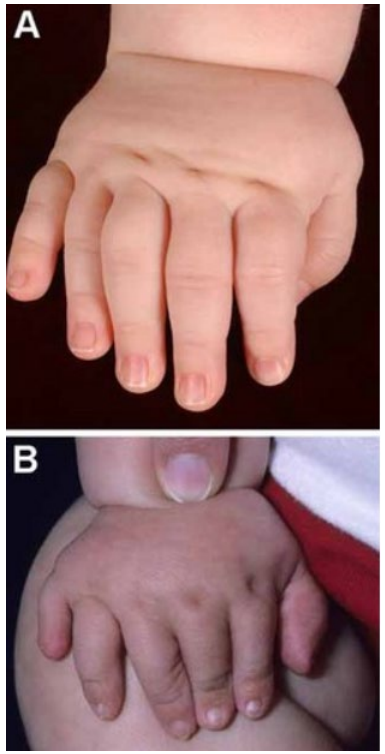
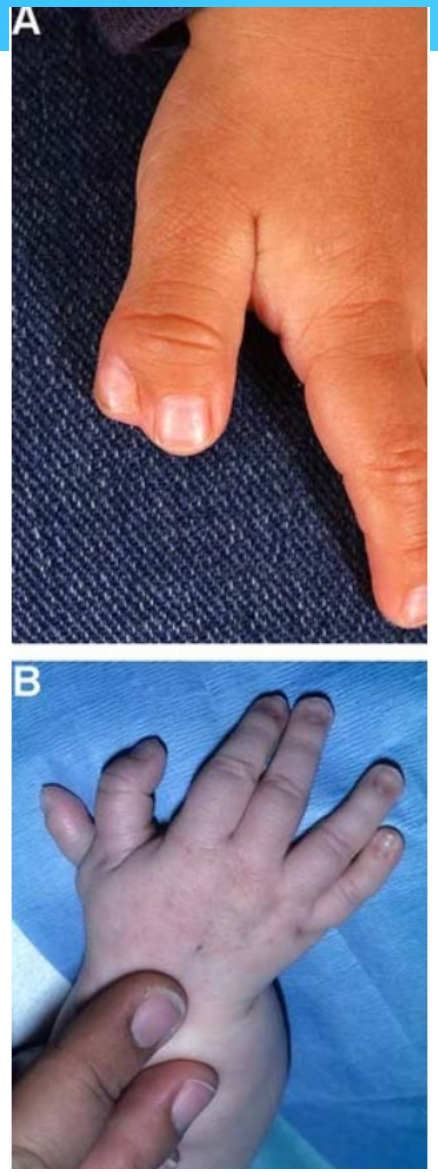
Polydactyly





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Feet

Rocker-bottom foot: The presence of both a "prominent heel" and a "convex contour of the sole"

Overlapping Toes: Describes a foot digit resting on the dorsal surface of an adjacent digit when the foot is at rest

Sandal Gap: A widely spaced gap between the first toe (the great toe) and the second toe

Pes Cavus: The presence of an unusually high plantar arch

Pes Planus: A foot where the arch is in contact with the ground or floor when the individual is standing

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Creases

Deep Plantar Crease: Narrow, paramedian longitudinal depressions in the plantar skin of the forefoot

Single Palmar Crease: The distal and proximal transverse palmar creases are merged into a single transverse palmar crease

- Bridged Palmar Crease: A crease that connects the proximal and distal transverse palmar creases

- Absent Palmar Crease: The absence of a major crease of the palm (distal transverse crease, proximal transverse crease, or thenar crease)

Nails

Thick Nails: Nail that appears thick when viewed on end

- Small Nails: A nail that is diminished in length and width

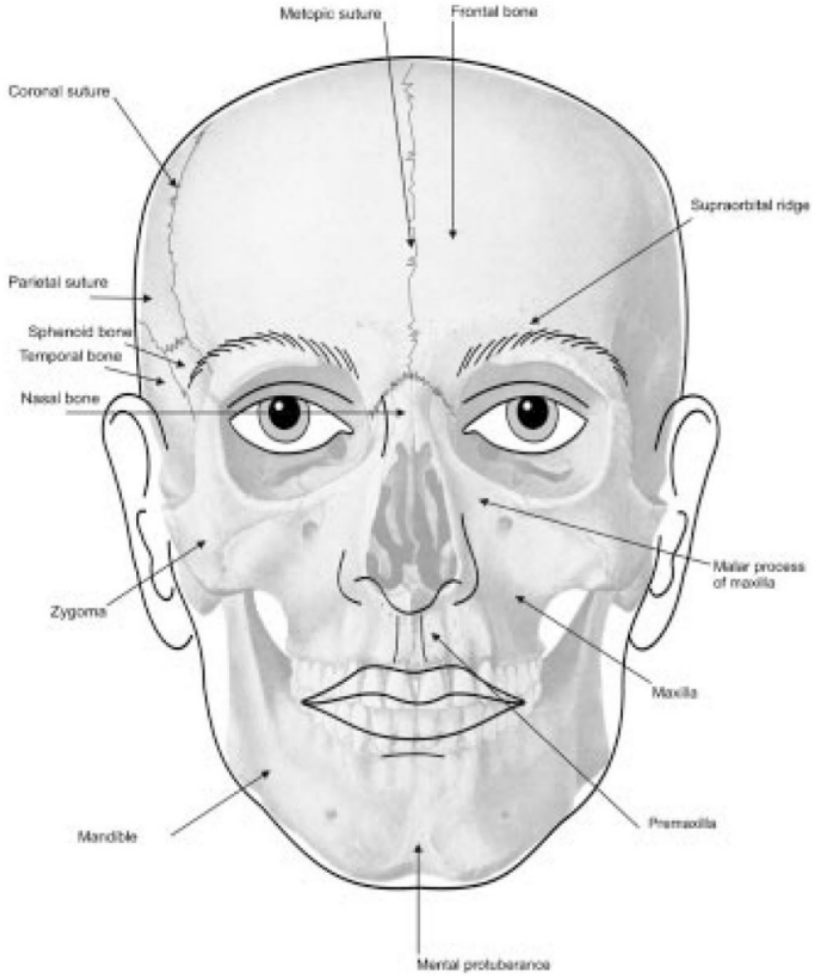
- Nail Pits: Small (typically about 1 mm or less in size) depressions on the dorsal nail surface

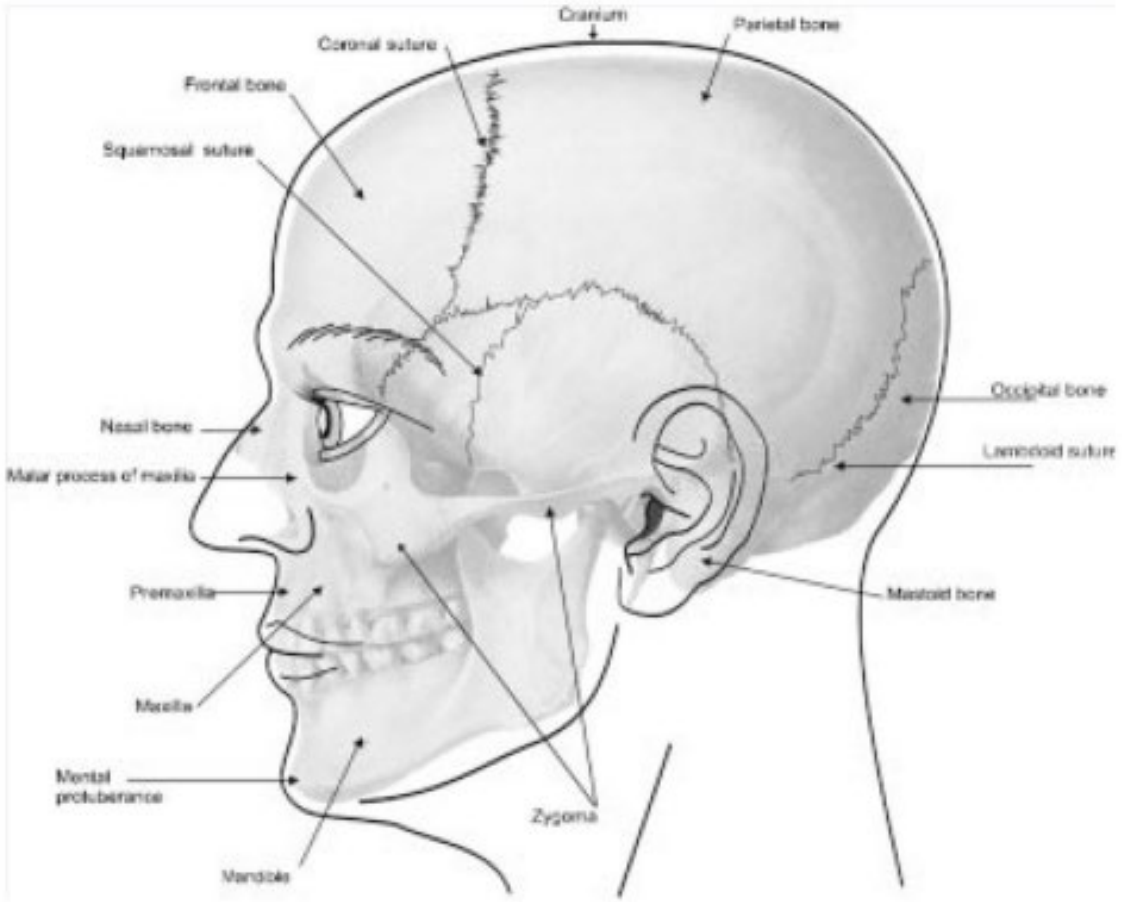
- Concave Nails: The natural longitudinal (P/D) convex arch is not present or is inverted (spoon-shaped)

- Clubbing

Head and Face

Landmarks and Anatomy

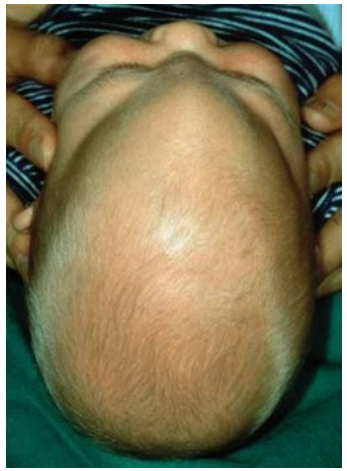




Head Shape

Dolichocephaly: increased antero-posterior length of the head compared to width

- Trigonocephaly: wedge-shaped, or triangular head, with the apex of the triangle at the midline of the forehead and the base of the triangle at the occiput



- Brachycephaly: shortened antero-posterior length of the head compared to width

- Turricephaly: tall head relative to width and length

Head Shape

Cloverleaf skull: Trilobar skull configuration when viewed from the front or behind

- Macrocephaly: Occipitofrontal (head) circumference greater than 97th centile compared to appropriate, age matched, sex-matched normal standards
- Microcephaly: Occipito-frontal (head) circumference (OFC) less than 3rd centile compared to appropriate, age matched, normal standards

- Plagiocephaly: Asymmetric head shape, which is usually a combination of unilateral occipital flattening with ipsilateral frontal prominence, leading to rhomboid cranial shape

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Face

Coarse

- Triangular

- Long

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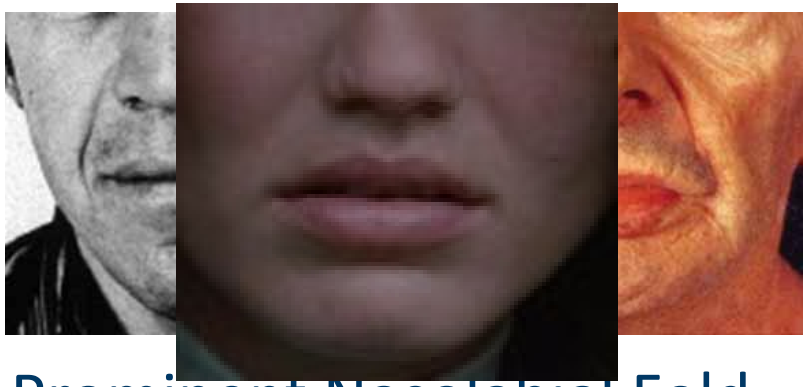
Face

Midface retrusion



- Malar Flattening

- Prominent Nasolabial Fold

- Underdeveloped Nasolabial Fold

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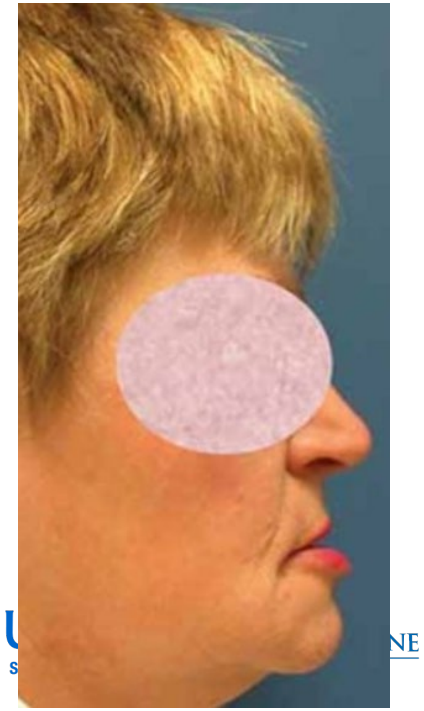
Jaw

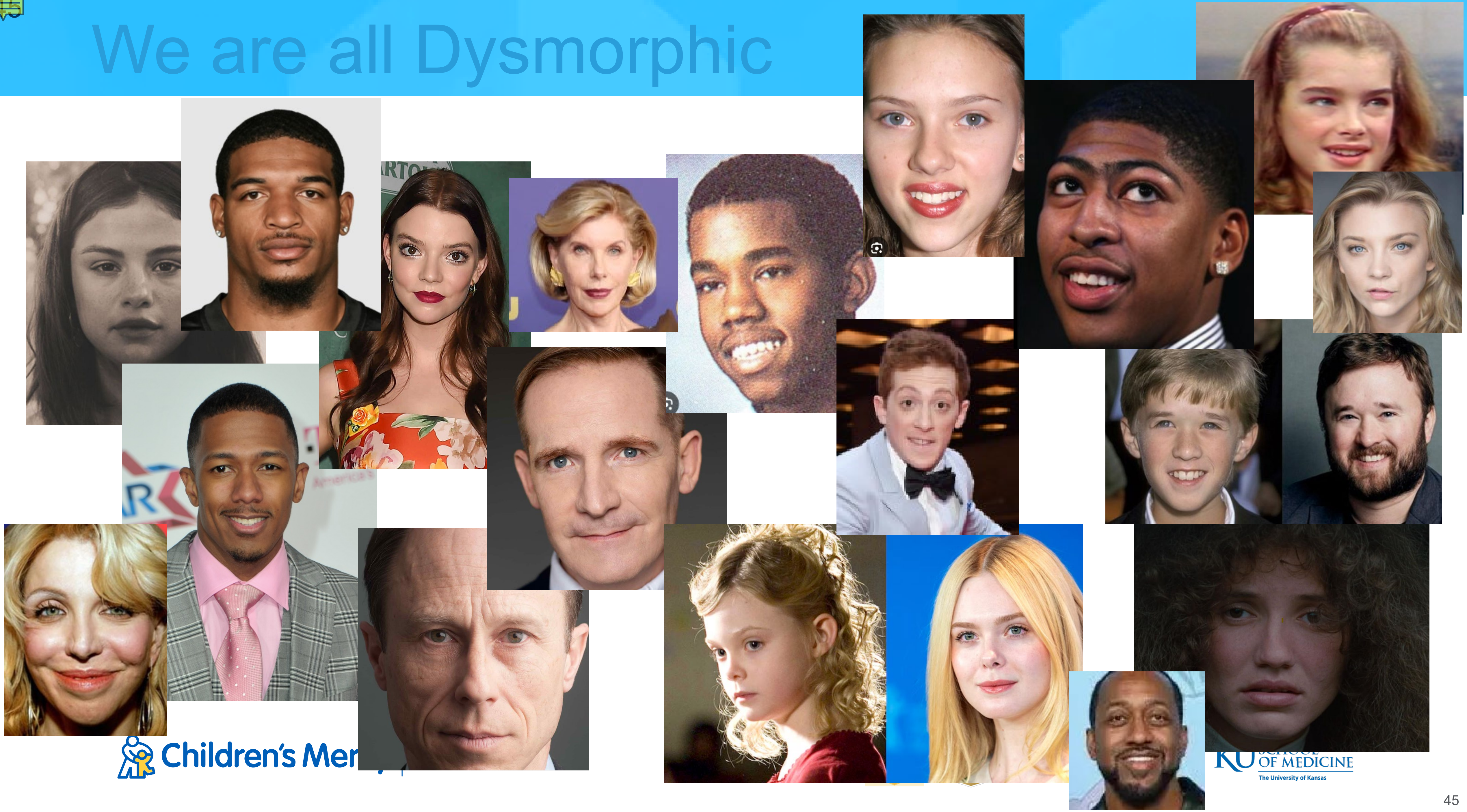
Micrognathia: Apparently reduced length and width of the mandible when viewed from the front but not from the side

- Retrognathia: Posteriorly positioned lower jaw, which is set back from the plane of the face when viewed from the side but not from the front

- Prognathism: Anterior protrusion of the mandibular alveolar ridge beyond the vertical plane of the maxillary alveolar ridge, best appreciated in profile



We are all Dysmorphic



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