

FACT SHEET ON BIRTH DEFECTS

What are birth defects?

Any abnormalities of body structure or function present at birth. Some birth defects have genetic (hereditary) causes, whereas in others the causes are non-genetic. There are many birth defects, ranging from minor abnormalities (e.g. Red-green colour blindness) to serious, sometimes fatal disorders. Some are visible at birth (e.g. Down syndrome, cleft-lip, club foot); others are not immediately obvious and only manifest themselves later on in life (e.g. diabetes, porphyria). Whereas some birth defects are conspicuous (e.g. dwarfism), others are hidden (e.g. Congenital heart defects). Some are rare, while others occur relatively frequently.

What are examples of birth defects?

Mental retardation	blindness	muscular dystrophy
Cleft lip	Down syndrome	diabetes mellitus
Spina bifida (open spine)	albinism	porphyria
Club-foot	haemophilia	cystic fibrosis
Dwarfism	Tay-Sachs disease	Thalassaemia
Hypercholesterolaemia	colour blindness	deafness

How often do birth defects occur?

In South Africa it is estimated that 1 in every 40 babies has one or another birth defect and 1 in 10 will develop an inherited disorder during their lifetime.

What are the causes of birth defects?

The causes of many birth defects are still unknown. Where the causes are known, birth defects can be classified into three broad categories, viz. Genetic, environmental, and multi-factorial.

- Genetic disorders are caused by genetic or hereditary factors, i.e. they can recur in the same family and can be passed from one generation to the next.
- Some disorders are caused by external or environmental factors, such as viral infections, medicines, and alcohol that harm the unborn baby; they are not hereditary.
- Multi-factorial disorders arise from the interaction between genetic factors and environmental influences; they can recur in families.

Can birth defects be cured?

At present birth defects cannot be cured. However, in some of these disorders much can be done to alleviate or prevent certain disabilities that could manifest themselves.

Can birth defects be prevented?

Yes, an increasing number of birth defects can be prevented:

- By taking 0.4 mg folic acid before falling pregnant and during pregnancy;
- By avoiding harmful substances during pregnancy, e.g. alcohol;
- By diagnosing them in the unborn baby, e.g. by means of amniocentesis; or
- By diagnosing them and starting treatment, if available, as early as possible after birth.

What is genetic counselling?

Information about the prevention and treatment of birth defects can be obtained by way of genetic counselling. This involves providing information about the nature and implications of a specific birth defect; giving psychosocial support; and referring the family to appropriate facilities. The aim of genetic counselling is to provide the information needed for rational decision-making.

Who should seek genetic counselling?

It is advisable to contact a genetic counsellor

- If you or your spouse has a birth defect;
- If you have a child with a birth defect;
- If you have a family history of some genetic disorder;
- If you are married to wish to marry a close relative (consanguinity);
- If you have had a series of miscarriages or a stillbirth for which no medical reason could be found;
- If you are in your late thirties or older and are planning a pregnancy or are pregnant;
- If you have been exposed during pregnancy that could have a harmful effect on your unborn baby (e.g. alcohol, irradiation, infections)

Where can genetic counselling be obtained?

Genetic counselling clinics are found in the major centres in South Africa, at e.g. certain large hospitals, universities with a Human Genetics department; provincial offices of the Department of Health.

Here are some telephone numbers for Genetic Services of the Department of Health:

<u>Western Cape:</u>	(021) 483-2686	<u>Northern Cape:</u>	(053) 830-0707
<u>Eastern Cape:</u>	(040) 609-3577	<u>Free State:</u>	(051) 403-3856/9
<u>KZN:</u>	(031) 332-2425	<u>Gauteng:</u>	(011) 355-3311/41
<u>Mpumalanga:</u>	(013) 752-8085 x 2117	<u>North West:</u>	(018) 387-5231/4
<u>Northern Province:</u>	(015) 290-9000 x 9127	<u>Dept of Health:</u>	(012) 312-0000