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Ministry of Health

Down Syndrome

Definition:

Down syndrome is a congenital non-pathological condition resulting from an extra chromosome and causing delays in mental and physical development. It varies in severity and leads to mental disability and developmental delay. There is no evidence that Down syndrome is caused by environmental factors or any specific practices before or during pregnancy.

Other names:

21 Trisomy - Mongolism - Mongolia

Types:

- 21 Trisomy: It makes up about 95 percent of cases. In this type, one has three copies of chromosome 21, instead of two.
- Mosaicism: One of the rarest types. It occurs with of an extra copy of chromosome 21 as a result of abnormal cell division after fertilization.
- Translocation: In this type there are the usual two copies of chromosome 21, but part of it is attached to another chromosome.

Cause:

Human cells typically contain 23 pairs of chromosomes inherited from both parents. Down syndrome occurs when an individual has a complete or partial extra copy of the chromosome 21, resulting in varying degrees of mental and physical disability. The cause of the full or partial extra chromosome remains unknown.

Risk factors:

Any pregnancy has little risk of delivering a child with Down's syndrome, but the risk increases as the mother ages.

Symptoms:

There are some distinguishing traits, although some do not have them. The most common are:

- Short stature and neck.

- Flattened face and occiput.
- Small head, ears and mouth.
- Upward slanting eyes with smaller palpebral fissure.
- Weak muscles.
- Short fingers and small hand and feet.

When to see a doctor:

Children are usually diagnosed before or at birth when there are any questions about the pregnancy or the child's development.

Complications:

People with Down syndrome are more likely to have certain health problems, including:

- Heart diseases (such as: congenital heart diseases).
- Hearing and vision problems.
- Thyroid problems (such as: hypothyroidism).
- Recurrent infections (such as: pneumonia).
- Sleep apnea.
- Obesity.
- Spinal problems.
- Mental illnesses.

Diagnosis:

Before birth:

- Screening Tests: Ultrasound and blood tests. Most screening tests measure amounts of different substances in the mother's blood.
- Diagnostic Tests: Sometimes a child is diagnosed with Down syndrome during pregnancy when screening tests indicates the likelihood of having the syndrome. Other tests can be carried out, including:
- Taking a small sample of the placenta, usually in the first trimester between the 11^{th} to 14^{th} weeks of pregnancy.
- Testing a sample of the amniotic fluid usually between the 15^{th} to 20^{th} weeks of pregnancy.

Screening during pregnancy and diagnostic tests are routinely performed for women of all ages.

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At birth:
Down syndrome is usually detected at birth by the presence of some physical traits in newborns. Chromosome analysis or "karyotyping" is carried out to confirm the diagnosis using a blood sample.

Treatment:
Although there is no cure for Down syndrome, early intervention in infants and children can make a difference in improving their quality of life because of the difference seen among patients and the dependency of treatment on individual needs to develop patient's potentials, as each stage of life may require different services.

Prevention:
There is no way to prevent Down syndrome, but when there are risk factors, a person may need to consult a genetic counselor before pregnancy.

Frequently Asked Questions:

- **Is Down syndrome a hereditary disease?**

Down syndrome is not a hereditary disease and is mainly caused by an abnormal cell division in the egg or the sperm or during the embryonic development.

Misconceptions:

- **People with Down syndrome are unable to learn.**
  Fact: People with this condition are able to learn when motivated.

Clinical Health Education Department
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