

The MANGO Study



Overview of Congenital Anomalies

Outline

- Definitions
- Overview of major congenital anomalies

Learning Objectives

By the end of this lesson, participants will be able to:

Skills
Describe the ways that congenital anomalies are classified
Recognize different types of major congenital abnormalities

Types of Congenital Anomalies (CAs)

- Major structural anomalies focused on.
 - Cause most deaths, morbidity & disability
 - Minor CAs can accompany Major CAs
- Definition of major CAs
 - ...structural changes that have significant medical, social or cosmetic consequences for the affected individual, and typically require medical intervention.

Examples of Major CAs

- Anencephaly
- Cleft lip
- Cleft palate
- Cleft palate with cleft lip
- Craniorachischisis
- Encephalocele
- Exomphalos/omphalocele
- Gastroschisis
- Hypospadias
- Iniencephaly
- Reduction defects of upper and lower limbs
- Spina bifida
- Talipes equinovarus/clubfoot

Detection of CAs

- Neonatal surface examination
- Internal examination through imaging
- MANGO will focus mainly on neonatal surface exam

Documenting CAs

- Legal framework (Check Kenya Data Protection Act 2018, also HIPAA and GDPR)
- Privacy and confidentiality considerations for PHI
 - Use of unique identifiers for neonates with CAs
 - Data aggregated and de-identified
 - Informed consenting to sharing of data / info

Communicating with parents

- Task reserved for qualified clinical service providers and NOT RAs
 - Referral services for urgent consults
 - May need written info for later after grieving period if death occurs
 - Support group
 - Genetic counseling

CA- Metrics

$$\text{Birth prevalence} = a/b \times 10\,000$$

a: Number of live births and fetal deaths (stillbirths) with a specific congenital anomaly (e.g. spina bifida) counted among the source population in a given year.

b: Number of live births and fetal deaths (stillbirths) (during the same year).

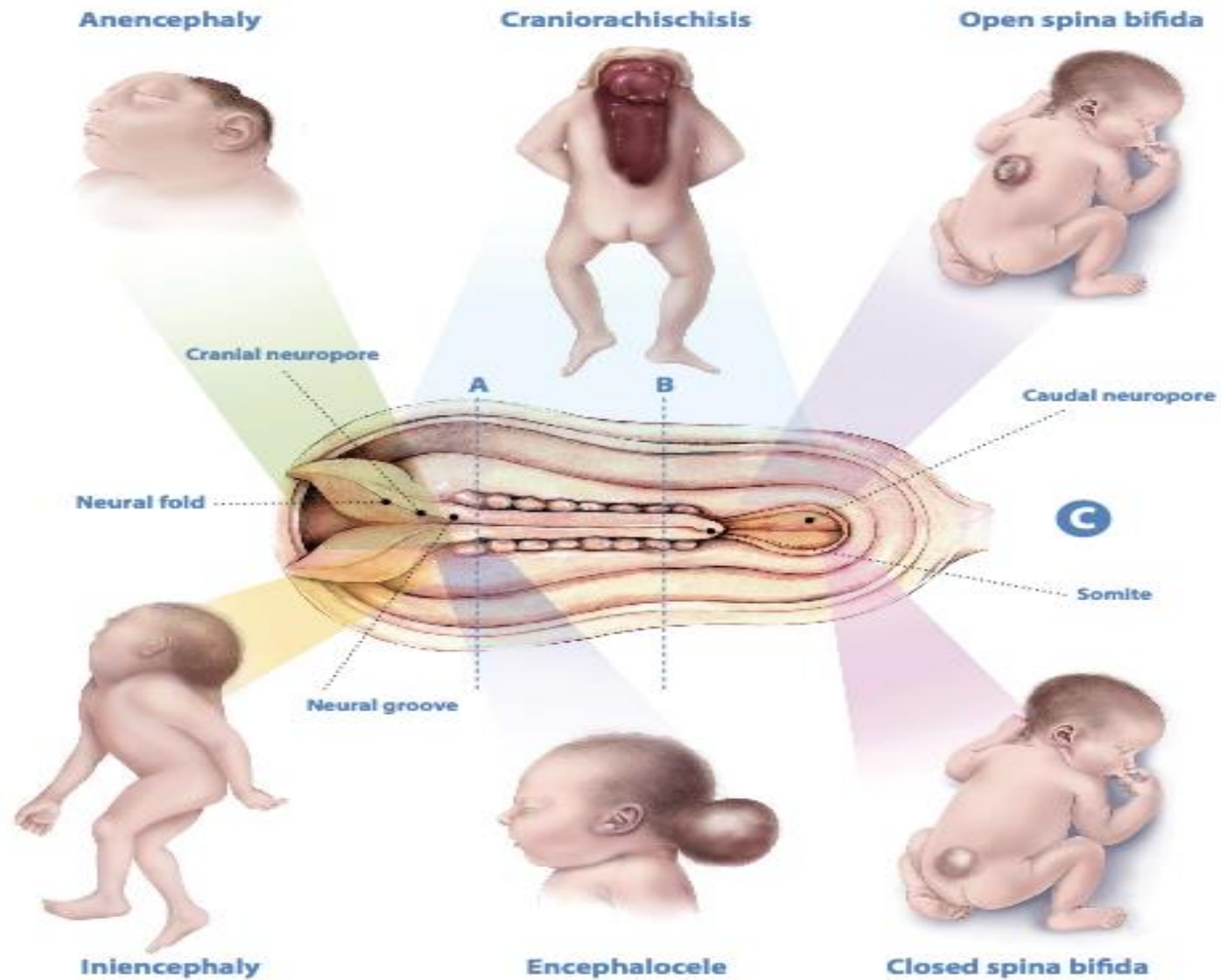
$$1. \quad \text{Live birth prevalence of congenital anomalies} = \frac{\text{live birth cases}}{\text{total live births}} \times 10\,000$$

$$2. \quad \text{Birth prevalence of congenital anomalies} = \frac{\text{live birth cases} + \text{fetal death (stillbirths) cases}}{\text{total live births} + \text{fetal deaths (stillbirths)}} \times 10\,000$$

$$3. \quad \text{Total prevalence of congenital anomalies} = \frac{\text{live birth cases} + \text{fetal death (stillbirths) cases} + \text{ETOPFA cases}}{\text{total live births} + \text{total fetal deaths (stillbirths)} + \text{total ETOPFA}} \times 10\,000$$

ETOPFA = elective termination of pregnancy for fetal anomaly.

CA-CNS



Variants of Encephalocele



Fig. 4.5 Encephalocele

a. Frontal encephalocele (Q01.0)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.



b. Nasofrontal encephalocele (Q01.1)

Photograph source: courtesy of Dr Jaime Frías (EE.UU.).



c. Occipital encephalocele (Q01.2)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Fig. 4.6. Spina bifida



d. Lumbar spina bifida



e. Cervical spina bifida without hydrocephalus (Q05.5)

Fig. 4.6. Spina bifida



a. Meningocele (Q05)



b. Myelomeningocele (Q05)



c. Myelocele or myeloschisis (Q05)

Photograph source (c): courtesy of CDC-Beijing Medical University collaborative project.



f. Lumbosacral spina bifida without hydrocephalus (Q05.7)



g. Lumbar spina bifida without hydrocephalus (Q05.7)

Cleft lip and cleft palate

Cleft lip with or without cleft palate, and cleft palate alone, are referred to collectively as orofacial clefts. Descriptions for each of these conditions follow. To aid understanding of the individual conditions, the structure of a normal palate is shown in Fig. 4.7.



Fig. 4.7. Normal palate

Cleft palate

(Q35, Q35.1, Q35.3, Q35.5, Q35.59, Q35.9, Q87.0)

Cleft palate is characterized by an incomplete fusion of the secondary palate and can affect the soft and the hard palate (see Fig. 4.8), or only the soft palate. Laterality of cleft palate is difficult to ascertain and some believe it does not exist.

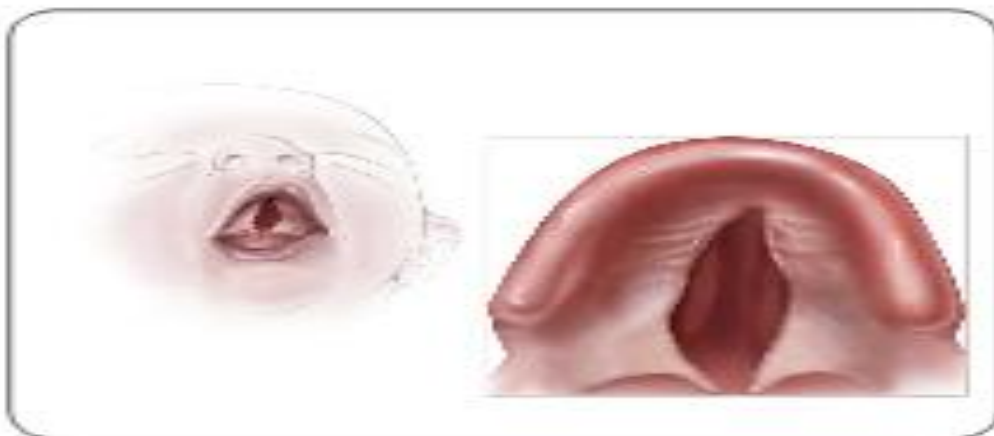


Fig. 4.8. Cleft hard palate with cleft soft palate (Q35.59)



Fig. 4.9. Cleft lip. a, b. unilateral (Q36.9) c. bilateral (Q36.0)

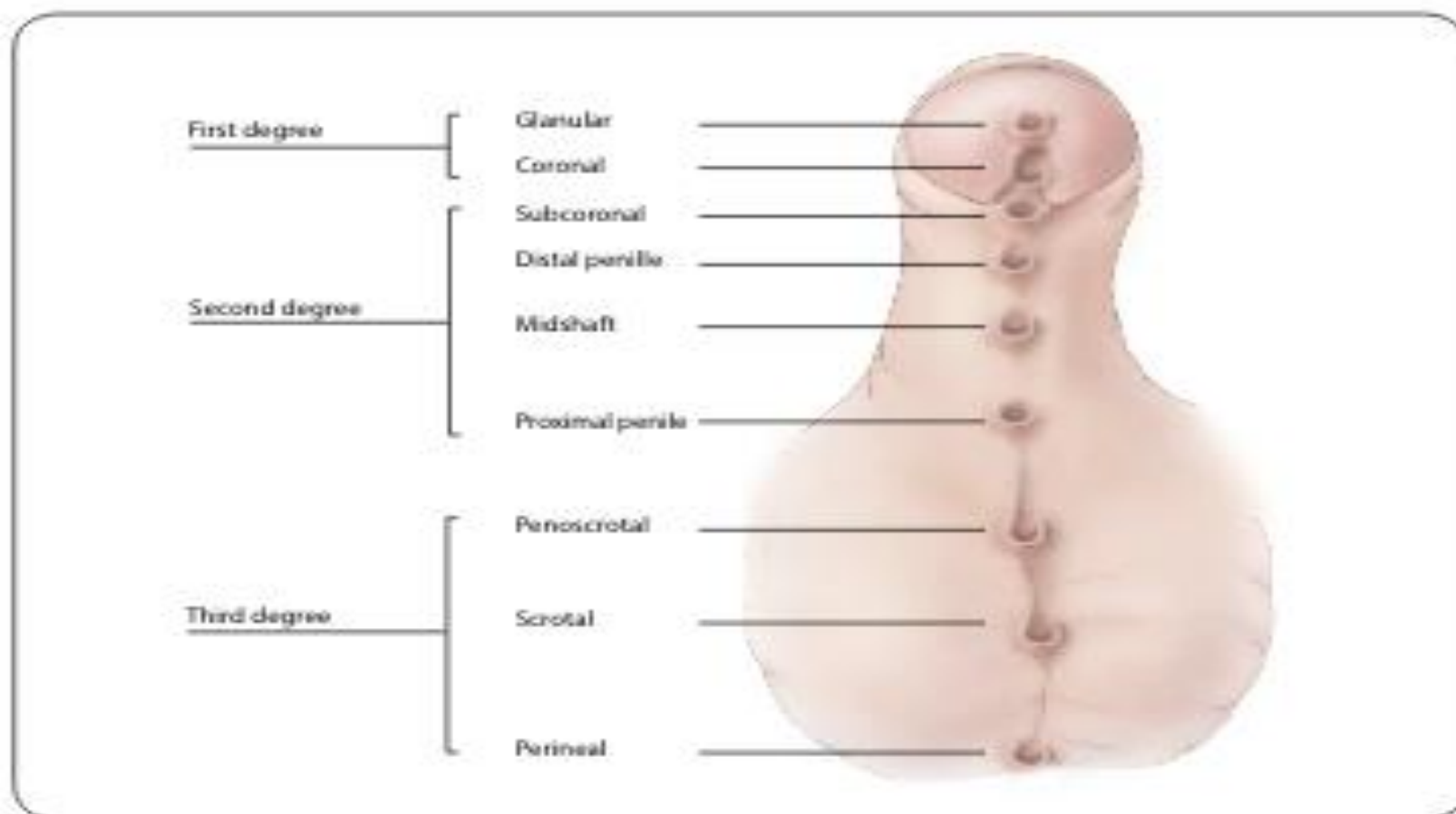
Photographs sources (b): courtesy of Dr Jaime Frías (EE. UU.). (c): courtesy of Dr Pedro Santiago and Dr Miguel Yáñez (EE. UU.).



Fig. 4.10. Cleft palate with unilateral cleft lip (Q37.10)

Photograph source: courtesy of Dr Pedro Santiago and Dr Miguel Yáñez (EE. UU.).

Fig. 4.11. Hypospadias (Q54, Q54.0, Q54.1, Q54.2, Q54.3, Q54.4, Q54.8, Q54.9)



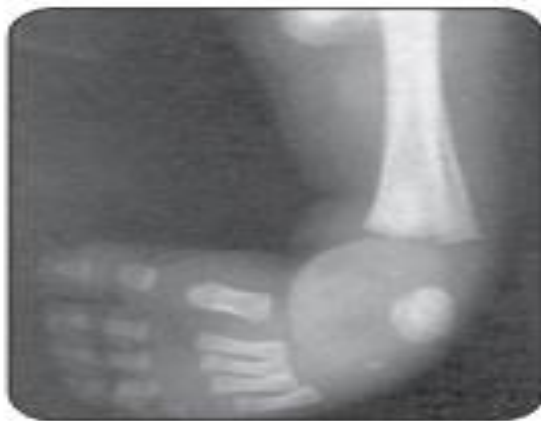




Fig. 4.13. Radial aplasia (Q71.4)



Fig. 4.16. Tibial aplasia (Q72.5)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.



Fig. 4.17. Split hand (Q71.6)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.



Fig. 4.18. Split foot (Q72.7)

Photographs source: courtesy of CDC- Beijing Medical University collaborative project.



Fig. 4.19. Congenital absence of forearm and hand (Q71.2)



Fig. 4.20



Fig 4.21

Fig. 4.20. Congenital absence of both forearm and hand (Q71.2)

Fig. 4.21. Congenital absence of lower leg and foot (Q72.2)

Photographs source: courtesy of CDC-Beijing Medical University collaborative project.



Fig. 4.22. Aphalangia of the hand. Partial absence of the phalanges (Q71.30)



Fig. 4.23. Aphalangia of the feet. Partial absence of the phalanges (Q72.30)

Photographs source: courtesy of John Wiley and Sons ©2009. Am. J. Med. Genet. A. 149A:93-127.



Fig. 4.24. Adactyly of the hand (Q71.30)

Photographs and X-ray source: courtesy Dr E. Gene Deune, Johns Hopkins Department of Orthopedic Surgery, Division of Hand Surgery (EE, UU).



Fig. 4.25. Adactyly of the feet (Q72.30)

Photographs source: courtesy of John Wiley and Sons ©2009. Am. J. Med. Genet. A. 149A:93–127.



Fig 4.26



Fig 4.27

Fig. 4.26. Oligodactyly of the hand (Q71.30)

Fig. 4.27. Oligodactyly of the foot (absent hallux) (Q72.31)

Photographs source: courtesy of John Wiley and Sons ©2009. Am. J. Med. Genet. A. 149A:93–127.



Fig 4.28



Fig 4.29

Fig. 4.28. Amelia upper limb (Q71.0)

Fig. 4.29. Amelia of the lower limb (Q72.0)

Photographs source: courtesy of CDC-Beijing Medical University collaborative project.



Fig. 4.30. Reduction defects of upper arm and forearm with hand present (Q71.1)

Photograph source: courtesy of Dr Jaime Frias (EE. UU.).



Fig. 4.31. Reduction defects of thigh and lower leg with foot present (Q72.1)



Fig. 4.32. a, b. Omphalocele (Q79.2) and c. with ruptured membrane

Photographs Source: courtesy of CDC-Beijing Medical University collaborative project.



Fig. 4.33. Gastroschisis (Q79.3)

Documentation thru Photography

- MANGO RAs will be responsible but will work in close collaboration with the clinical providers
- Only for those whose mothers provide a specific informed consent allowing it.
- identifiers will be avoided and if facial pictures, some kind of obscuring mechanism will be applied

Study expectations from the clinical team

- Routine detailed neonatal exam
- Accurate & complete documentation of the clinical findings of neonatal exam
- The RAs may identify areas of incomplete entry of patient information and liaise with the service providers to rectify
- Prompt referral of cases to the neonatologist as needed

Moving forward