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\textsuperscript{rd}s
– 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

• Turricephaly - 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
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

- outer canthal distance
- inner canthal distance
- interpupillary distance
- lacrimal punctum
- palpebral fissure length

Types of Orbital Spacing:
- 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 Ridge

Depressed Nasal Bridge

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 vermillion 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

- Helix
- Antihelical Scapha
- Antihelix
- Antitragus
- Antihelical Crura
- Cymba conchae
- Cavum conchae
- Tragus
- Lobule
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
- Uplifted

Anterior creases
Helical Variation

Lop ear

Satyr ear
Ear Tags
Ear Pits

Auricular pit

Helical pit

preauricular 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 fisted 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
- Mesoaxial
- Mirror Image
- Postaxial
Finger Variation

Overlapping fingers

Tapered fingers

Short fingers

Radial deviation
Foot Variation

- Rocker bottom
- Sandal Gap
- Metatarsus adductus
- 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
- Rhizomelic
- Mesomelic
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

Dysmorphology Tools for NICU Providers

- 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