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BIRTH DEFECT SURVEILLANCE & IMPORTANCE OF GOOD QUALITY DATA

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What is birth defect (BD) surveillance?

What is BD surveillance?	What does this mean?
Ongoing, systematic collection, analysis, and interpretation of health data on BDs.	Keeping careful records of babies born with BDs and making sense of the information
- Estimates prevalence of BDs - Monitors trends - Identifies clusters	- How often are babies born with BDs in a specific population? - How do numbers change over time? - Are BDs more/less common in certain areas/ populations/ circumstances?
Identifies risk factors	Under which circumstances are BDs more likely? What might be causing the BDs?
To inform prevention and healthcare policies	The information should be used to decide: - Which programs or guidelines to put in place to detect, manage or prevent the BDs.

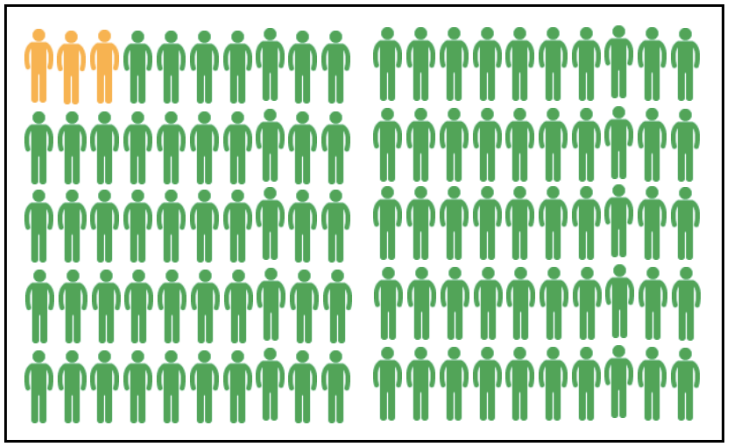
What is "prevalence" of BDs?

What's the difference between "number" and "prevalence" of BDs?

3 BD cases

Is it a lot?
Is it a little?

3 BD cases
out of 10
births



3 BD cases
out of 100 births

Prevalence (also sometimes called "rate") of a particular BD = number of babies born with that BD *out of the total number of babies in the population you're looking at*.

Often expressed as number of BDs per 1000 babies born

E.g. "7 per 1000 liveborn babies have major BDs identifiable on surface exam in the WC"

Why is BD surveillance important?

The most significant part of a public health surveillance program is how the data collected will be used to *promote the health of the population.*

In South Africa, BDs are vastly under-reported

Why is BD surveillance important?

For example:

What can we learn from BD surveillance?

How many babies are born with specific BDs, and where?

→ What is needed to care for these babies?
e.g.
- More staff, services and budget needed?
- Establish referral pathways
- Inform training for nurses and doctors

→ Improved care and management for babies with BDs

How can this impact the health of the population?

Why is BD surveillance important?

For example:

What can we learn from BD surveillance?		How can this impact the health of the population?
Is the prevalence of specific BDs higher when pregnant women take a particular medication?	→ Detects medications that might be increasing the risk of BDs. →	Amend guidelines for safer use of products in pregnancy – helps reduce number of babies born with this BD

Why is BD surveillance important?

For example:

What can we learn from BD surveillance?

How can this impact the health of the population?

NTDs were noted to be increased in women with folic acid deficiency.

→

- Add folic acid to staple foods
- Update pregnancy care guidelines
- Give folic acid supplements in pregnancy.

→ Fewer babies born with NTDs

Ongoing BD surveillance can help monitor the response to the intervention: Were babies born less often with NTDs after the interventions were put in place?

Why is BD surveillance important?

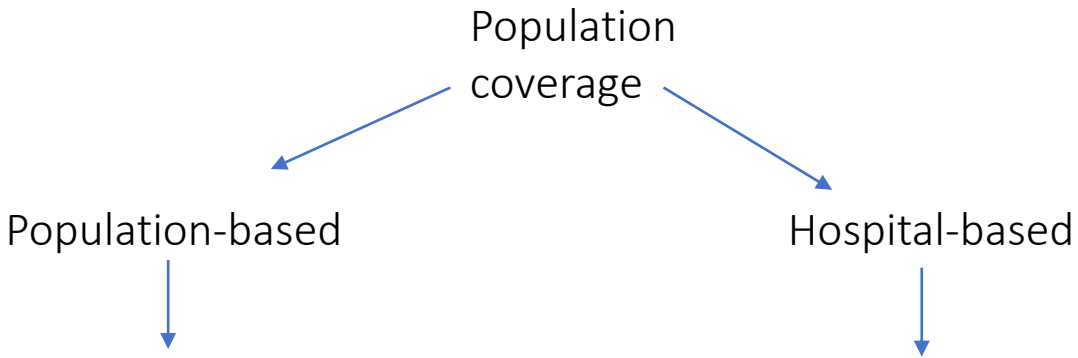
For example:

What can we learn from BD surveillance?

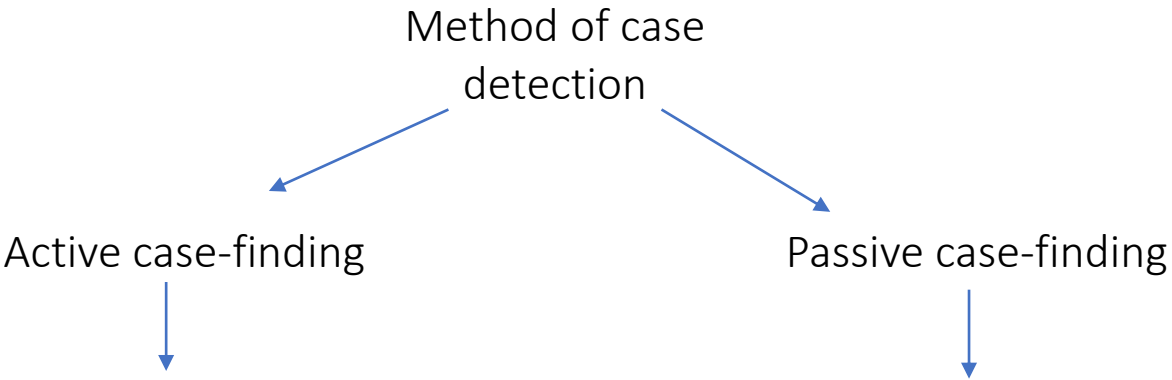
Are BDs much higher in areas where population is exposed to environmental toxins e.g. Mining chemicals → Detects environmental toxins that might be causing the BDs → Interventions can be put in place to improve safety of communities

How can this impact the health of the population?

Types of birth defect surveillance systems



Types of birth defect surveillance systems



Optimising data quality

BD surveillance is only as good as the quality of data collected!

How do we optimise BD data quality?

Optimising data quality

Health staff training on BDs

Clubfoot

Fixed, abnormal position.
Foot cannot be gently stretched into correct position.
Needs referral.

Major birth defect

vs

Positional deformity

Foot can gently be stretched into a normal position.
Will self correct with time.
Does not need referral.

Not a Birth Defect

Optimising data quality

Health staff training on BDs

Polydactyly

Give detailed description of polydactyly

- Fingers or toes
- Both sides or one side only
- Pre- or post-axial (thumb side or baby finger side)
- Bone or no bone

Entire additional finger/s, with a bone

OR

Just a skin tag, with no bone

Major birth defect

Minor birth defect

Optimising data quality

Health staff training on BDs

Multiple minor BDs might indicate an underlying syndrome, which is a major BD.

E.g. Short neck
Low-set, externally-rotated ears
Small chin

Minor birth defects

…in a baby with Noonan's Syndrome

Major birth defect

Suspect syndrome especially if >2 minor anomalies

https://commons.wikimedia.org/wiki/File:InfantWithNoonan2.jpg

Optimising data quality

Health staff training on newborn surface examination

- Always do thorough surface examination
- In liveborn and stillborn (SB) babies (prevalence of BDs is higher in SBs). Major BDs are sometimes identifiable, even in macerated stillbirths (MSB's).
- Complete tick-chart in MCR

FIRST EXAMINATION OF NEONATE (includes examination of stillborn babies)

Baby allowed to be placed skin to skin ☐ Time _____

General	Well	Sick			Comment
Appearance	Well nourished	Obese	Wasted	Dysmorphic	
Behaviour	Responsive	Lethargic	Irritable	Jittery	
Cry	Normal	Hoarse	High-pitched	Absent	
Colour	Pink	Blue	Plethoric	Pale	
Skin	Intact	Jaundice	Rash / Purpura	Bruising	
Temperature	36-37°C	Hypothermic	Hyperthemic		
Odour	Normal	Offensive			
Head shape	Normal	Asymmetrical	Caput	Haematoma	
Fontanelles	Normal	Bulging	Large		
Sutures	Mobile	Overriding	Widened	Fused	
Face	Symmetrical	Asymmetrical	Abnormal		
Eyes	Normal	Infected	Small / Large	Slanting	
Ears	Normal	Abnormal	Low position		
Nose	Patent	Blocked			
Mouth	Normal	Smooth philtrum	Cleft lip		
Palate	Intact	Cleft soft	Cleft hard		
Tongue	Normal	Lip-tie, tongue tie	Large	Protruding	
Chin	Normal	Small			
Neck	Normal	Swellings	Webbed		
Apex beat	120-160/min	Tachycardia	Bradycardia		
Chest - nipples	Normal	Accessory			
Chest – clavicles	Intact	Swelling	Crepitus		
Chest movement	Symmetrical	Asymmetrical	Shallow		
Chest indrawing	Absent	Costal	Sternal		
Respiratory rate	40 – 60 pm	Fast	Slow		
Breath sounds	Quiet	Grunting	Noisy		
Arms	Normal	Not moving	Fracture L/R		
Palmar creases	Normal	Single			
Fingers	Normal	Polydactyly	Syndactyly		
Abdomen	Normal	Distended			
Umbilicus	Normal	Moist	Flare	Bleeding	
Hips	Normal	Dislocated	Dislocatable		
Legs	Normal	Not moving			
Toes	Normal	Polydactyly	Syndactyly		
Feet position	Normal	Position Deformity	Clubbed		
Back	Normal	Meningocoele	Dimple / Hair tuft	Scoliosis	
Anus	Patent	Imperforate			
Femoral pulses	Present	Absent			
Genitalia: Male	Testes down	Undescended L/R	Hydrocoele	Inguinal hernia	
Genitalia: Female	Normal	Ambiguous			
Muscle tone	Normal	Hypotonic	Hypertonic		
Moro reflex	Present & equal	Asymmetrical	Weak	Absent	
Grasp reflex	Present	Weak	Absent		
Suck reflex	Present	Weak	Absent		
Urine	Passed	Not passed			
Meconium	Passed	Not passed			
Assessment:					
Examined by:				**Date and time:**	
Checked by:				**Date and time:**	

* **If any birth defects noted, please complete the birth defects notification form.**

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Optimising data quality

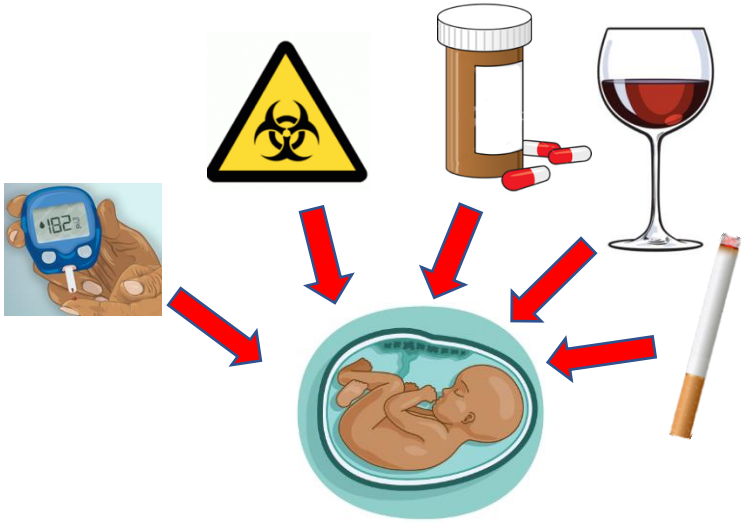
Health staff training on good exposure history

- Assists with identifying risk factors

- Take a good medical history of maternal conditions, medical exposures (medicines, vaccines, OTC and traditional remedies) and other risk factors (e.g. drugs, alcohol)

- Refer and follow up diagnosis and record

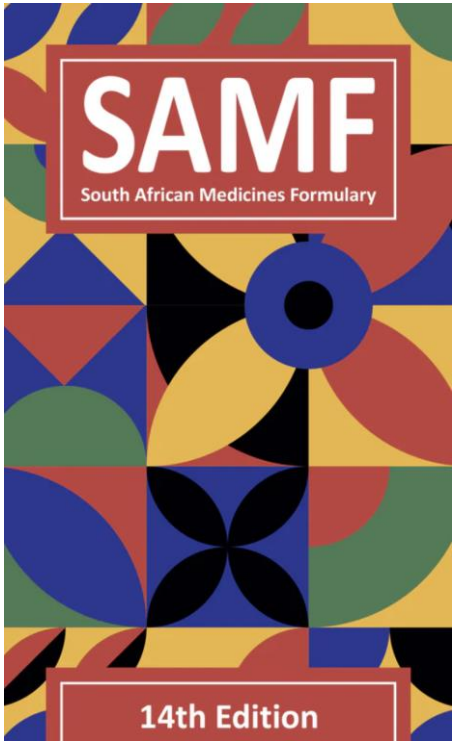
- Record diagnostic tests
  eg. Chromosomal tests, xrays



Resources for BD detection

Information on health products that may increase the risk of BDs:

SAMF



Medicines Information Centre

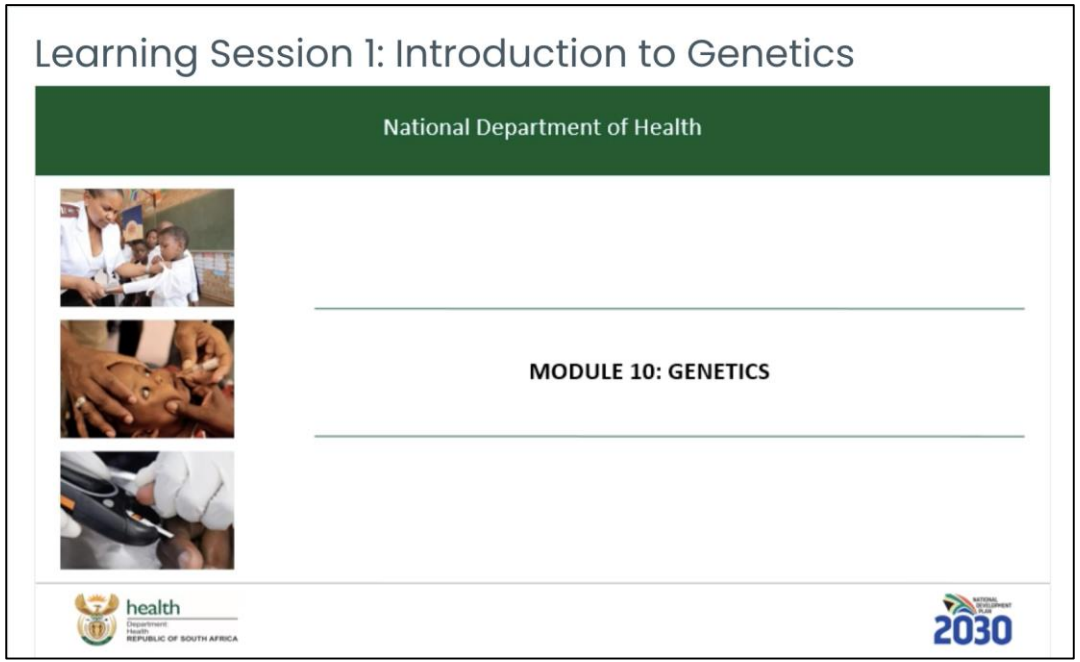
Resources for BD detection

Ultrasound assessment:

In-utero identification of major birth defects
Optimal if done at 20-23 weeks gestation, but can be detected earlier/later.

Resources for BD detection

Module 10 of Knowledge Hub: Introduction to Genetics



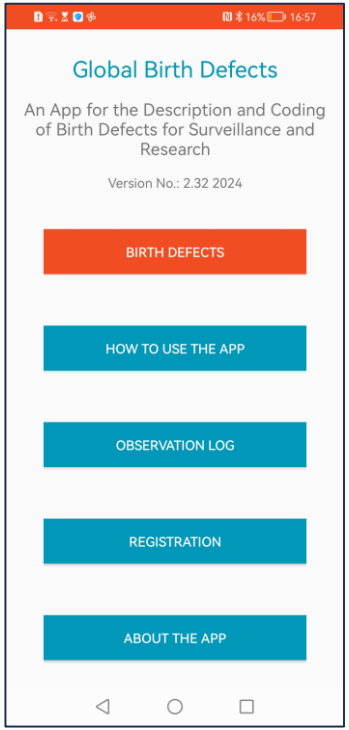
Resources for BD detection

- Training on surface examination of new/still-born, birth defect detection and management

www.ubomibuhle.org.za

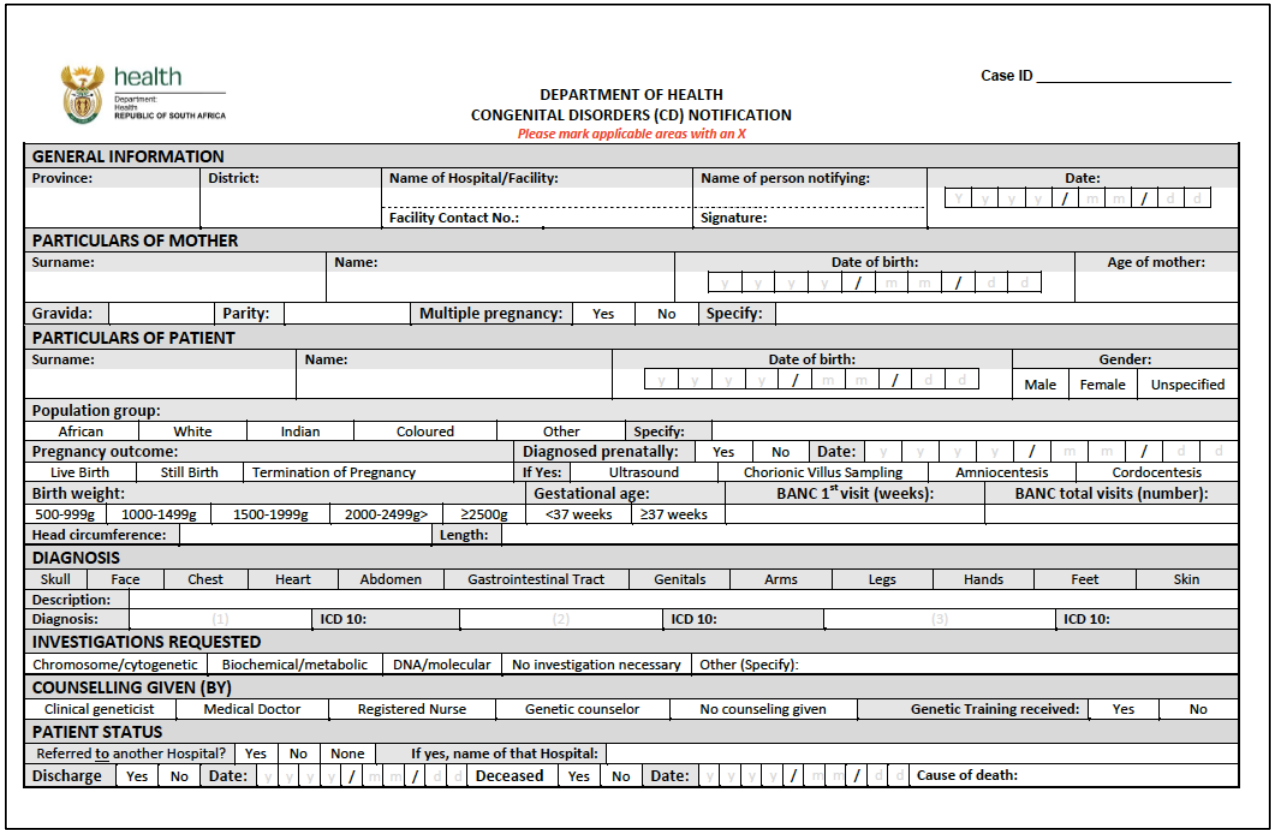
Resources for BD detection

- Global Birth Defects App – basic version
- Training module: www.ubomibuhle.org.za
- Download app: https://globalbirthdefects.tghn.org/



Notification of major BDs to NDOH

- Major BDs are a notifiable condition
- Notification form available in MCR
- Notify, even if only suspected
- Notify in liveborn *and stillborn* babies



health
Department: Health
REPUBLIC OF SOUTH AFRICA

Case ID _____

DEPARTMENT OF HEALTH
CONGENITAL DISORDERS (CD) NOTIFICATION
Please mark applicable areas with an X

GENERAL INFORMATION

Province:	District:	Name of Hospital/Facility:	Name of person notifying:	Date:
		Facility Contact No.:	Signature:	y y y y / m m / d d

PARTICULARS OF MOTHER

Surname:	Name:	Date of birth:	Age of mother:
		y y y y / m m / d d	

Gravida:	Parity:	Multiple pregnancy:	Yes	No	Specify:

PARTICULARS OF PATIENT

Surname:	Name:	Date of birth:	Gender:		
		y y y y / m m / d d	Male	Female	Unspecified

Population group:					
African	White	Indian	Coloured	Other	Specify:

Pregnancy outcome:			Diagnosed prenatally:	Yes	No	Date: y y y y / m m / d d
Live Birth	Still Birth	Termination of Pregnancy	If Yes:	Ultrasound	Chorionic Villus Sampling	Amniocentesis	Cordocentesis

Birth weight:				Gestational age:		BANC 1st visit (weeks):	BANC total visits (number):
500-999g	1000-1499g	1500-1999g	2000-2499g>	≥2500g	<37 weeks	≥37 weeks	

Head circumference:	Length:

DIAGNOSIS

Skull	Face	Chest	Heart	Abdomen	Gastrointestinal Tract	Genitals	Arms	Legs	Hands	Feet	Skin

Description:						
Diagnosis:	(1)	ICD 10:	(2)	ICD 10:	(3)	ICD 10:

INVESTIGATIONS REQUESTED

Chromosome/cytogenetic	Biochemical/metabolic	DNA/molecular	No investigation necessary	Other (Specify):

COUNSELLING GIVEN (BY)

Clinical geneticist	Medical Doctor	Registered Nurse	Genetic counselor	No counseling given	Genetic Training received:	Yes	No

PATIENT STATUS

Referred to another Hospital?	Yes	No	None	If yes, name of that Hospital:	
Discharge	Yes	No	Date: y y y y / m m / d d	Deceased	Yes	No	Date: y y y y / m m / d d	Cause of death:
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In closing...please remember…

YOU play a very important role in:

- Identifying mothers at high risk for having babies with BDs and contributing to prevention
- identifying and notifying babies with birth defects
- Referring babies with BDs for specialist care
- Supporting families that are affected by babies born with BDs

Thank you!