

Dysmorphology

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www.rch.unimelb.edu.au/nets/handbook

Diagnostic Dysmorphology, Aase



Dysmorphology
Assessment

Algorithm

Are the features familial?

no

yes

Recognised
syndrome

AD/XL
syndrome

No syndrome dx,
familial features

yes

no

Confirmatory
testing

Search:

- ✓ New/old photos
- ✓ Published literature
- ✓ Databases
- ✓ tests
- ✓ record them – publish/other
- ✓ Watchful waiting



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



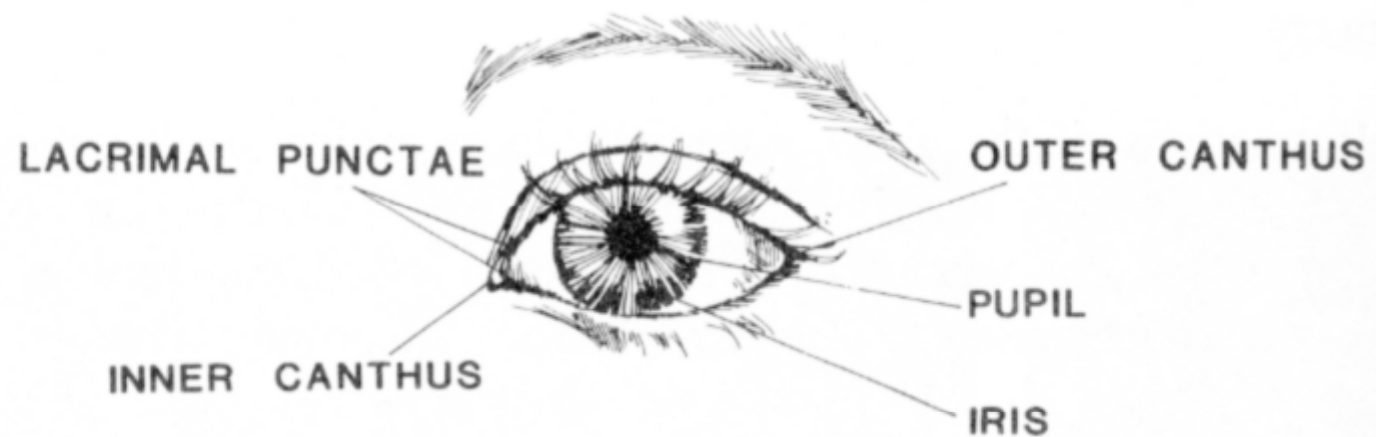
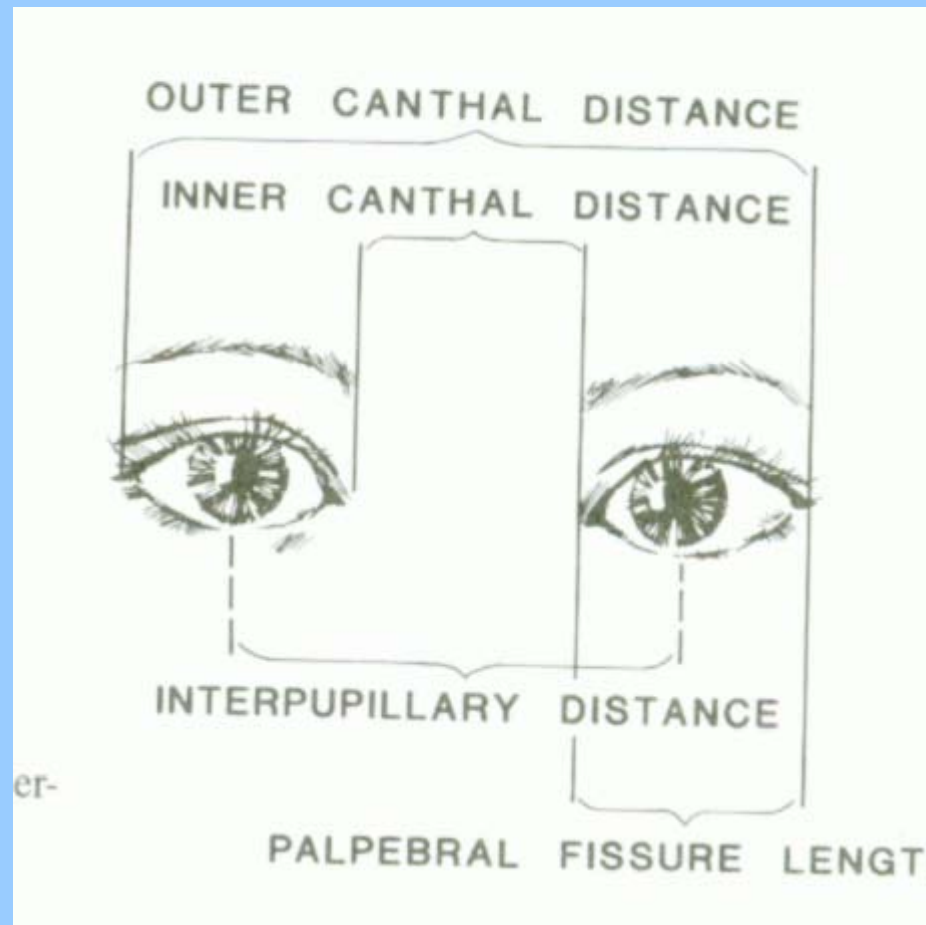


Figure 4.4.1. Superficial anatomy and landmarks of the eye and periocular region.





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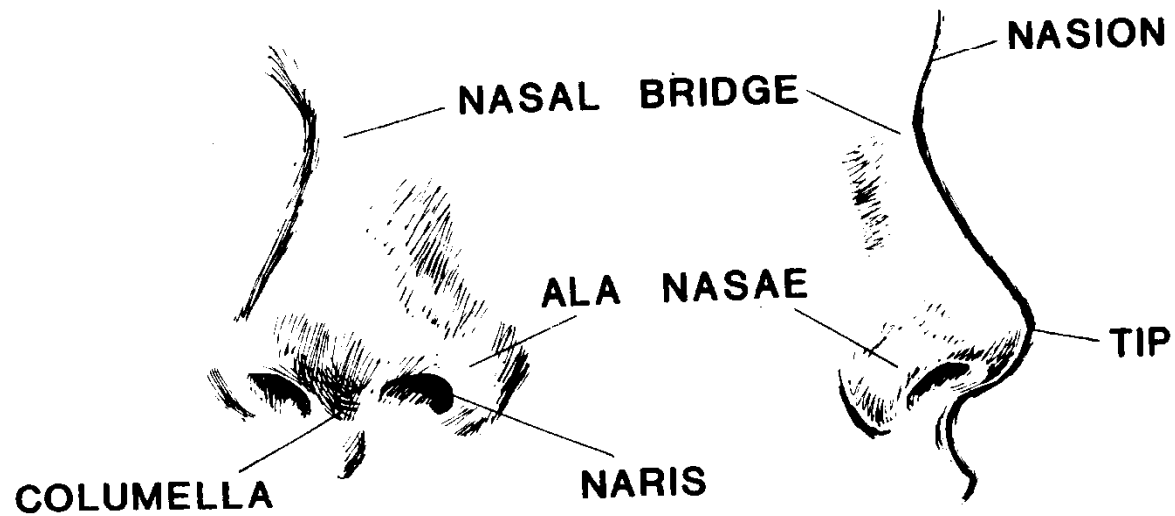


Figure 4.6.1. Superficial anatomy and landmarks of the nose.



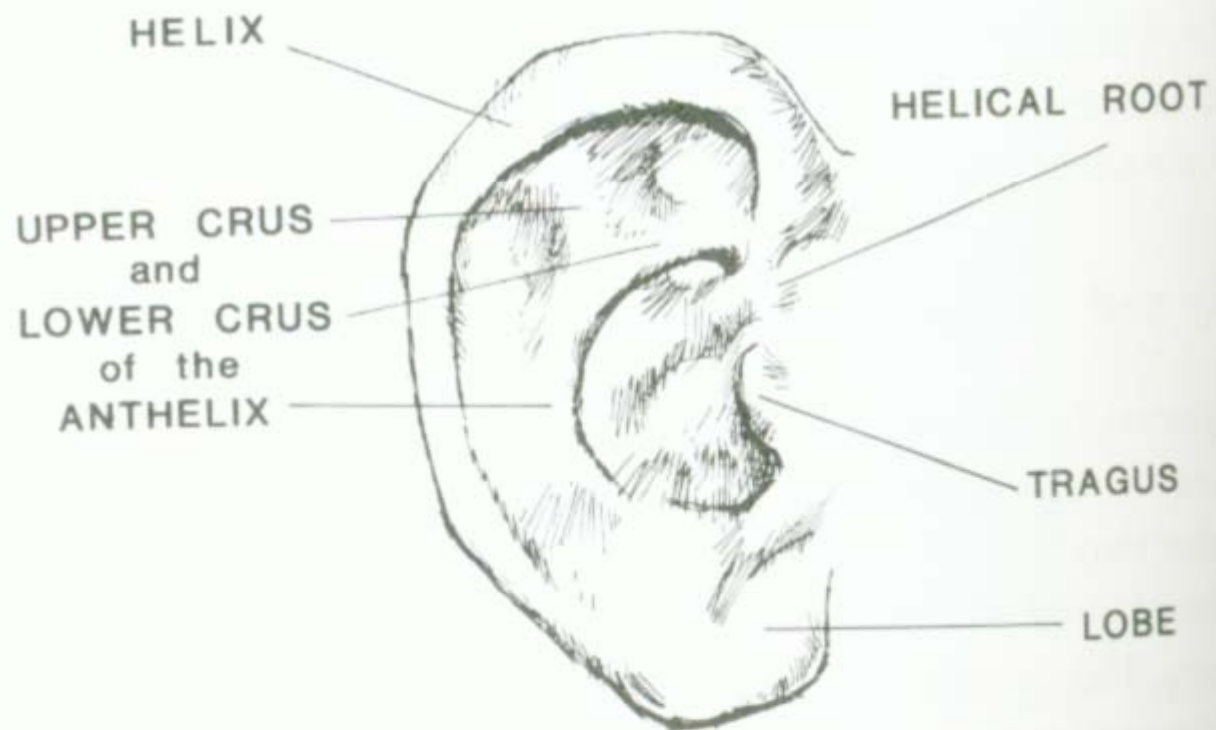


Figure 4.3.2. Normal anatomy and landmarks of the auricle.



- 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

