Working to improve the lives of people with Down syndrome and other intellectual disabilities

WHAT IS DOWN SYNDROME?

Down syndrome is a genetic disorder that is characterised by typical features. It is caused by an abnormality in the genetic material and it affects all races and all economic groups equally.

WHAT IS THE CAUSE OF DOWN SYNDROME?

Down syndrome is caused by an excess of genetic material. The genetic material is present in every human body cell and is arranged in tiny structures called chromosomes. They are arranged in pairs and humans have 46 chromosomes or 23 pairs of chromosomes.

People with Down syndrome have an extra chromosome 21. Down syndrome is therefore also called trisomy 21. Trisomy 21 refers to the presence of three copies of chromosome 21.

THE TYPICAL FEATURES OF DOWN SYNDROME

Babies with Down syndrome look different and they can therefore usually be identified at birth.

The most common features include: -

FACE

- eyes that slant upwards (up slanting palperbral fissures)
- folds on the inside of the eyes (epicanthal folds)
- small nose with a broad, flat nose bridge
- small mouth, making the tongue appear large
- small low set ears

HANDS AND FEET

- single line on the palm of the hand
- broad hands with short fingers
- inclining pinkie (clinodactyly)
- gap between the big toe and second toe (sandle gap)

OTHER

- low muscle tone (hypotonia)
- unusual looseness of the joints
- loose skin folds at the back of the neck
- heart defects occur in about 50% of cases
- eye defects occur in about 60% of cases
- hearing defects may occur and can affect speech and language
- thyroid deficiency and leukaemia, are more common than in people without Down syndrome

INTELLECT

- developmental delay
- the number of common features present in an individual however is not an indication of the level of developmental delay or the potential of the person

TO WHOM CAN THIS HAPPEN?

Down syndrome can occur in any family. It is not caused by food or medication or any other event. No one is to blame or should feel guilty.

However, the risk to have a baby with Down syndrome has been shown to increase with the age of the mother.

For a woman who is 20 years of age, the risk is about 1 in 1700, while for a woman of 40 years of age the risk is about 1 in 100.

Although the risk of having a child with Down syndrome is less for young mothers than for older women, babies with Down syndrome are also born to young mothers.

CAN DOWN SYNDROME BE DETECTED?

Down syndrome can be detected in the unborn baby.

Different tests can be performed to advise the parents of the status of the baby. Such tests include a maternal blood screening test, ultrasound, amniocentesis, chorionic villus sampling and cordocentesis.

These tests are performed at different stages of the pregnancy and there are different complications associated with each test.

It is therefore essential that any test should **only** be performed *after genetic counselling* has been obtained, when all the facts, risks and consequences have been fully discussed.

IS THERE A CURE?

There is currently no cure of Down syndrome.

However, there is much that can be done to help a baby with the condition.

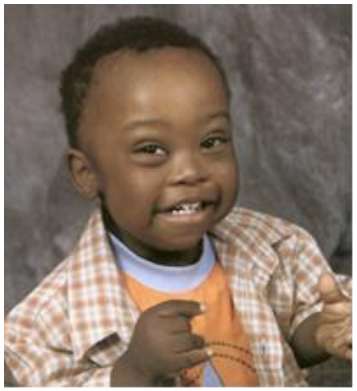
Early referral for the detection of complications (heart, hearing and eye defects) is encouraged.

Early intervention and stimulation programmes have been developed to encourage the child to reach his/her full potential.

It is very important to realise that children with Down syndrome are born with many qualities and abilities.

It is up to the parents and caregivers to see that these are developed to the fullest.

People with Down syndrome and other intellectual disabilities, benefit from and prosper, in a loving caring family environment.



WHERE TO GET MORE INFORMATION

For further advice and support you can contact: -

The Down Syndrome Association Gauteng

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