

# Congenital disorders

## Walking with parents

Parents whose baby has recently been diagnosed with a congenital disorder may feel overwhelmed, and speaking to a well-informed mom or dad who has already walked a similar path can be hugely helpful, says Dr Helen Malherbe, Chairperson of Genetic Alliance South Africa

### What are congenital disorders?

Congenital disorders (CDs), also known as birth defects, are defined by the World Health Organization as abnormalities in structure or function that are present from birth, including metabolic disorders.

Some CDs are obvious at birth, such as club feet or spina bifida while others, such as haemophilia, are present at birth but only become obvious and diagnosed as the child grows. Other CDs only manifest later in life, such as Huntington's disease. CDs may be caused by genetic (pre-conception), environmental (post-conception) or unknown factors.

In South Africa, one in every 15 live births (6.8% of births) is affected by a CD.

When a baby is born with a congenital disorder (CD) it is important that his caregivers receive the right psychosocial support. In addition to genetic counselling, families facing a CD should be referred to a support group of families with children or family members who have been diagnosed with the same condition.

These carer- or parent-led groups can help by connecting families with others in the same situation. Such groups not only provide information on CDs, but help to prevent isolation, offering unique support and relief that cannot be provided by the medical community.

Support comes in many forms, and connections can be made online, over the telephone, and through in-person, local support groups. However, finding the right point of contact can be a challenge.

### Genetic Alliance South Africa

Genetic Alliance South Africa (GA-SA) is a non-profit, membership organisation that unites patient support groups, healthcare professionals and others relevant to the care and prevention of CDs.

GA-SA's vision is to improve the lives of those affected by CDs and a key aim is to provide a network of support for individuals, families and caregivers affected by CDs. A list of support groups is available on their [website](#). For conditions where there is no listed support group, GA-SA helps connect patients with other affected families where possible. They also encourage families to start support groups where there are none existing and offer advice on how to do this successfully.

GA-SA's other aims include:

- Facilitating **advocacy** activities to promote accessible, effective and relevant medical genetic services for the care, prevention and treatment of CDs
- **Education** of medical health professionals through training for improved diagnosis and treatment of CDs; the Medical Genetics Education Programme (MGEP) is a post-graduate distance learning programme that equips nurses and other healthcare professionals with genetics knowledge and skills; MGEP uses the *Congenital Disorders* course book from Bettercare.
- Raising the general public's **awareness** on CDs.
- Facilitating **research** related to the cause, prevention, treatment and monitoring, and surveillance of CDs in South Africa



For more information about GA-SA:

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The *Congenital Disorders* course book is an updated version of the Birth Defects book from the Perinatal Education Programme. Special attention is still given to modes of inheritance, medical genetic counselling, and congenital disorders caused by chromosomal or single gene defects, teratogens or multifactorial inheritance, but there is an additional chapter on the role of support groups. As the book will be used in the Medical Genetic Education Programme, practical sessions are included. The book is designed for group learning so that professional healthcare workers can improve their knowledge and understanding of both physical and functional disorders which are present from birth but may only manifest later.



The book can be viewed and bought on the [Bettercare website](#)