Dysmorphology Assessment: Recognizing “Normal” & “Abnormal” Physical Differences & Common Genetic Syndromes

Sharon Anderson, MSN, NNP-BC
Rutgers University-School of Nursing and Robert Wood Johnson Medical School

Program Objectives:
Dysmorphology assessment requires a critical eye and pursuit of clues that may suggest an underlying condition. At the completion of the program, the participant will be able to:

- Perform a dysmorphology assessment and recognize common major and minor congenital anomalies.
- Differentiate physical differences that are cosmetically insignificant and/or normal population variants from those of clinical significance.
- Describe common physical differences differences using appropriate genetic terminology.
- Recognize phenotypic characteristics of several common genetic syndromes.

Head to Toe Assessment Topical Outline

- **Head**
  - Fontanels & sutures
  - Size of skull
    - Macrocephaly
    - Microcephaly
  - Shape of skull
    - Brachycephaly
    - Trigonocephaly
    - Positional plagiocephaly
    - Dolicocephaly/scaphocephaly
  - Size & shape of forehead
    - Acrocephaly/oxycephaly/turriccephaly
    - Frontal bossing
    - Broad/Narrow

- **Hair**
  - Hairlines
  - Patterns/Distribution
    - Alopecia/Sparce
    - Hair whorls
    - Hirsuitism/Hypertrichosis
    - Synophrys
  - Color & Texture
    - Albinism
• White forelock
• Wiry/Coarse
• Fine/Thin

• **Face**
  • Symmetry
  • Shape
    • Short
    • Round
    • Triangular
    • Square
  • Hypoplasia/Hyperplasia

• **Chin**
  • Micrognathia
  • Retrognathia
  • Retromicrognathia
  • Prognathism

• **Eyes**
  • Spacing
    • Hypertelorism
    • Hypotelorism
    • Telecanthus
  • Palpebral fissure length
    • Short/narrow
    • Long
  • Fissure Slant
    • Up
    • Down
  • Orbital depth
    • Deep set
    • Epicanthal folds
  • Ptosis
  • Sclera
  • Size
    • Microphthalmos
  • Iris/Pupil
    • Brushfield spots
    • Coloboma
    • Heterochromia

• **Ears**
  • Anatomy (configuration)
  • Position
    • Set
    • Rotation
  • Shape, size & contours
    • Ear pit
    • Preauricular skin tag
    • Folded pinna/lob ear
    • Satyr, Spock or Vulcan ear
    • Microtia
    • Prominent
    • Cup ear
• Hypoplastic ear

• **Nose**
  • Wide variability in size & shape
  • Anatomy (landmarks)
  • Shape/ Configuration
    • Upturned nose/ anteverted nares
    • Beaked
    • Bulbous

• **Mouth, Lips & Teeth**
  • Size, shape, symmetry
  • Mouth
    • Macrostomia
    • Macroglossia
    • Ankyloglossia
    • Cleft palate
    • Bifid uvula
  • Lips
    • Thin/ Full
    • Cupid’s bow
    • Tent ed upper lip
    • Cleft lip
    • Pits
  • Teeth
    • Hyperdontia
    • Hypodontia
    • Peg
    • Pitting, hypocalcification, enamel abnormalities

• **Neck**
  • Shape & symmetry
    • Webbing
    • Short
    • Nuchal redundancy

• **Chest**
  • Shape & symmetry
    • Barrel
    • Short sternum
    • Pectus excavatum (funnel chest)
    • Pectus carinatum (pigeon chest)
  • Nipples/ Breasts
    • Spacing
    • Supernumerary nipples
    • Gynecomastia

• **Abdomen**
  • Shape & symmetry
  • Muscle tone
    • Diastasis recti
    • Prune belly syndrome
  • Umbilicus
    • Omphalocele
    • Hernia
• **Genitalia**
  - Male
    - Urethral meatus
      - Hypospadias
      - Epispadias
    - Phallus
      - Micropenis/Macropenis
    - Testes
      - Cryptorchidism
      - Macroorchidism
    - Hypogonadism
  - Female
    - Labia
    - Clitoris
    - Virilization
    - Hypogonadism

• **Skin**
  - Inspection
    - Pigmentation
    - Lesions
      - Café au lait macules
      - Neurofibroma
      - Axillary freckling
      - Port wine stain
      - Stork bite/Angel kiss
      - Hemangioma
  - Palpation
    - Elasticity
    - Texture

• **Limbs/Extremities**
  - Size, shape & symmetry
    - Creases
    - Length
    - Hemihypertrophy
  - Upper extremities
    - Radial limb defect
    - Polydactyly
    - Syndactyly
    - Clinodactyly
    - Ectrodactyly
    - Hypodactyly/oligodactyly
  - Lower extremities
    - Sandle gap
    - Rocker bottom feet
    - Lymphedema

• **Back/Spine**
  - Inspection
    - Scoliosis
    - Kyphosis
    - Lordosis
    - Spinal dysraphism