

Tips for medical history taking during pregnancy with a focus on genetic history taking

Samuel Kabwigu

History taking during pregnancy

- The medical history is a structured assessment to get a comprehensive picture of a participant's health and health problems before and during her pregnancy.
- Health problems, medical treatment, and general health both before and during pregnancy should be assessed. Outcomes of prior pregnancies are important as are other risk factors for adverse outcome of this pregnancy including lifestyle (smoking and alcohol use) and family health issues.

Specific issues

- Need a setting with adequate privacy
 - Admission of prior pregnancy termination or abnormal outcome may be sensitive
 - May have had pregnancies that current partner or family are not aware of and need to be kept confidential
 - May be afraid to disclose behaviors not deemed socially acceptable during pregnancy
 - Family history of birth defects may also be sensitive
 - Unknown paternity (not sure who the baby's father or even her biologic father might be) can be very sensitive, but getting a history regarding someone else is not helpful, need to sort out as possible

Tips for the history-taker

- Be accepting and non-judgmental
- Use empathy
- Be supportive
- Be careful with sensitive issues and potentially embarrassing or disturbing topics

Genetic screening history

- One major limitation is to identify appropriate local terminology.
- In most cases describe the disease in local vernacular.

Genetic History Screening Tool

TERM	EXPLANATION/DEFINITION	NOTES
Cleft Lip or Palate	A child with cleft lip is born with part or the entire upper lip split or missing. In cleft palate, part or the entire roof (top) of the baby's mouth (inside) is missing at the time of birth.	
Heart Defect	The shape of the heart or connections of the blood vessels to the heart are not normal, and thus the heart does not move blood properly. The baby may look normal on the outside, but may look blue, grey or pale all of the time or just with activity or	This can sometimes be seen on ultrasound, especially a special ultrasound called echocardiogram. It is not diagnosed just by
Spina Bifida	A baby with spina bifida (also known as open spine) may have a lump sticking out of the middle or lower part of the back or the area may just be open with no skin or covering. The baby may not be able to feel things normally (abnormal sensation) and to move	
Muscle Disease/ Muscular Dystrophy	The muscles are very weak and may get weaker over time. Sometimes, this may occur with other problems in the body such as stomach problems, learning problems, etc.	
Mental Retardation	The child does not learn like typical children. They learn skills that are normal for children their age.	
Down Syndrome	A child is born with a very small chin, weak muscles, a tongue that may rest outside of the mouth, a short neck, a very round face, and mental retardation (see above).	
Cystic Fibrosis	This disease causes difficulty breathing and frequent infections of the lungs. Children may have other problems in the body such as blockage of the bowels from thick mucus and not grow properly.	
Kidney Disease	The body needs kidneys to help clean the blood and make urine. A person born with kidney disease may have problems growing, often, have side or back pain, or may have no symptoms.	
Sickle Cell Anaemia	This is a disease of the blood that causes people to have episodes of very severe pain, due to their blood not flowing properly. These painful episodes can be triggered by a fever or infection.	
Haemophilia	The body has difficulty controlling bleeding inside and from the skin. Even a small cut or minor surgery may cause a lot of bleeding. People with haemophilia may have cuts that will start bleeding again after stopping at first.	
Thalassaemia	This is a disease of the blood that can cause fatigue. It is a form of anemia that can be mild or severe. It is not the common form of anemia seen in pregnancy or in people who lack iron in their diet. The liver and spleen may be enlarged.	



Cleft Lip



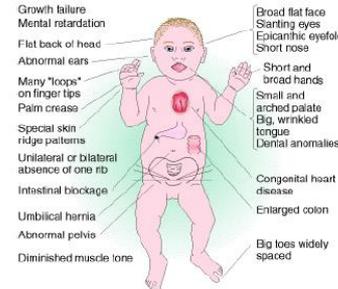
Spina bifida



People with Down syndrome



Muscular dystrophy



Some features of Down syndrome

MTN-016 Staff Guide for Explaining Conditions on GSH-1 CRF

For internal use only. Image Credits: US Centers for Disease Control, Down Syndrome South Africa, Netter, US NIH

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Staff may use this tool to aid in discussions with participants while in the clinic.

GSH cont

- Examples of description from GSH CRF:
 1. cleft-lip or palate: birth defect(congenital) of the upper part of the mouth. Cleft=split/separated.
Use pictures if helpful. Describe as an opening or hole in the middle of the upper lip.

GSH cont

2. Heart defects: a problem(s) with the heart's structure and function that is present at birth.

Ask whether heart problem was found at birth or later and any further description. Ask if it is a problem with the walls in the heart (like a hole in the heart or septal defect) or with the valves.

3. Spina bifida: *latin* spina=spine; bifida=split

a congenital defect of the spine in which part of the spinal cord and its meninges are exposed through a gap in the backbone.

Ask about any swelling, lump or opening on the back at birth (along vertebral column).

GSH cont

4: Muscle disease/muscular dystrophy; is a group of ***muscle diseases*** that weaken the musculoskeletal system and hamper locomotion.

Ask about weak or floppy arms and legs and inability to walk

5. Mental retardation: lifelong condition of impaired or incomplete mental development. IQ <70.

Ask about inability to learn new tasks as compared to others in same setting (siblings or neighborhood children)

6. Down syndrome: a genetic disorder, associated with the presence of an extra chromosome 21, characterized by mild to severe mental impairment, weak muscle tone, shorter stature, and a flattened facial profile.

Use of pictures may be helpful. May be known as mongoloid or mongol.

GSH cont

7. Cystic fibrosis: a disease passed down through families that causes thick, sticky mucus to build up in the lungs, digestive tract, and other areas of the body.
8. Kidney disease
9. Sickle cell anaemia
10. Hemophilia: h/o bleeding tendencies
11. Thalassaemia (Mediterranean/Cooley's anaemia)

Conclusion

- Its vital to get the nomenclature for each disease in local language if possible.
- Describe the disease appropriately if no single term known locally.
- Use visual aids as much as possible.