

## Down syndrome, Down's syndrome, or trisomy 21

### Definition:

**Down Syndrome** or trisomy 21 is a chromosomal disorder. It is a condition in which extra genetic material causes delays in the way a child develops, both mentally and physically. It affects about 1 in every 650 babies.

### Causes

The chromosome abnormality that causes **Down Syndrome** is trisomy 21, a extra copy of chromosome number 21. It is due to a nondisjunction of chromosome 21 during the original cell division, resulting in an extra chromosome 21. These children have a total of 47 chromosomes instead of the usual 46.

### Signs:

Kids with **Down Syndrome** tend to share certain physical features such as:

- a single transverse palmar crease
- an upward slant to the eyes,
- upslanting palpebral fissures,
- a flat facial profile,
- a rounded, brachycephalic skull, a broad, flattened bridge of the nose, small ears, and a large or protruding tongue,
- Smaller stature,
- Low muscle tone (called hypotonia)

Children with **Down Syndrome** may face health complications beyond the usual childhood illnesses. Problems such as:

- A higher sensory threshold (higher pain tolerance)
- Vision impairment (strabismus, nearsightedness, farsightedness)
- metabolic imbalance (reduce level of vitamins)
- thyroid dysfunctions
- immunologic concerns (increased chance of developing infections)
- congenital malformations of the heart
- seizure disorders
- greater chance of developing leukemia
- faster aging process (hearing and vision loss, Alzheimer's sometimes referred to dementia or senility).

### Diagnosis

Diagnosis is usually suspected at birth, when the characteristic physical signs of **Down Syndrome** are noted. Once this suspicion has been raised, genetic testing (chromosome analysis) can be undertaken in order to verify the presence of the disorder.

### Treatment

Treatment is directed at addressing the individual concerns of a particular patient. For example, to treat cardiomyopathy, epilepsy, diabetes, orthopedic disorders...

Early stimulation and special education are recommended as well as speech therapy, psychomotricity and occupational therapy

### Expectations (prognosis):

The prognosis in **Down Syndrome** is quite variable, depending on the types of complications.

**Life expectancy has greatly increased due to progress in medicine and surgery.**