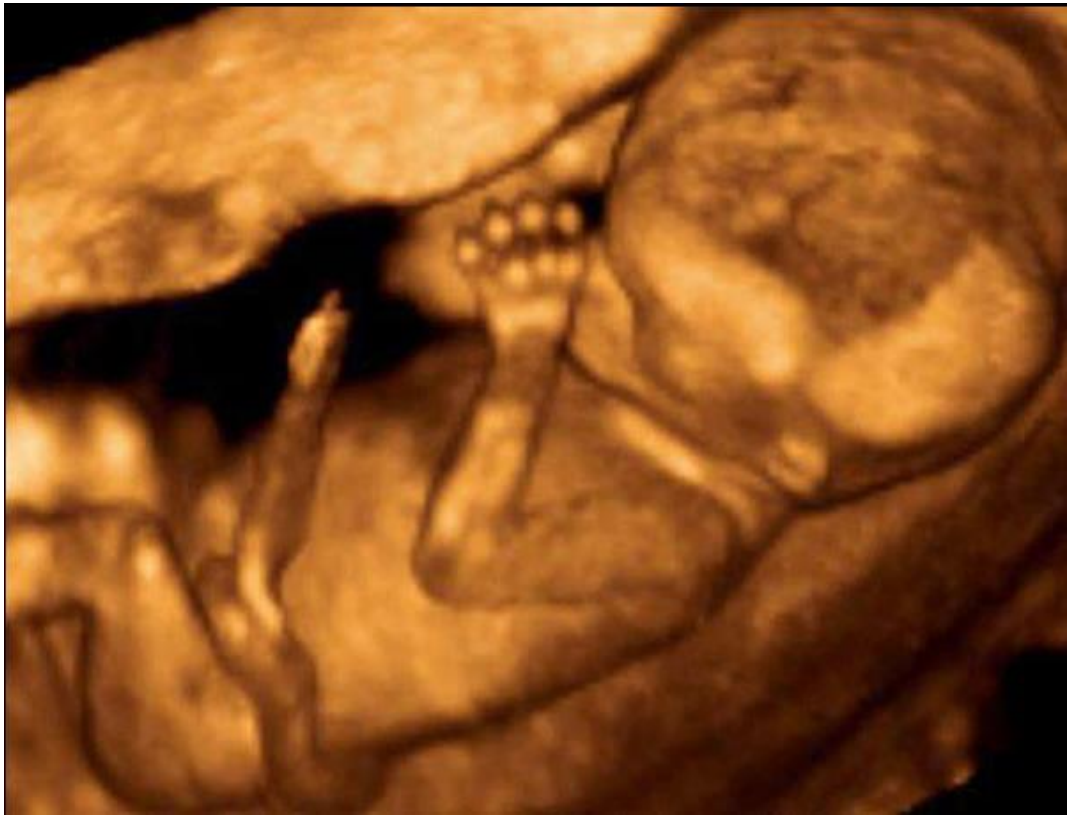
St George's
University of London

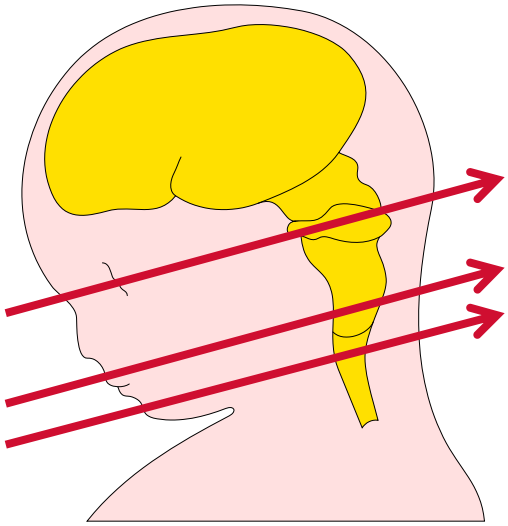
Face, Hands and Feet

Asma Khalil
St George's Hospital
London, UK

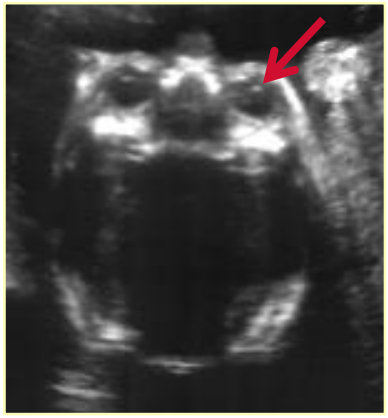


The Fetal Medicine Foundation

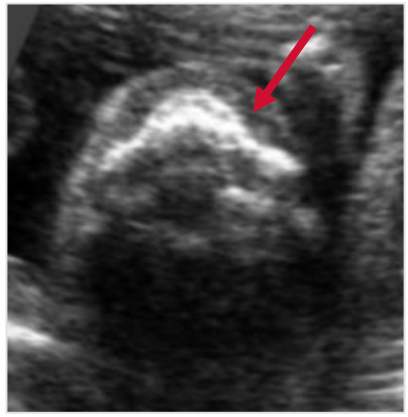
The 20-22 wks scan



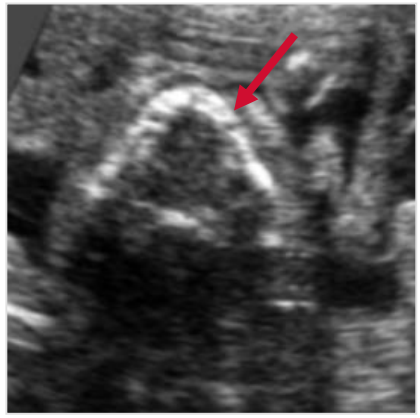
orbits

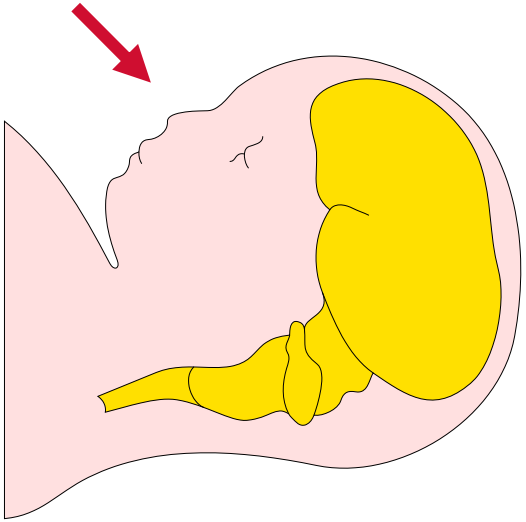


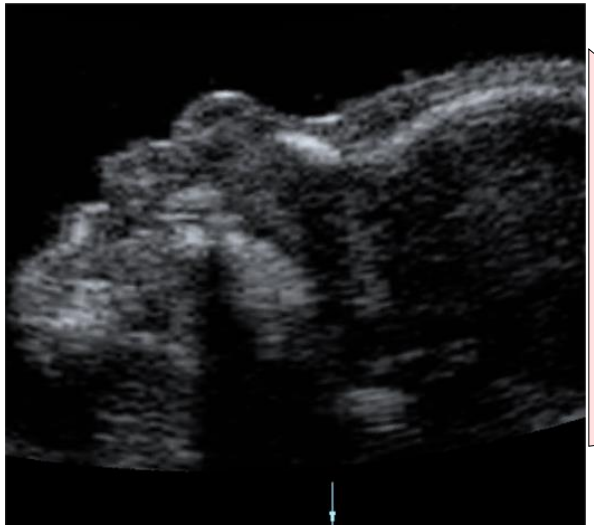
maxilla

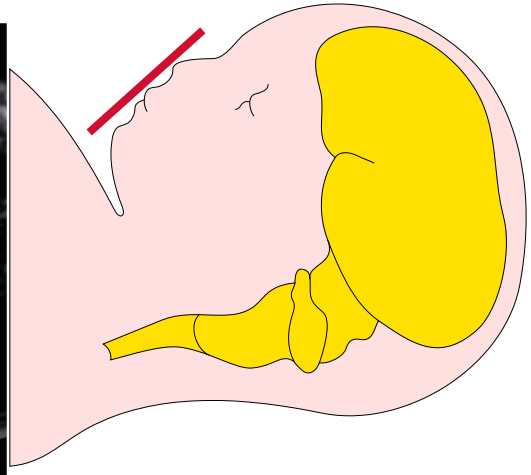


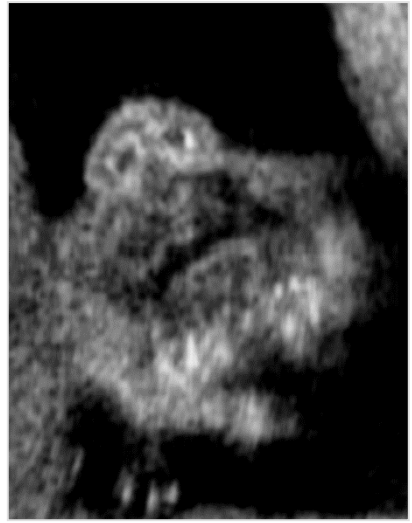
mandible

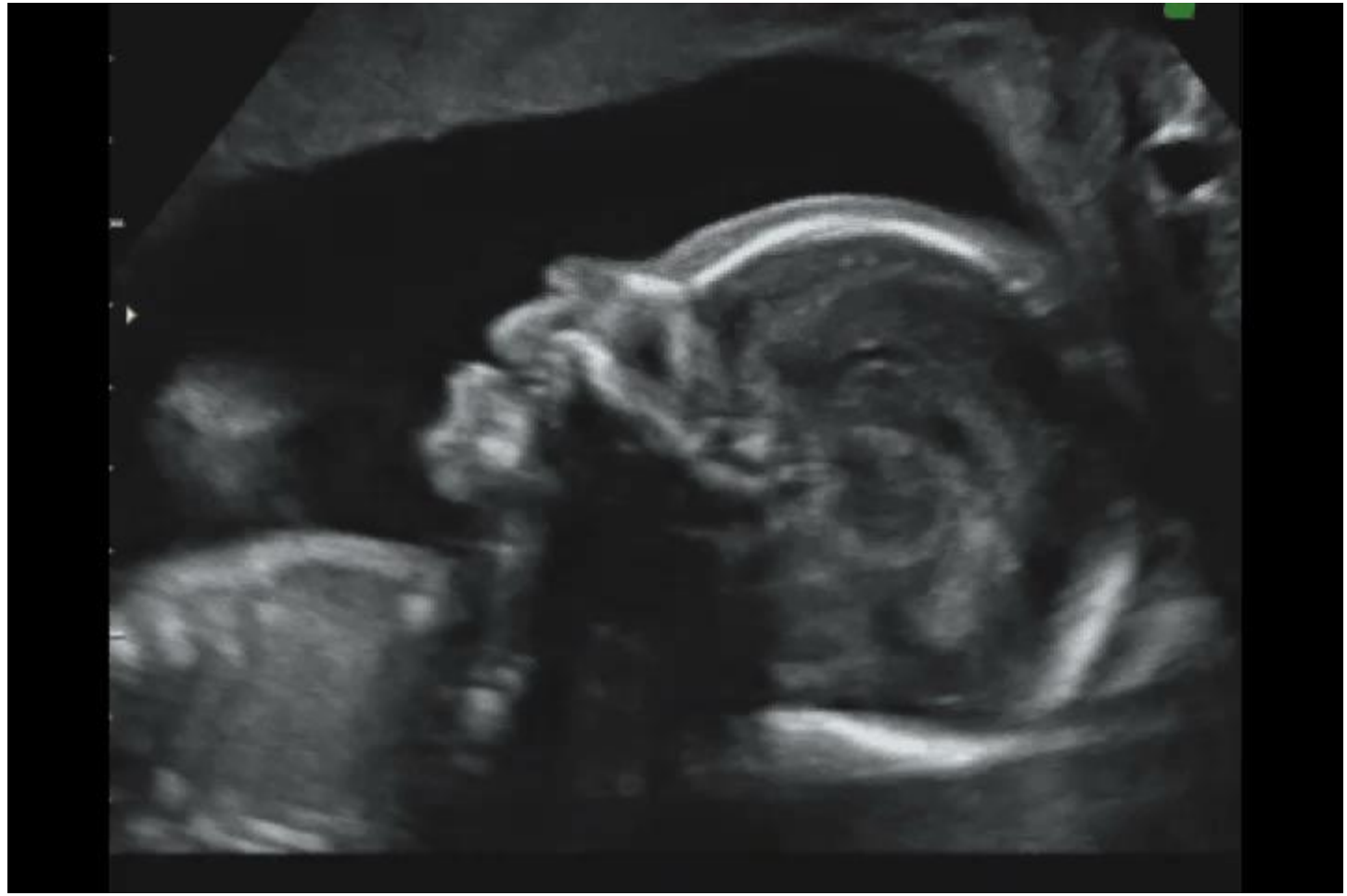






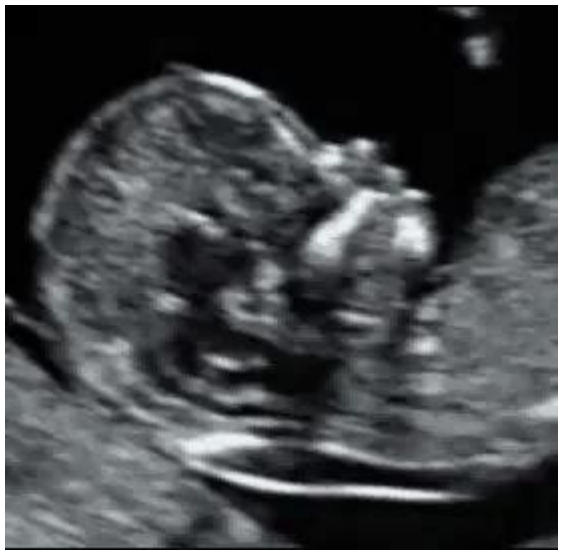


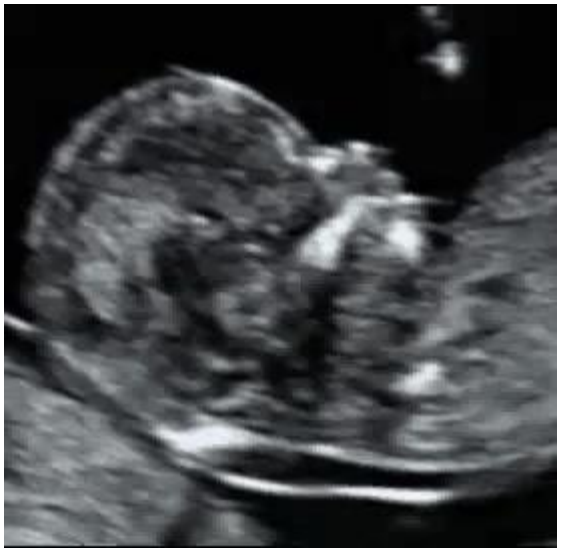




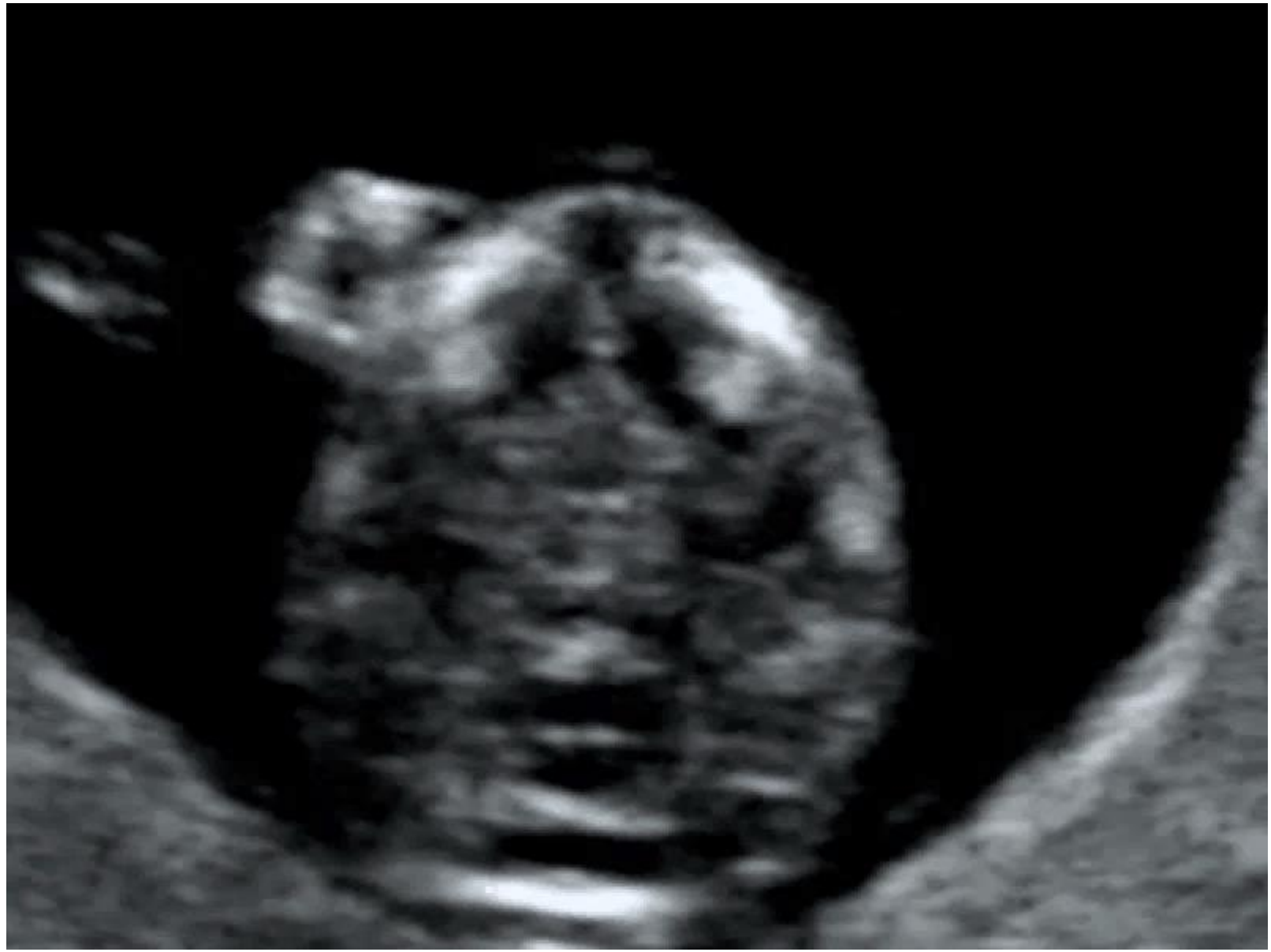
St George's University of London

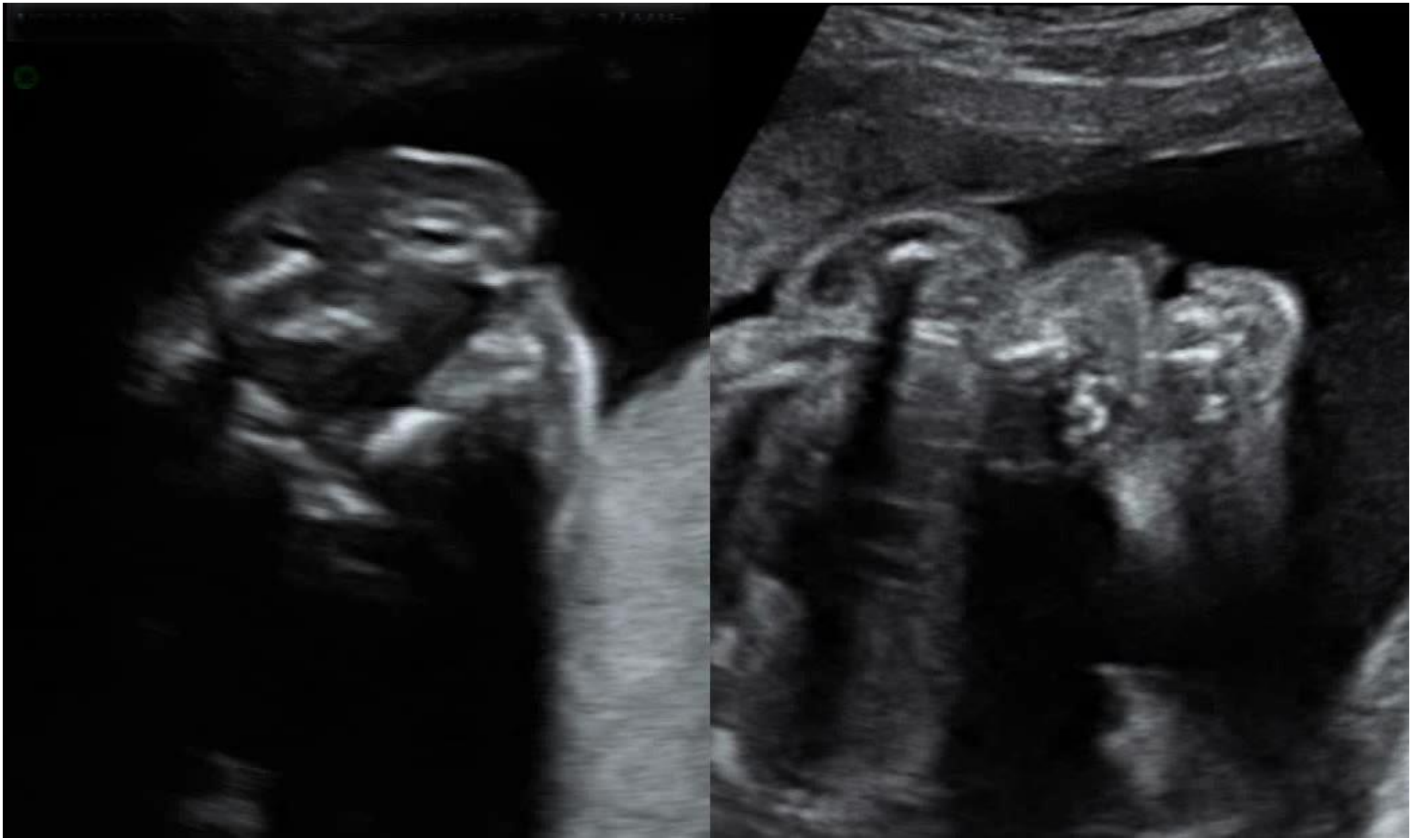
ISUOG Guideline

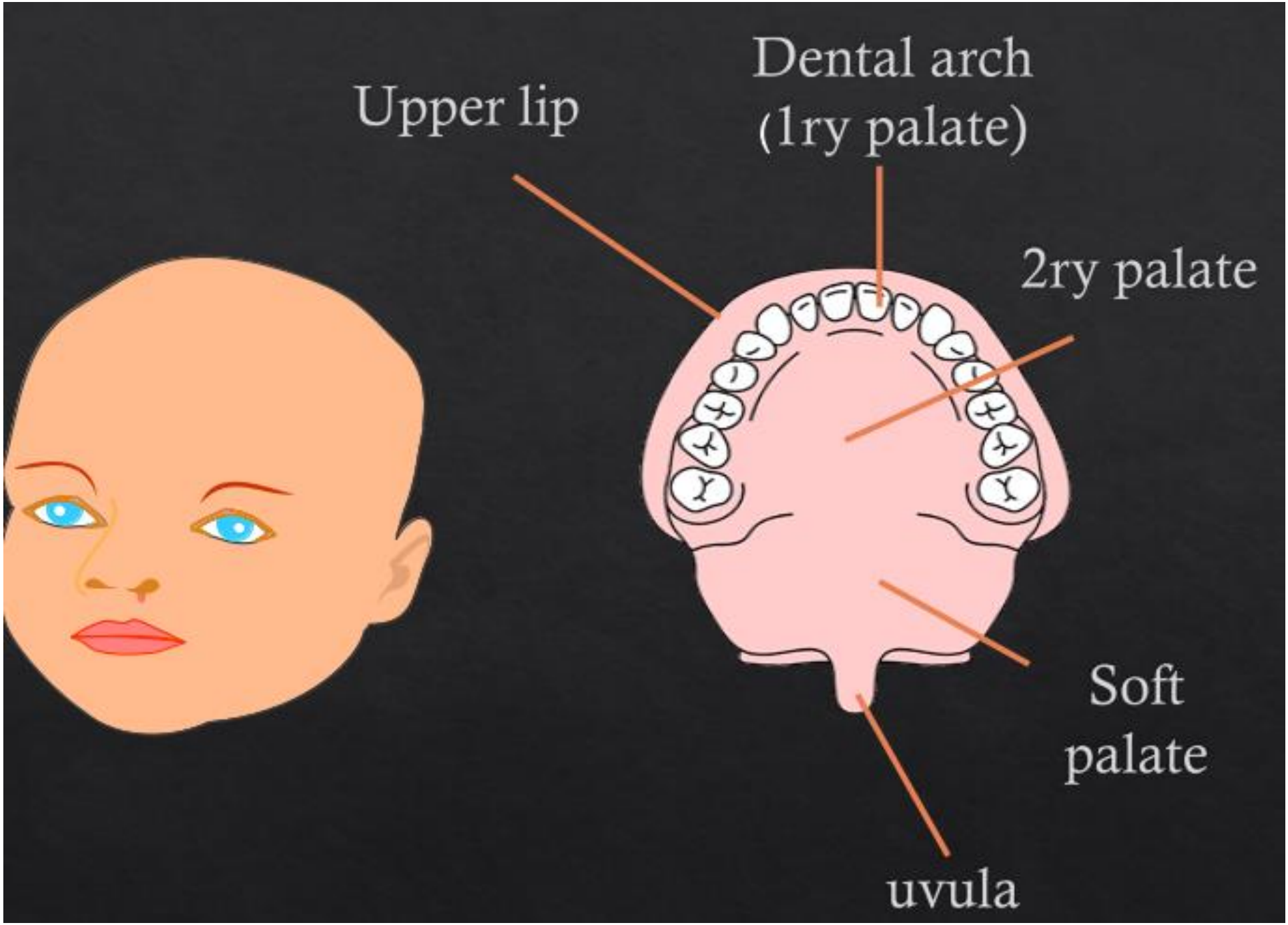




Upper lip
Orbits
? Midline profile
? Nose and nostrils
(if technically feasible)

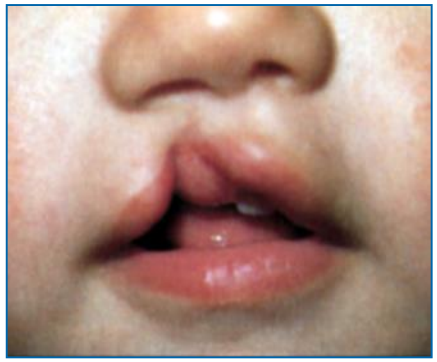


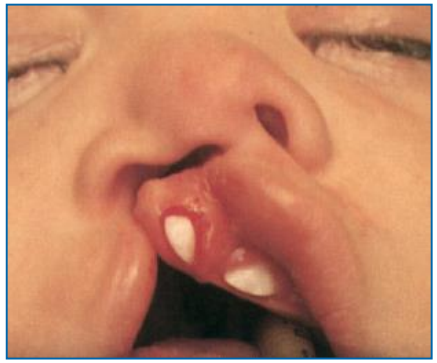


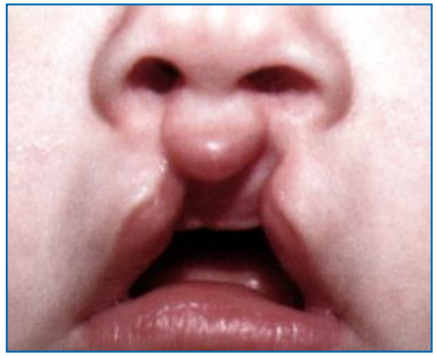

Upper lip
Dental arch
(1ry palate)
2ry palate
Soft
palate
uvula

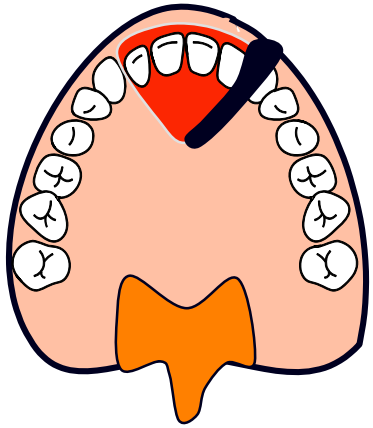
The Fetal Medicine Foundation

Facial cleft

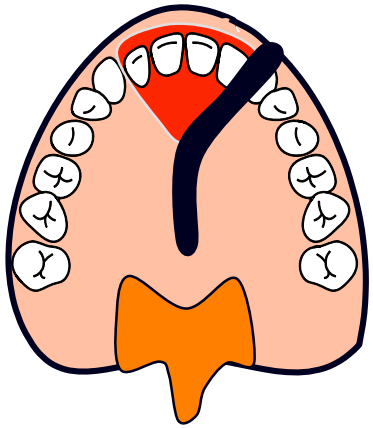




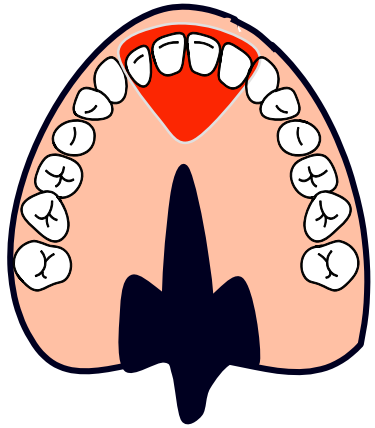




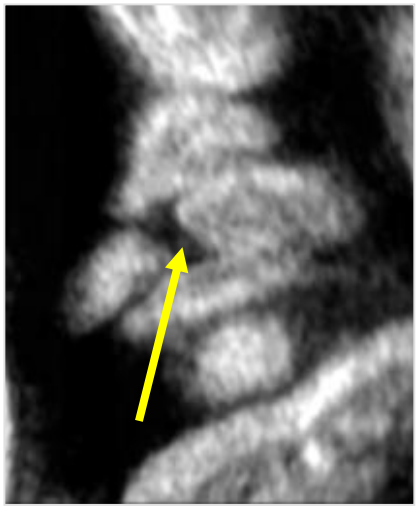
Cleft lip

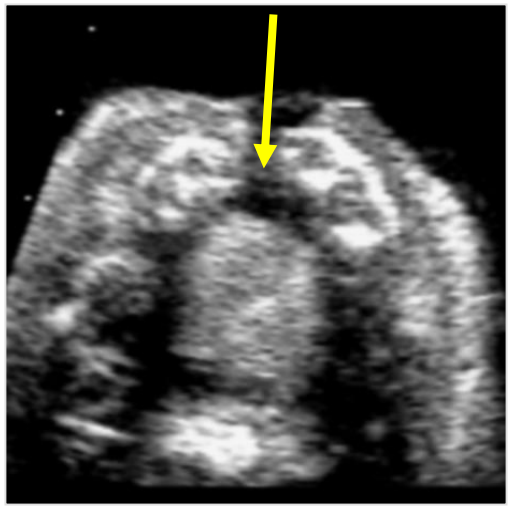


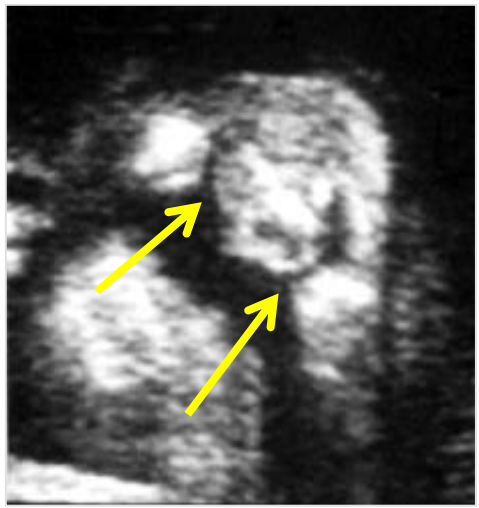
Cleft lip & palate

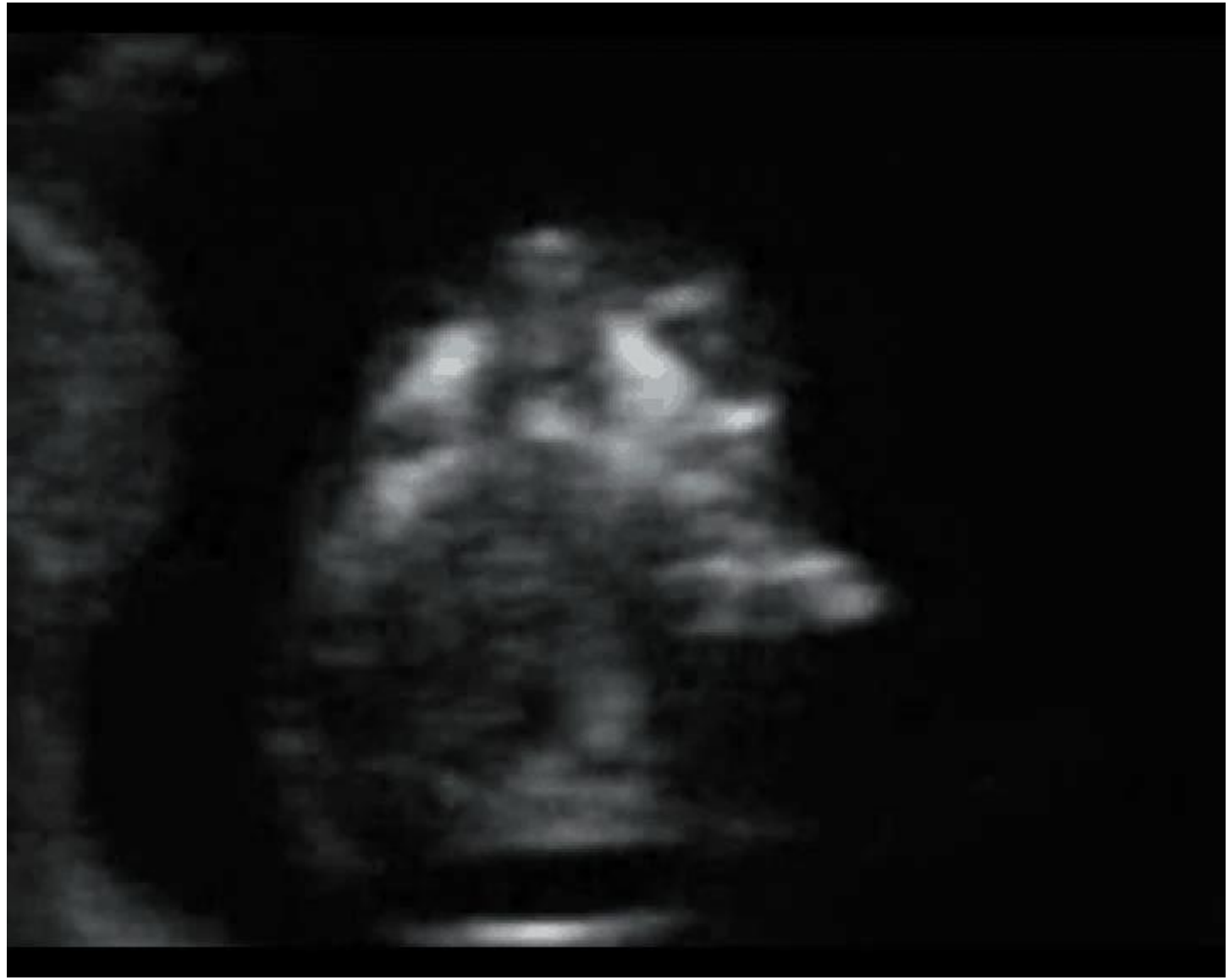


Cleft palate









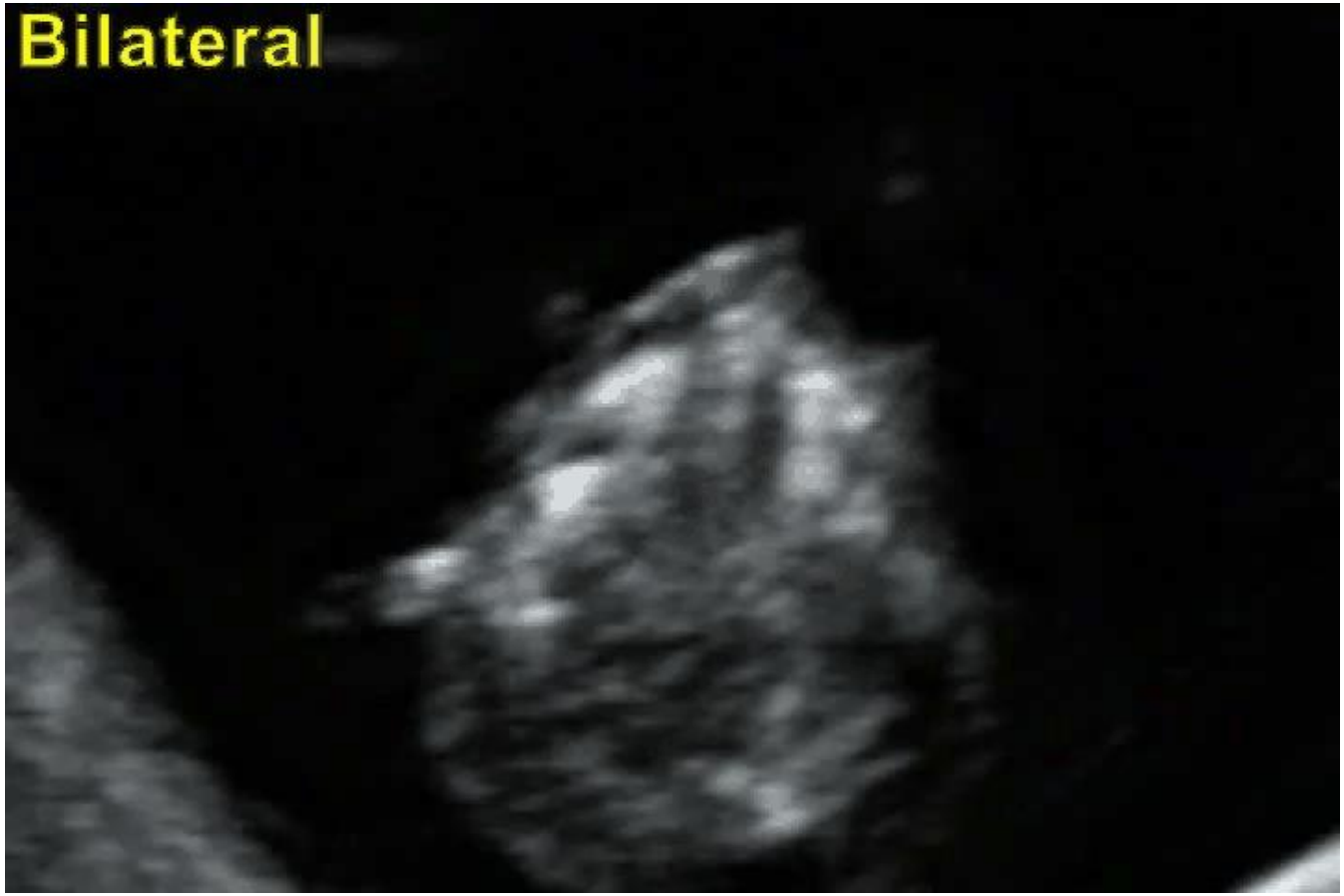
Epidemiology

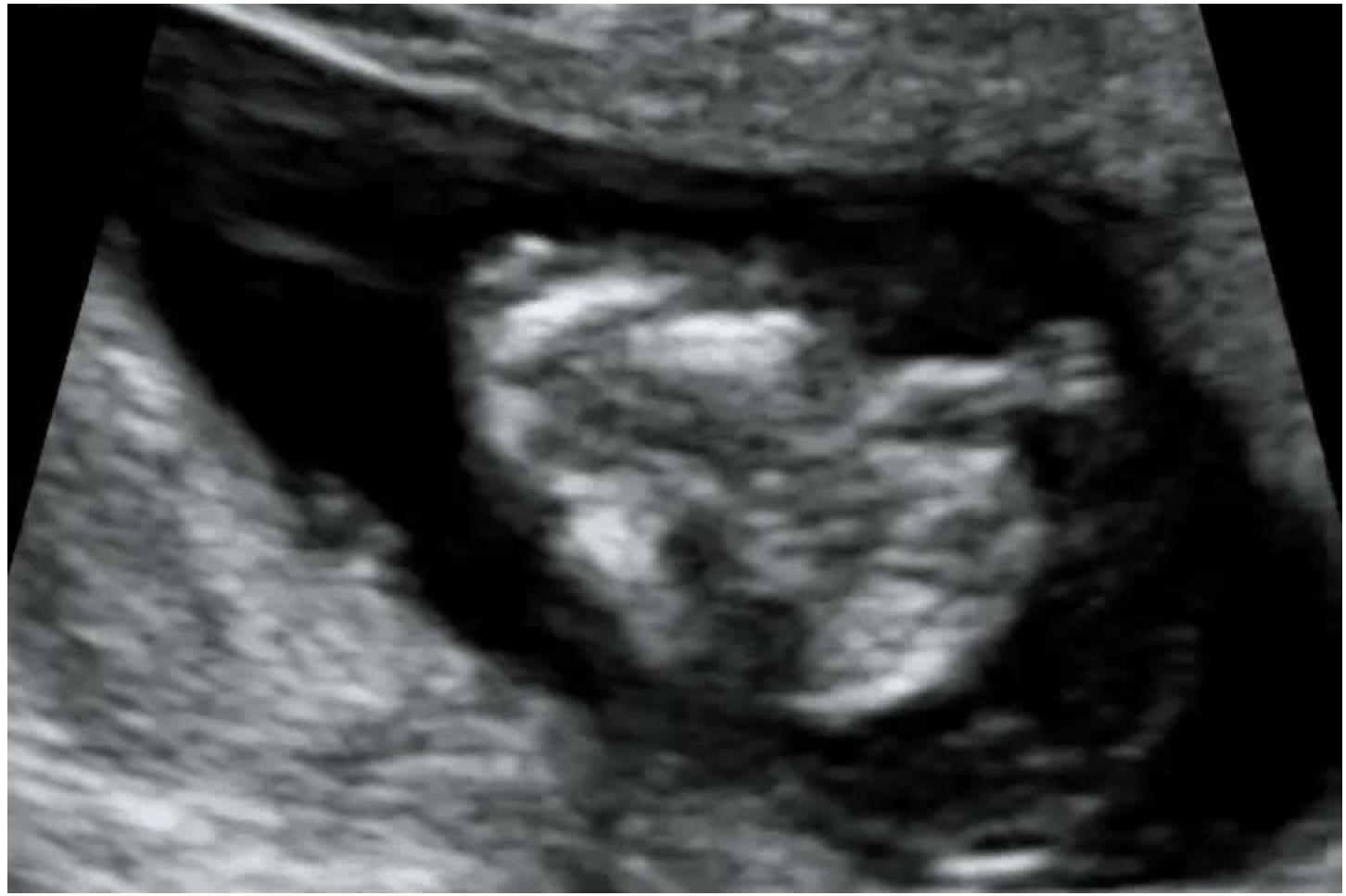
- Prevalence: 1 in 700 births
- More common in males than females and in Whites than Blacks
- In 50% of cases, both the lip and palate are affected
- In 25% only lip
- In 25% only palate
- Unilateral in 75% of cases (more common on left side) and bilateral in 25%

St George's University of London

Facial cleft at 11-13 weeks

Retronasal triangle in a coronal view and the maxillary gap in the standard mid-sagittal view of face





Associated abnormalities

- Chromosomal abnormalities
  - Mainly trisomies 13 and 18
  - Found in 1–2% of cases
  - Unilateral cleft lip is not associated with chromosomal abnormalities

Associated abnormalities

- **Syndromes**

  Associated with >400 syndromes in 30% of cases

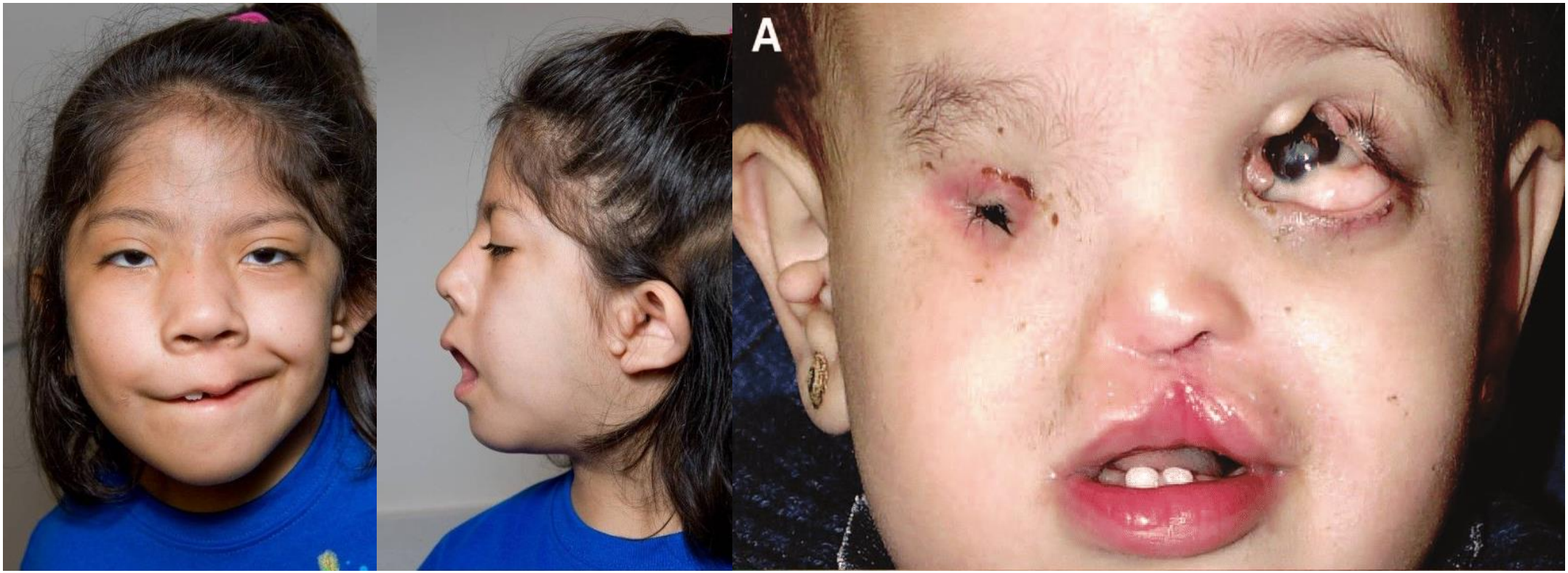
  The most common are:

  - **Goldenhar syndrome** (sporadic; anophthalmia, ear defects, facial cleft, facial macrosomia)
  - **Treacher–Collins syndrome** (AR or AD with 60% *de novo* mutations; hypoplasia of the maxilla and zygomatic bone, micrognathia, cleft palate, malformed or absent ears)
  - **Pierre–Robin Anomalad** (micrognathia or retrognathia, cleft palate and glossoptosis.

St George's University of London

Goldenhar syndrome

Anophthalmia, ear defects, facial cleft, facial macrosomia



St George's University of London

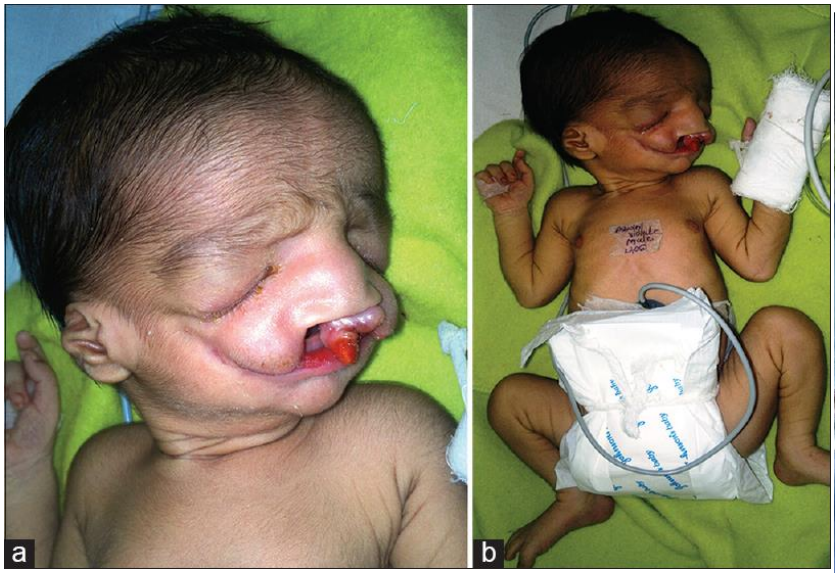
Treacher–Collins syndrome

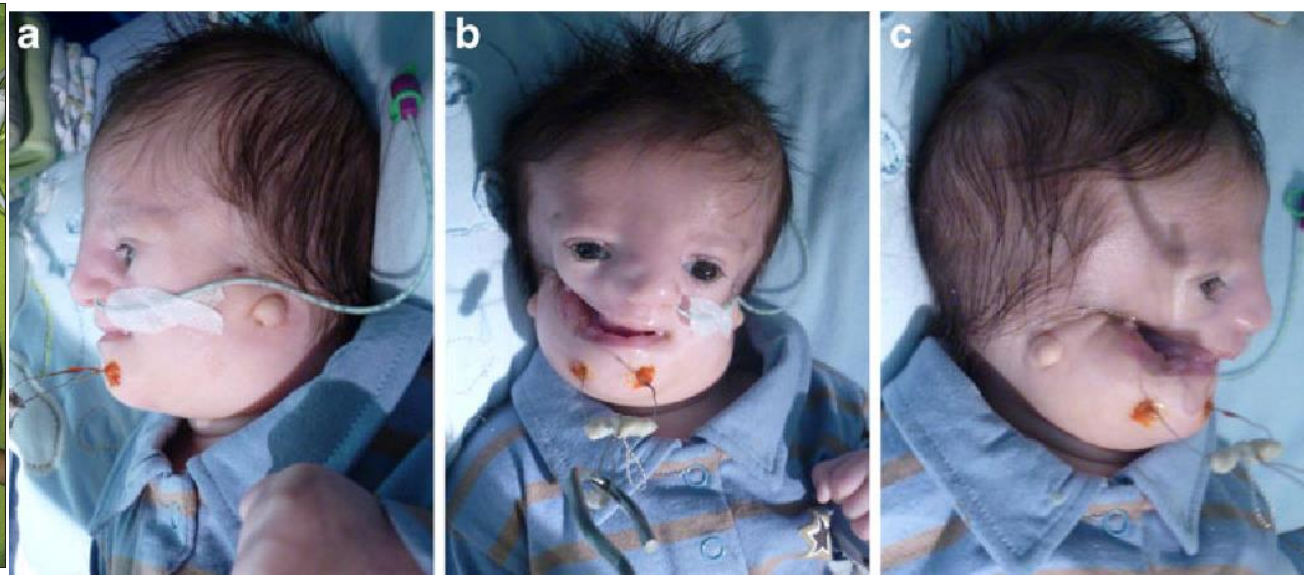
Hypoplasia of the maxilla and zygomatic bone

Micrognathia

Cleft palate

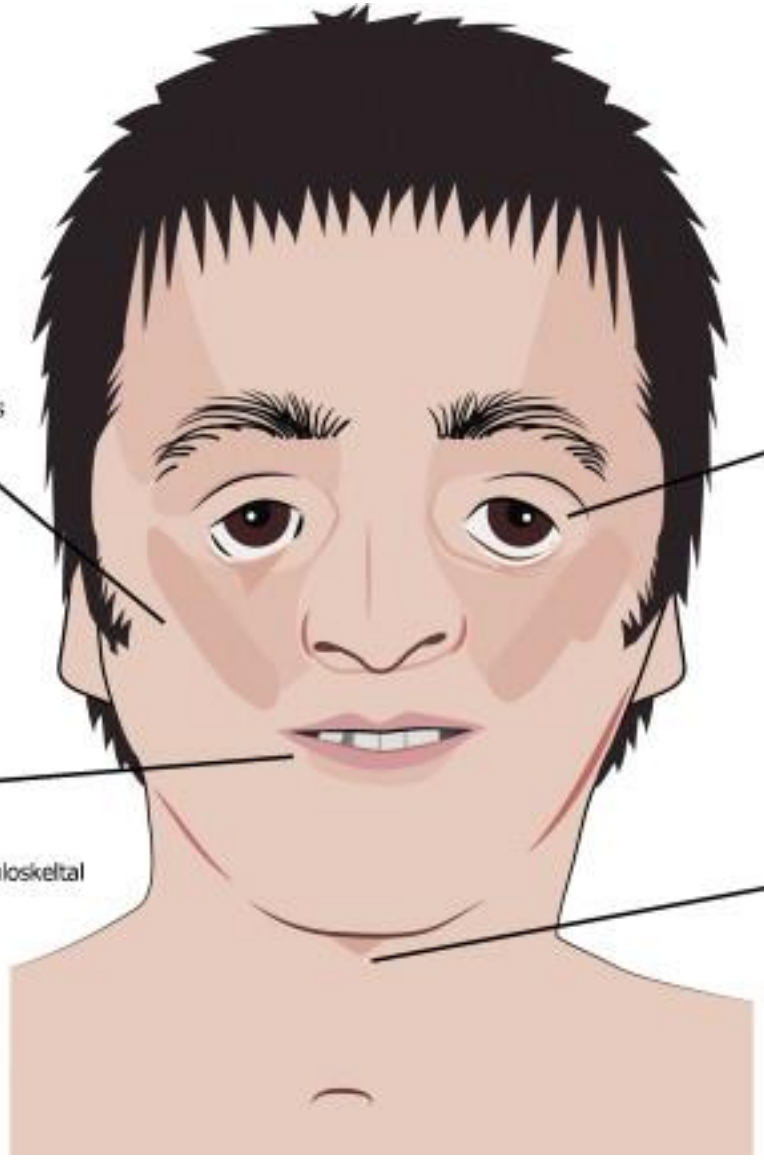
Malformed or absent ears





St George's University of London

Treacher–Collins syndrome



Face
Facial characteristics
- usually present bilaterally and symmetrically

Parotid gland hypoplasia/ aplasia
Pseudo macrorhinia
- apparent large beak like nose because of lack of malar development and hypoplastic supraorbital ridges

Sideburn hair on cheek 25%

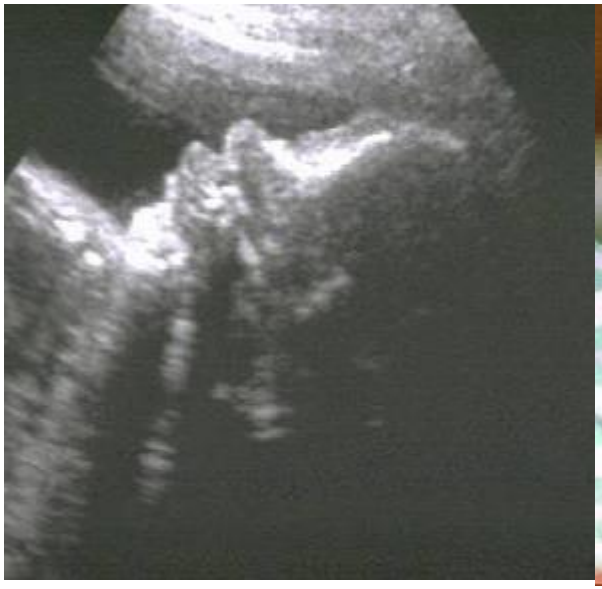
Eyes/ eyelids
Lower lid eyelashes aplasia
Short and downslanting palpebral fissures
Hypertelorism
Notched upper/ lower eyelids (coloboma)
Euryblepharon

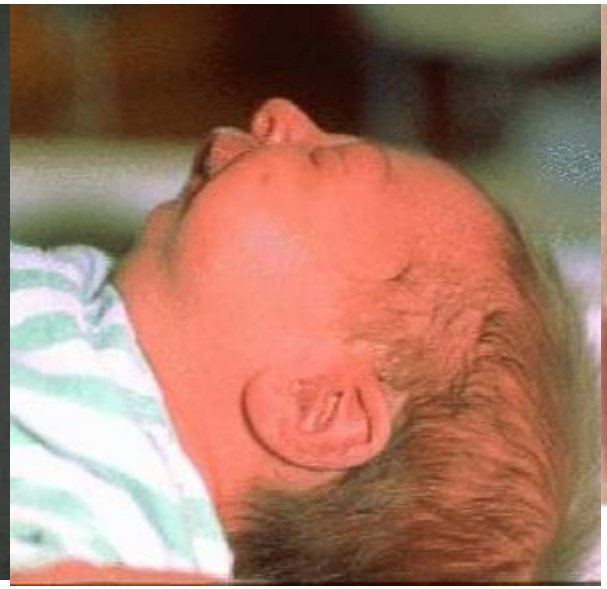
Mouth
Macrostomia 15%
Cleft palate 33%
Velopharyngeal incompetence
Difficulties with swallowing and feeding secondary to musculoskeltal underdevelopment and cleft palate
High-arched palate
Dental anomalies 60%
tooth agenesis 33%
enamel deformities 20%
malposition of maxillary first molars 13%
Hypoplastic and retropositioned tongue

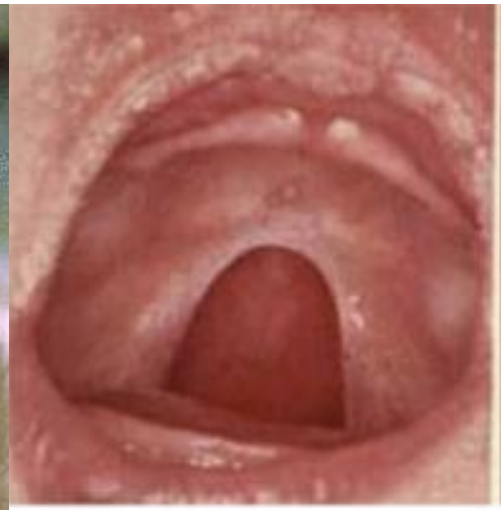
Airways
Airway problems secondary to mandibular hypoplasia
Pharyngeal hypoplasia
Choanal atresia
Tracheo-oesophageal fistula
Small or obstructed nasal passages

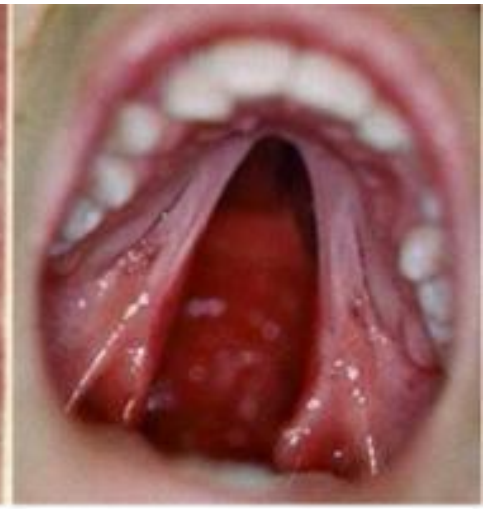
St George's University of London

Pierre–Robin Anomalad syndrome







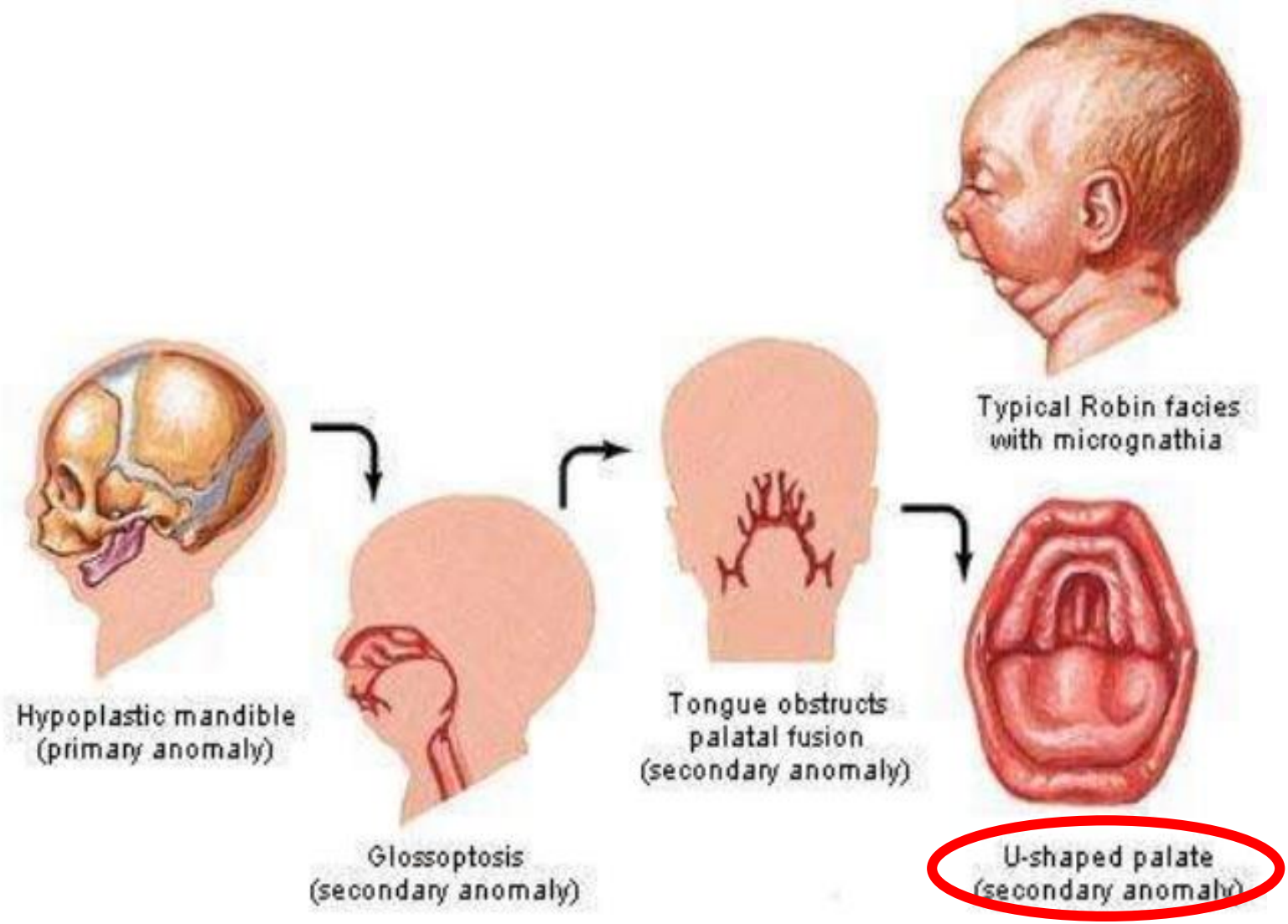


"U" shaped cleft palate

"V" shaped cleft palate

St George's University of London

Pierre–Robin Anomalad syndrome



Management

Investigations:

- Detailed ultrasound examination
- Invasive testing for karyotyping and array

Follow-up:

- Standard
- Prenatal consultation with multidisciplinary team

Delivery:

- Standard obstetric care and delivery

Prognosis

- Depends on the associated anomalies
- Isolated:
  - Good prognosis and normal survival
  - Surgical repair is at 3-6 months of age
  - Long-term in children with cleft lip and palate:
    - Dental abnormalities
    - Hearing and olfactory problems
    - Midface hypoplasia
    - Psychological problems

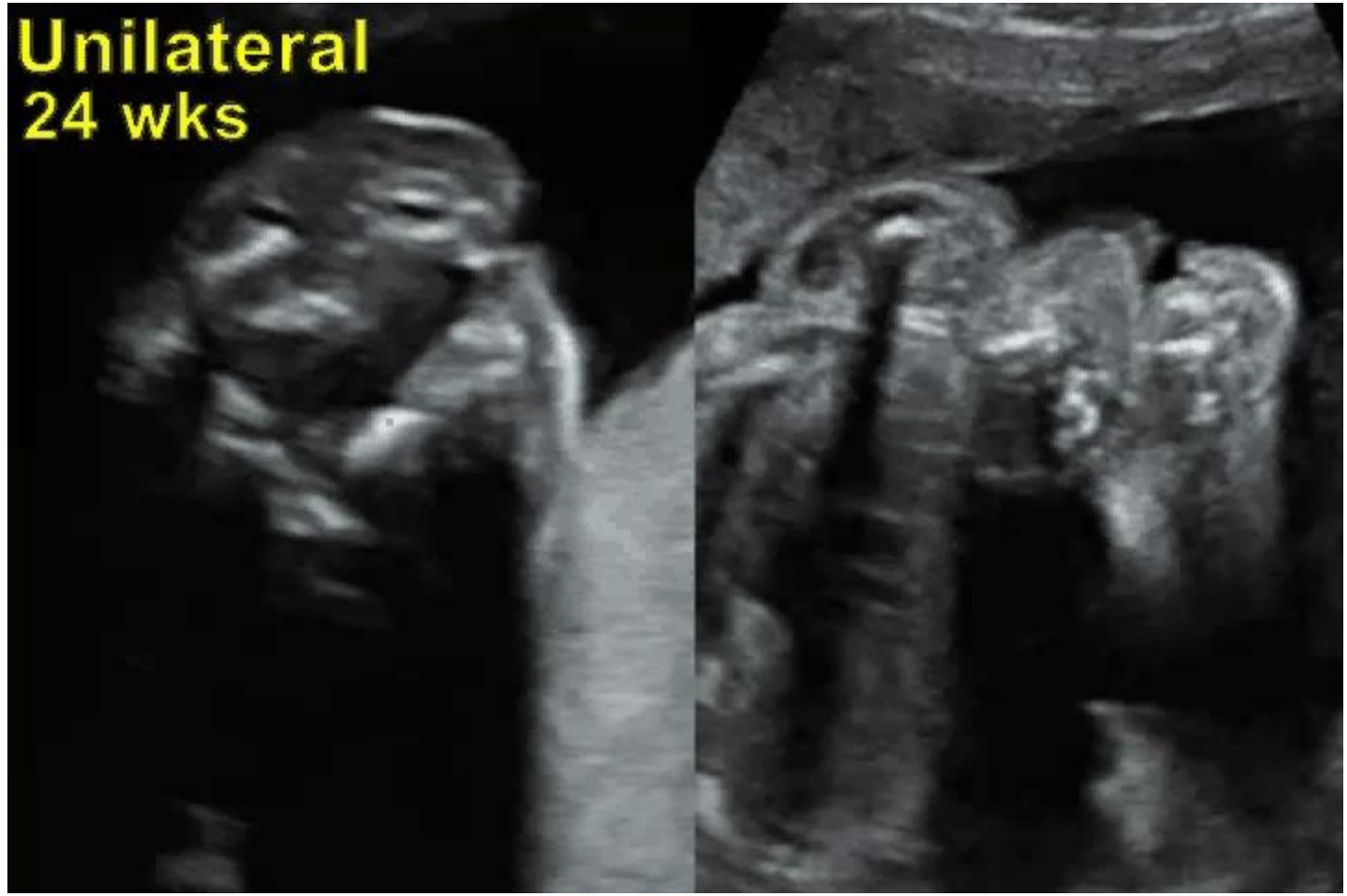
Long-term problems in children with Cleft lip & palate

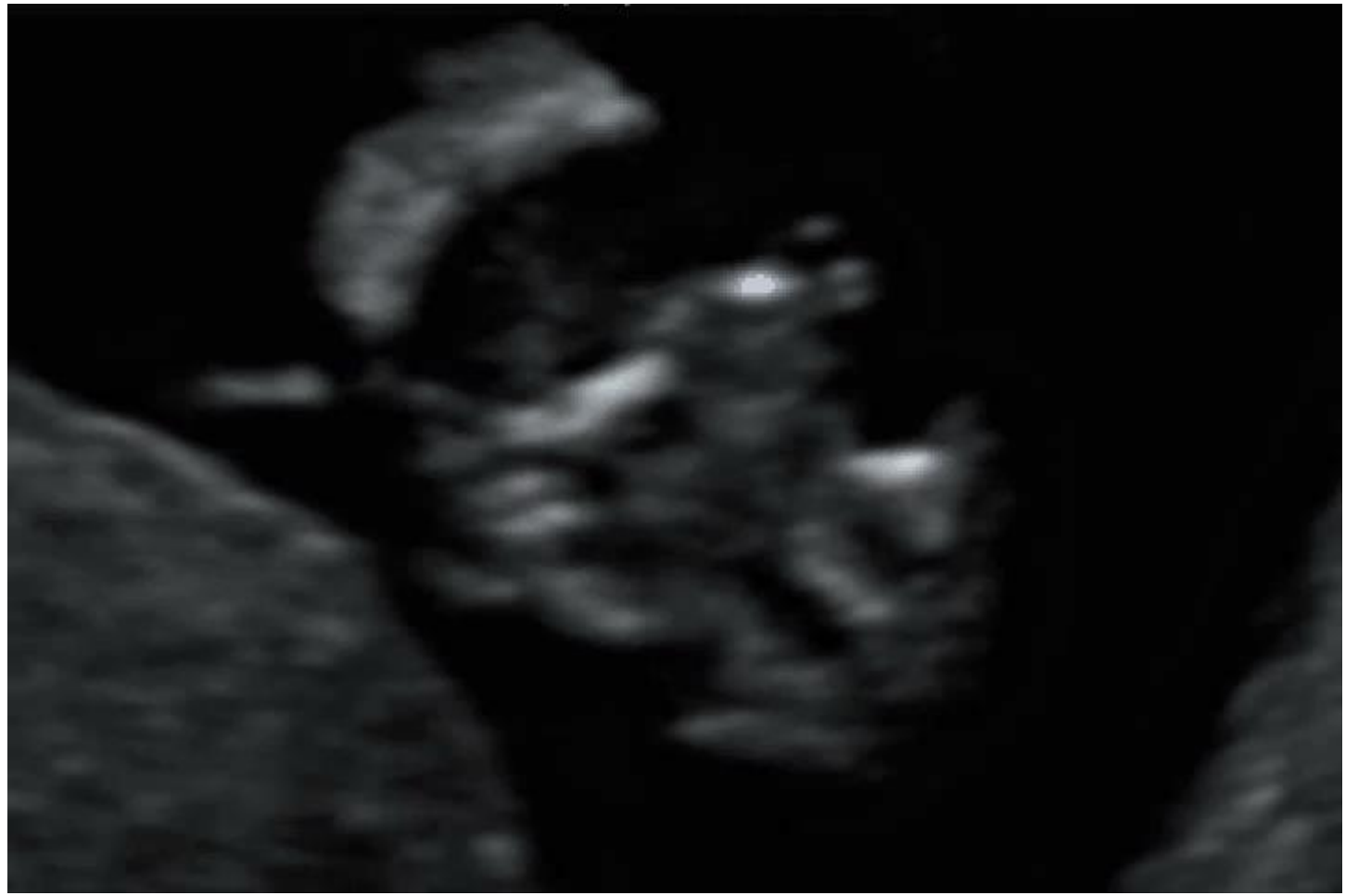
- **25% have speech abnormalities requiring secondary palate surgery and speech therapy**
- **Dental anomalies:**
  - Missing, extra, or malpositioned teeth
  - Require braces on permanent teeth
- **Hearing abnormalities: may require myringotomy with placement of bilateral tympanotomy tubes**
- **Regular psychological screening:**
  - Cognitive development, behaviour, and self-image

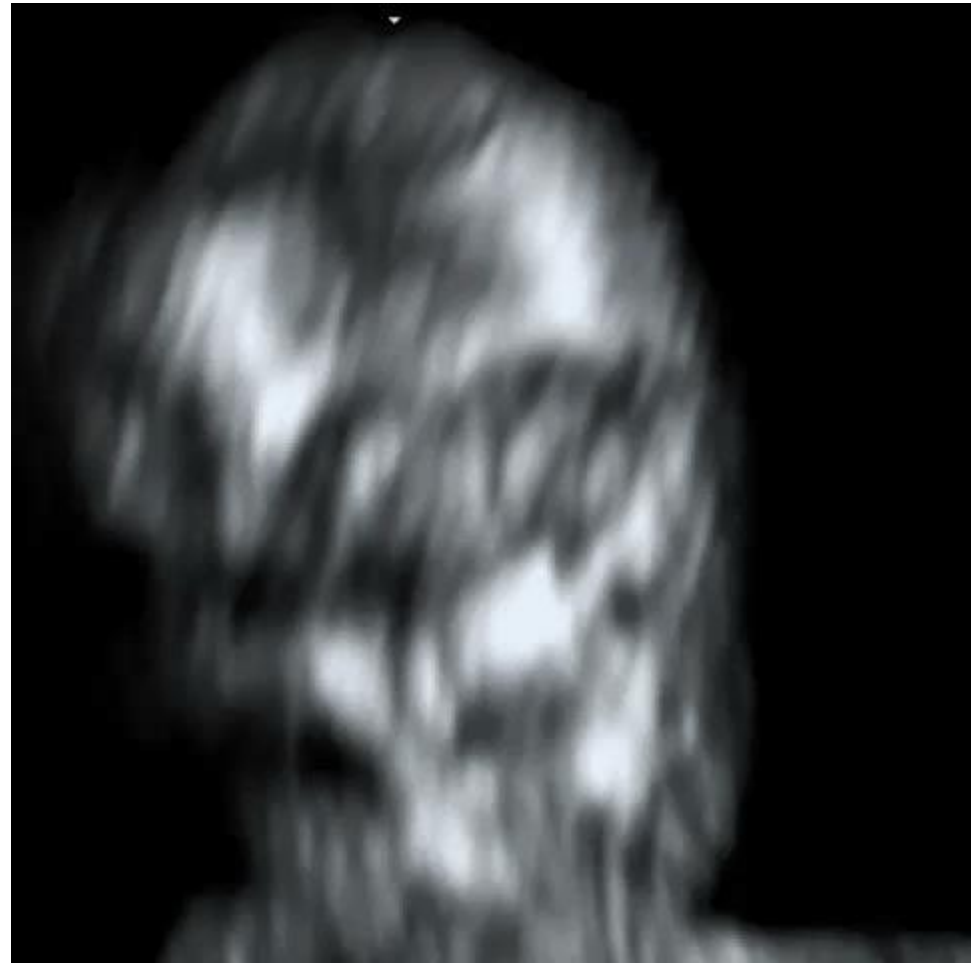
Facial cleft

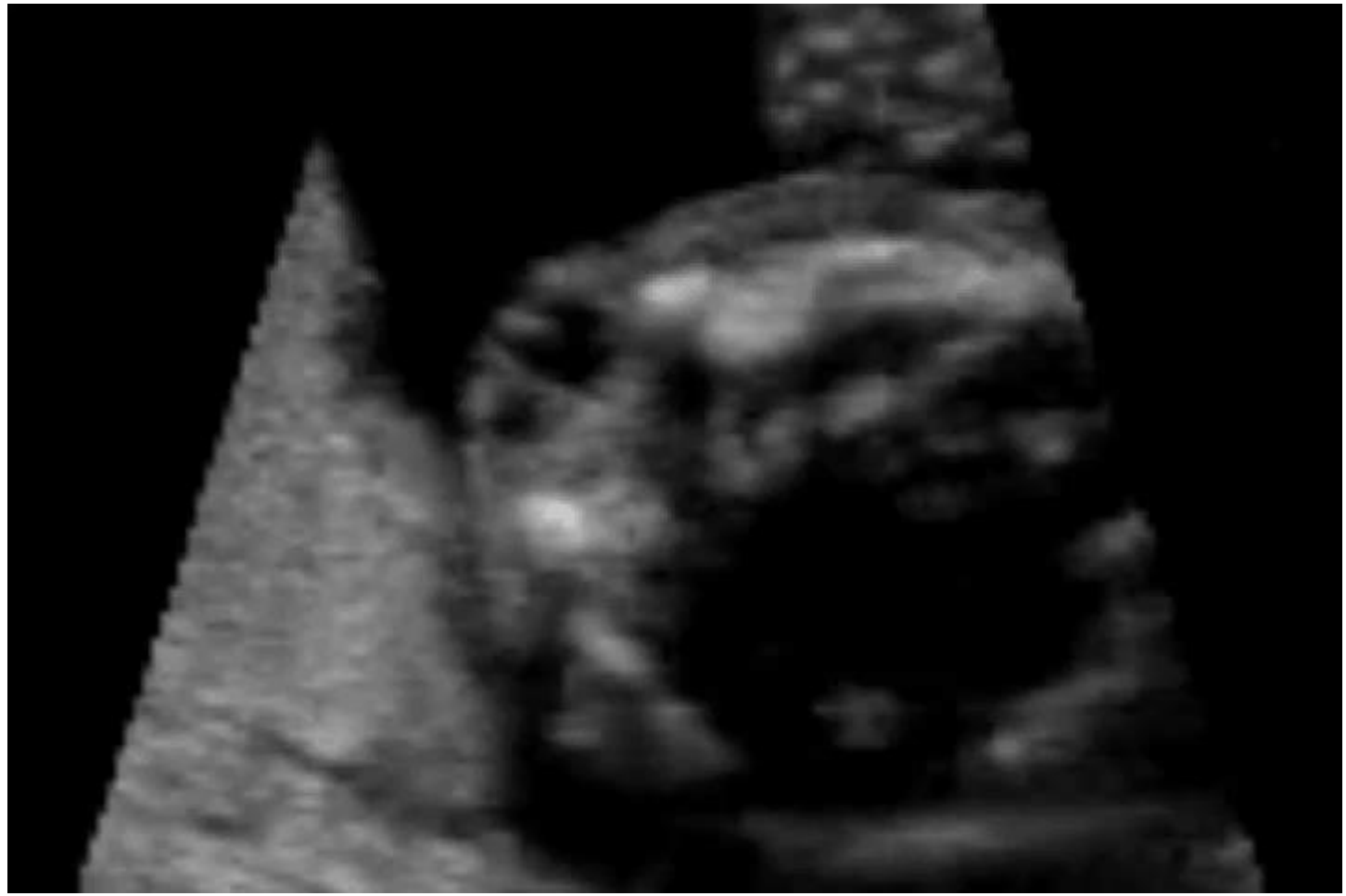
Recurrence

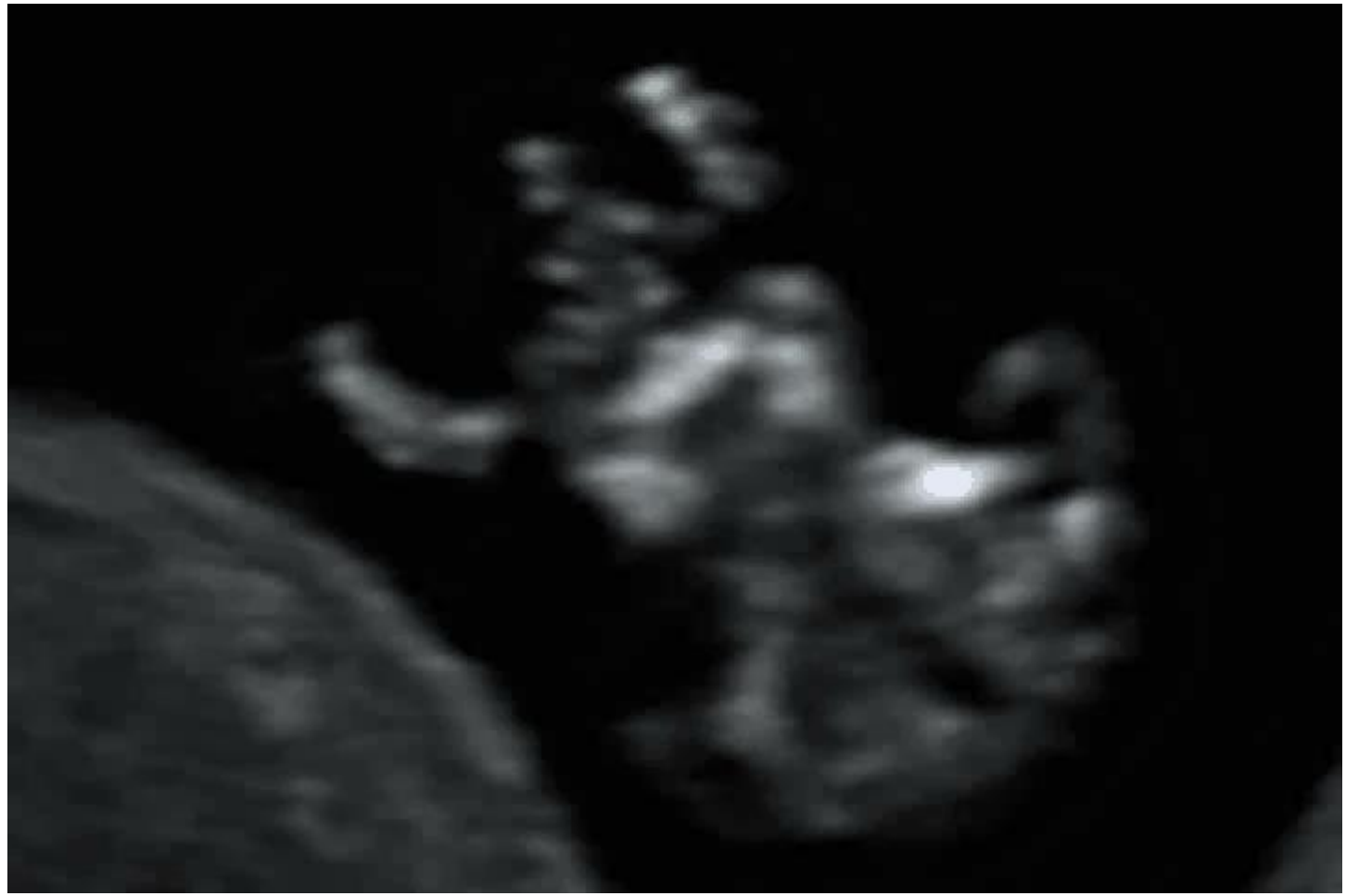
- Isolated:
  - 5% if 1 sibling or parent is affected
  - 10% if 2 siblings are affected
- Syndromic: varies


Unilateral
24 wks

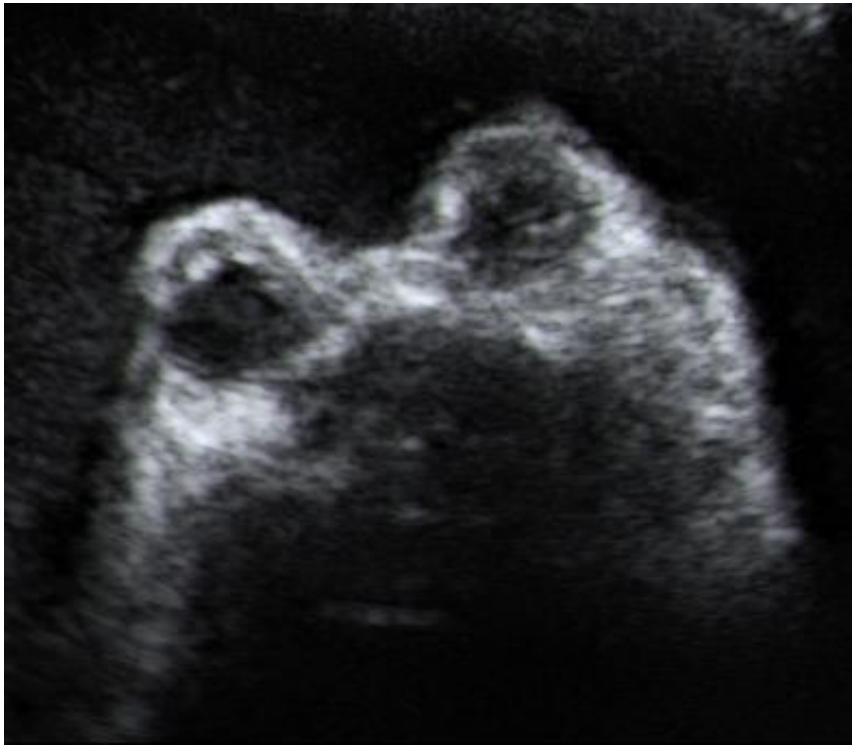


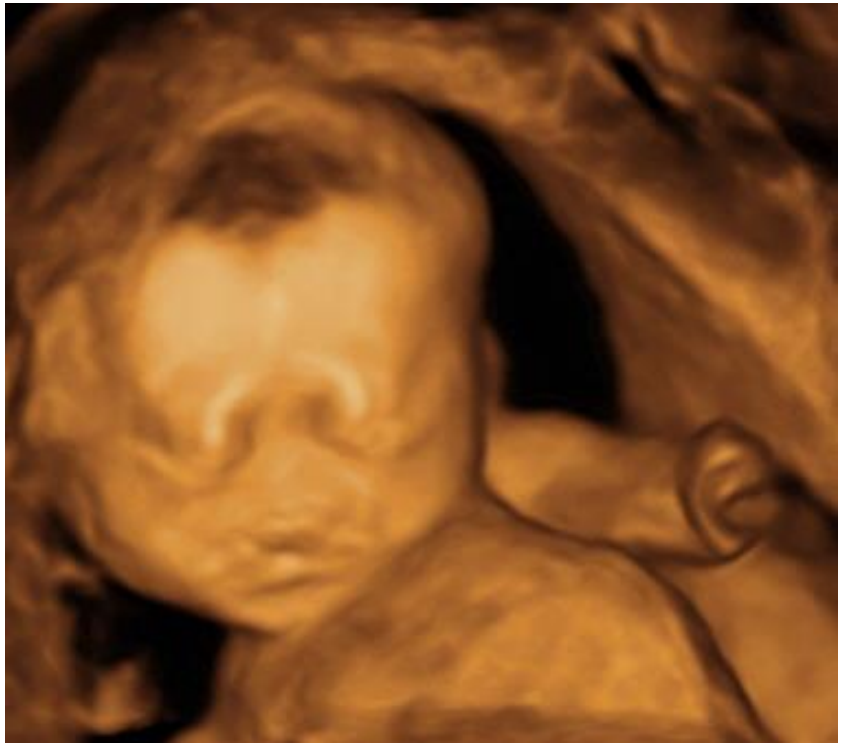




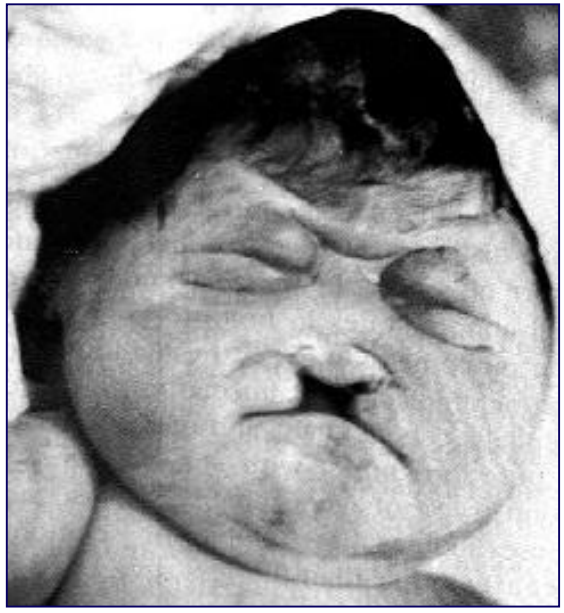


Holoprosencephaly: facial defects

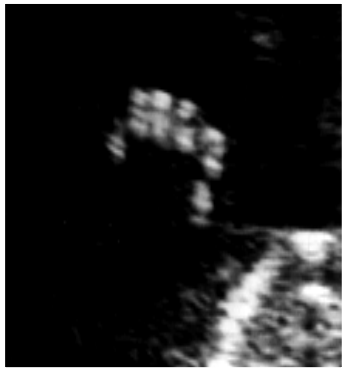


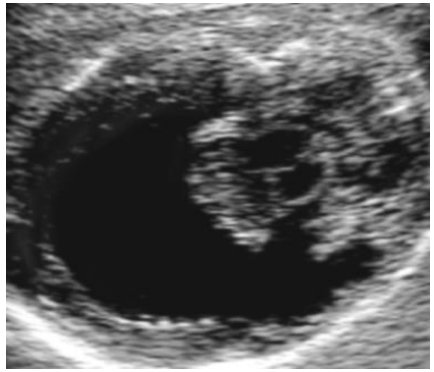


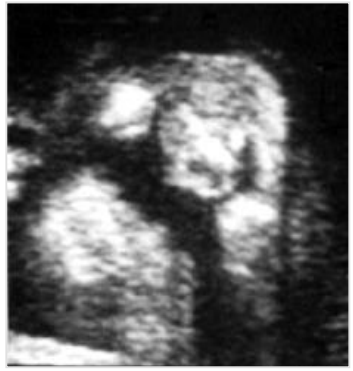
Trisomy 13

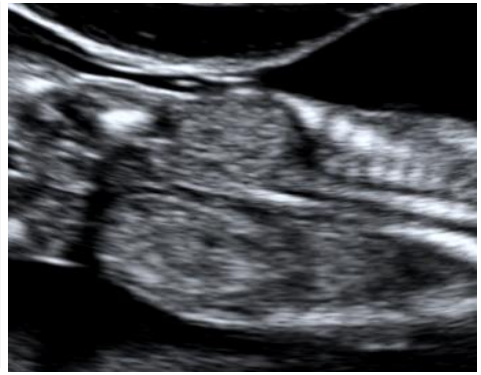


- Holoprocencephaly
- Anophthalmia/microphthalmia
- Abnormal nose
- Facial cleft
- Cardiac abnormalities
- Exomphalos
- Renal abnormalities
- Postaxial polydactyly
- Myelomeningocele





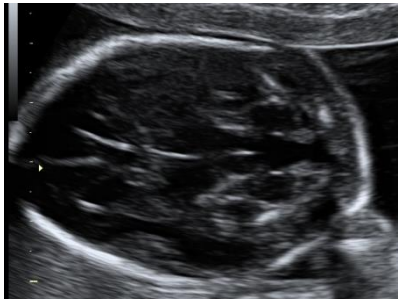


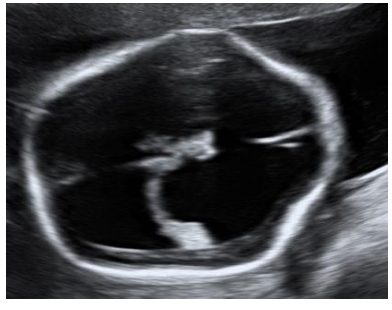


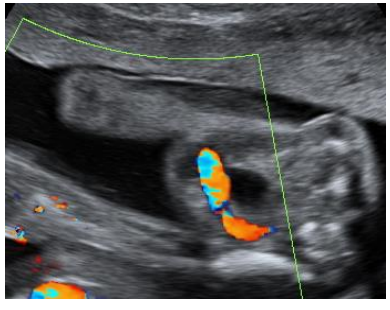
The Fetal Medicine Foundation

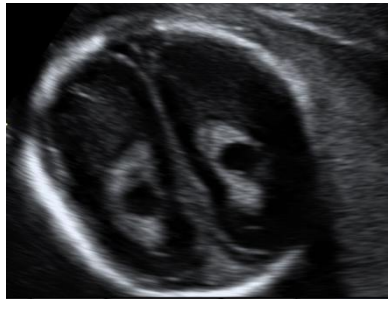
Trisomy 18

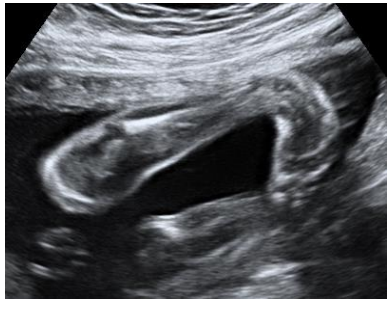
- Strawberry-shaped head
- Choroid plexus cysts
- Absent corpus callosum
- Ventriculomegaly
- Dandy-Walker complex
- Facial cleft
- Micrognathia
- Nuchal edema
- Cardiac defects
- Diaphragmatic hernia
- Esophageal atresia
- Exomphalos
- 2-vessel cord
- Renal defects
- Myelomeningocele
- Short limbs
- Radial aplasia/hypoplasia
- Overlapping fingers
- Talipes / rocker bottom feet
- Growth restriction

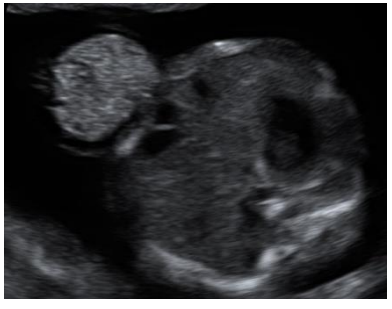


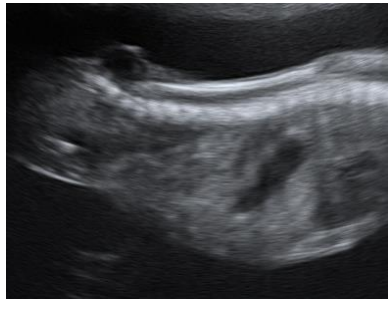








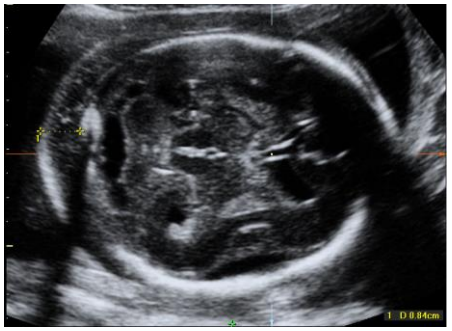


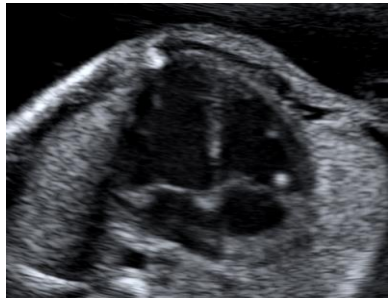


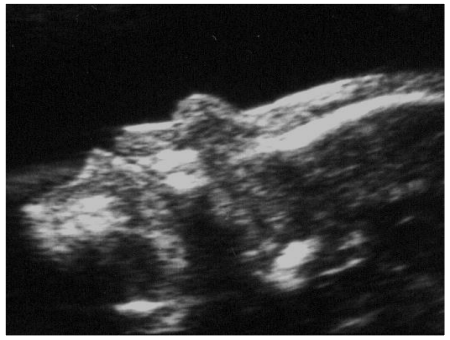
The Fetal Medicine Foundation

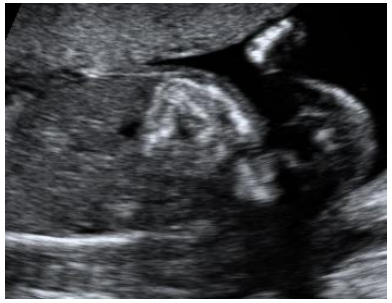
Trisomy 21

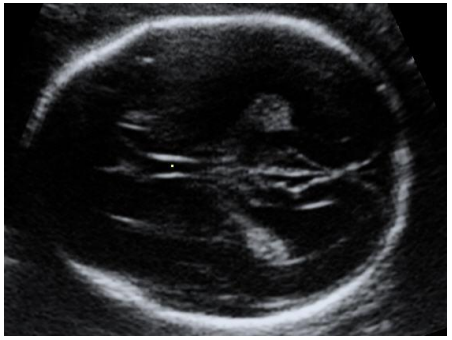
- Brachycephaly
- Mild ventriculomegaly
- Nasal hypoplasia
- Nuchal edema
- Cardiac defects (AVSD)
- Echogenic focus
- Duodenal atresia
- Hyperechogenic bowel
- Shortening of femur
- Shortening of humerus
- Saldal gap
- Clinodactily

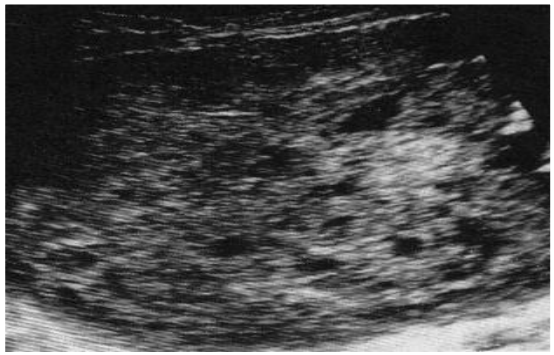


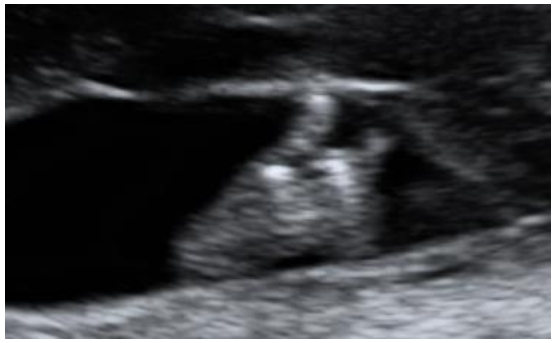


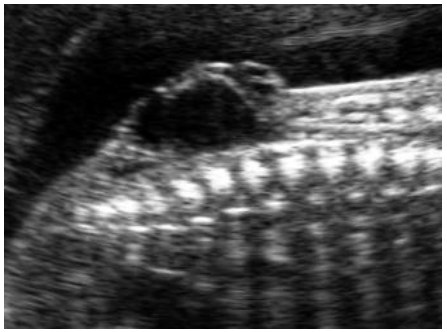






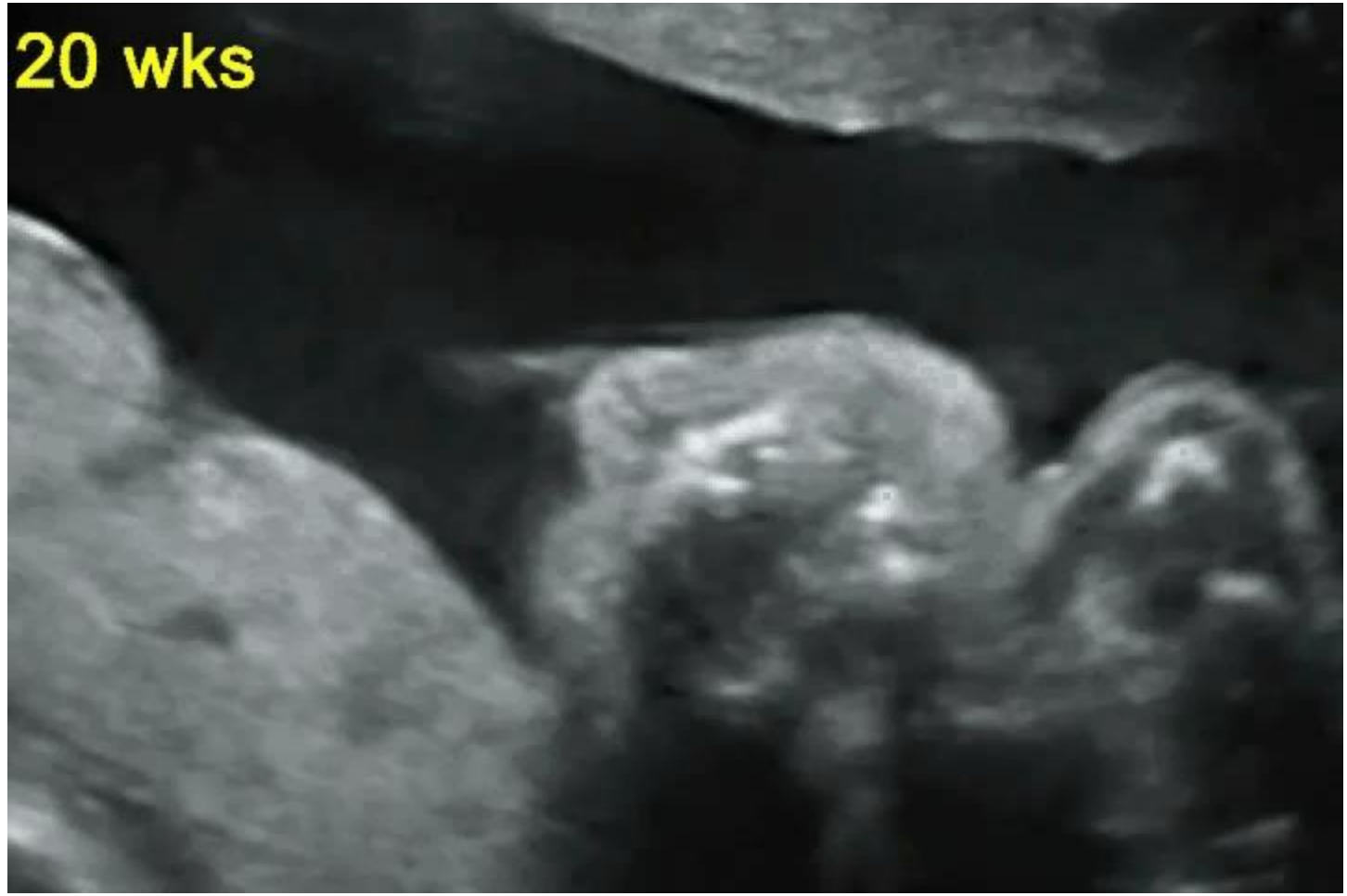






Triploidy

- Molar placenta
- Asymmetrical growth restriction
- Ventriculomegaly (mild)
- Micrognathia
- Cardiac abnormalities
- Exomphalos
- Myelomeningocele
- 3rd-4th fingers syndactyly
- “Hitch-hiker” toe deformity


20 wks

St George's University of London

Hypertelorism

- **Prevalence:** 1 in 20,000 births
- **Diagnosis:** interorbital diameter >95th percentile
- **Associated abnormalities:**
- Chromosomal defects (mainly trisomy 13) are very rare
- Genetic syndromes are found in >50% of cases. The most common are:
  - Frontonasal dysplasia (sporadic; hypertelorism, midline facial cleft, abnormalities of the nose, cranium bifidum occultum)
  - Craniosynostosis (Apert, Carpenter, Crouzon)
  - Neu-Laxova syndrome (AR; hypertelorism, microcephaly, ACC, contractures in the upper and lower limbs, FGR)
- Associated defects: frontal encephalocele and ACC

St George's University of London

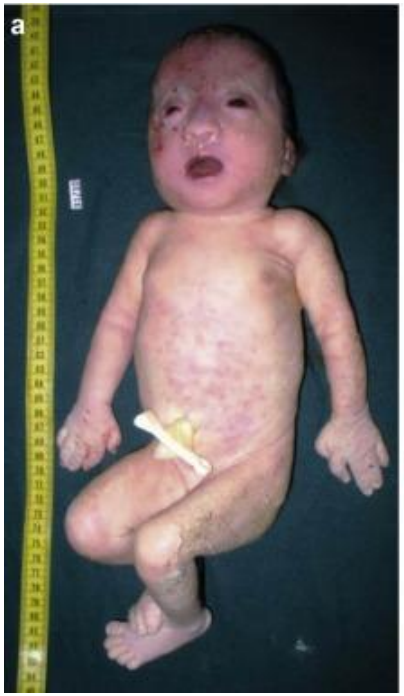
Neu-Laxova syndrome

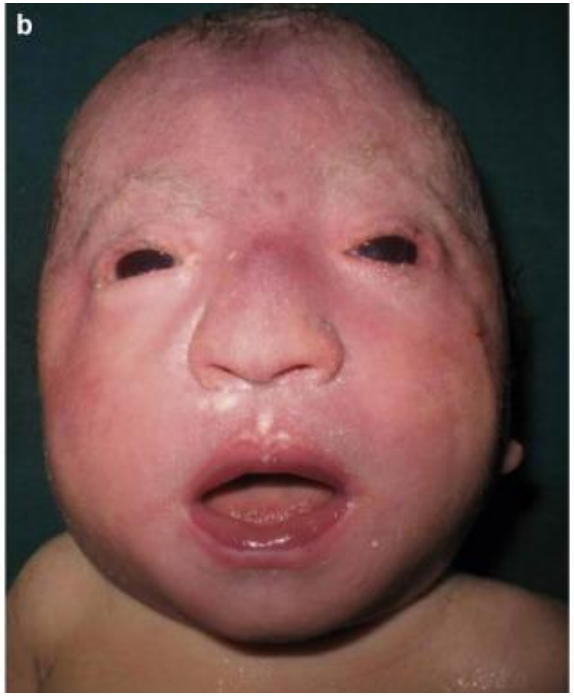
Hypertelorism

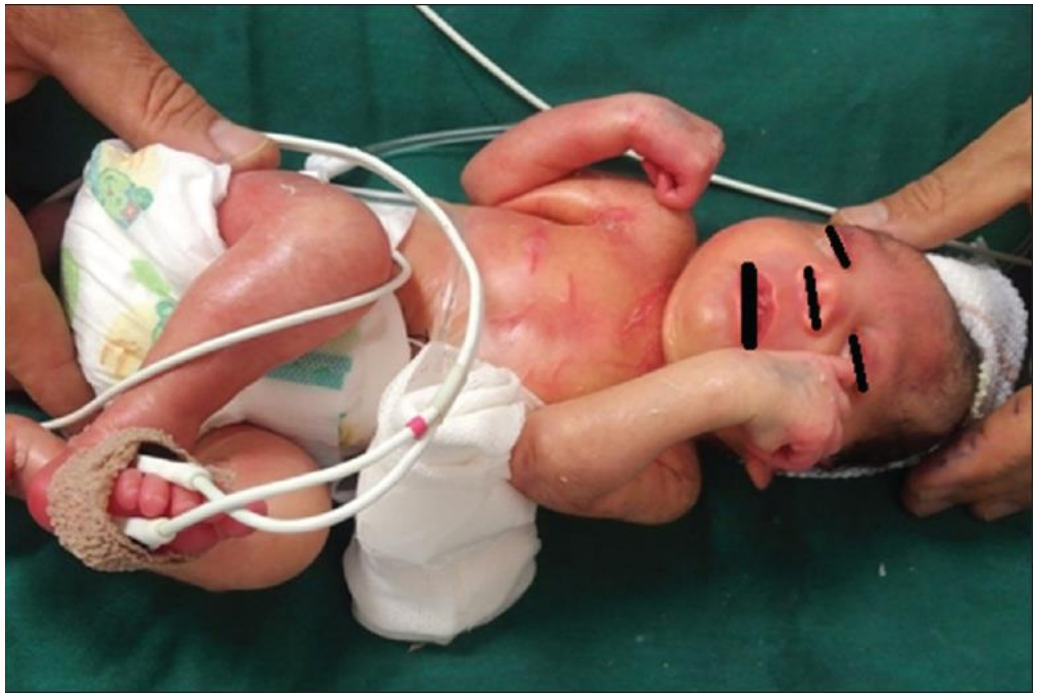
Microcephaly

Contractures in the upper and lower limbs

Fetal growth restriction

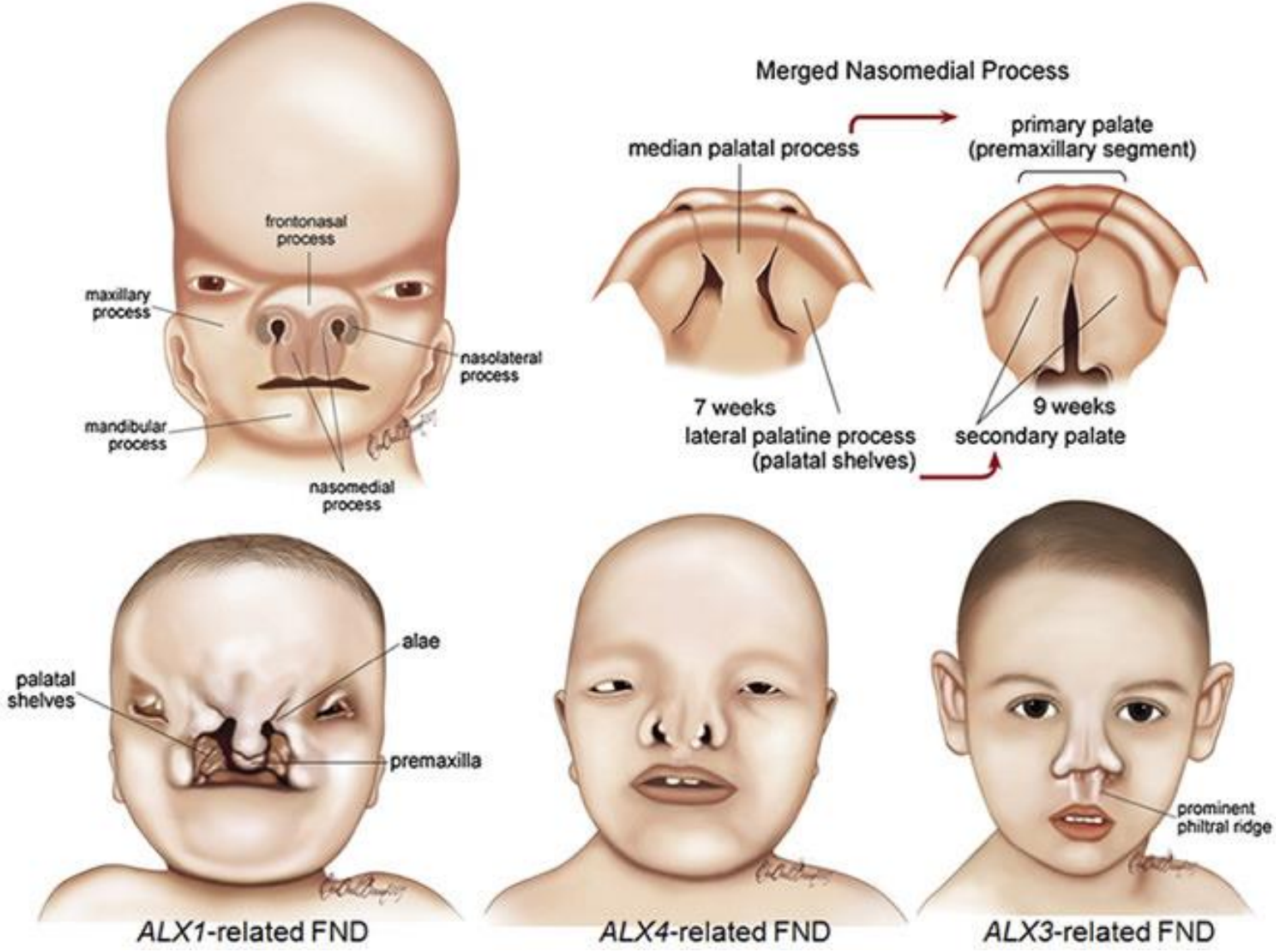






St George's University of London

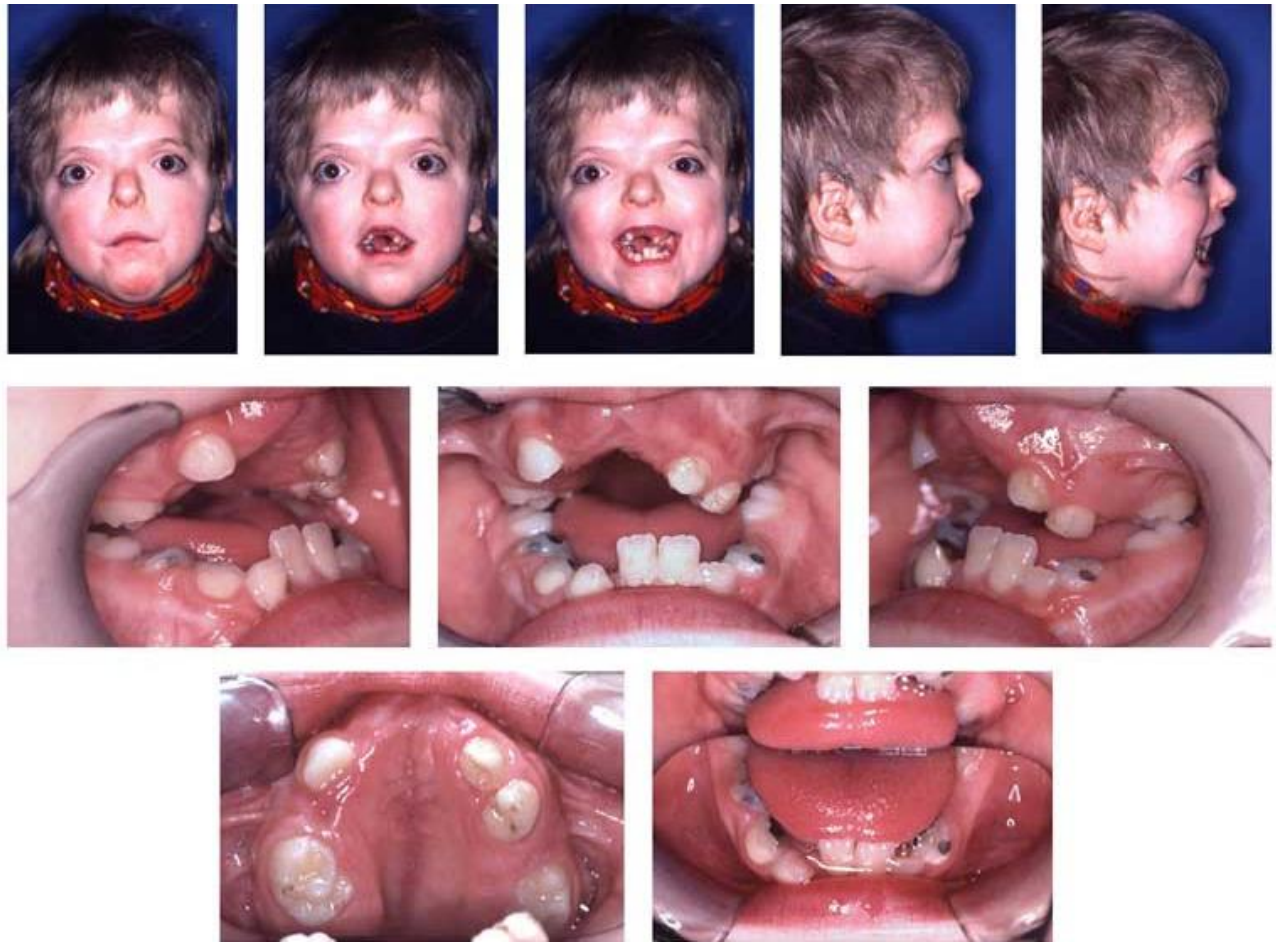
Frontonasal dysplasia



St George's University of London

Apert craniosynostosis

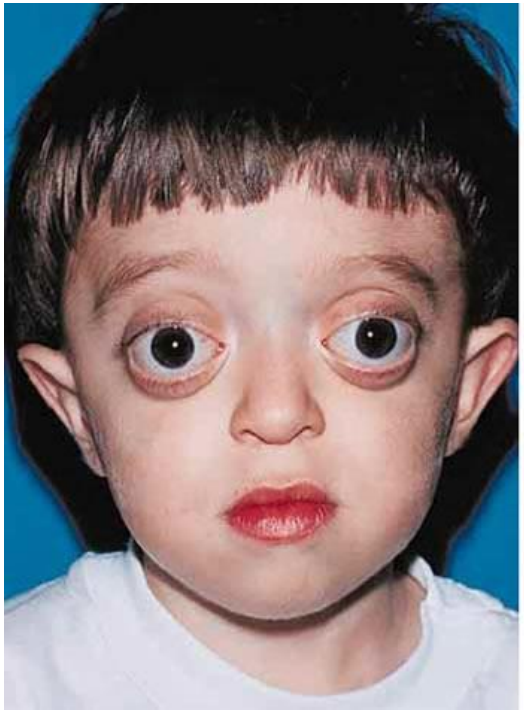
AD, syndactyly of hands and feet, heart defects

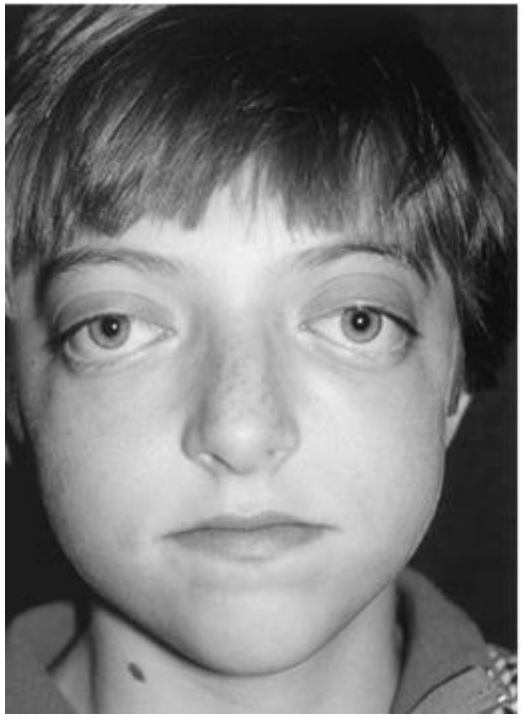
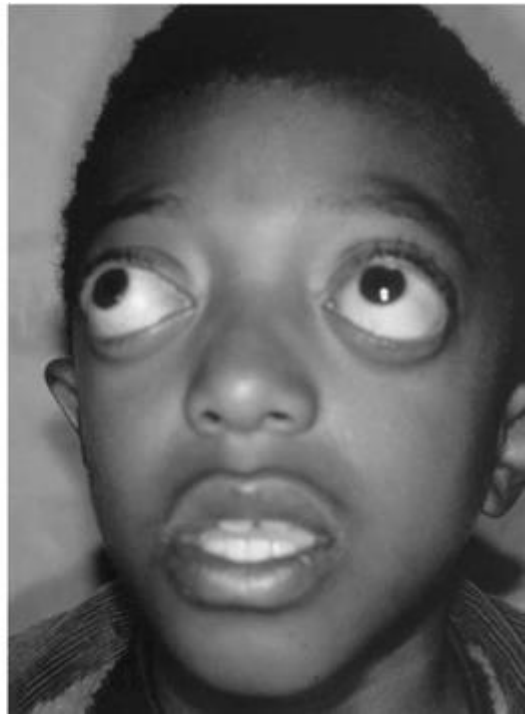


St George's University of London

Craniosynostosis

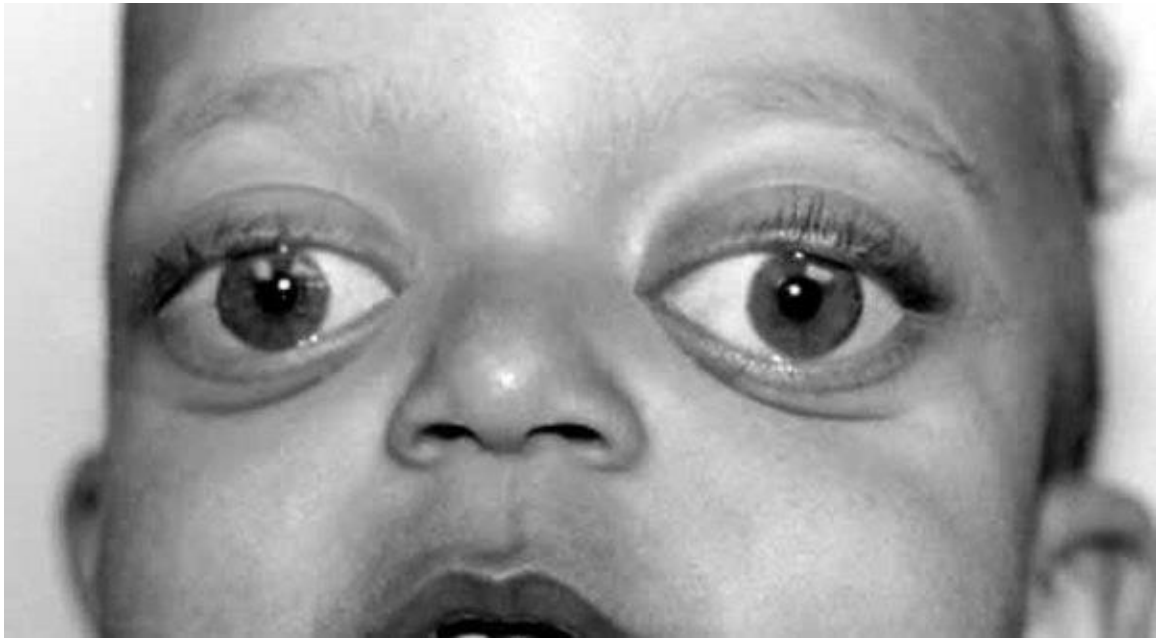
Crouzon craniosynostosis



Carpenter craniosynostosis

AR, polysyndactyly



St George's University of London

Hypertelorism

Management

Investigations:

- Detailed ultrasound examination
- Invasive testing for karyotyping and array

Follow-up:

- Isolated: follow-up should be standard
- Syndromic: antenatal care should be adjusted according to the risks of the condition

Delivery:

- Standard obstetric care and delivery

St George's University of London

Hypertelorism

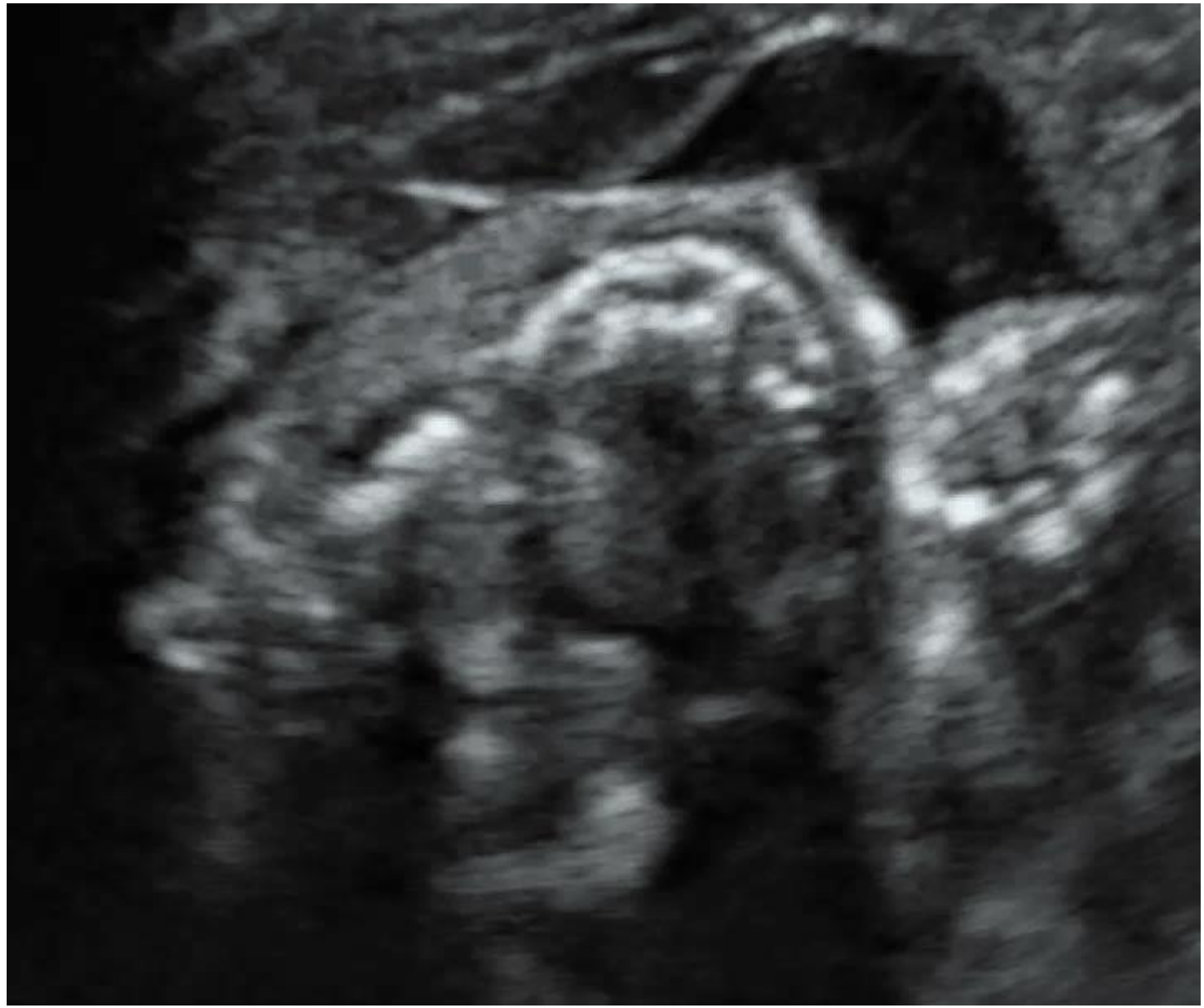
Prognosis

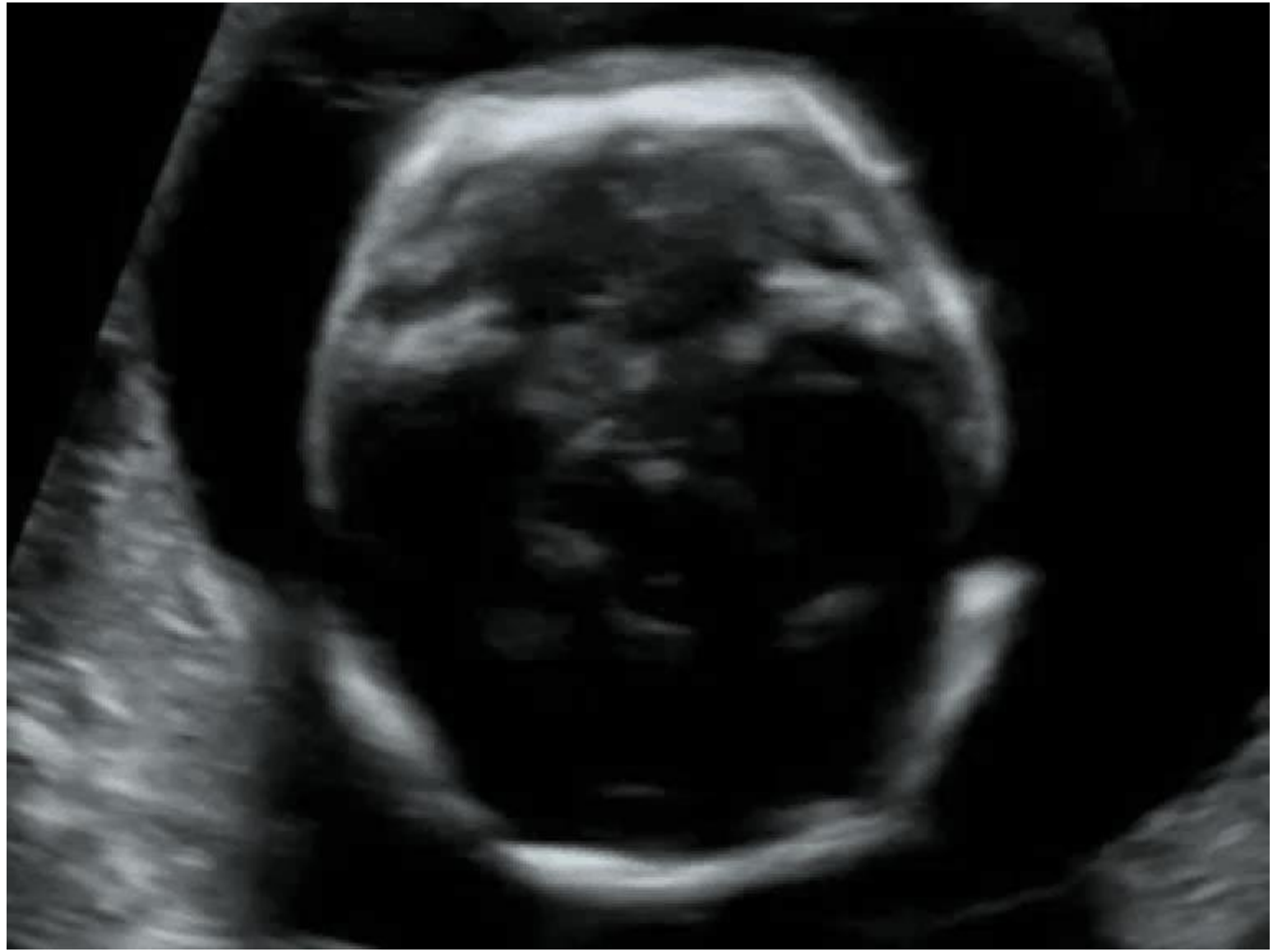
- **Isolated:**
  generally good but in severe cases the cosmetic implications are important.
  There might be impaired stereoscopic binocular vision

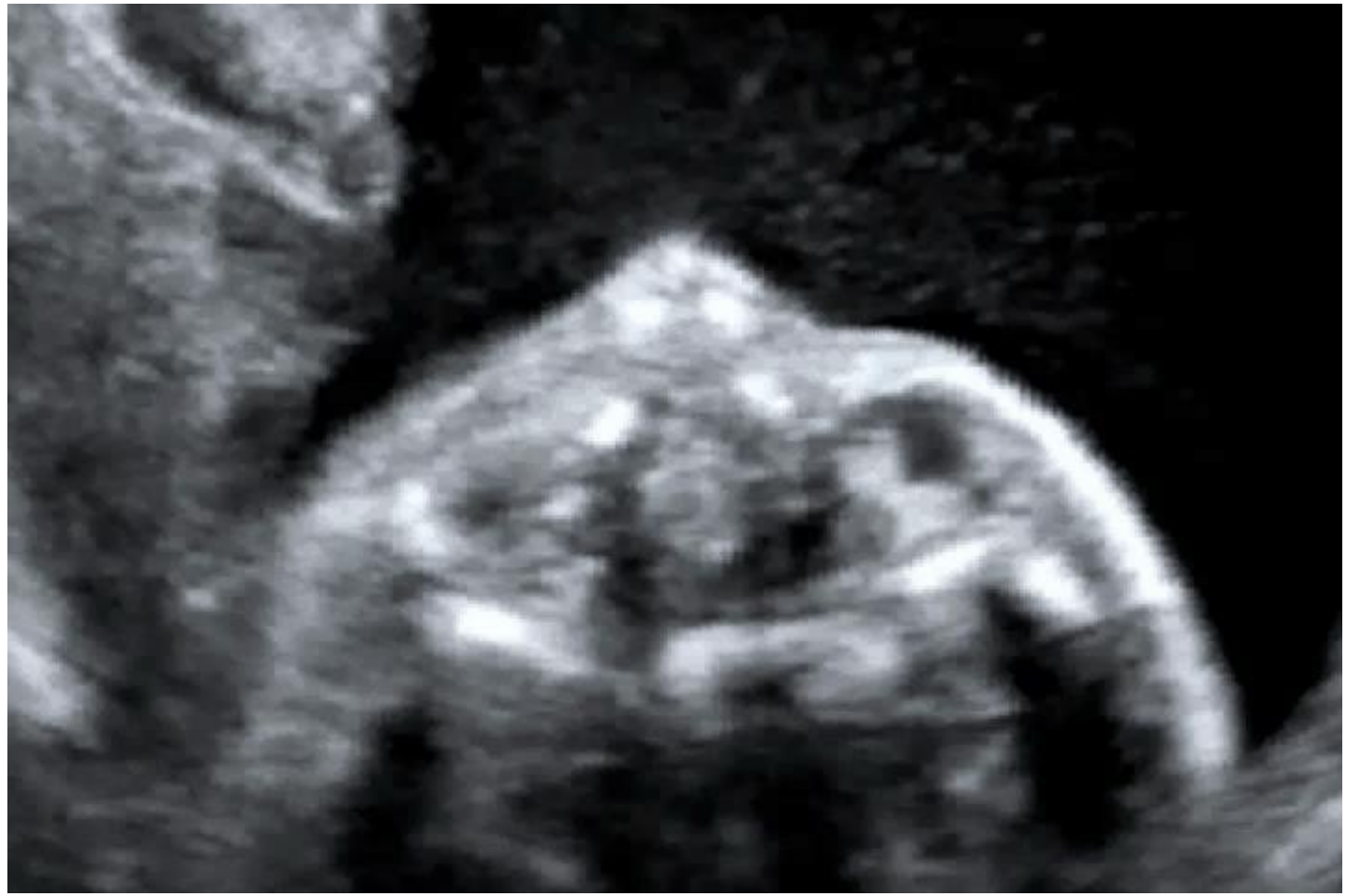
- **Syndromic:**
  generally poor with high risk of neurodevelopmental delay

Recurrence

Isolated: no increased risk of recurrence







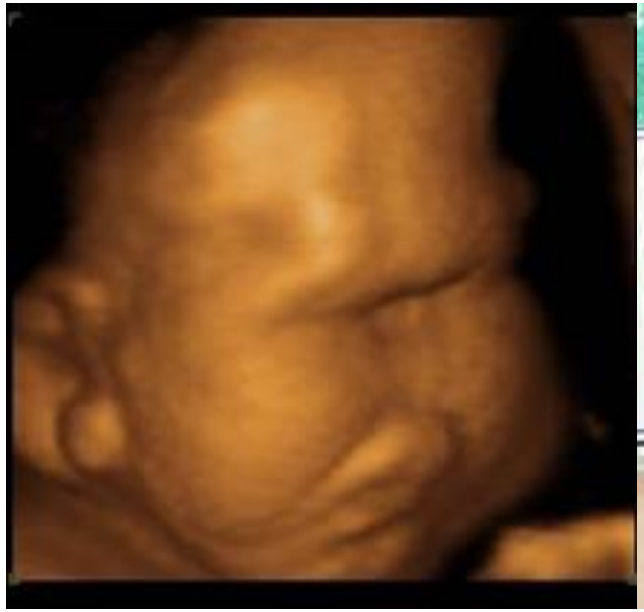
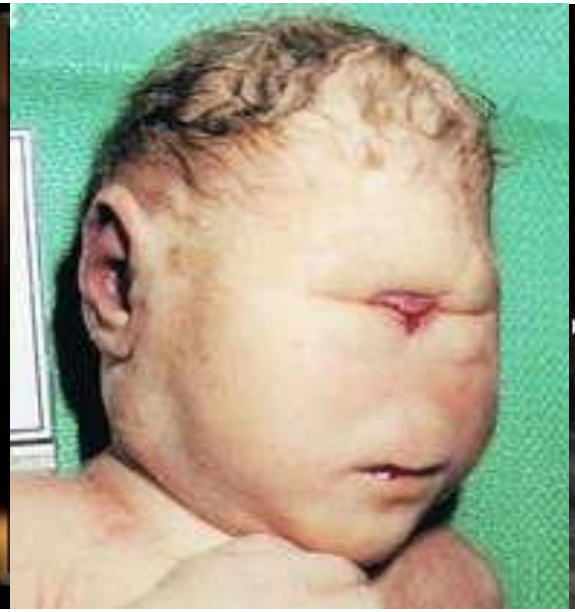
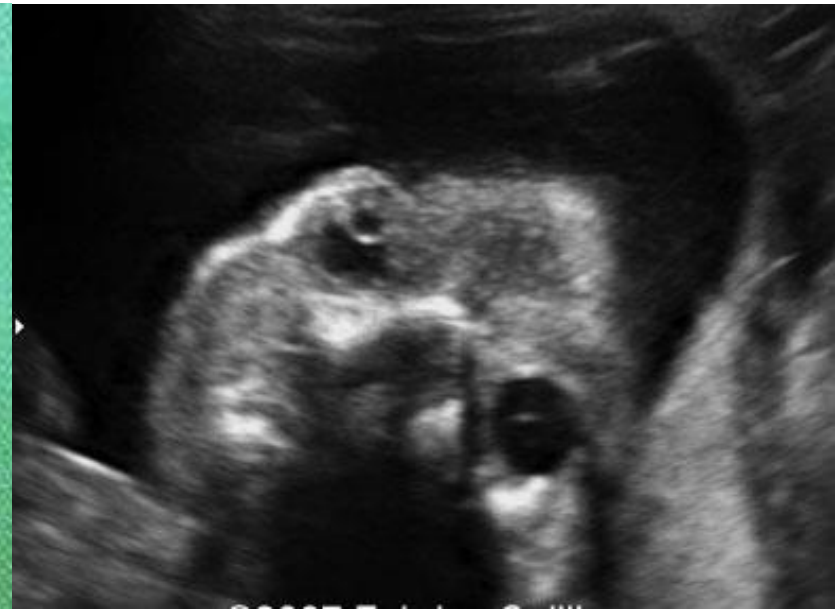
St George's University of London

Hypotelorism

- **Prevalence: 1 in 20,000 births**
- **Diagnosis: inter-orbital diameter <5th centile**

  Part of the midline migration defects together with holoprosecephaly (which is almost always present)

  The degree of hypotelorism can be extreme as in cyclopia

St George's University of London

Hypotelorism

Associated abnormalities:

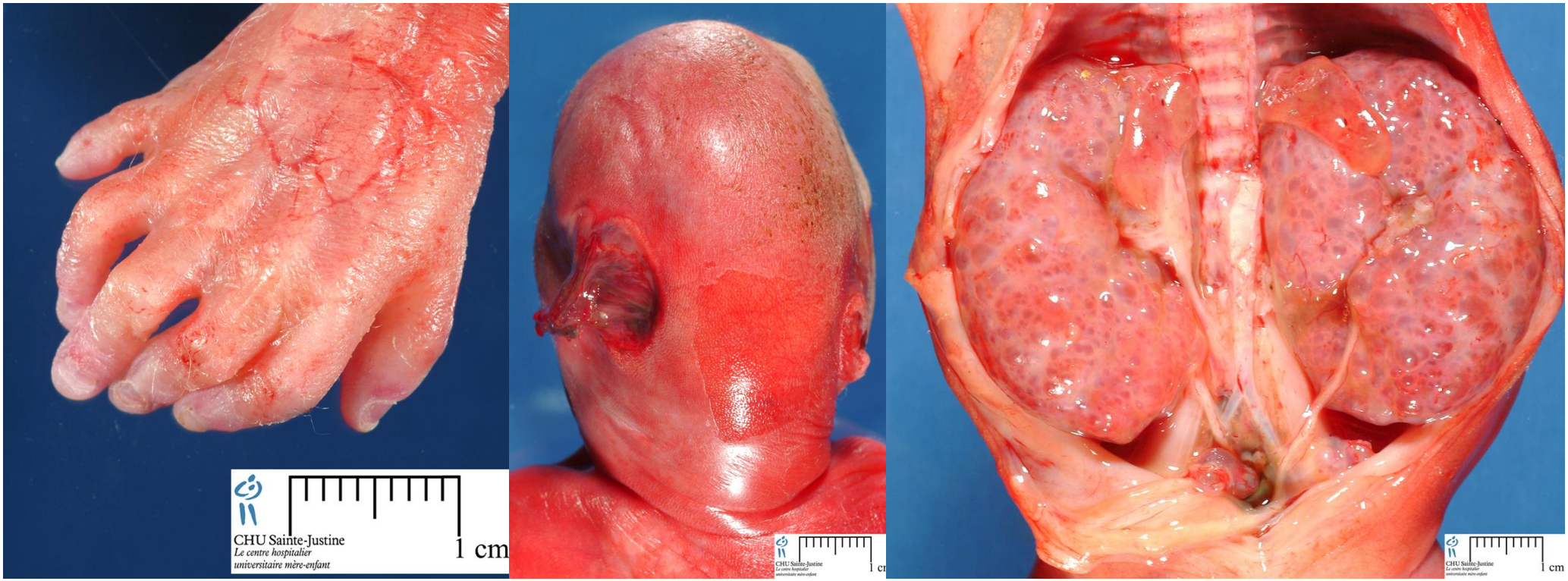
- **Chromosomal defects (mainly trisomy 13) found in 50% of cases**
- **Genetic syndromes are very common**

The most common is:

Meckel-Gruber syndrome (AR, lethal, occipital encephalocele, multicystic kidneys and post-axial polydactyly)

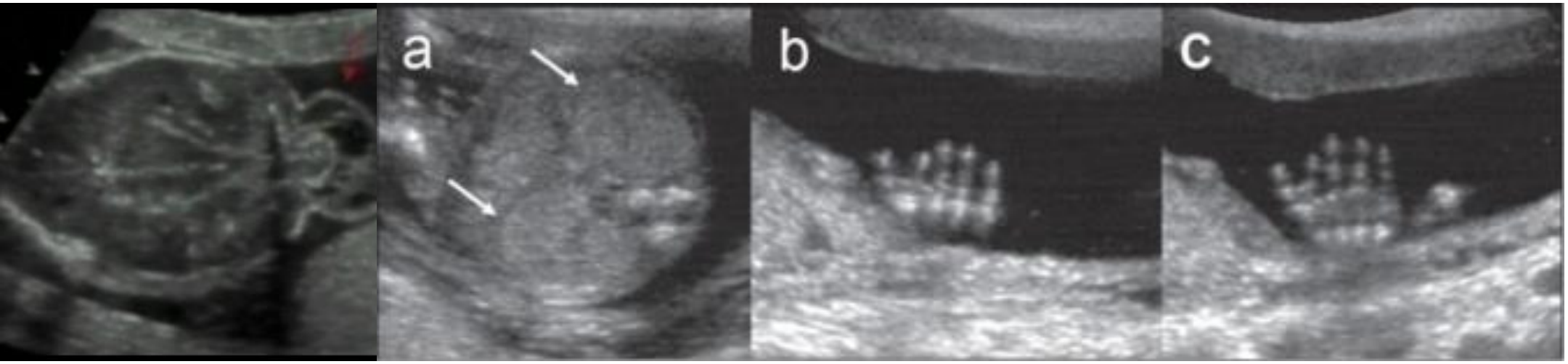
St George's University of London

Meckel-Gruber syndrome



St George's University of London

Meckel-Gruber syndrome



St George's University of London

Hypotelorism

Management

Investigations:

- Detailed ultrasound examination (NEUROSONOGRAPHY)
- Invasive testing for karyotyping and array

Follow-up:

- Follow-up should be standard

Delivery:

- Standard obstetric care and delivery

St George's University of London

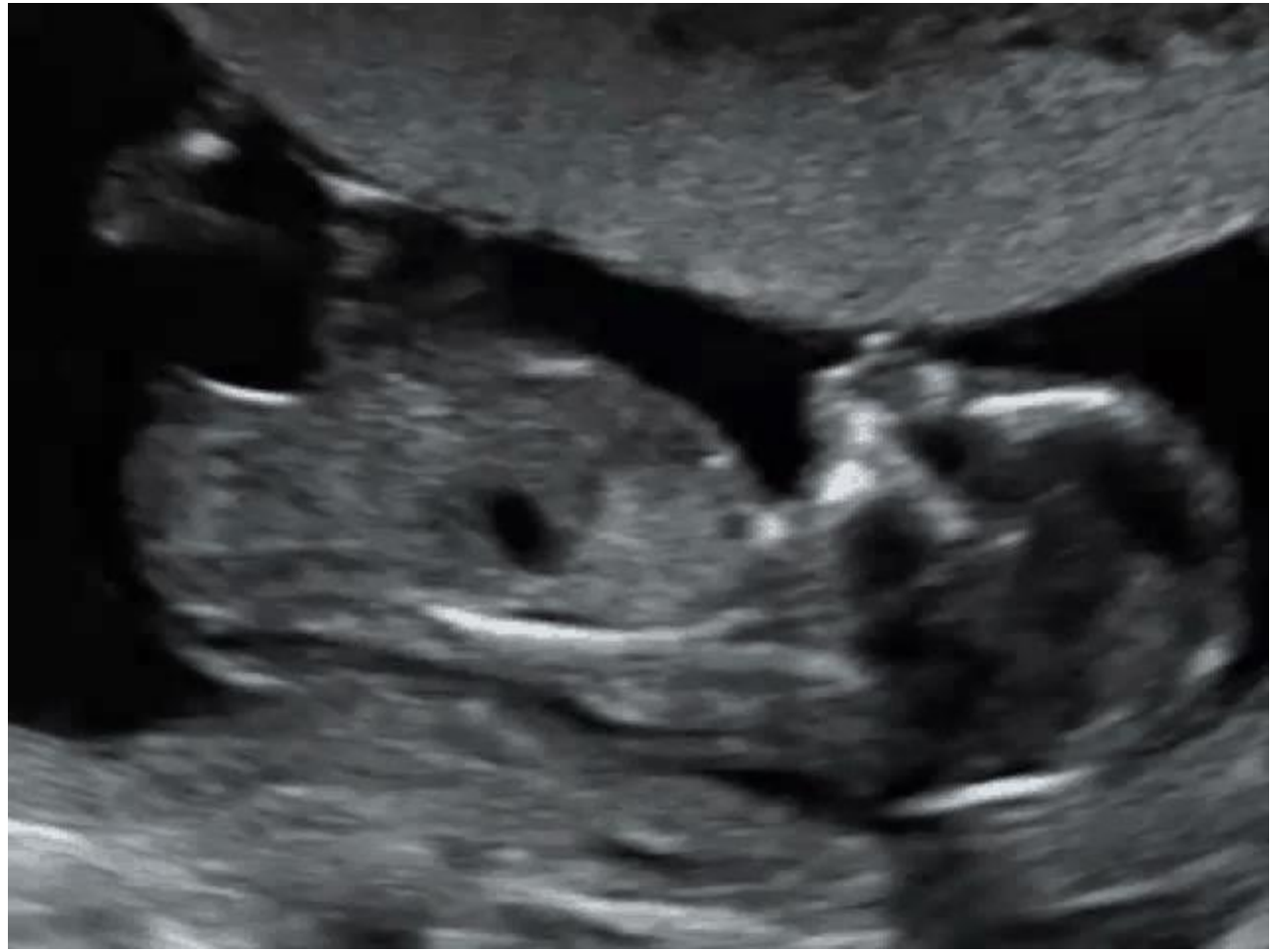
Hypotelorism

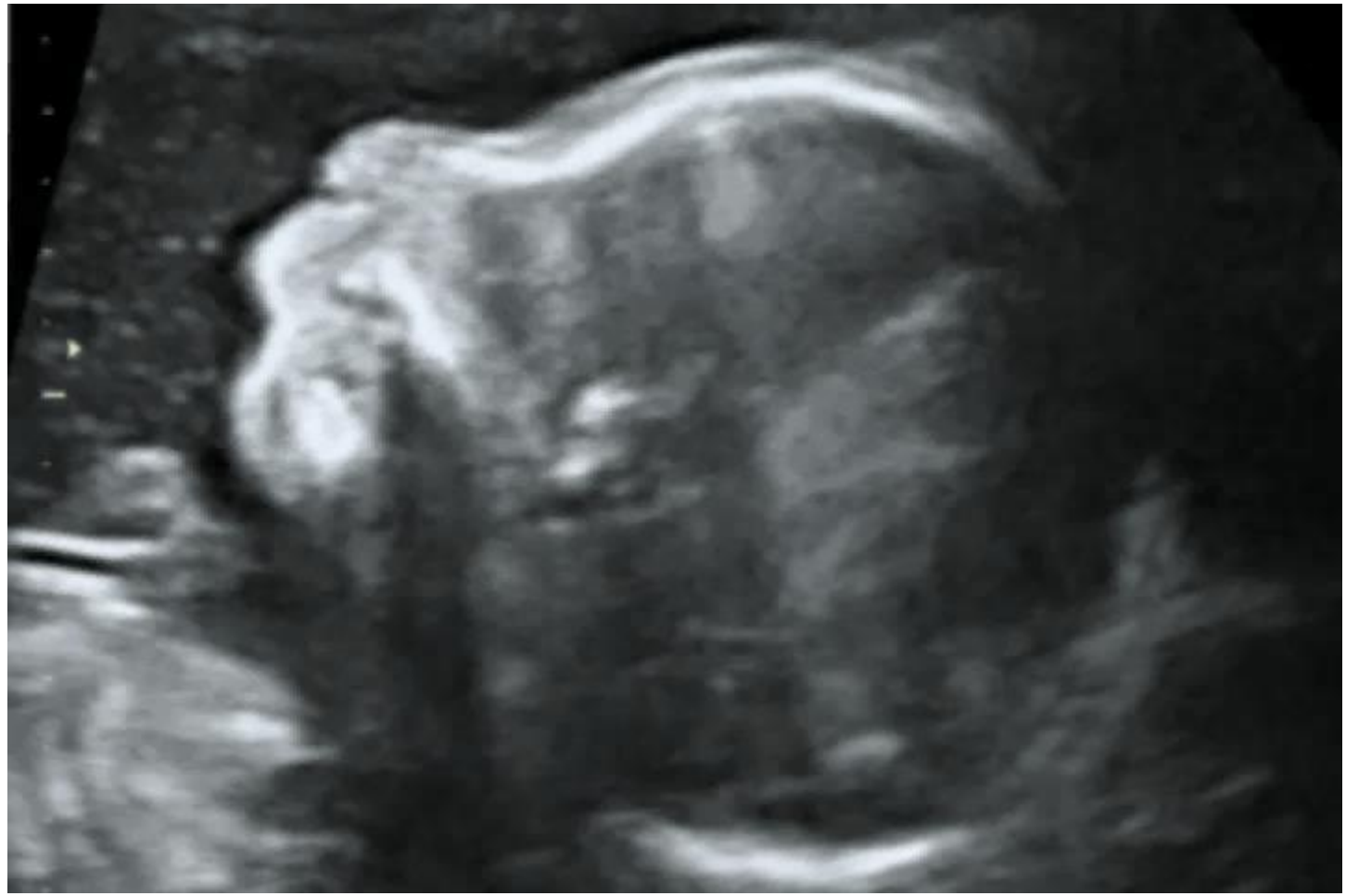
Prognosis

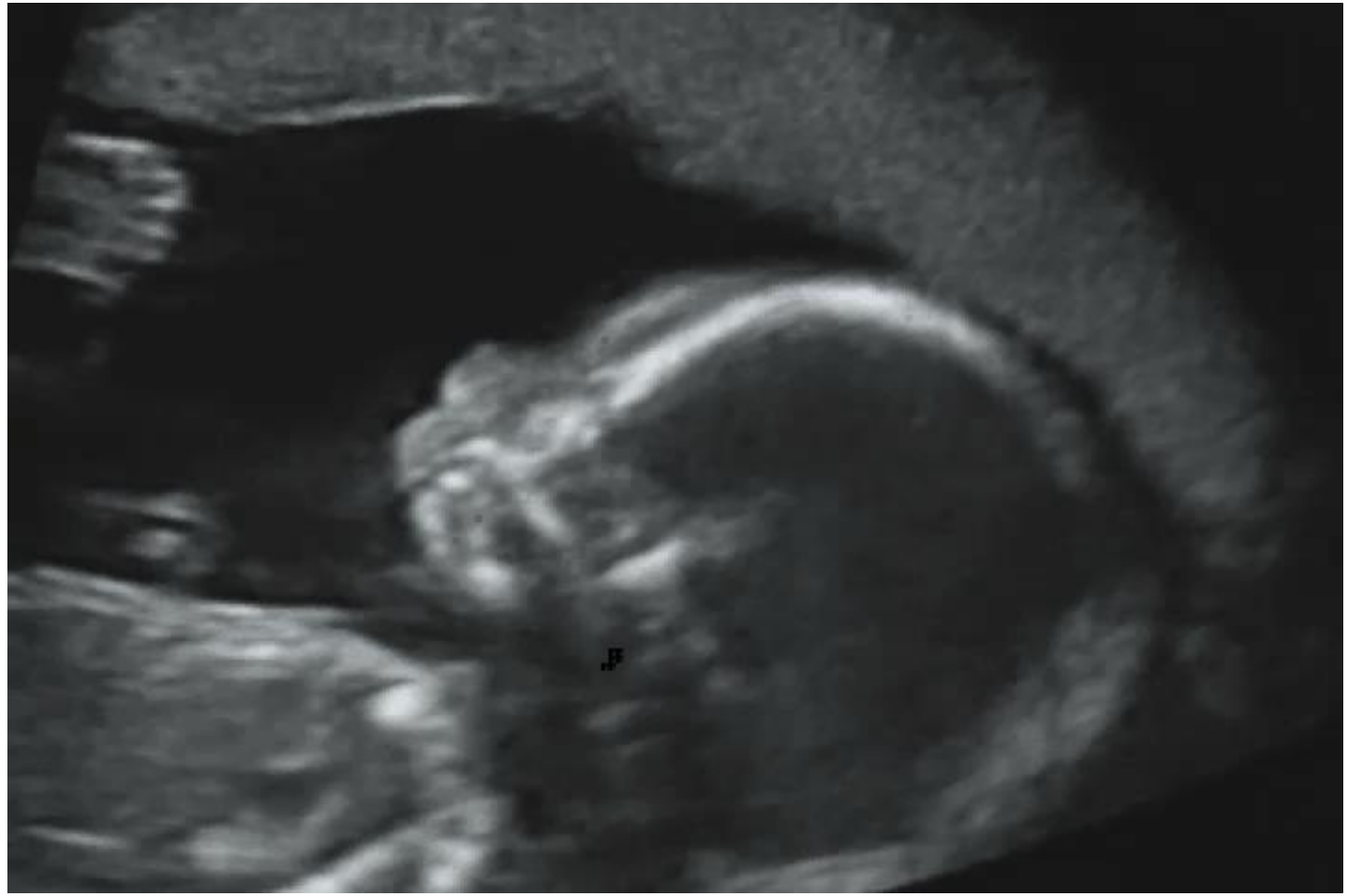
- Part of trisomy 13: lethal
- Normal karyotype: high risk of neurodevelopmental delay depending on the degree of holoprosencephaly

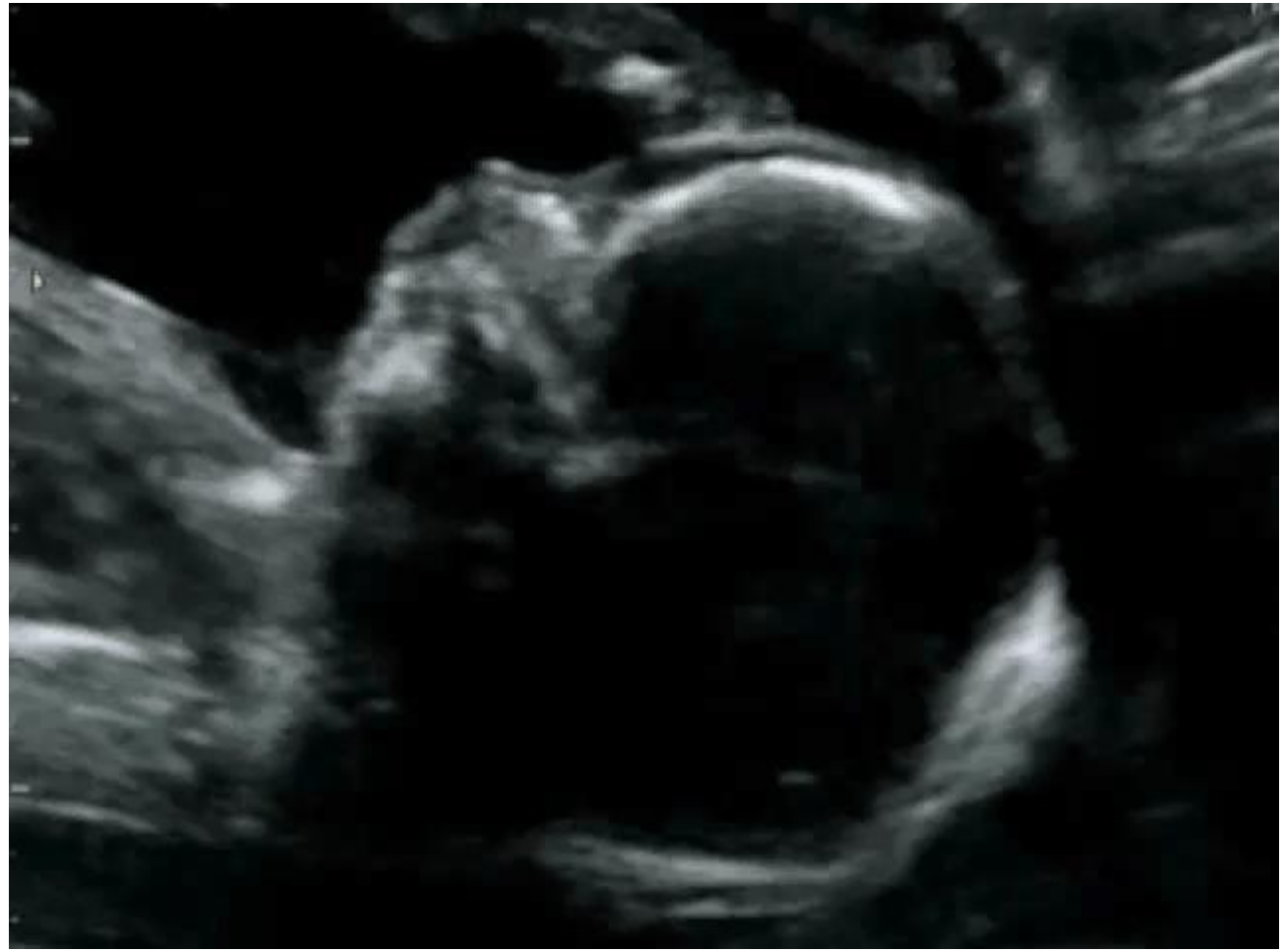
Recurrence

- Isolated: no increased risk of recurrence
- Part of trisomy 13: 1%
- Part of an autosomal recessive condition: 25%









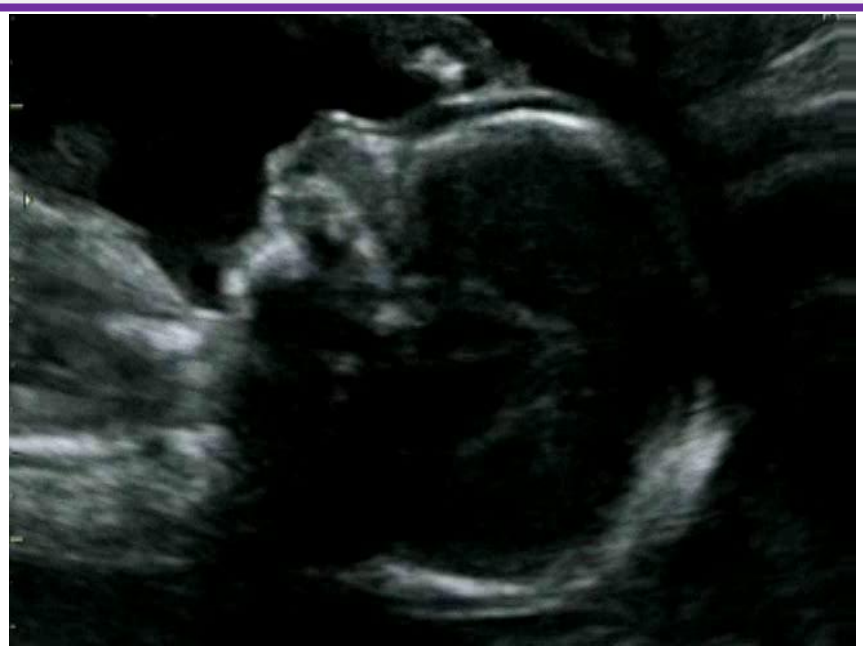
St George's University of London

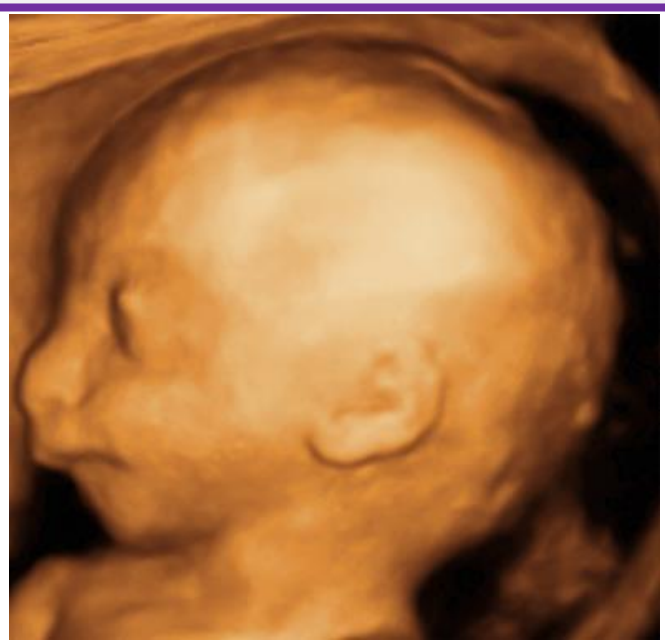
Micrognathia

- **Prevalence:** 1 in 1,500 births
- **Diagnosis:**

  Prominent upper lip and receding chin in the mid-sagittal view

  Polyhydramnios (>25 weeks) due to glossoptosis (normal tongue obstructing small oral cavity)





St George's University of London

Micrognathia

Micrognathia or retrognathia

- **Micrognathia**: small mandible
- **Retrognathia**: normal dimensions of the mandible but posteriorly displaced
- Difficult to differentiate prenatally
- Often concomitant

St George's University of London

Micrognathia

Associated abnormalities:

- Associated defects: frontal encephalocele and ACC
- Chromosomal abnormalities, mainly trisomy 18 and triploidy, are found in about 30% of cases
- Associated with >50 genetic syndromes, including:
  - Pierre–Robin Anomalad syndrome
  - Treacher Collins syndrome
  - Otocephaly: sporadic: severe micrognathia or agnathia, and mid-line defects, including holoprosencephaly, anterior encephalocele, cyclopia, aglossia, and mid-facial location of the ears

St George's University of London

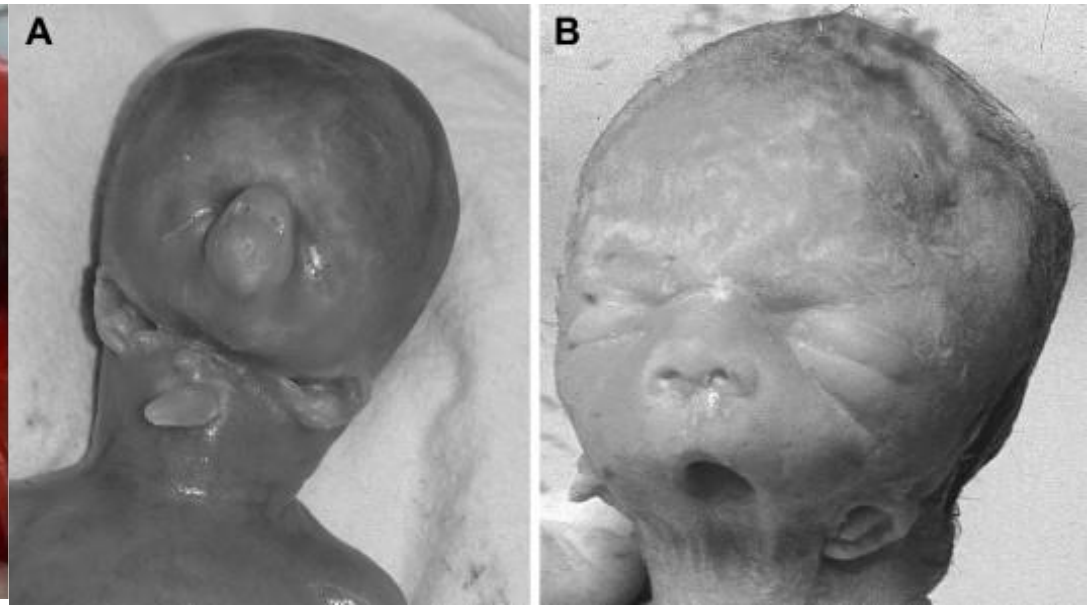
Otocephaly

Severe micrognathia or agnathia

Mid-line defects, including holoprosencephaly, anterior encephalocele, cyclopia

Aglossia

Mid-facial location of the ears



Micrognathia

Management

Investigations:

- Detailed ultrasound examination
- Invasive testing for karyotyping and array

Follow-up:

- Ultrasound every 4 weeks (AFV)

Delivery:

- **Place**: hospital with facilities for neonatal intensive care.
- **Time**: 38 weeks.
- **Method**: induction of labor aiming for vaginal delivery.
- A paediatrician should be present in the delivery room and be prepared to intubate the neonate.

St George's University of London

Micrognathia

Prognosis

- Neonatal mortality: >80% due to associated abnormalities
- In Pierre–Robin anomalad survival is good

Recurrence

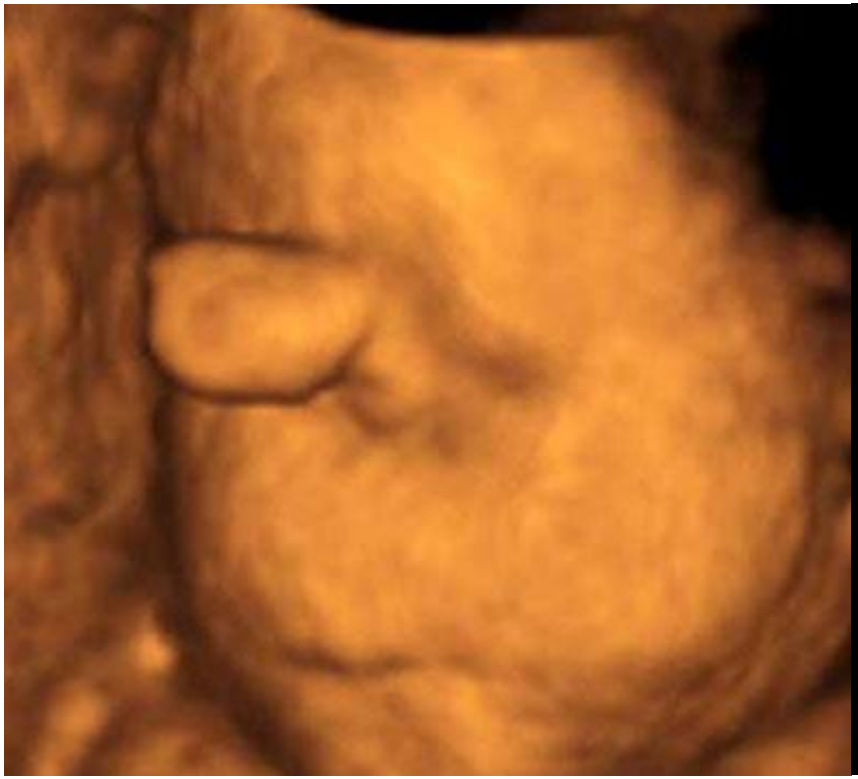
- Isolated: no increased risk of recurrence
- Part of trisomies: 1%
- Part of genetic syndromes: 25% to 50%

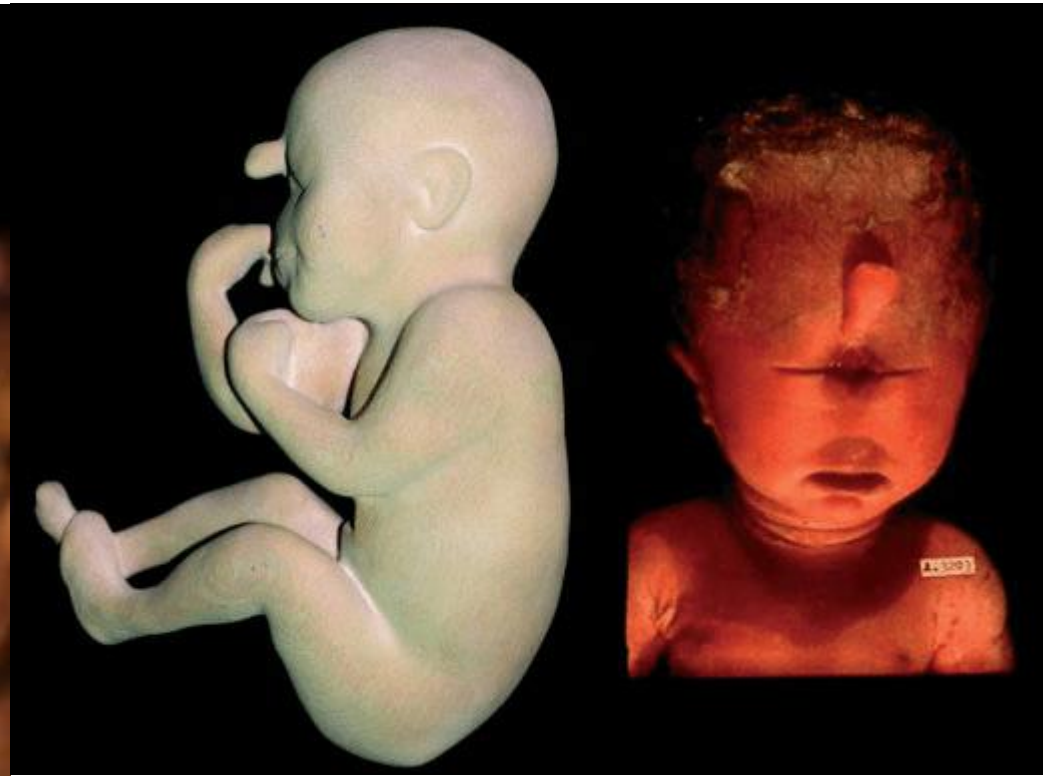
St George's University of London

Nasal abnormalities

Proboscis

soft tissue appendage projecting from just below the forehead

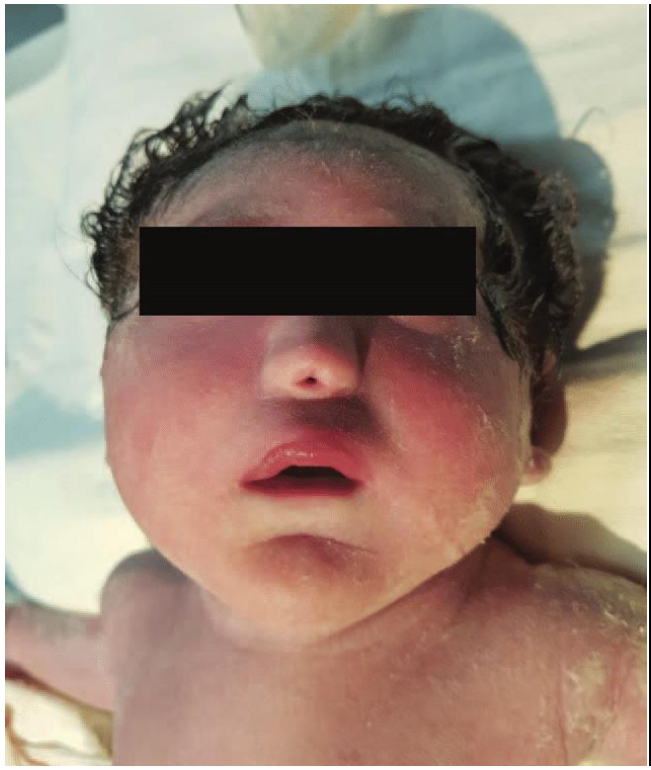


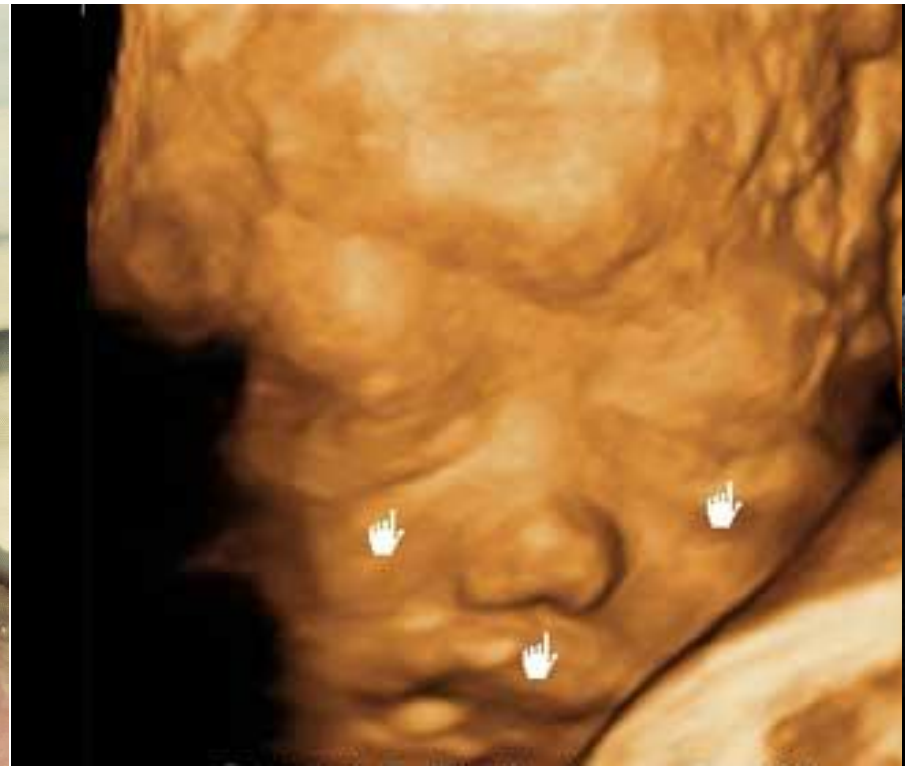


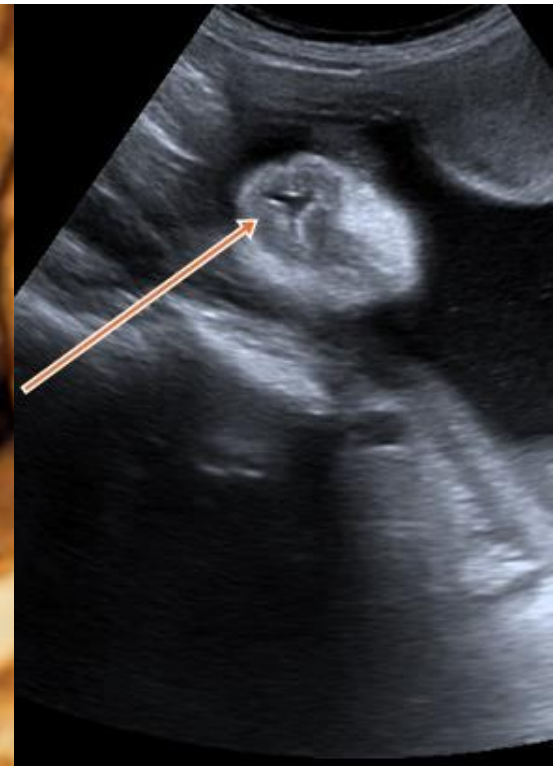
St George's University of London

Nasal abnormalities

Single nostril



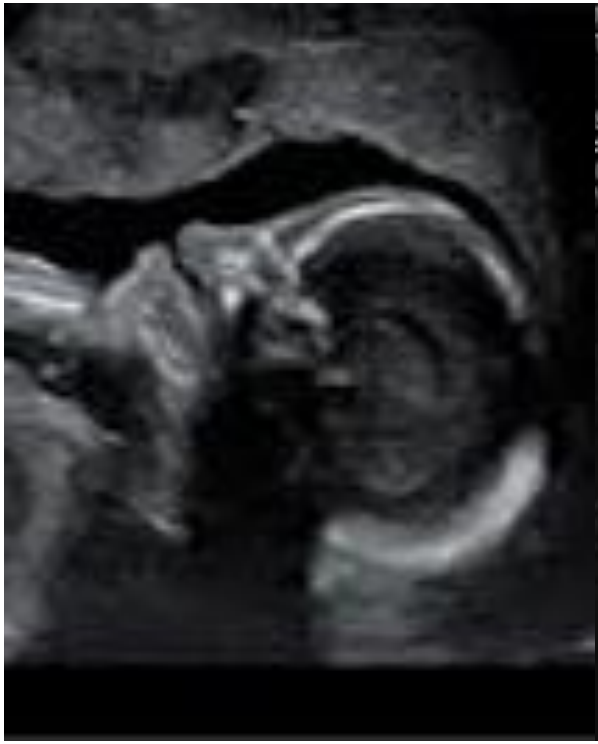


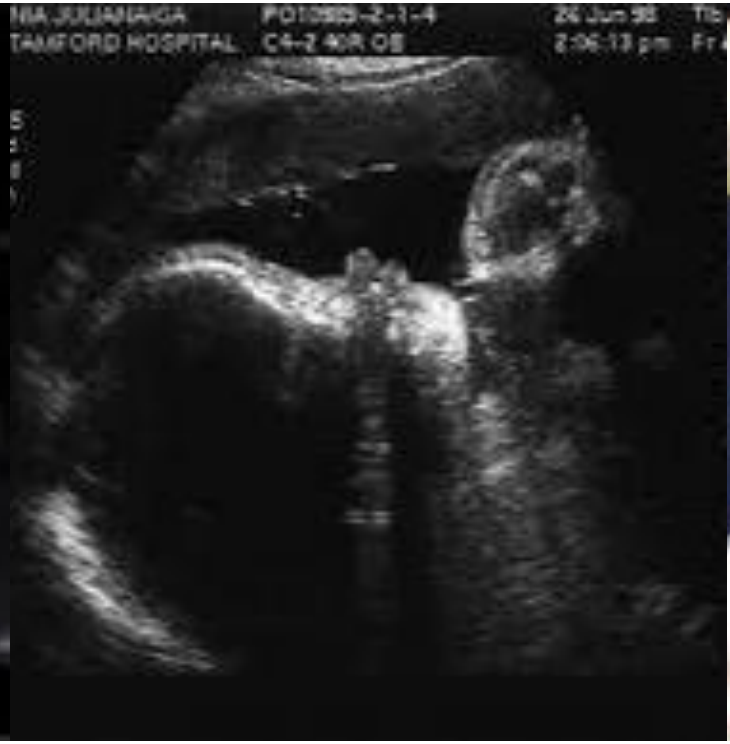


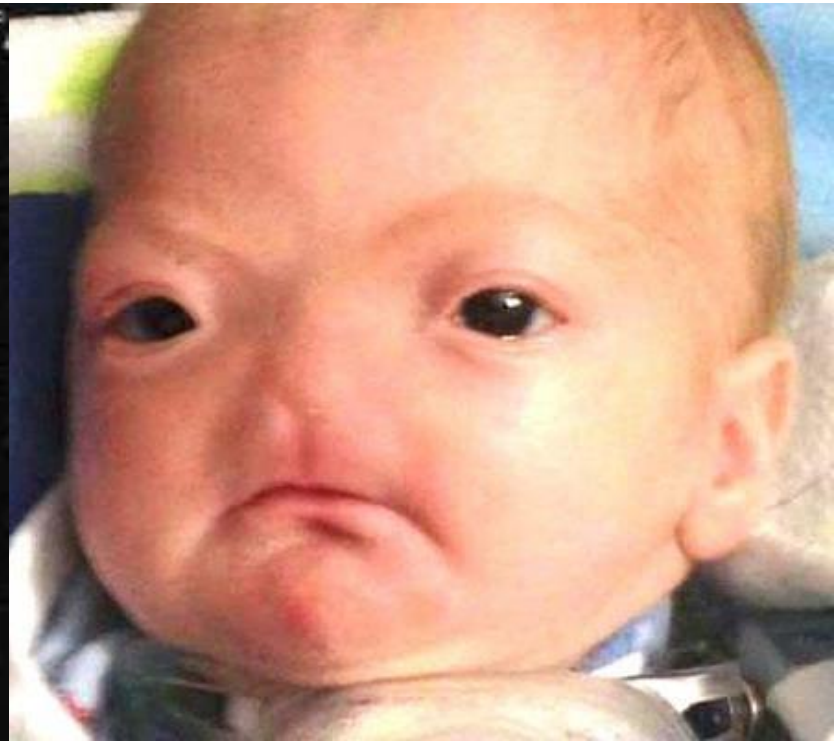
St George's University of London

Nasal abnormalities

Arhinia



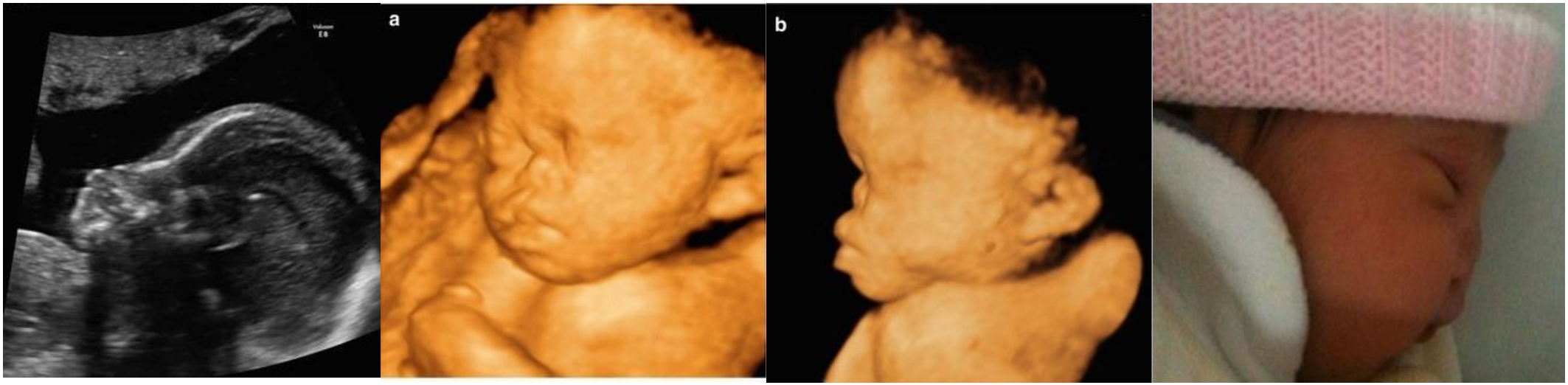


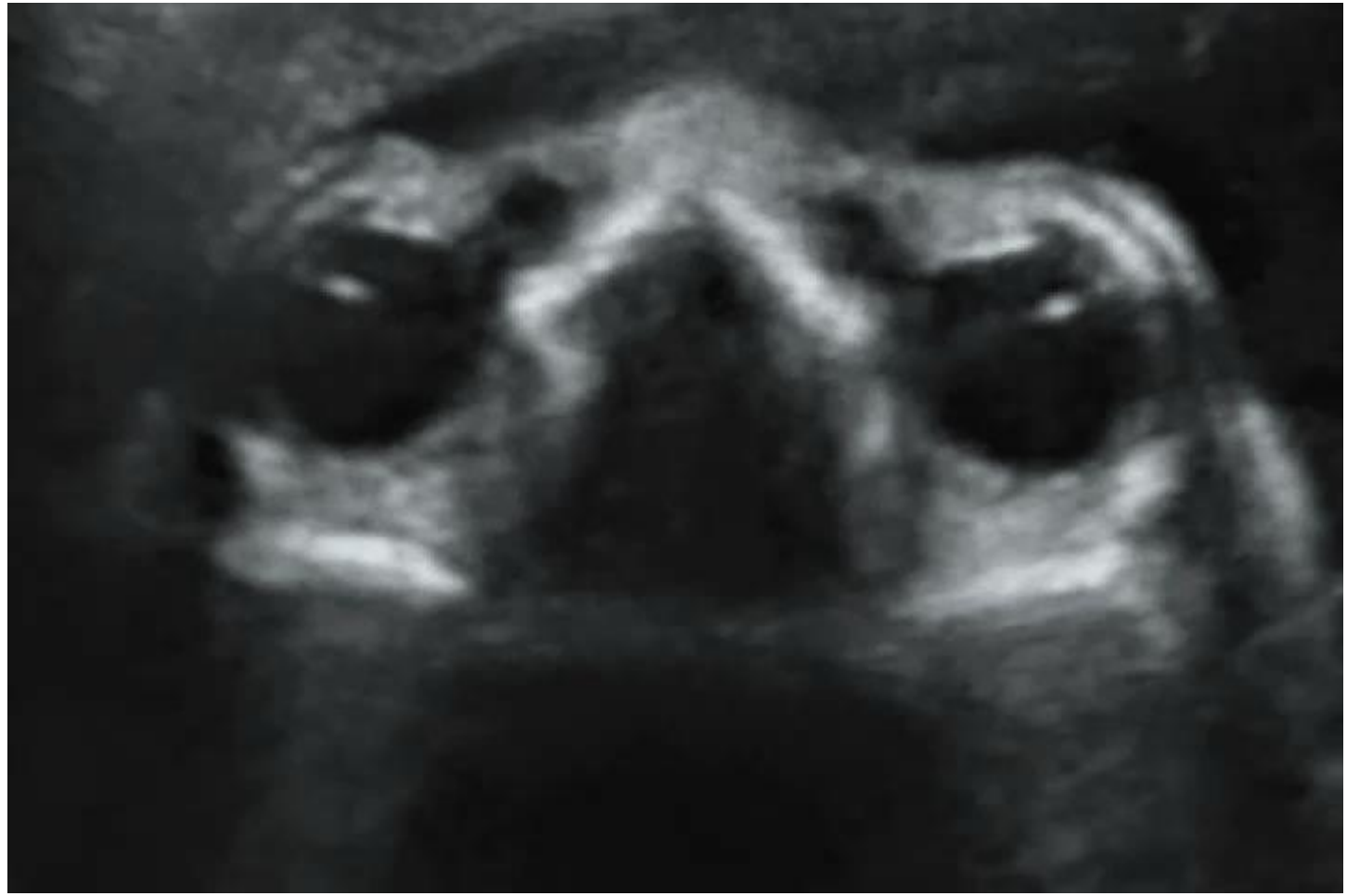


St George's University of London

Nasal abnormalities

Binder syndrome (maxillo-nasal dysplasia or maxillo-nasal dysostosis)

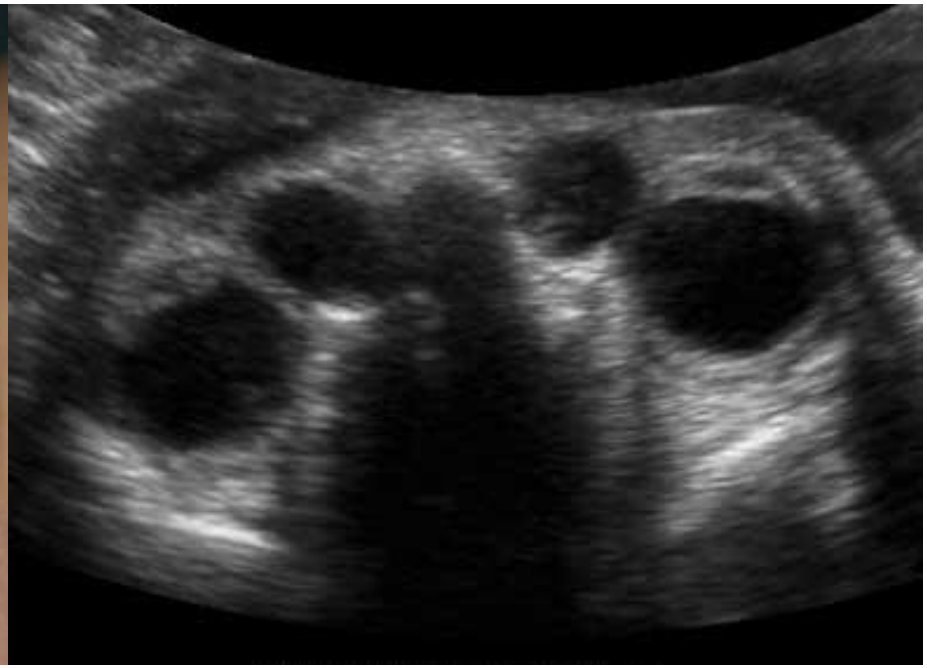




St George's University of London

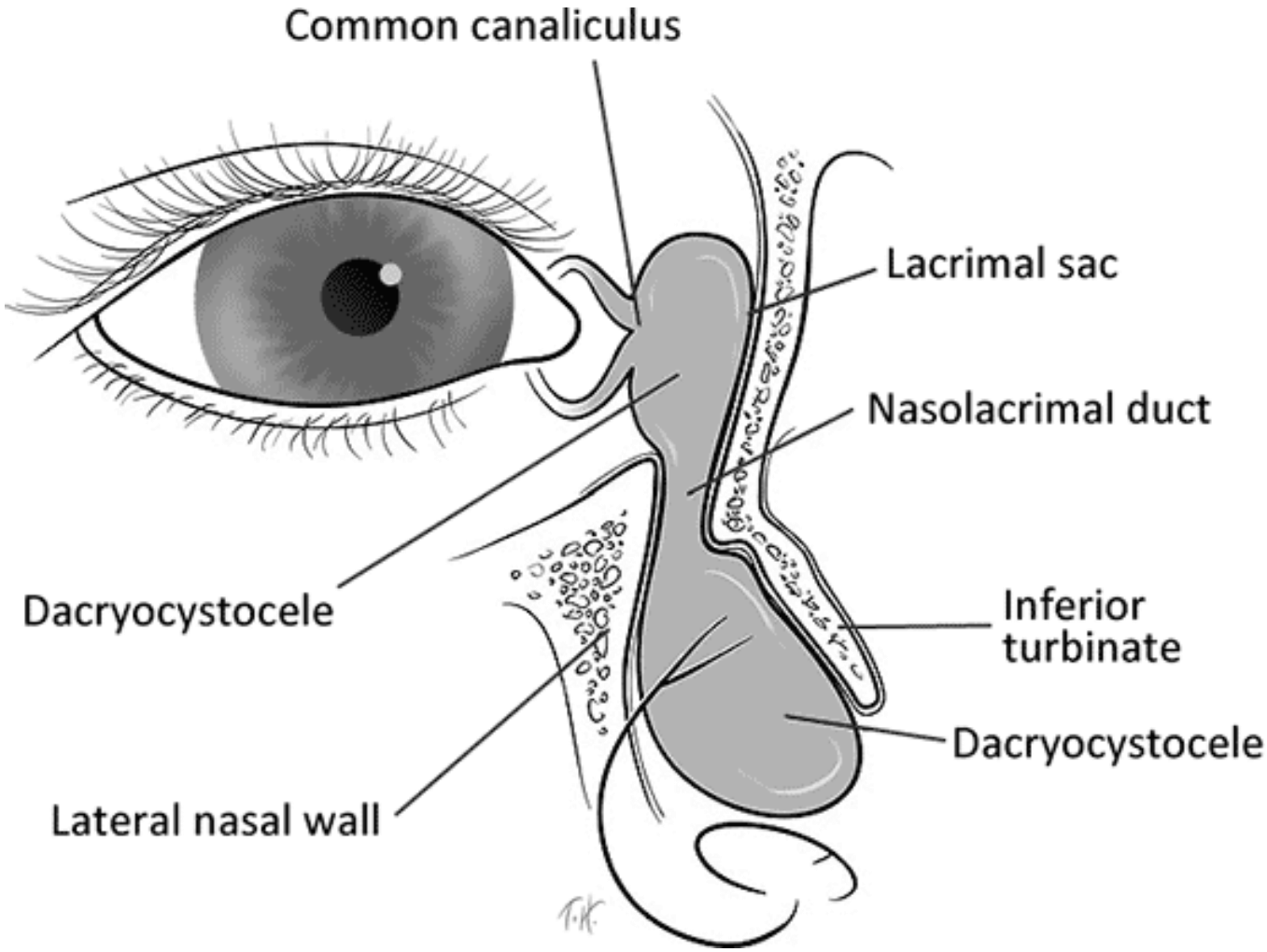
Dacryocystocele

- Cyst between the lower part of the orbit and the nose
- 75% unilateral and 25% bilateral
- 90% are due to delayed canalization of the lacrimal duct beyond 32 weeks



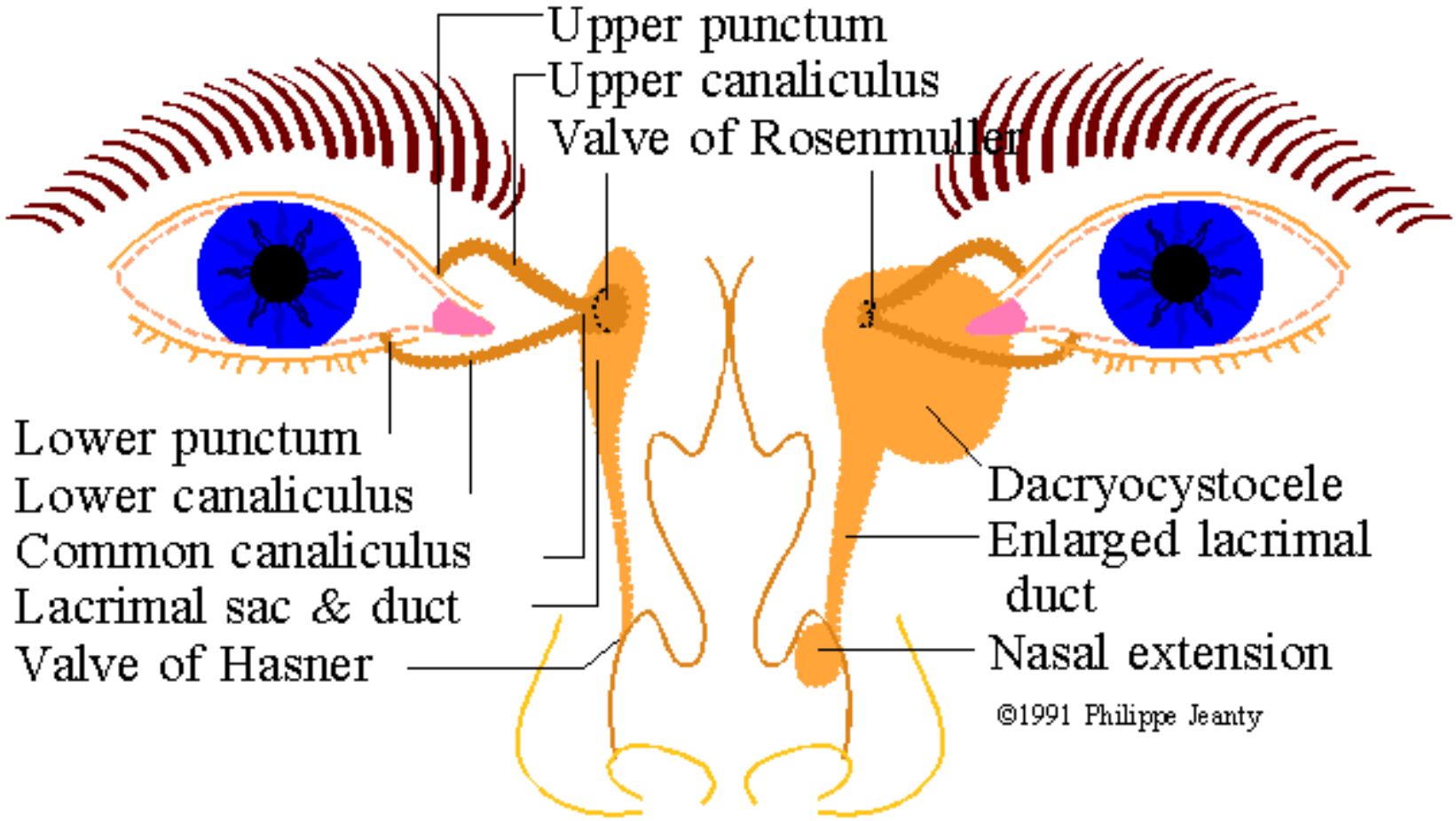
St George's University of London

Dacryocystocele



St George's University of London

Dacryocystocele

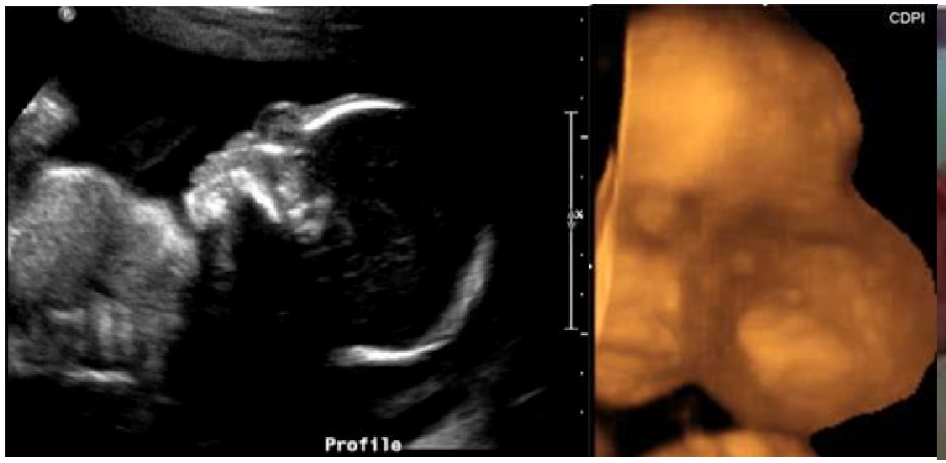


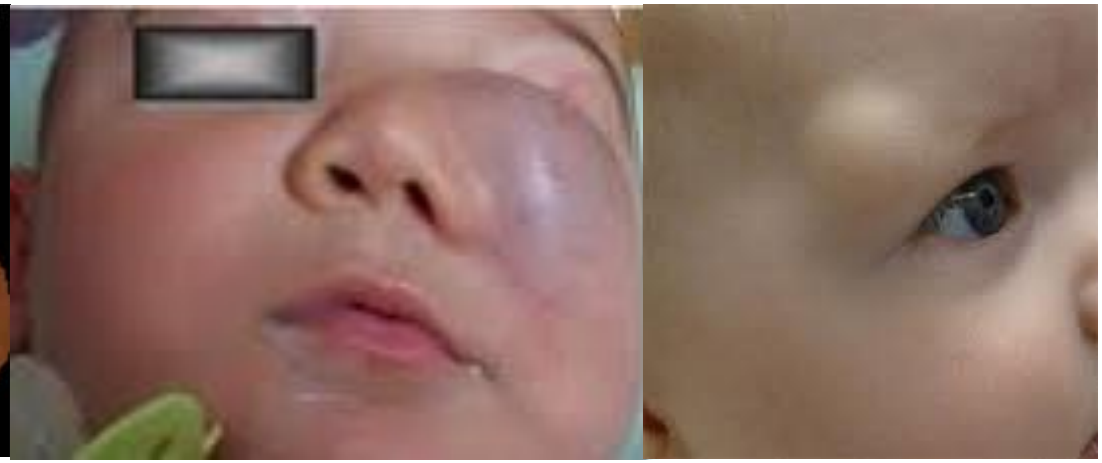
St George's University of London

Dacryocystocele

Differential diagnosis

- **Anterior encephalocele**
  Often associated with intracranial abnormalities, such as ventriculomegaly
- **Haemangioma**
  Usually solid or multiseptated
- **Dermoid cyst**
  Usually located superolaterally



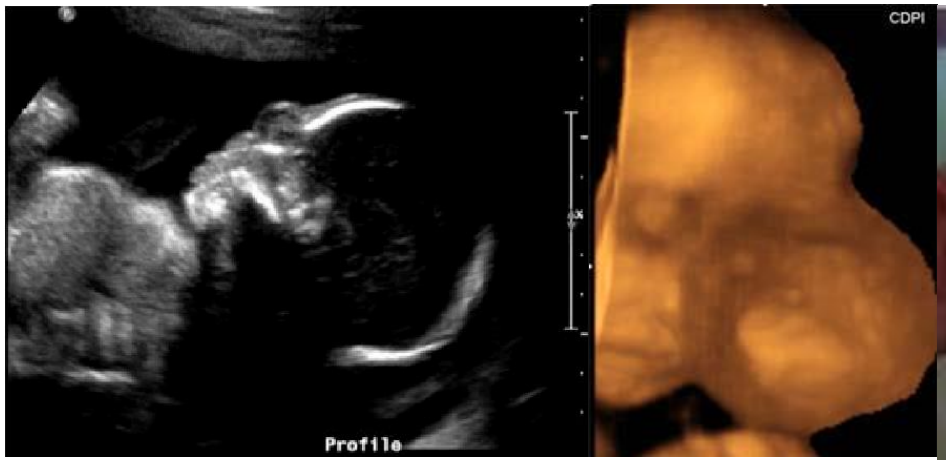


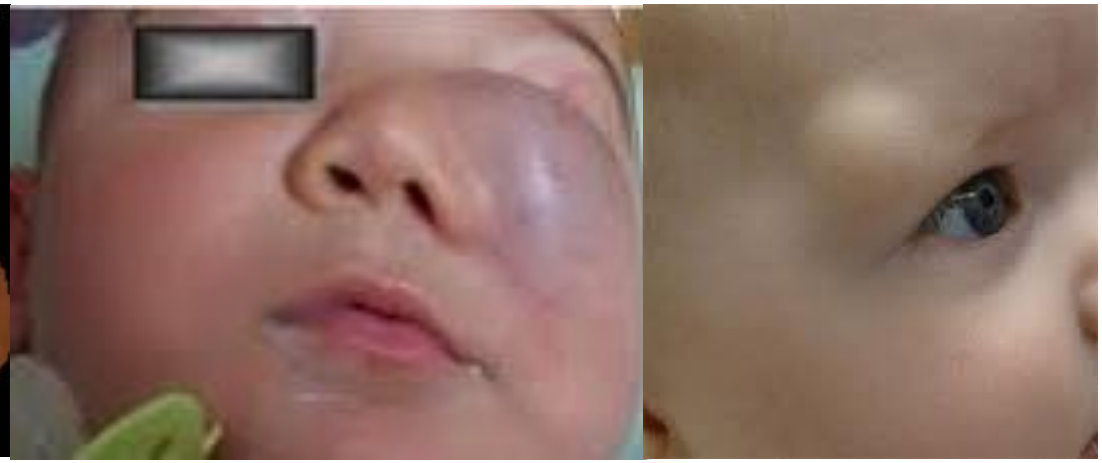
St George's University of London

Dacryocystocele

Differential diagnosis

- **Anterior encephalocele**
  Often associated with intracranial abnormalities, such as ventriculomegaly
- **Haemangioma**
  Usually solid or multiseptated
- **Dermoid cyst**
  Usually located superolaterally



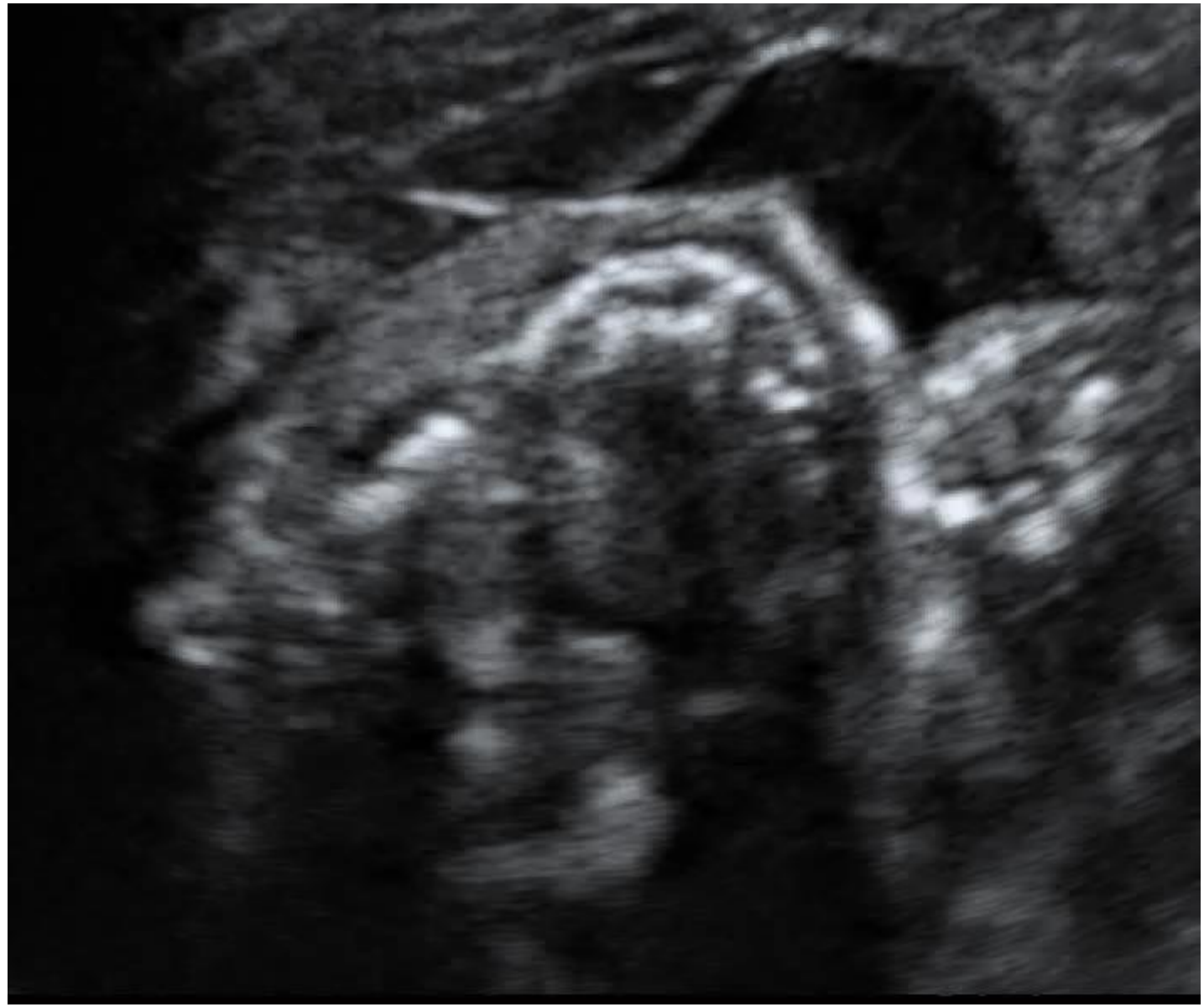


St George's University of London

Dacryocystocele

Counselling

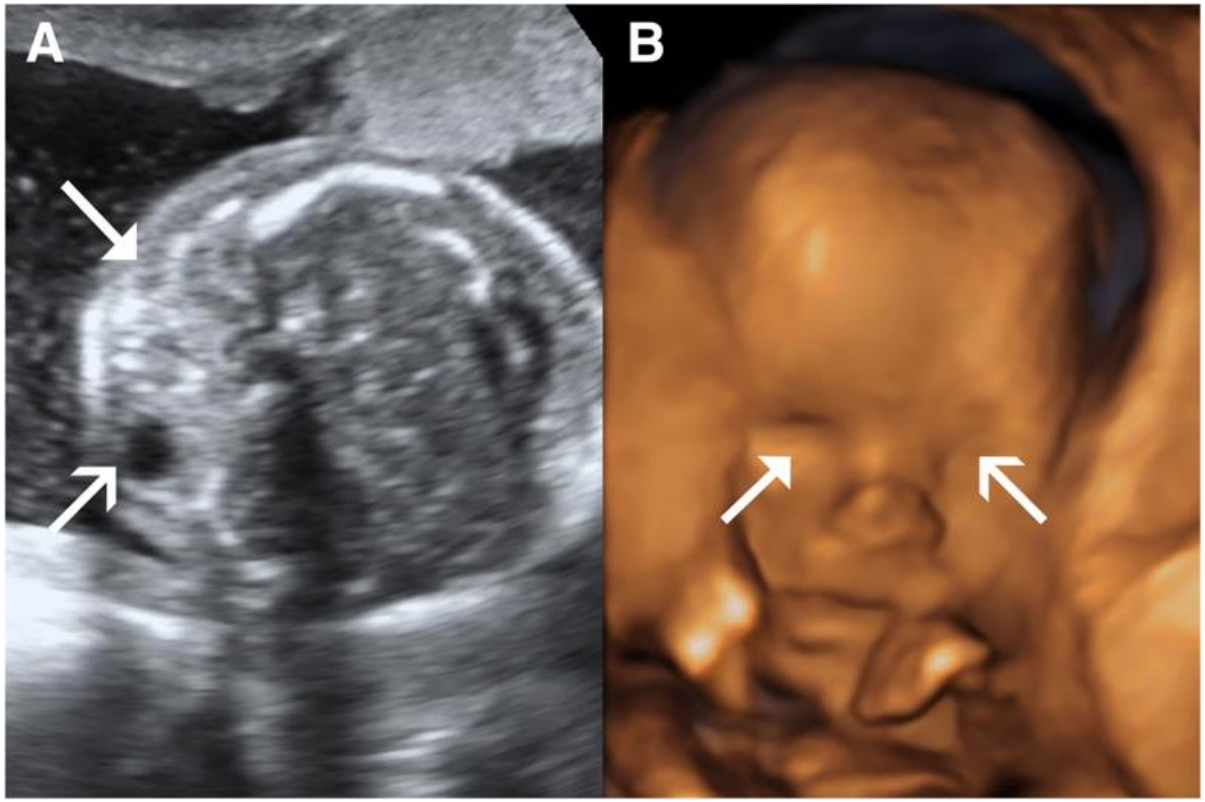
- The incidence of chromosomal abnormalities and genetic syndromes is not increased.
- Resolve spontaneously either in the 3rd trimester or within the first 6 months of life
- Occasionally, nasolacrimal duct probing may be required to open the obstruction
- No increased risk of recurrence

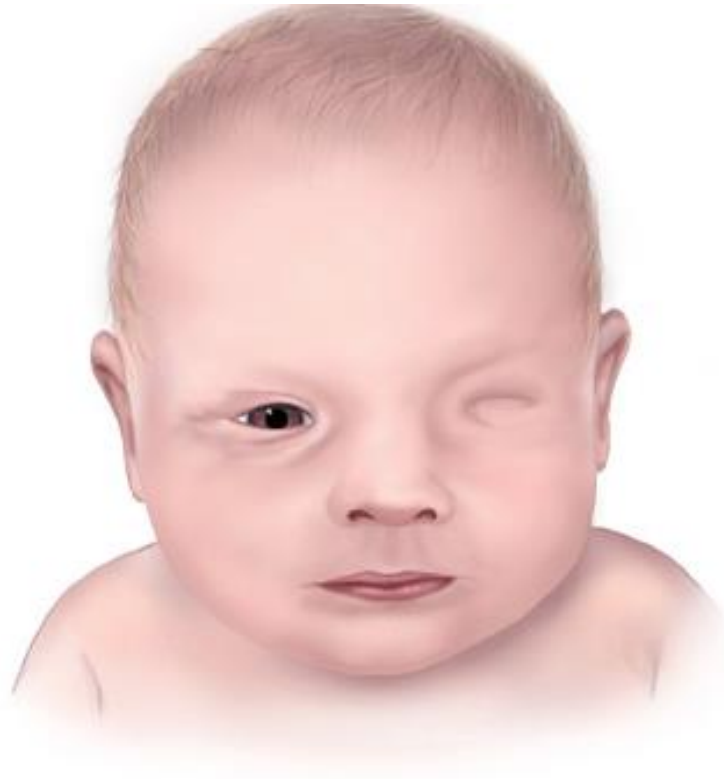


St George's University of London

Anophthalmia/Micophthalmia

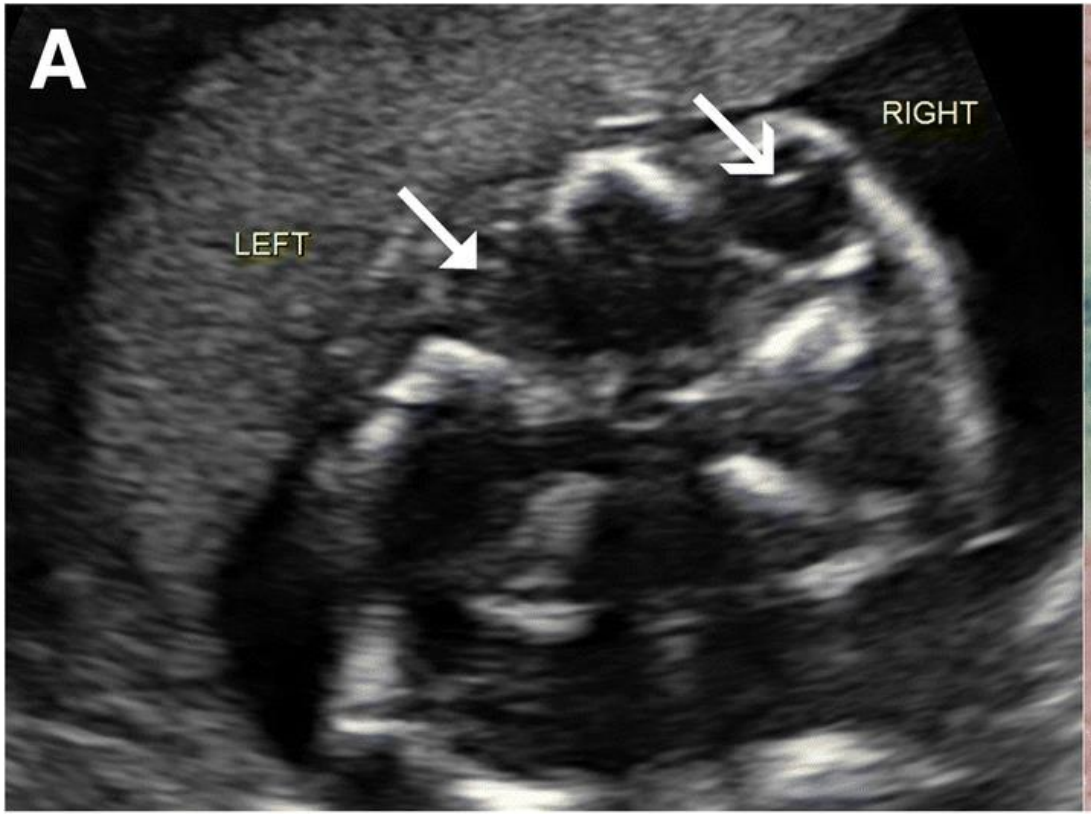
- Microphthalmia: small eyeball
- Anophtalmia: absence of the eyeball, optic nerve and chiasma
- Both can be unilateral or bilateral.

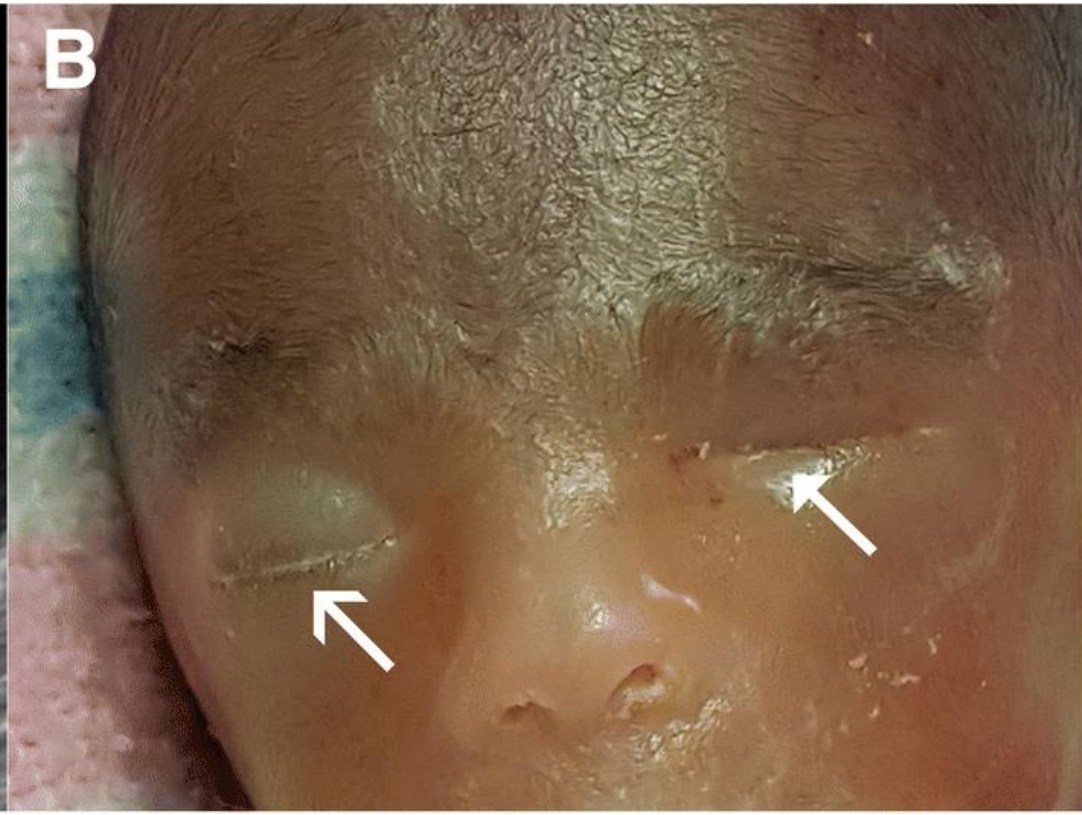




St George's University of London

Anophthalmia/Micophthalmia





St George's University of London

Anophthalmia/Micophthalmia

Associated abnormalities:

- **Chromosomal defects (mainly trisomy 13) found in >50% of cases**
- **Genetic syndromes are very common (>50% of cases)**

The most common is:

Goldenhar syndrome (sporadic; anophthalmia, ear defects, facial cleft, facial macrosomia)

Fraser syndrome (AR; microphthalmia, facial cleft, tracheal atresia, bilateral renal agenesis, heart defects, syndactyly or polydactyly)

Fryns syndrome (AR; anophthalmia, facial cleft, micrognathia, ventriculomegaly, diaphragmatic hernia)

St George's University of London

Goldenhar syndrome

Anophthalmia

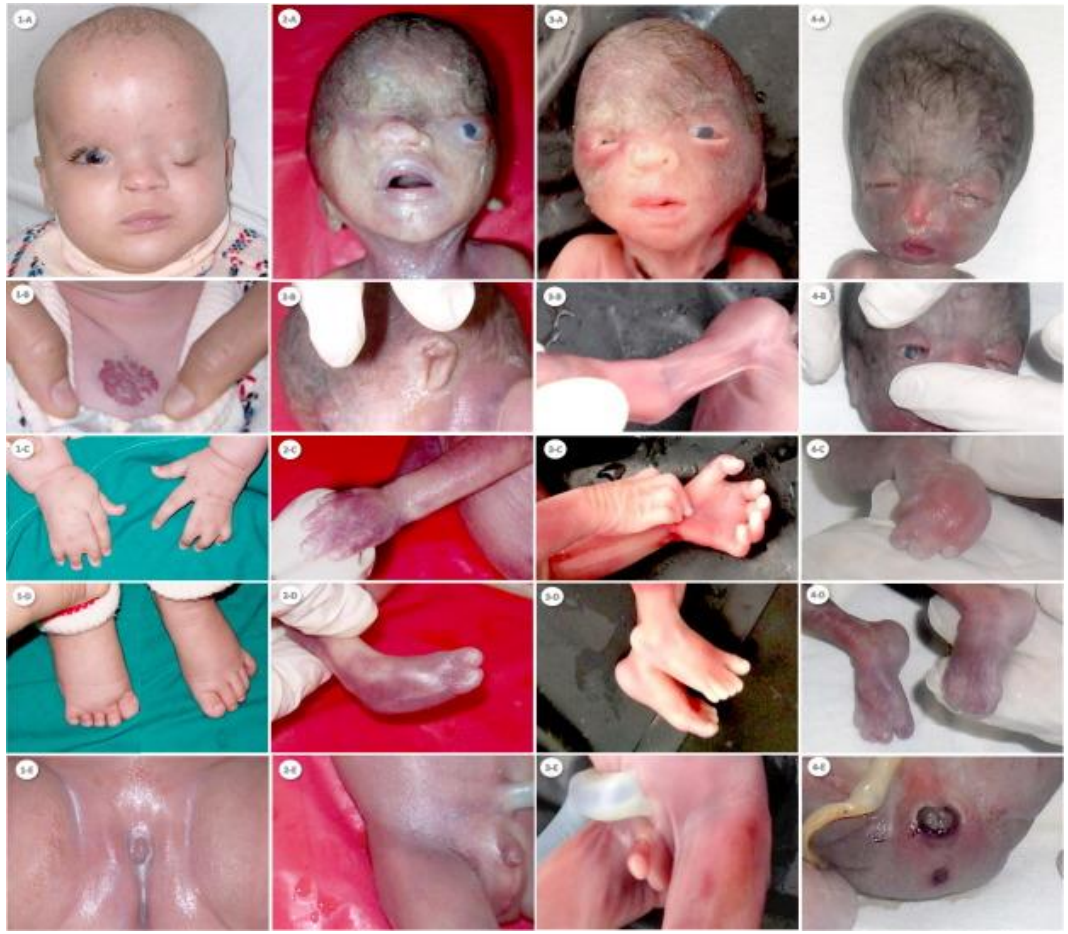
Ear defects

Facial cleft

St George's University of London

Fraser syndrome

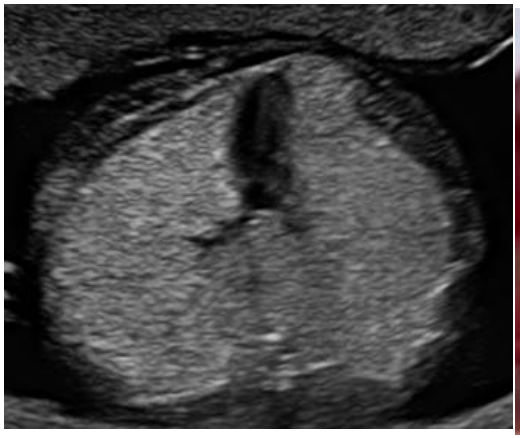
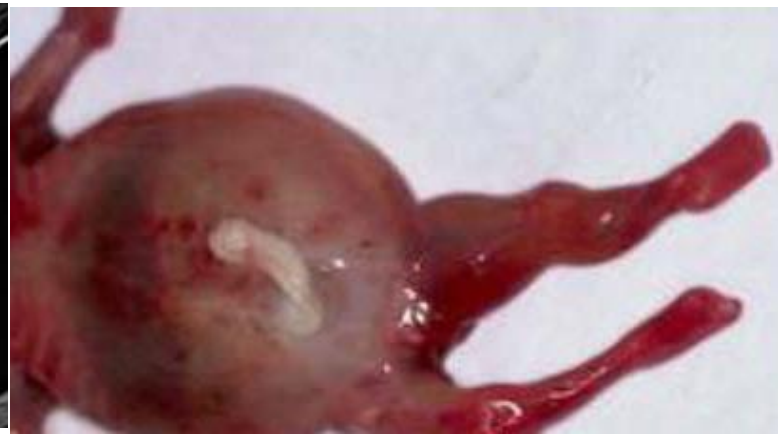
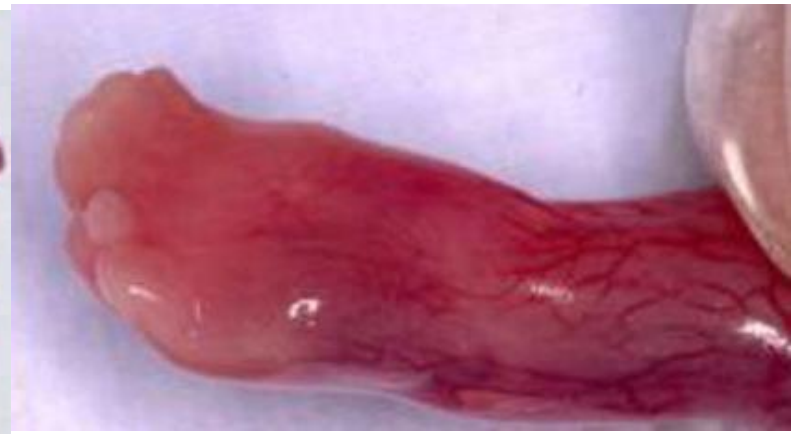
- Microphthalmia
- Facial cleft
- Tracheal atresia
- Bilateral renal agenesis
- Syndactyly or polydactyly
- CHD



St George's University of London

Fraser syndrome

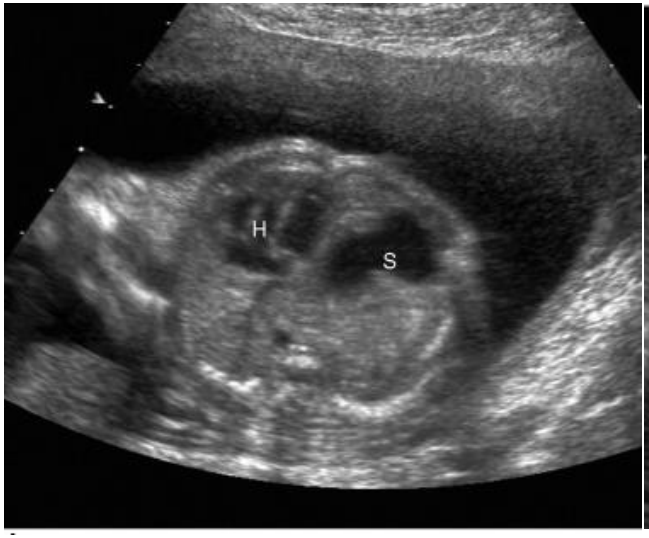
Microphthalmia
Facial cleft
Tracheal atresia
Bilateral renal agenesis
Syndactyly or polydactyly

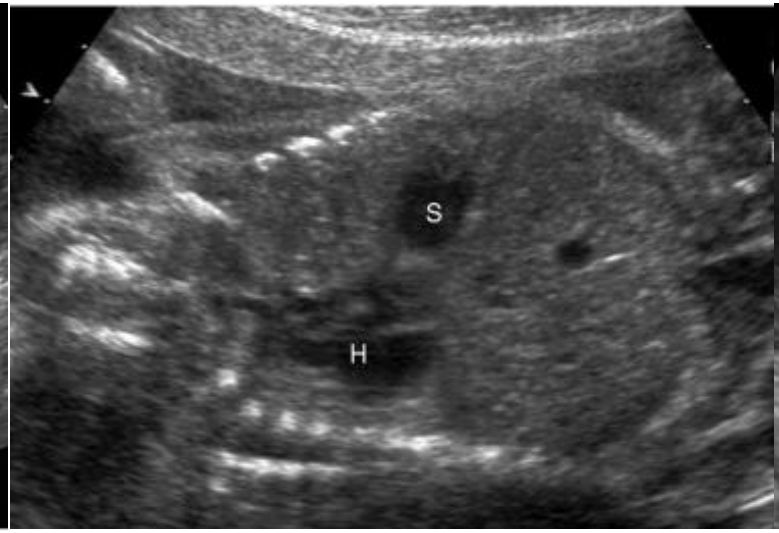




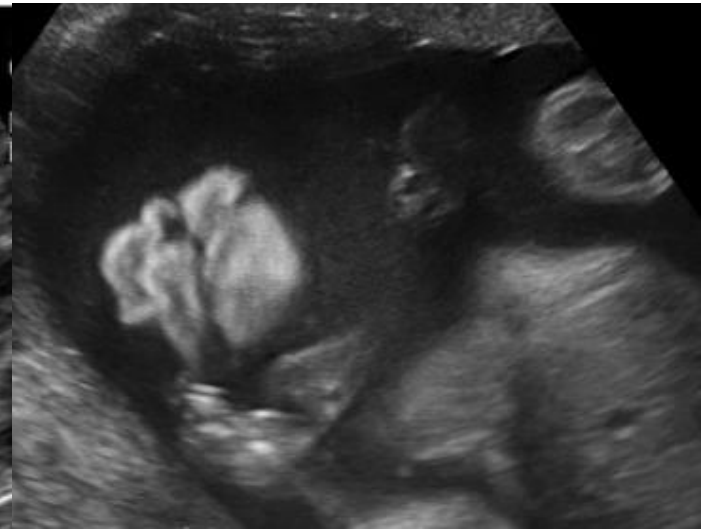
St George's University of London

Fryns syndrome

Anophthalmia
Facial cleft
Micrognathia
Diaphragmatic hernia



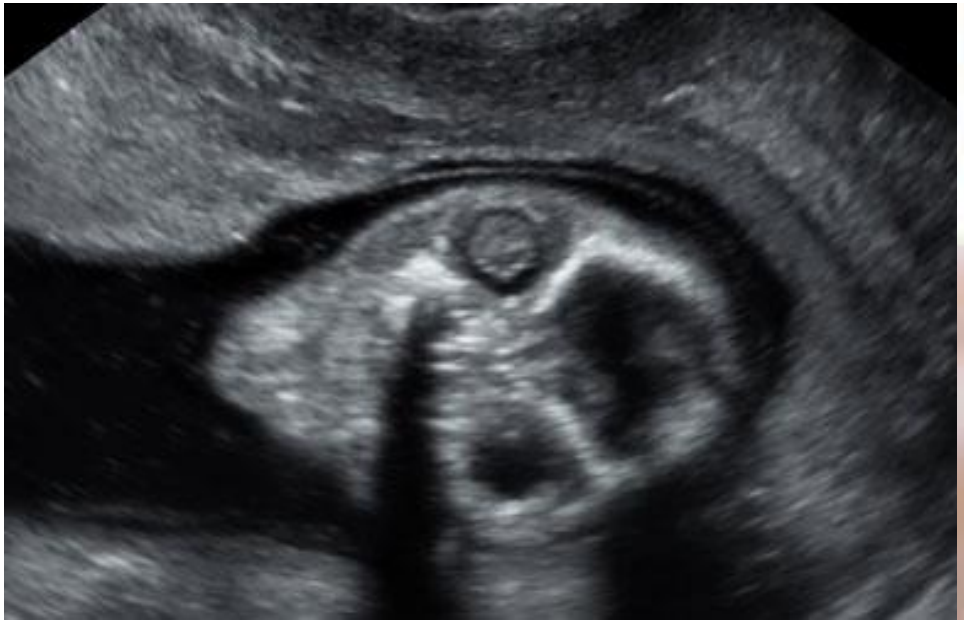




St George's University of London

Cataract

- Unilateral or bilateral opacity of the lens
- Bilateral lesions are usually syndromic, whereas unilateral are usually related to fetal infection



St George's University of London

Cataract

Associated abnormalities:

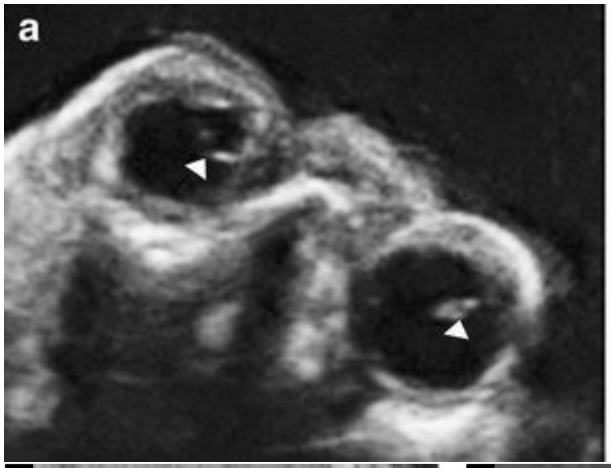
- **The incidence of chromosomal defects is not increased**
- **Genetic syndromes in 10% of cases**
- **The most common include:**

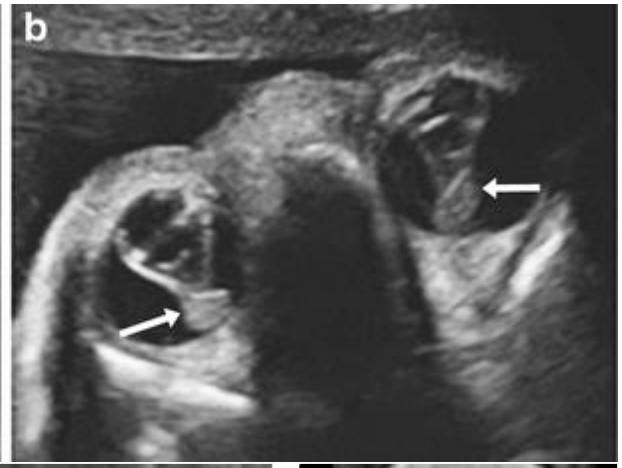
  Walker-Warburg (AR; type II lissencephaly, ACC, cerebellar malformations, cataract)

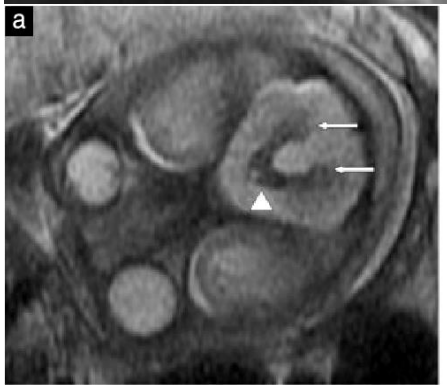
  Chondrodysplasia punctata (X-linked recessive; cataract, symmetric rhizomelic shortening and epiphyseal calcifications).
- **Congenital infection (especially rubella, toxoplasmosis, CMV) found in 30% of cases**

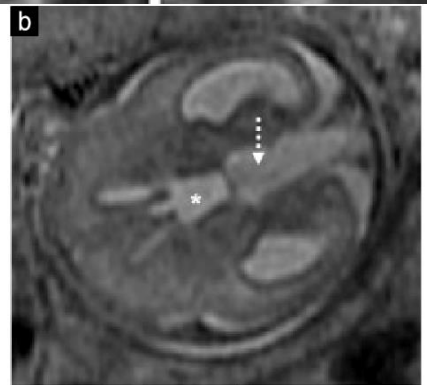
St George's University of London

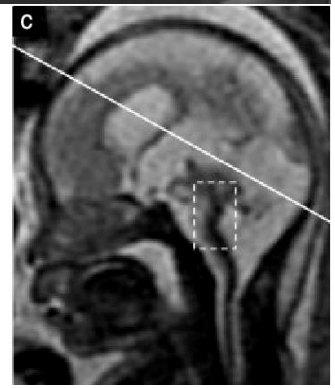
Walker-Warburg





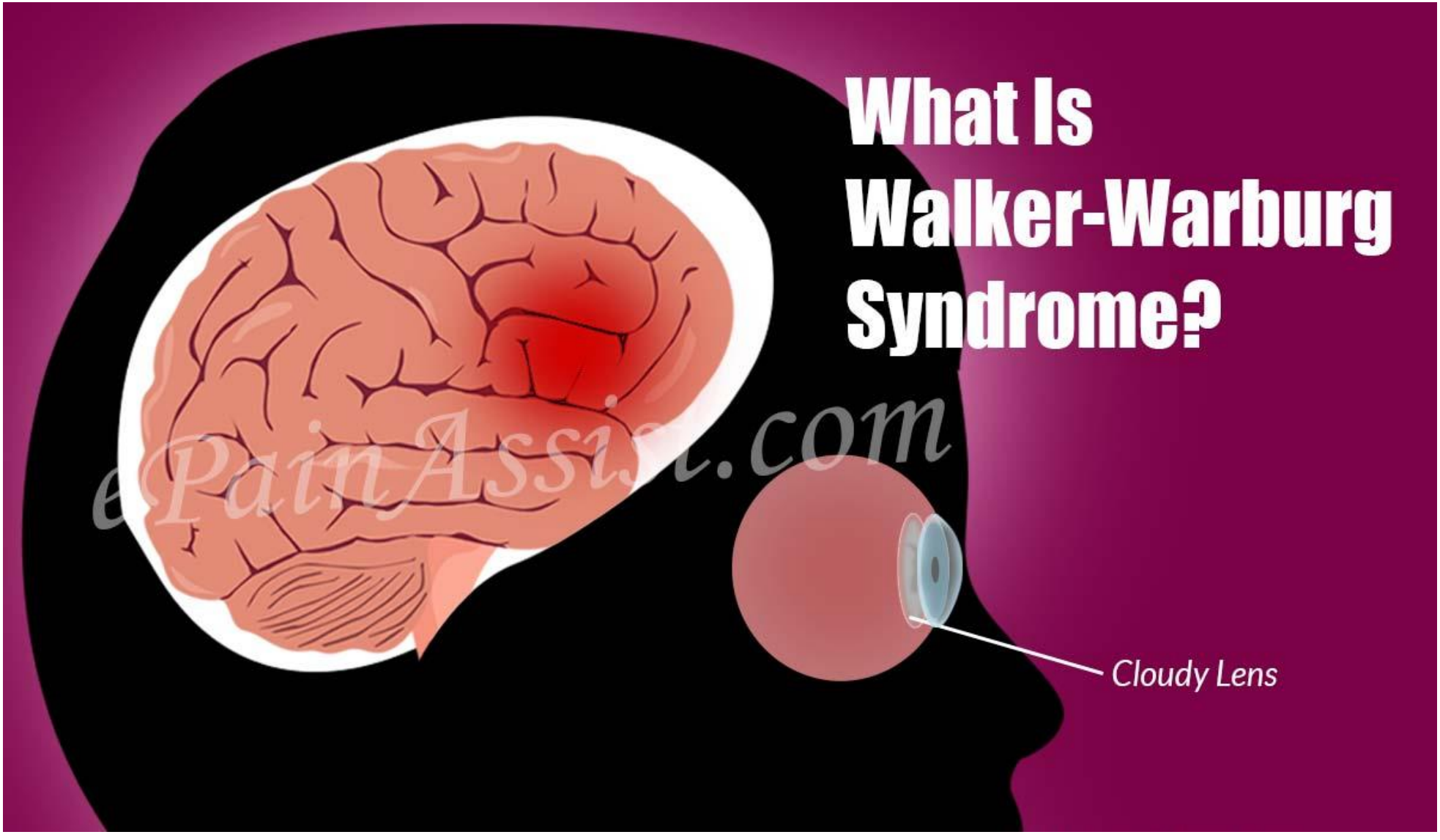






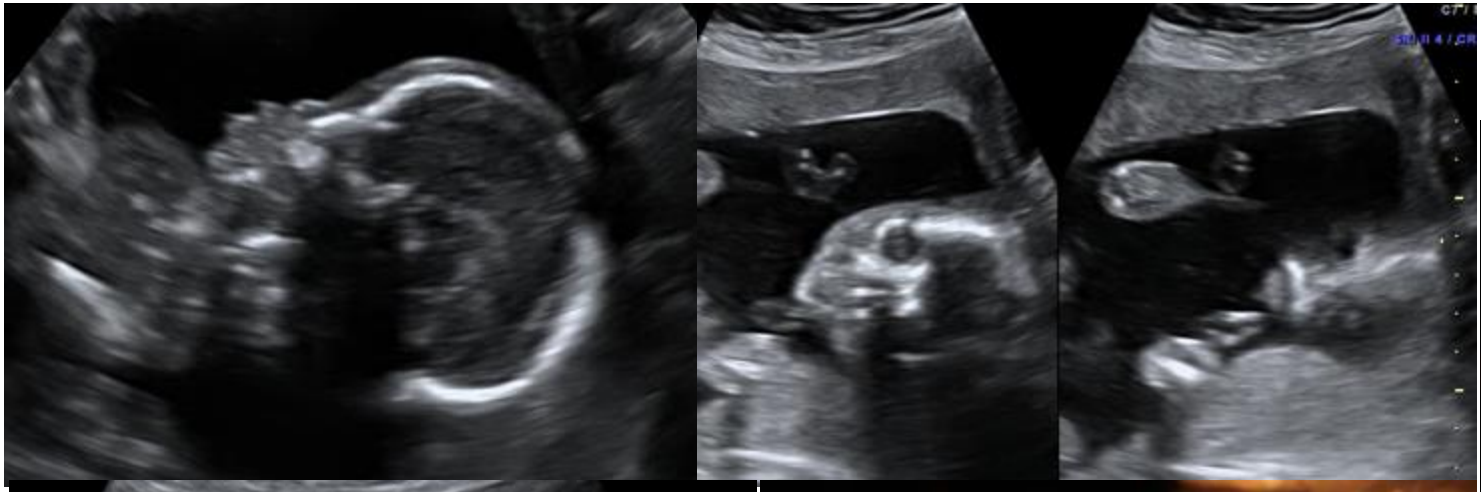
St George's
University of London

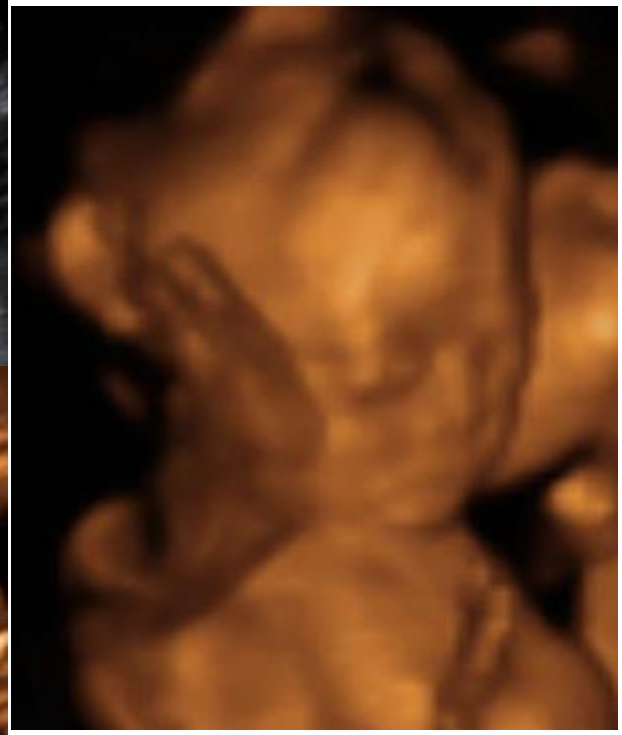
Walker-Warburg

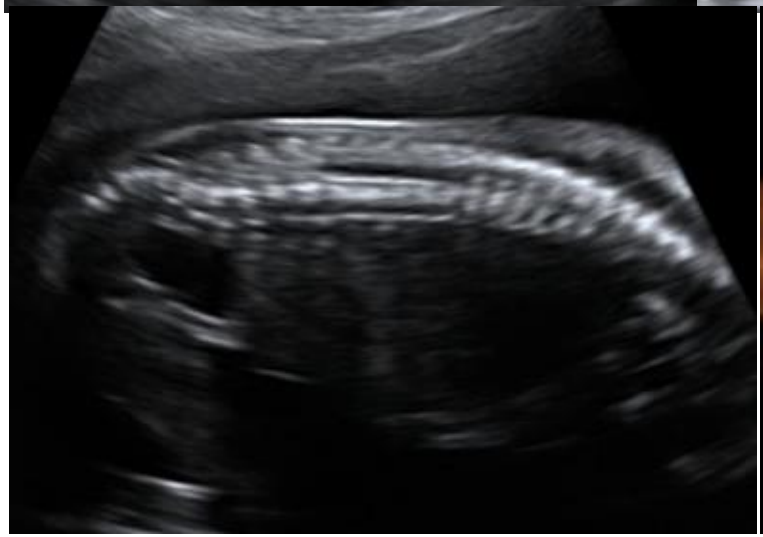


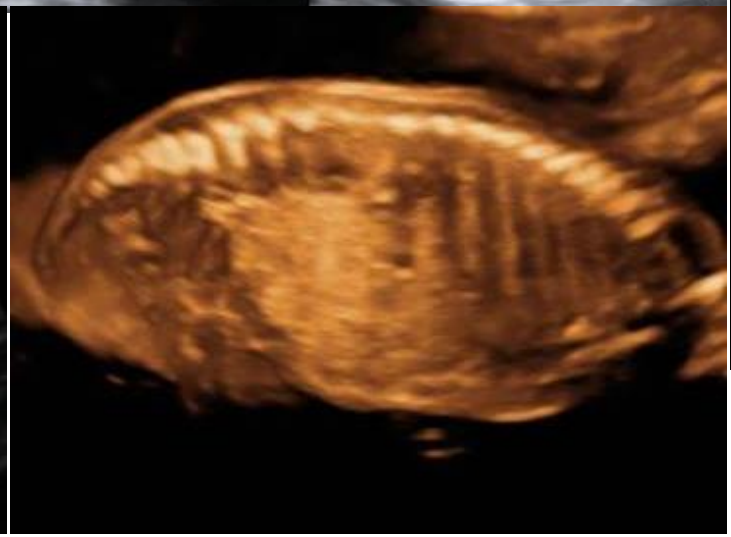
St George's University of London

Chondrodysplasia punctata





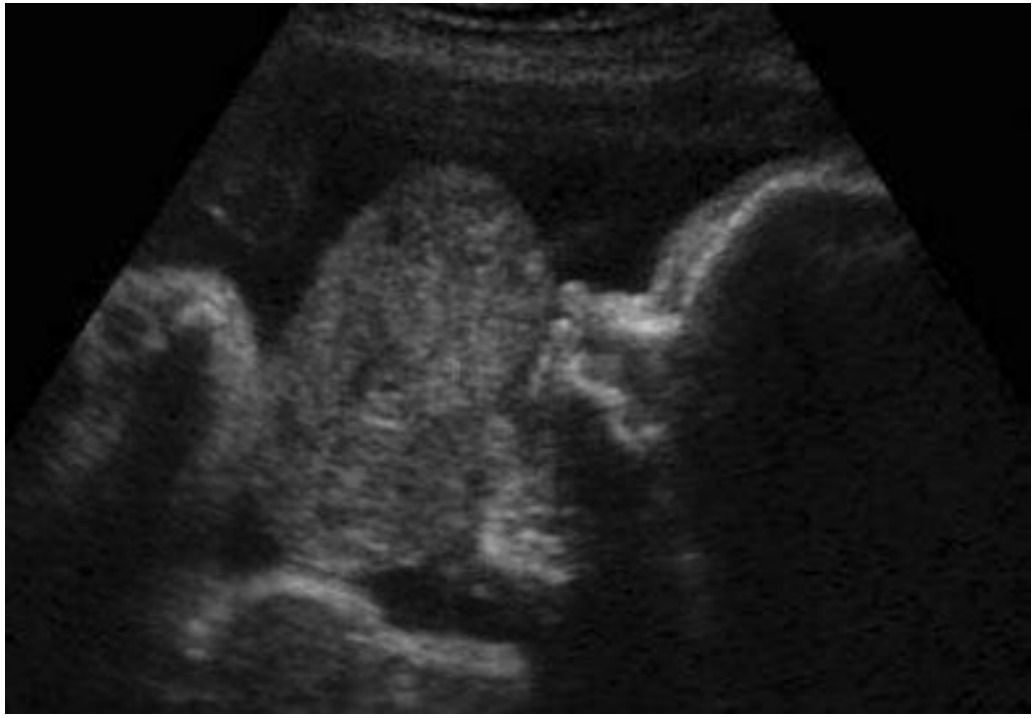


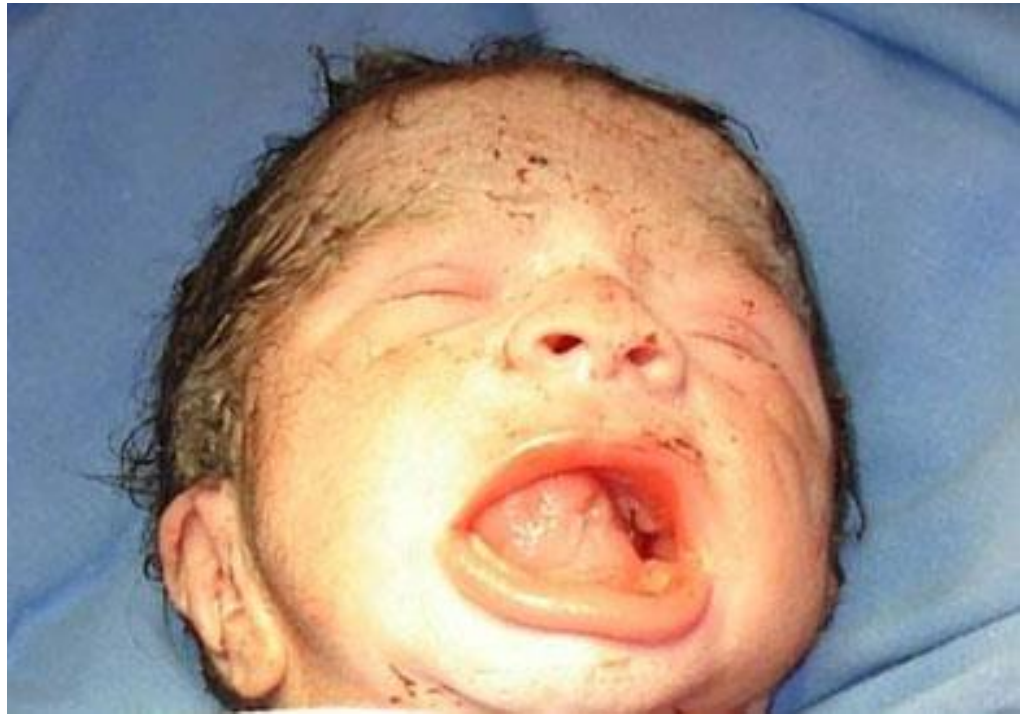


St George's University of London

Epignathus

- Solid tumor arising from the sphenoid bone, hard and soft palate, pharynx, tongue and jaw
- Grows into the oral or nasal cavity or intracranially
- Polyhydramnios (due to pharyngeal compression)



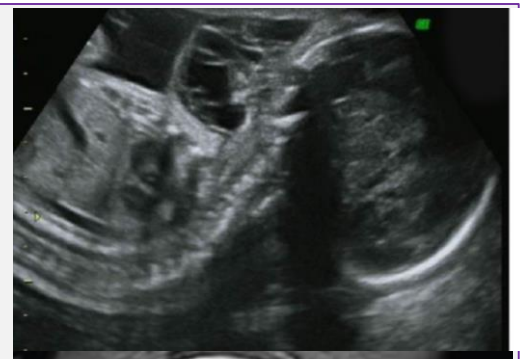


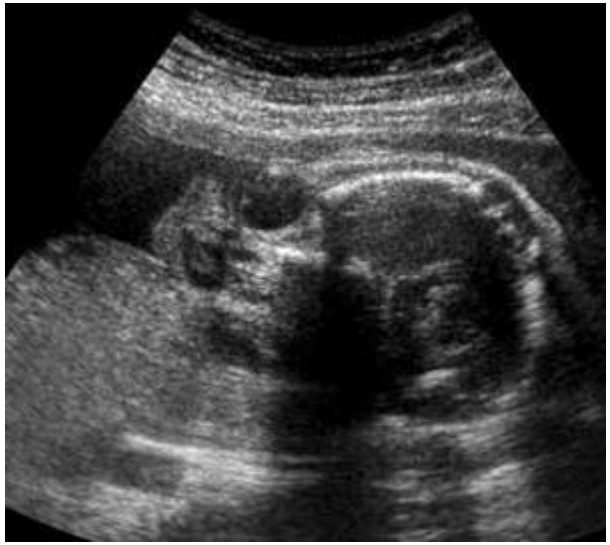
St George's University of London

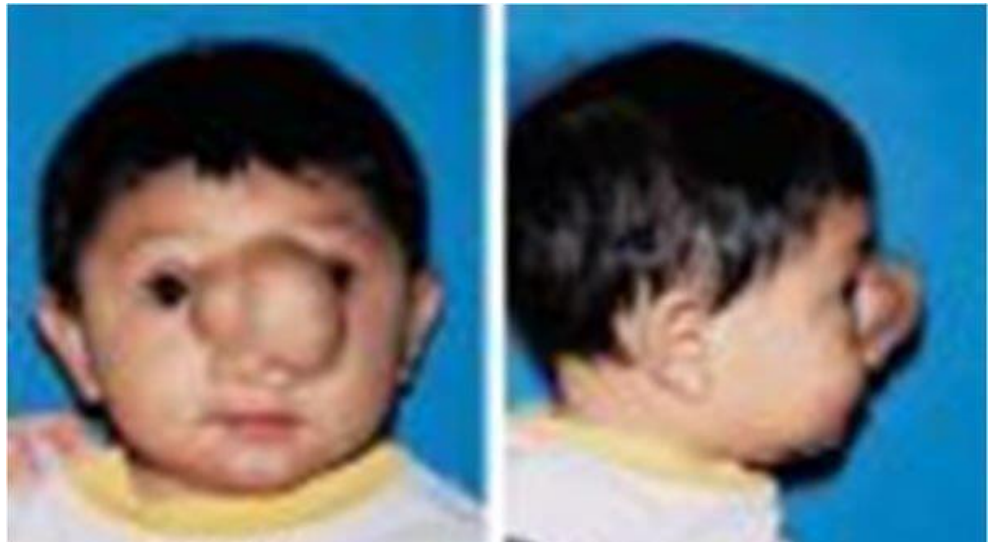
Epignathus

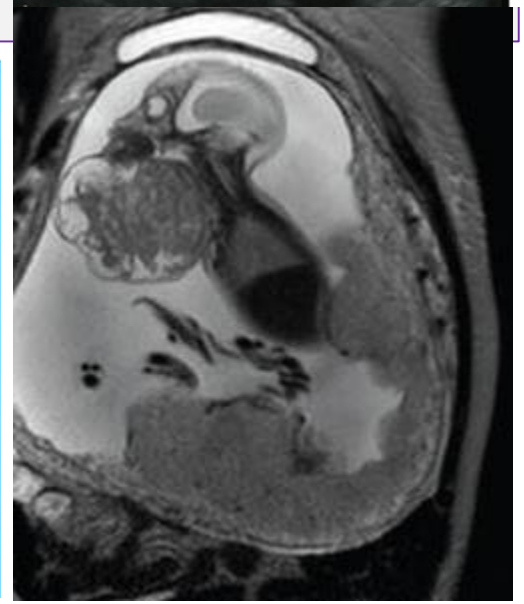
Differential diagnosis:

- Neck teratomas
- Encephaloceles
- Other tumors of the facial structures

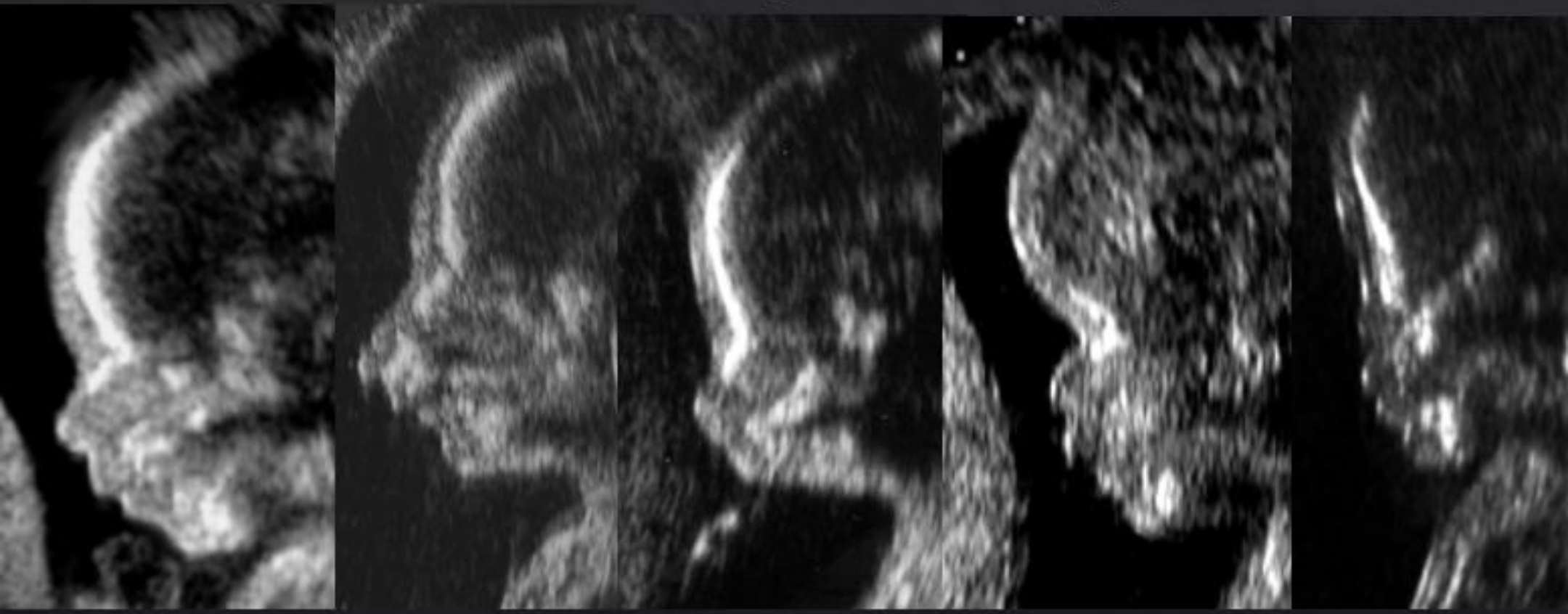






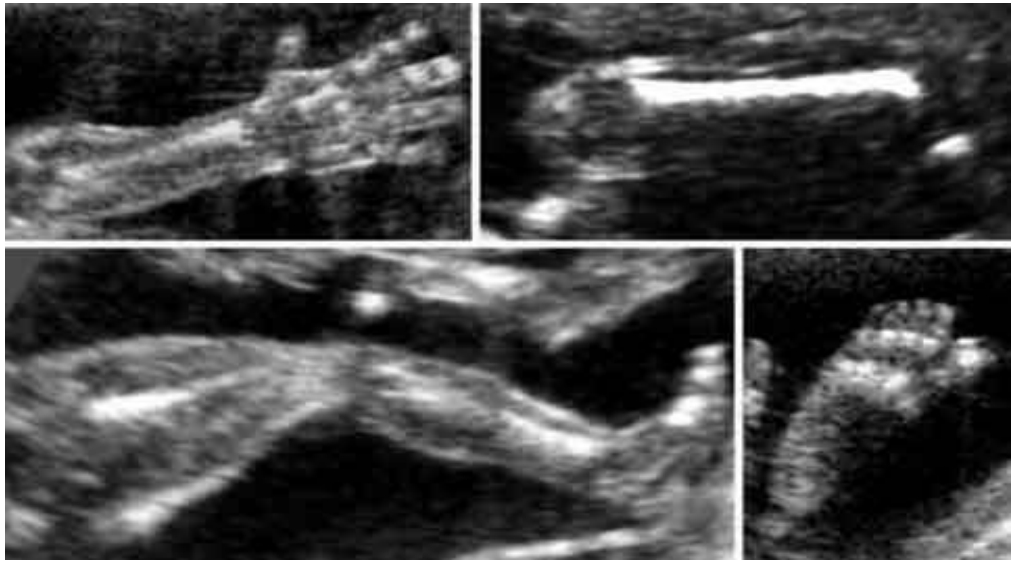


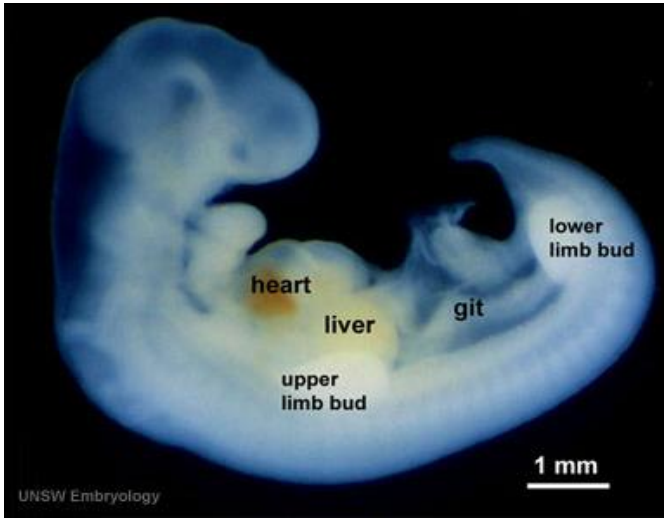
Fetal dysmorphism

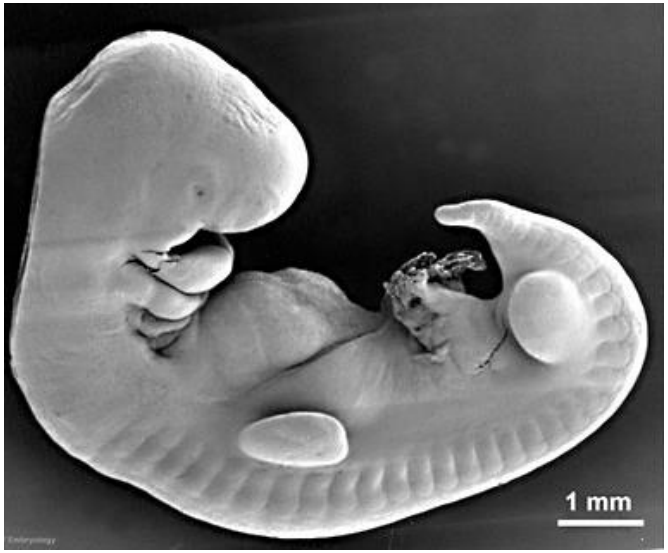


St George's University of London

Limb buds	8 wks
Femur & humerus	9 wks
Tibia/fibula & radius/ulna	10 wks
Digits of hands & feet	11 wks
Body movements	9 wks



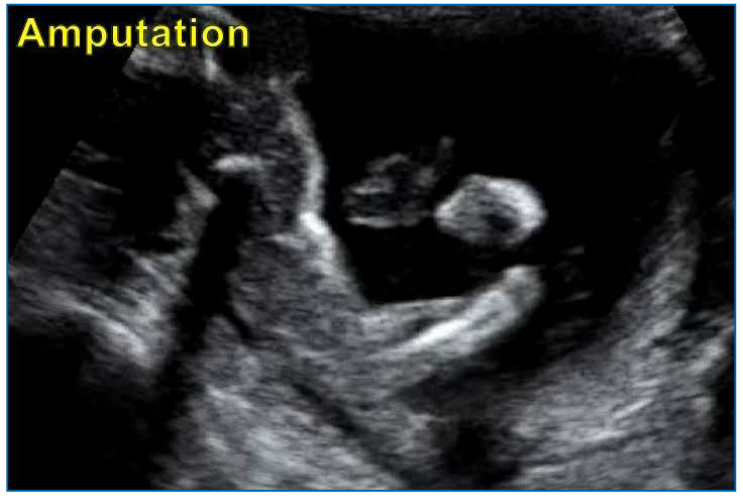


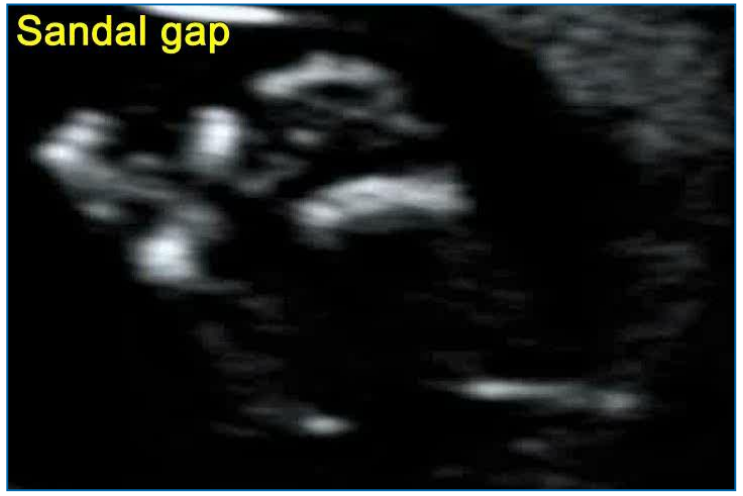


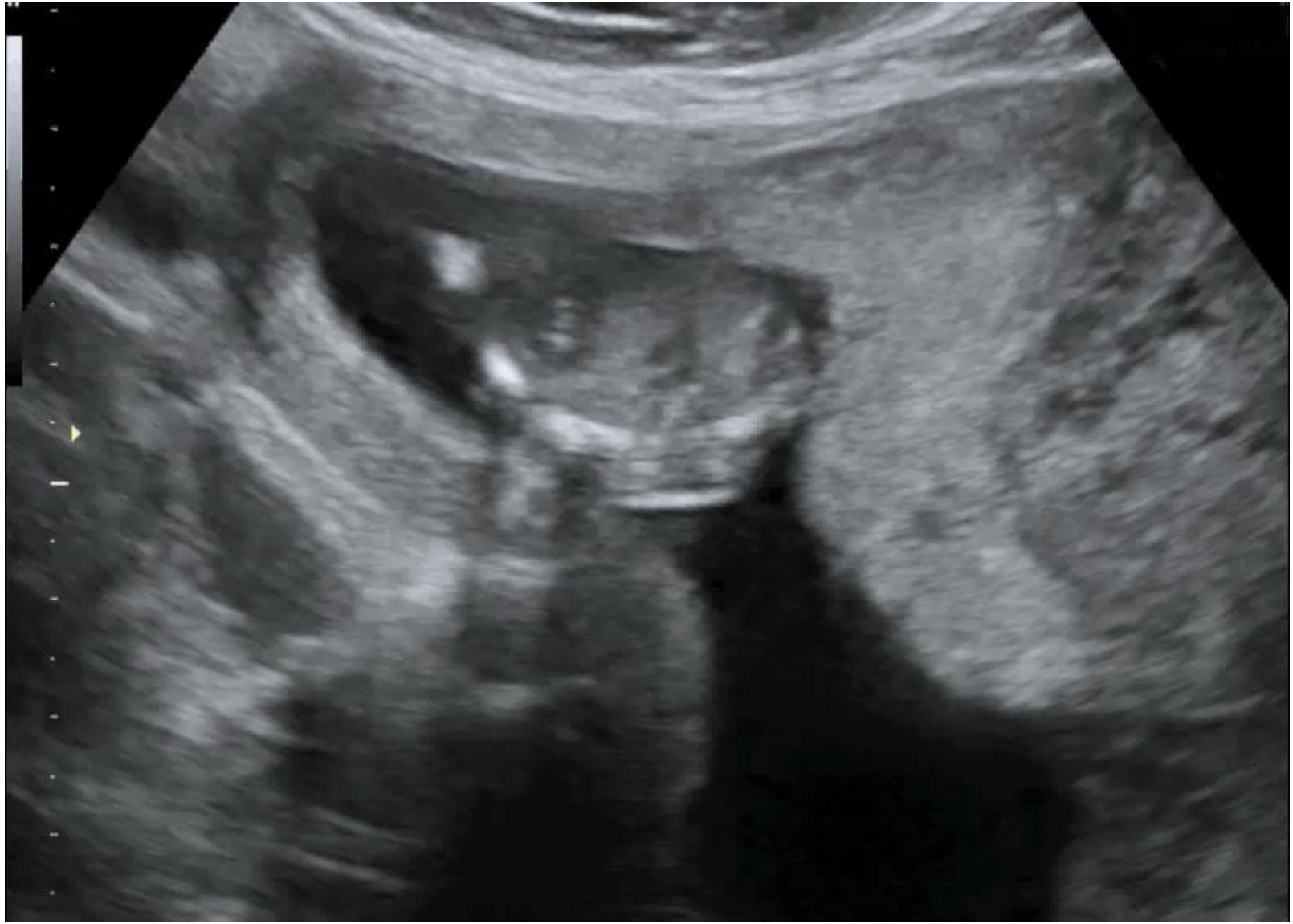
St George's University of London

Hand and foot abnormalities

Absent hands / feet	**62%**
Facial cleft	5%
Ventriculomegaly	9%
Spina bifida	14%
Major cardiac defect	33%
Diaphragmatic hernia	50%
Lethal skeletal dysplasia	50%







St George's University of London

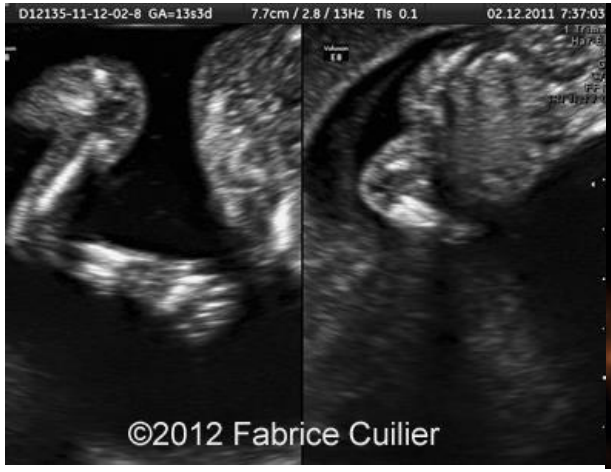
Limb Reduction Defects

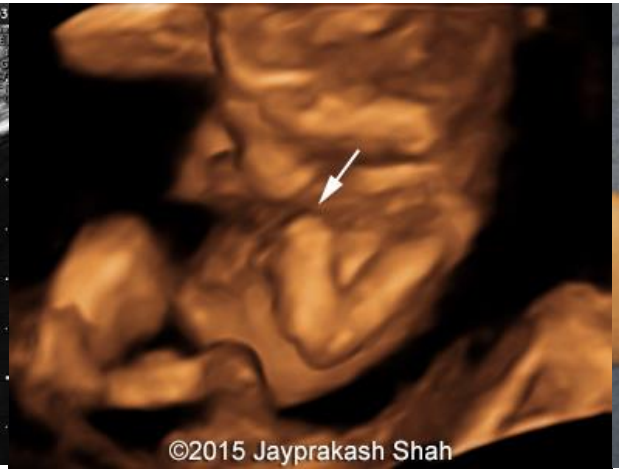
Amelia (complete absence of limb)

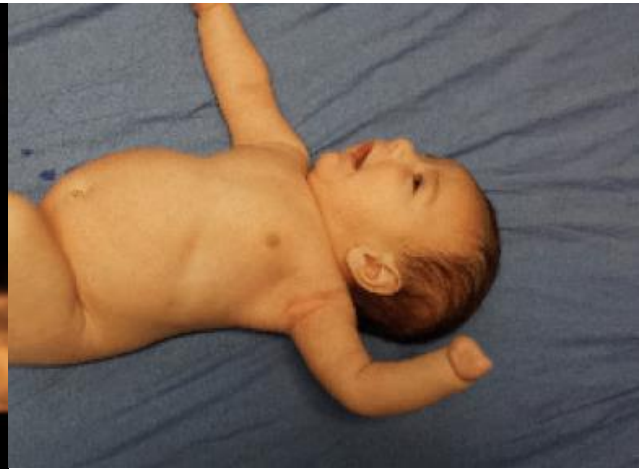
St George's University of London

Limb Reduction Defects

Acheiria (absence of the hand)







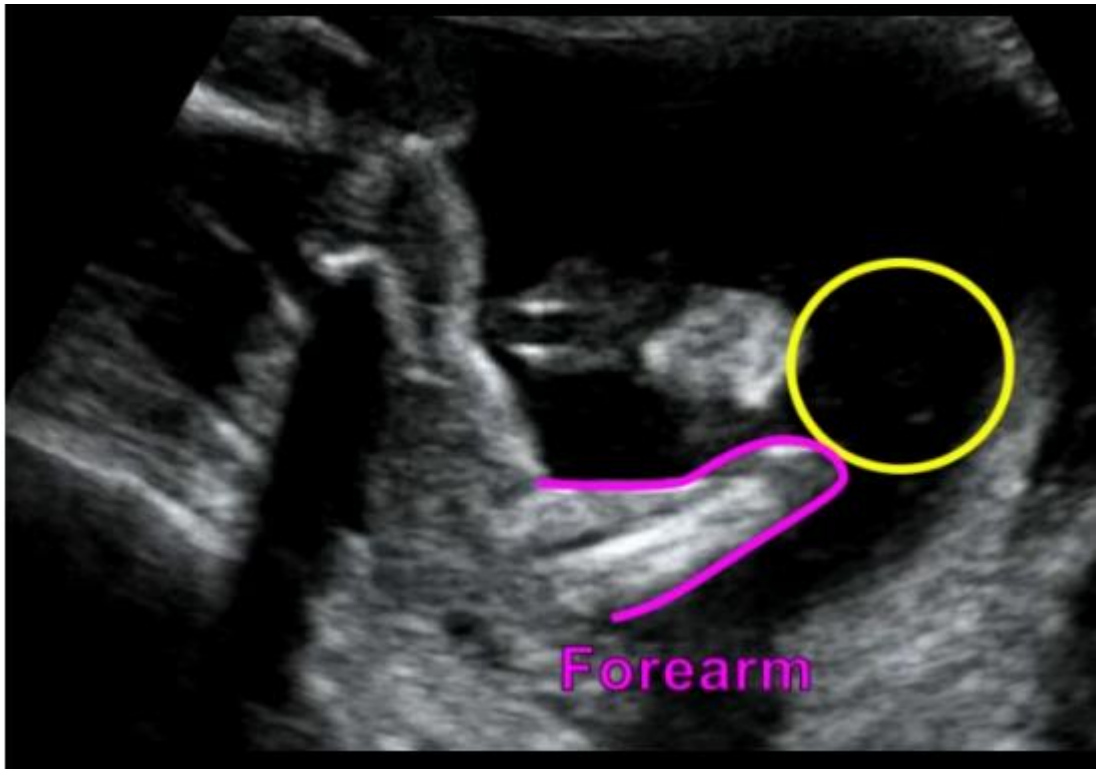
St George's University of London

Limb Reduction Defects

Acheiria (absence of the hand)

Limb Reduction Defects

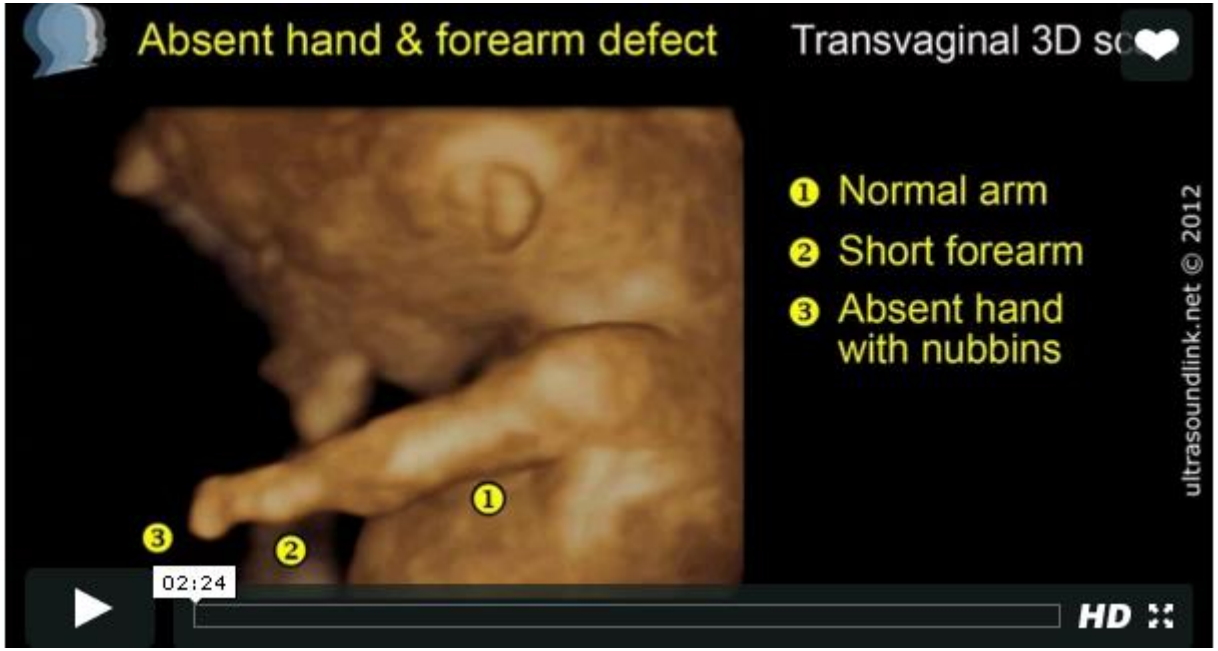
- 1 per 20 000 births
- 'limb deficiency' or 'congenital amputation'.
- 50% multiple
- 25% associated abnormalities
- Syndromes (spradic)
    - aglossia–adactylia syndrome
    - Moebius sequence
- Causes:
    - amniotic band syndrome
    - exposure to a teratogen
    - vascular accident



St George's University of London

Limb Reduction Defects

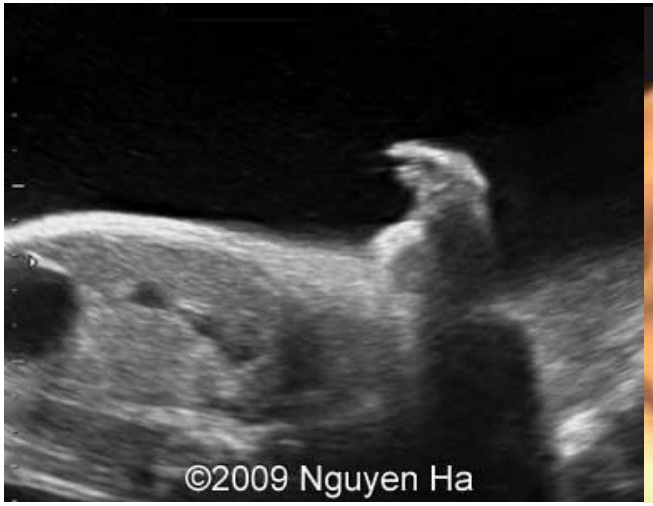
Acheiria (absence of the hand)

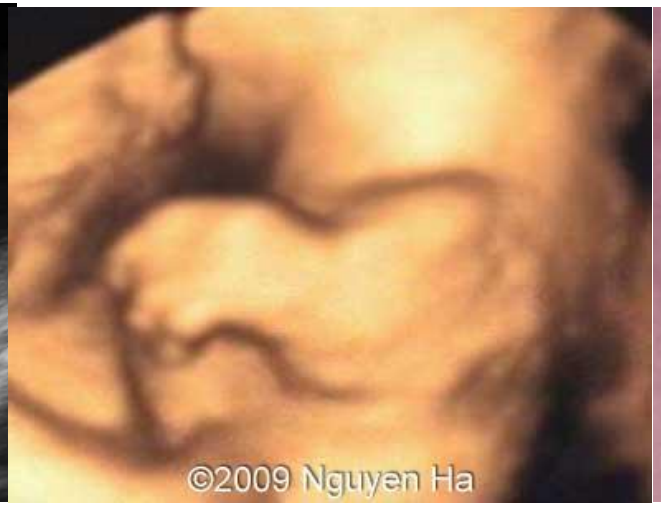


St George's University of London

Limb Reduction Defects

Phocomelia (seal limb)





- **Roberts syndrome** (AR, tetraphocomelia and facial clefting)
- **TAR syndrome**
- **Grebe syndrome** (AR, marked hypomelia of upper and lower limbs, lower limbs more affected than upper extremities)

St George's University of London

Limb Reduction Defects

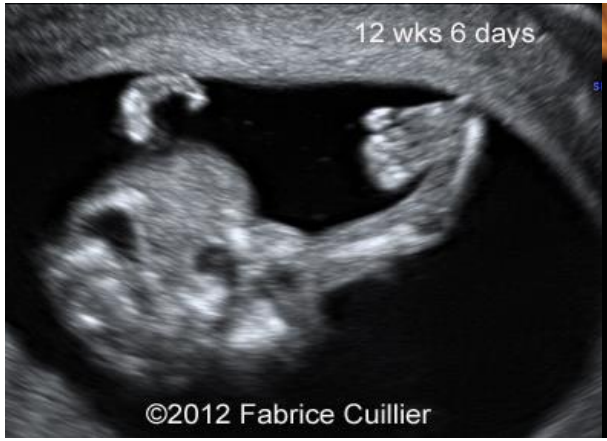
Drugs causing skeletal abnormalities

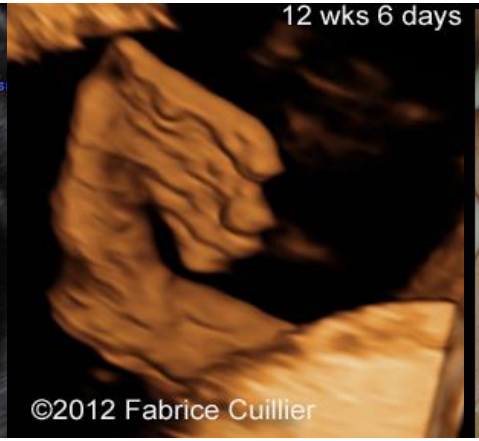
Drug	Skeletal	Other
Warfarin	Rhizomelia, stippled epiphyses, kyphoscoliosis	Depressed nasal bridge, renal cardiac, CNS, flat face
Sodium valproate	Reduction deformity arms, polydactyly, oligodactyly, talipes	Cardiac, CNS
Methotrexate	Mesomelia, hypomin skull, syndactyly, oligodactyly, talipes	CNS: NTD/Micrognathia
Vitmin A	Hypoplasia, aplasia arms bones	CNS, cardiac, NTD, cleft, DH, exomphalos
Phenytoin	Stippled epiphyses	Micrognathia, cleft, cardiac
Alcohol	Short long bones, reduction deformity arms, preaxial polydactyly arms, oligodactyly, stippled epiphyses	IUGR, cardiac
Cocaine	Reduction deformity arms +/- legs, hemivertebrae, absent ribs	CNS, cardiac, renal, ant abdo. Wall defects, bowel atresias

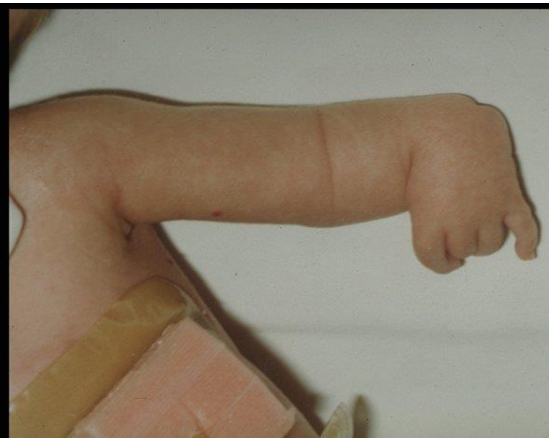
St George's University of London

Limb Reduction Defects

Aplasia–hypoplasia of the radius or ulna (club hand)







Radial clubhand

- frequently syndromatic
- absent thumb, thumb hypoplasia, thin first metacarpal, absent radius

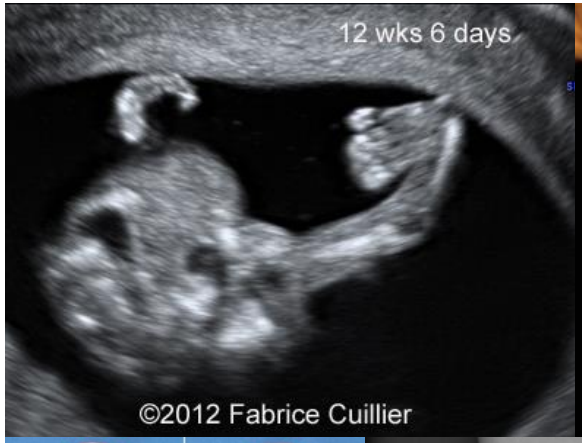
Ulnar clubhand

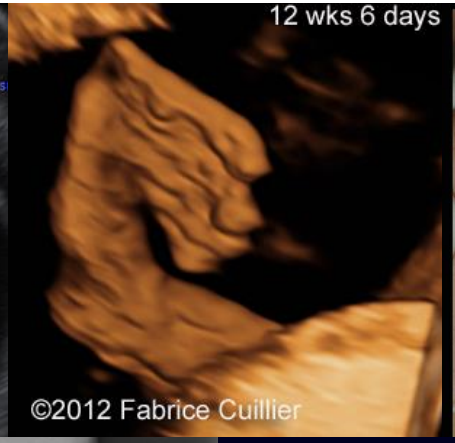
- usually isolated
- less common, ranges from mild deviations of the hand on the ulnar side of the forearm to complete absence of the ulna

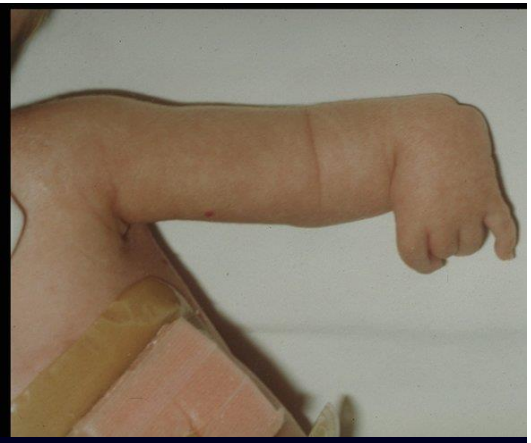
St George's University of London

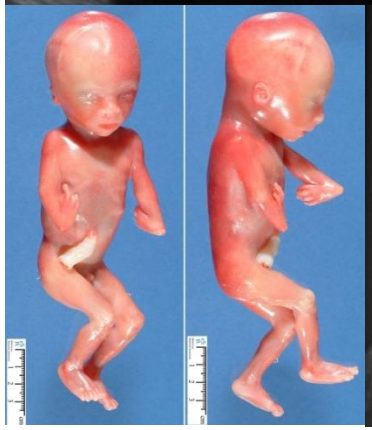
Limb Reduction Defects

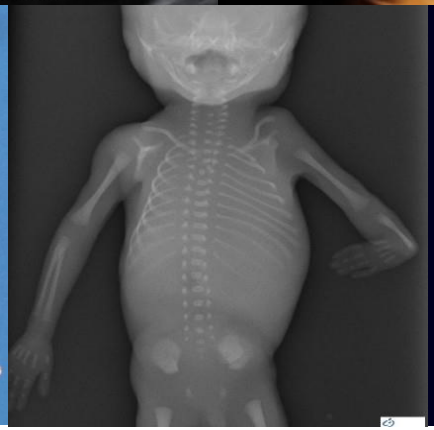
Aplasia–hypoplasia of the radius or ulna (club hand)









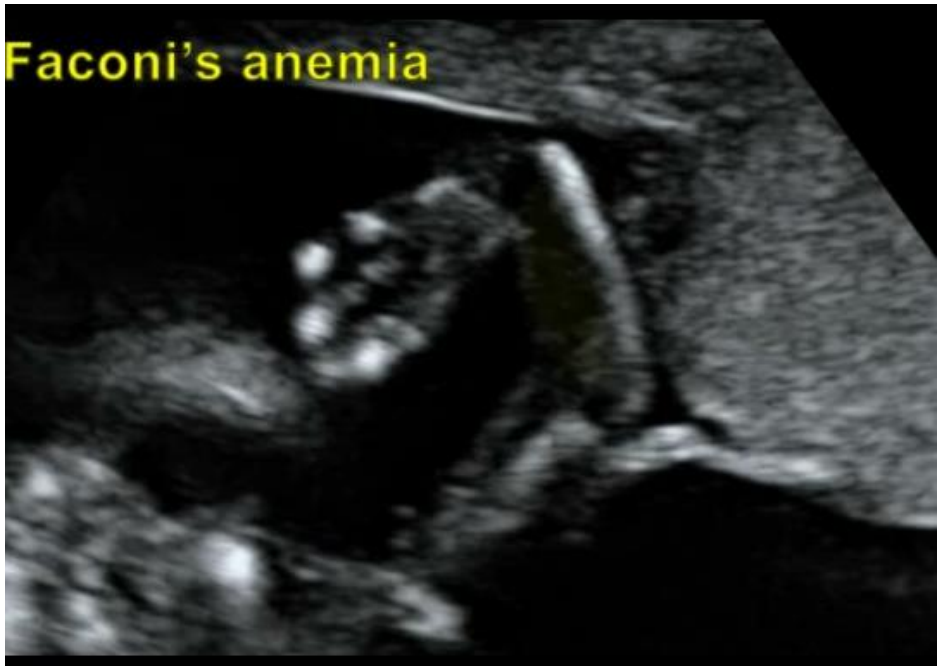


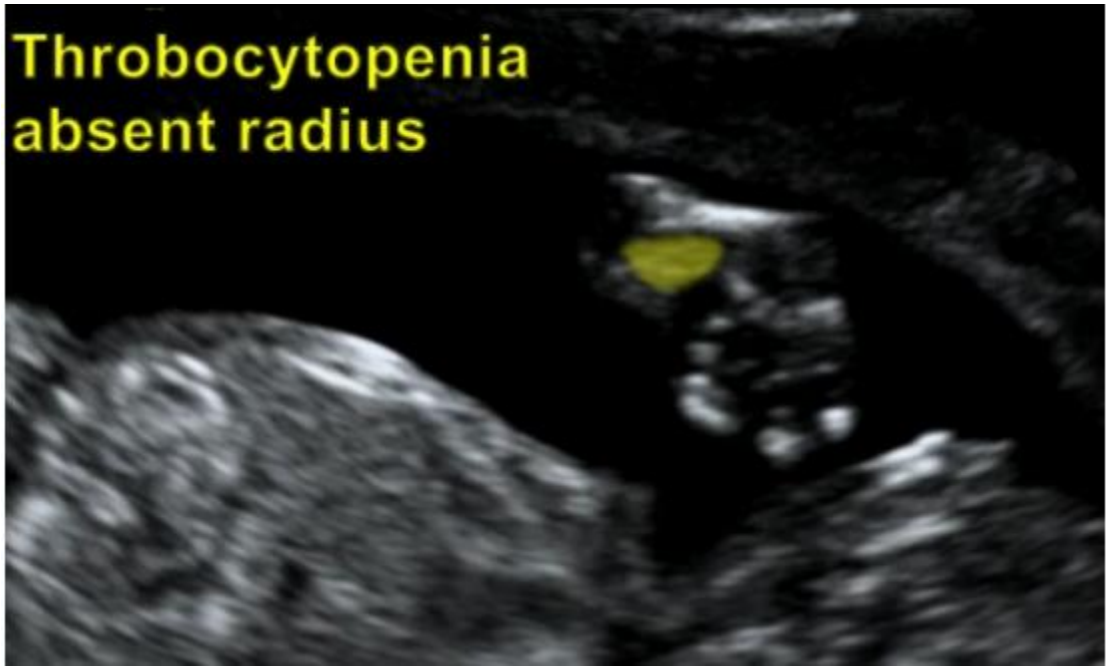
- Holt–Oram syndrome
- Fanconi anemia
- thrombocytopenia with absent radius syndrome (TAR)

St George's University of London

Limb Reduction Defects

Aplasia–hypoplasia of the radius or ulna (club hand)





St George's University of London

Limb Reduction Defects

Aplasia–hypoplasia of the radius or ulna (club hand)

Differential Diagnosis

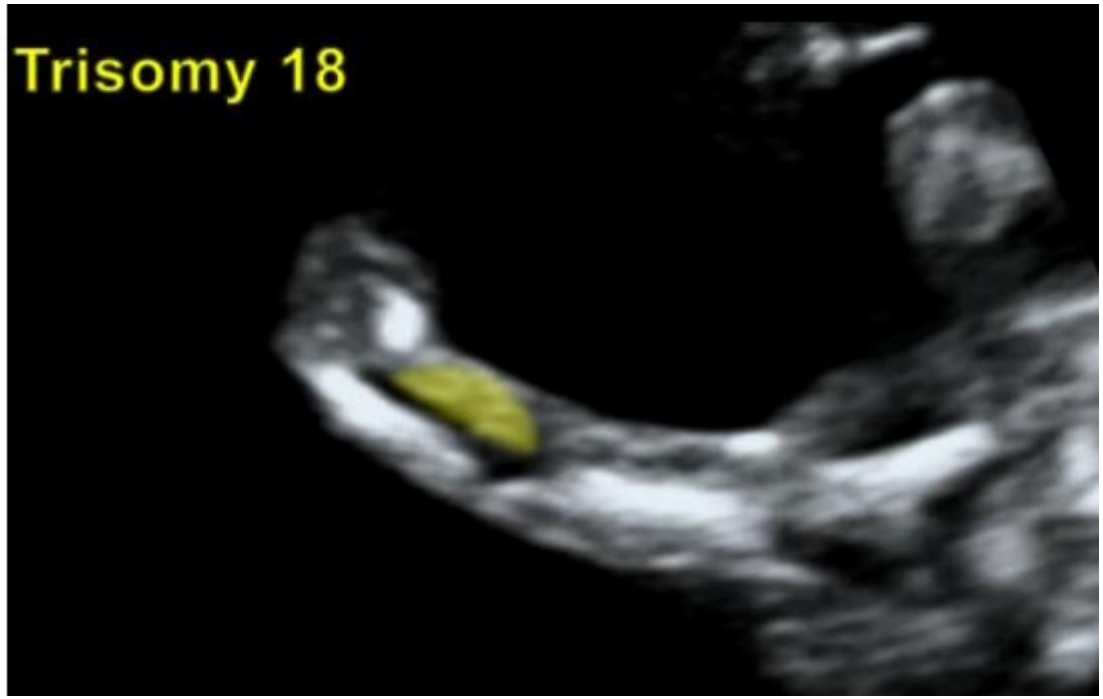
VATER

Vertebral

Anal atresia

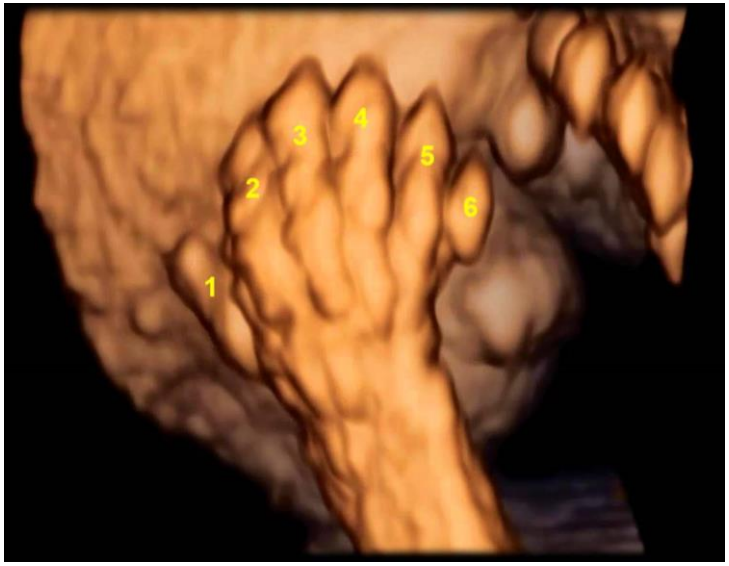
Tracheo-oesophageal fistula

Renal



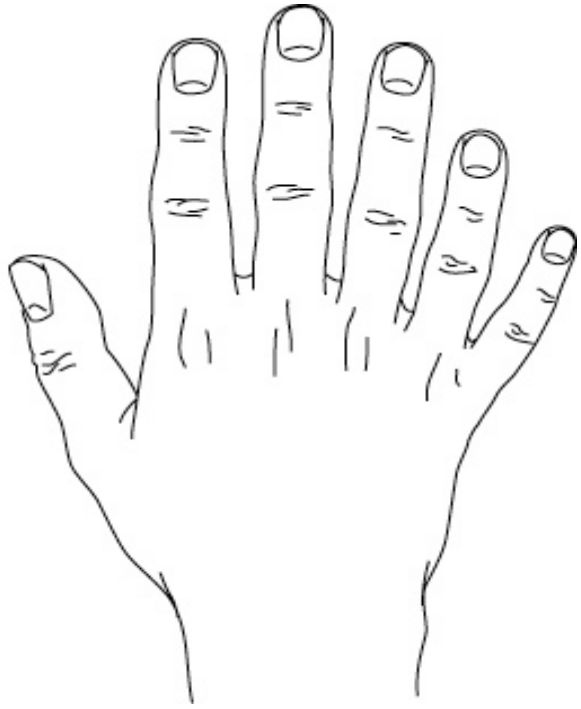
St George's University of London

Polydactyly

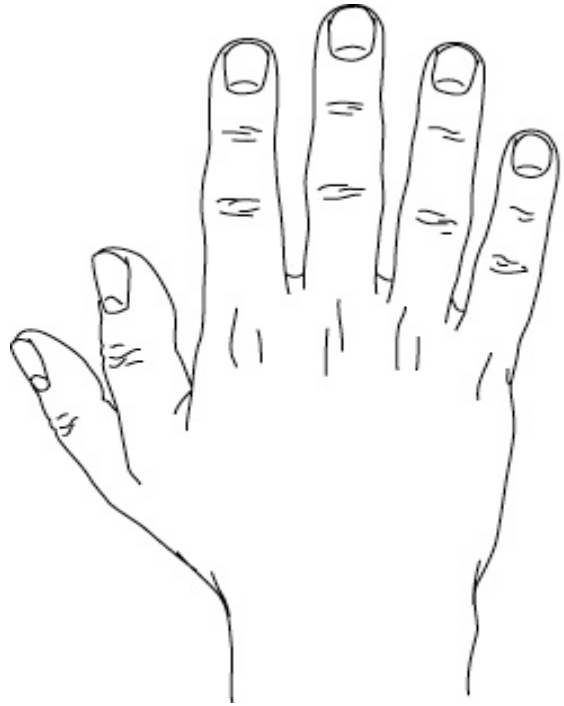


- Postaxial: isolated disorder AD
- More in AfroCaribbean
- Postaxial white: syndromic AR

Postaxial



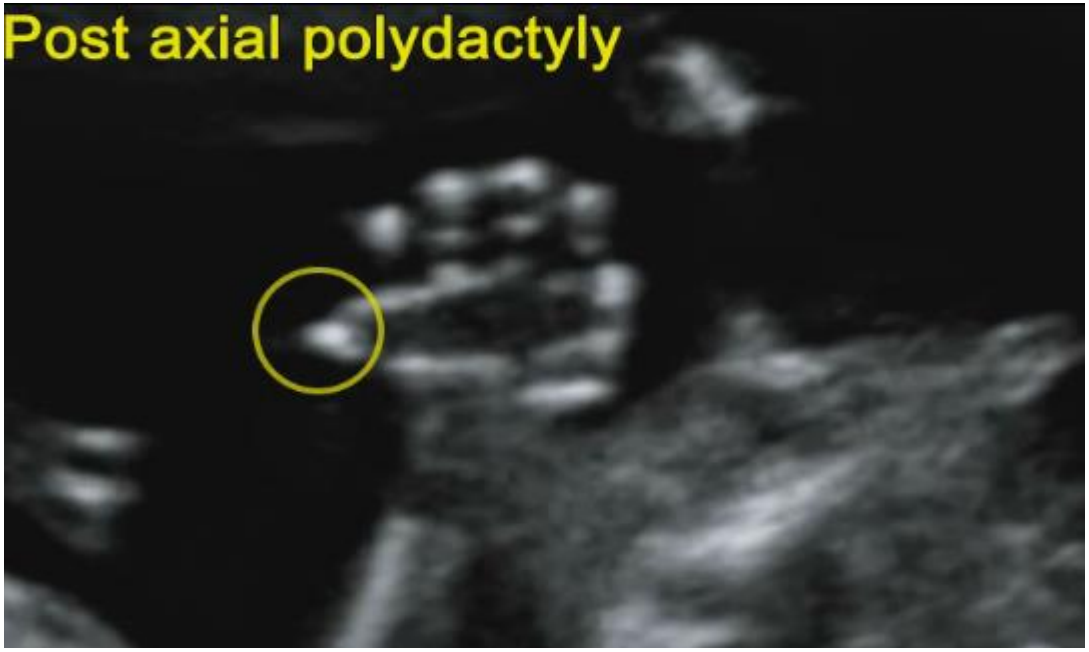
Pre-axial



St George's University of London

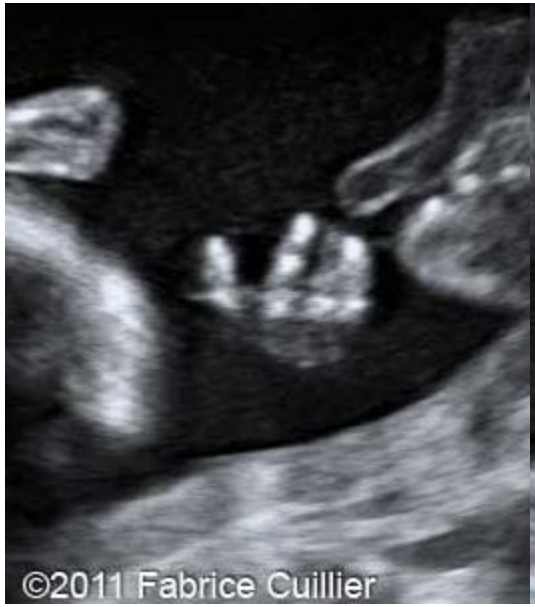
Polydactyly

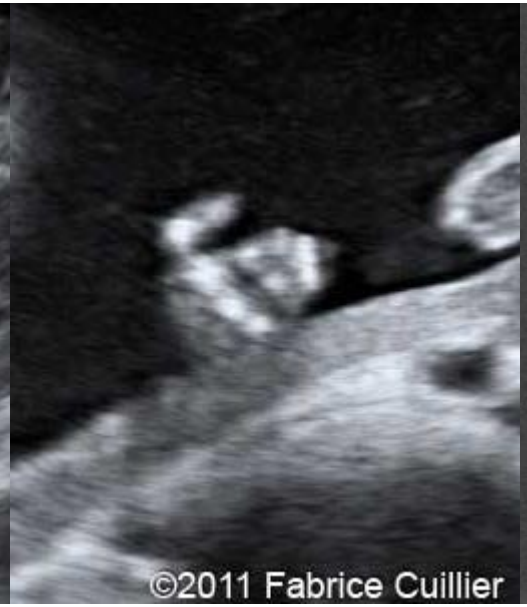
- **Fleshy nubbin or complete digit**
- **Central polydactyly**
  - usually hidden between the long and the ring finger
  - bilateral
  - associated with hand and foot malformations
  - AD

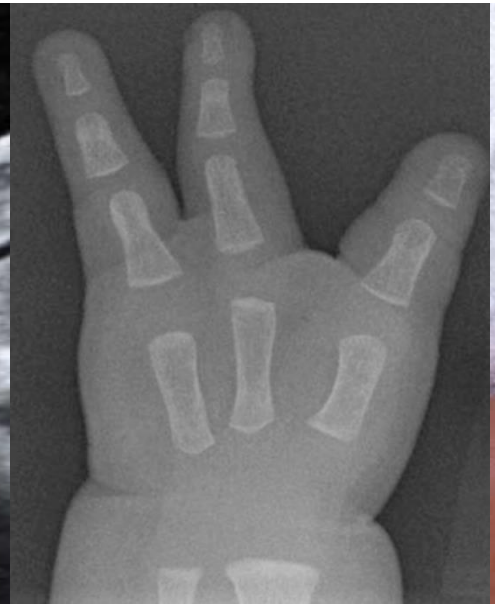


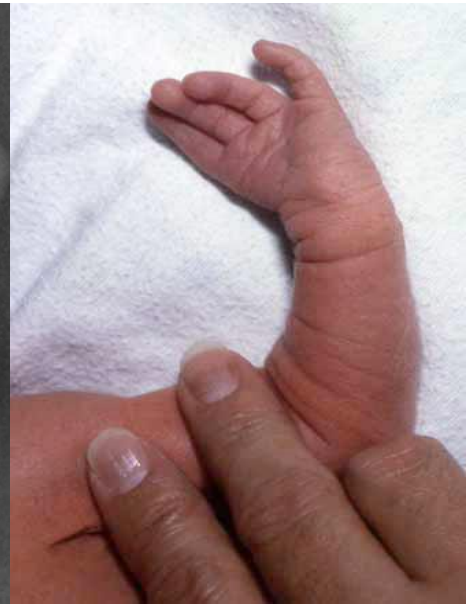
St George's University of London

Oligodactyly



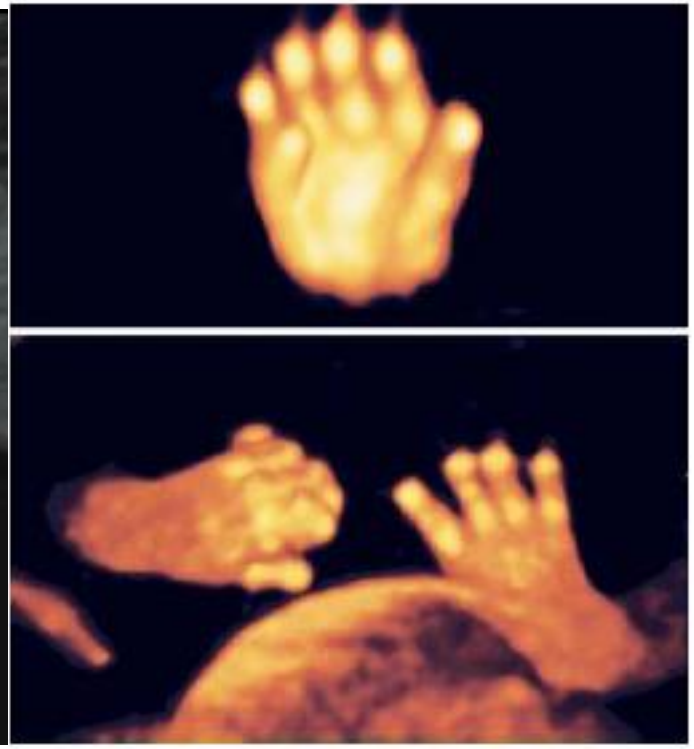


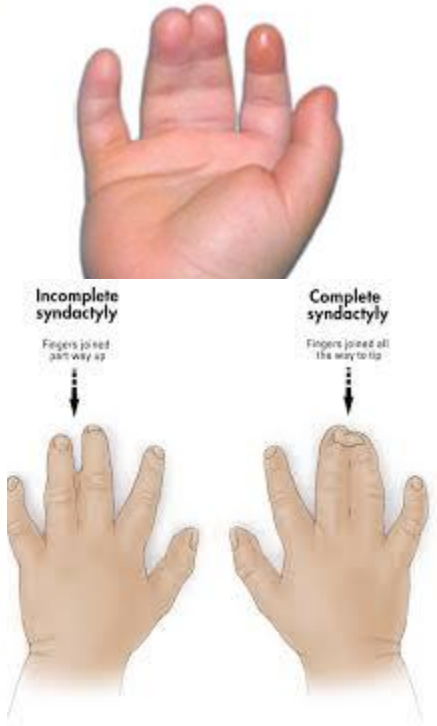




St George's University of London

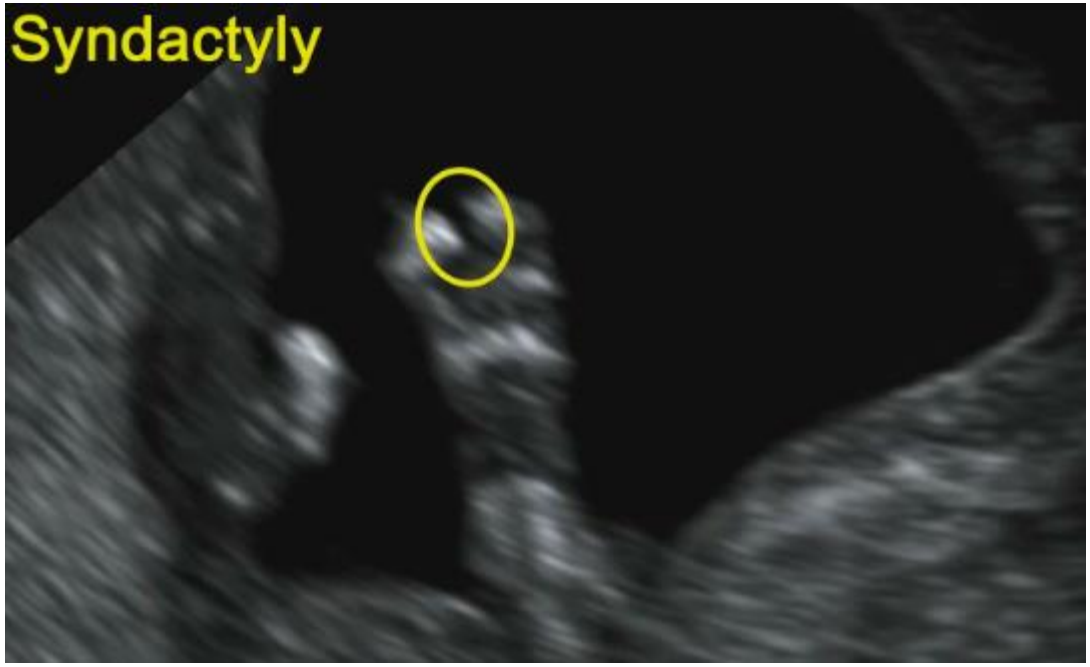
Syndactyly





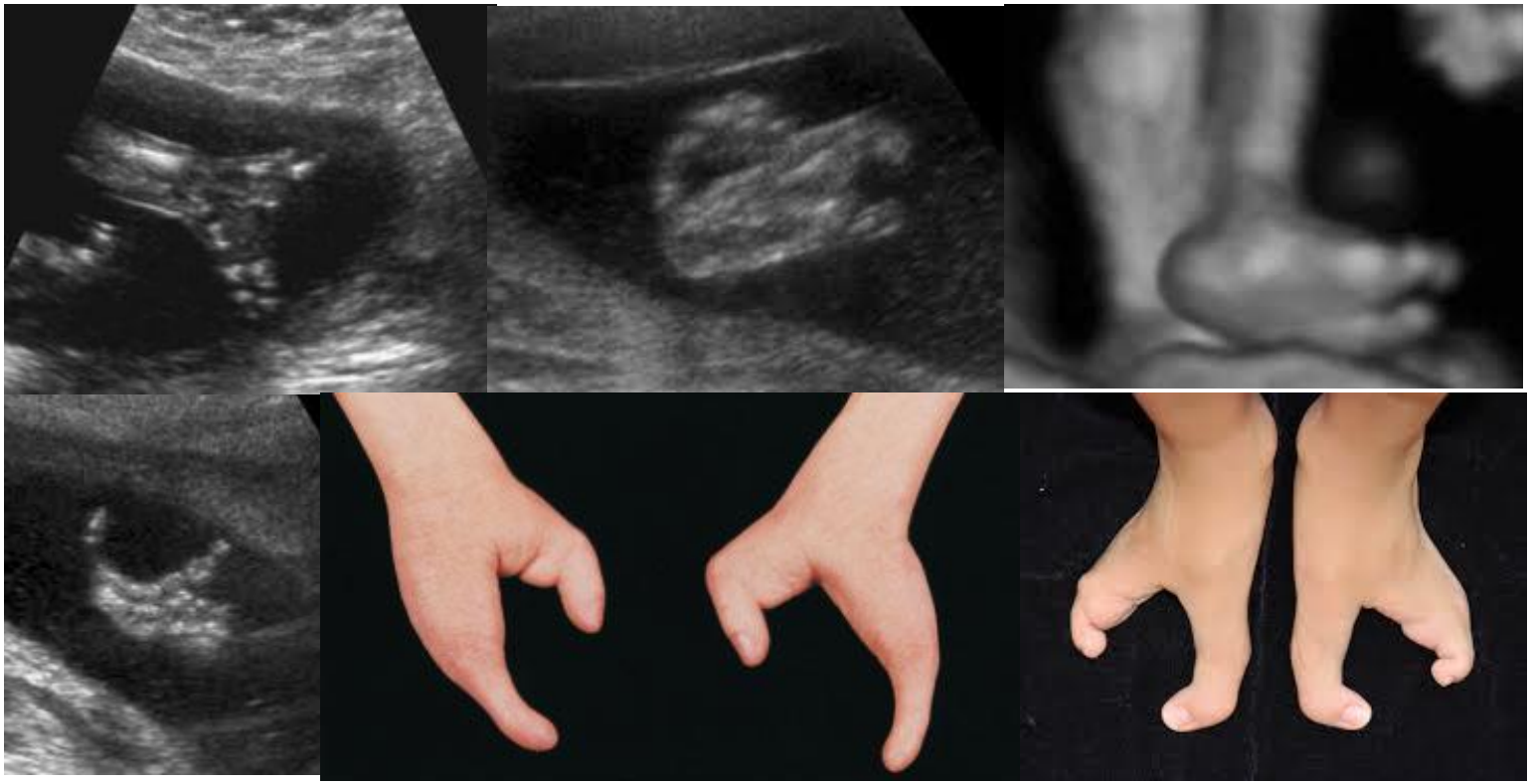
St George's
University of London

Syndactyly



St George's
University of London

Ectrodactyly

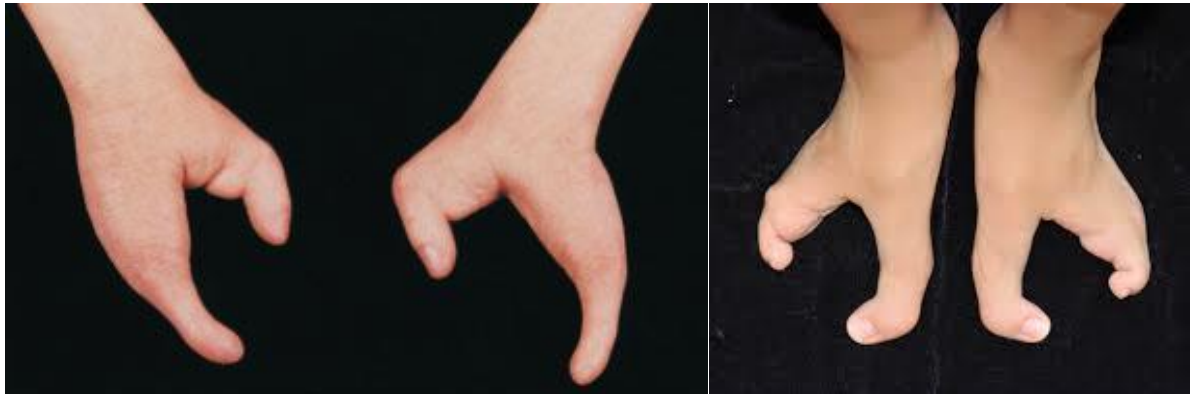


Ectrodactyly–ectodermal dysplasia–cleft syndrome
Split hand–split foot–ectodermal dysplasia–cleft syndrome
Lobster-claw deformity

St George's University of London

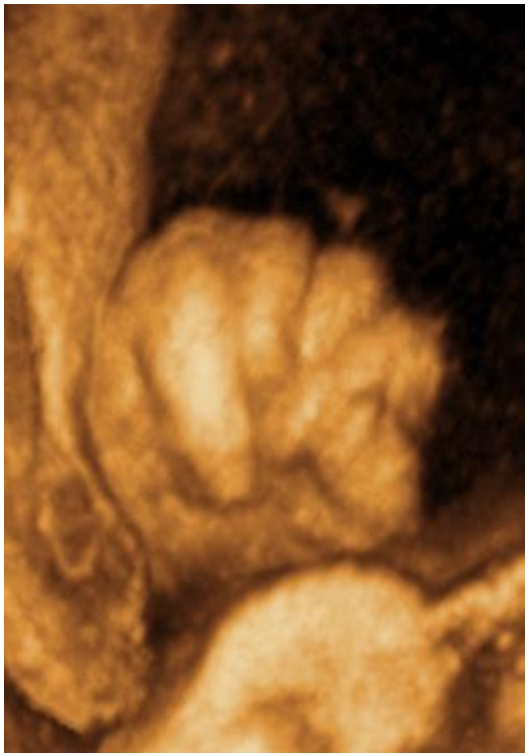
EEC syndrome

- AD
- four extremities
- more severe deformities of the hands
- wide spectrum of ectodermal defects
    - dry skin
    - sparse hair
    - dental defects
    - defects of the tear duct.



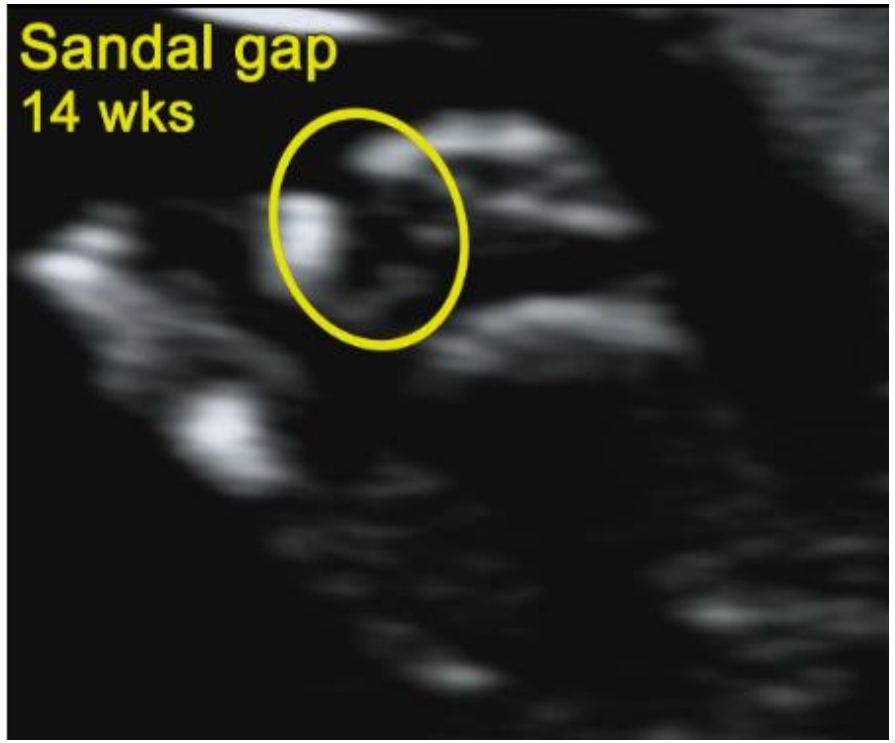
St George's University of London

Clenched hands



Trisomy 18

Sandal gap



Trisomy 21

St George's University of London

Challenges in prenatal diagnosis

- Rare conditions
- Few case reports
- Other investigations; associated syndromes
- Incidental finding
- Systematic examination