Dysmorphology Assessment

Are the features familial?

- **No**
  - Recognised syndrome
    - **Yes**
    - Confirmatory testing
    - Search:
      - New/old photos
      - Published literature
      - Databases
      - tests
      - record them – publish/other
      - Watchful waiting
    - **No**
  - **No**

- **Yes**
  - AD/XL syndrome
  - No syndrome dx, familial features
DYSMORPHOLOGY ASSESSMENT

- HISTORY CHECKLIST
- EXAMINATION CHECKLIST
- INVESTIGATIONS - WHEN TO DO WHAT?
- COMMUNICATION STRATEGIES WITH PARENTS
HISTORY CHECKLIST

- pregnancy history, noting particularly exposure to teratogens, amniotic fluid volume, ultrasound and amniocentesis/CVS, foetal movements
- family history of abnormalities
- consanguinity
Examination CHECKLIST

- **GROWTH**
  - Birth weight, length and head circumference
  - *Are the baby’s growth parameters in proportion and what are the centiles?*

- **ECTODERMAL FEATURES**
  - Skin – texture and colour, birthmarks, redundancy, defects
  - Hair – scalp hair and body hair: colour and distribution
  - Note position of anterior and posterior scalp hairline
Examination CHECKLIST

- **SKULL**
  - shape, symmetry
  - sutures (over-riding/normal/widely open)
  - Fontanelle size and number

- **FACE**
  - Overall face shape, symmetry, facial muscle movement
  - Forehead region:
    • Forehead shape – (broad/bitemporal narrowing/tall)
    • Eyes:
    • Palpebral fissure length (short/long)
Examination CHECKLIST

- EYES
- 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
- Iris colour
- Pupil shape
- Retina
- Globe position (assessed from lateral view: protruberant vs deep set globes)
Examination CHECKLIST

- **Midface region**
  - Nose:
    - Divide the nose into 3 sections from the lateral view from superior to inferior: nasal root, bridge and tip.
    - Root
    - Bridge (depressed/prominent/broad)
    - Tip
  - Columella (*the vertical ridge separating the nostrils*)
  - Nostrils – patency, position (*anteverted nostrils often reflect a short nose*)
Examination CHECKLIST

• Ears:
  • Ear position should be assessed relative to the face, from the lateral view.
  • Ear rotation is normally 15 degrees posterior to the vertical plane of the head.
  • Ear shape and structure
Examination CHECKLIST

- Oral region
  - Mouth size and shape
  - Lip shape, thickness
  - Gum thickness
  - Philtrum definition and length
  - Jaw position (*prognathia/micrognathia*)
  - Palate shape
  - Oral cavity – natal teeth/frenulum/tongue size and morphology
Examination CHECKLIST

- HANDS AND FEET
- Overall shape and size of hand and foot
- Digit number
- Digit shape (e.g. clinodactyly) and length
- Webbing between digits
- Palmar, plantar and digit creases
- Nail morphology
Examination CHECKLIST

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

- 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
Figure 4.4.1. Superficial anatomy and landmarks of the eye and periocular region.
Figure 4.6.1. Superficial anatomy and landmarks of the nose.
Figure 4.3.2. Normal anatomy and landmarks of the auricle.
• Examination of other family members (siblings and parents) may be crucial to determining whether any dysmorphic features noted are familial or syndromic.
Investigations – when to do what?

- **Tests in the syndrome work-up:**
- Renal ultrasound, echocardiogram and cranial ultrasound
- Eye examination
- Skeletal radiographs – a skeletal dysplasia or dysmorphic syndrome which can have skeletal abnormalities associated with it.
A genetic skeletal survey:
- AP and lateral X rays of the skull
- AP and lateral pelvis and spine (cervical to sacrum)
- AP of one arm, AP both hands, AP of one leg and AP of both feet
- In a neonate, it may be sufficient to obtain a “Baby-gram” (X-ray of the baby) and a separate X ray of the hands and feet.
• Blood chromosomes:
  – multiple congenital abnormalities +/- dysmorphic features
  – one congenital abnormality in the presence of dysmorphic features
    and/or growth retardation

• Typical chromosome picture: growth retardation and microcephaly, in association with dysmorphic features and congenital abnormalities.

• Normal chromosome analysis does not exclude a single gene mutation or a micro deletion syndrome

• Normal antenatal chromosome analysis does not completely exclude a chromosome abnormality

• A chromosome test takes 5 days

• Transfusion issues
• FISH for Trisomies 13/18/21

• arrays
Communication strategies with parents

- Raising issue of dysmorphic features
- Referral to genetics