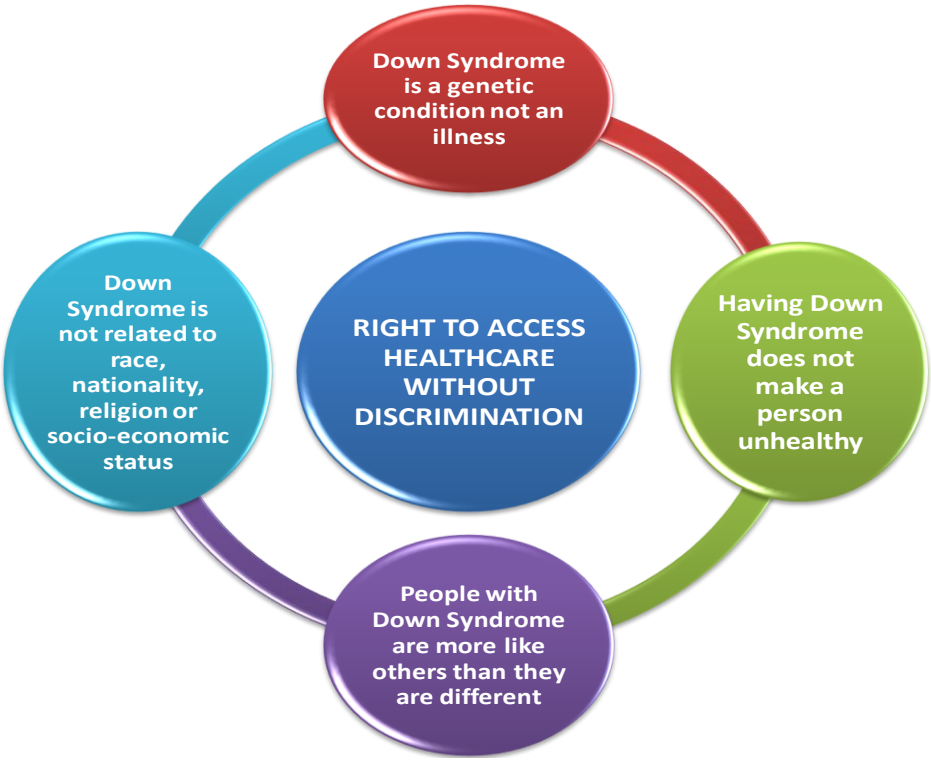
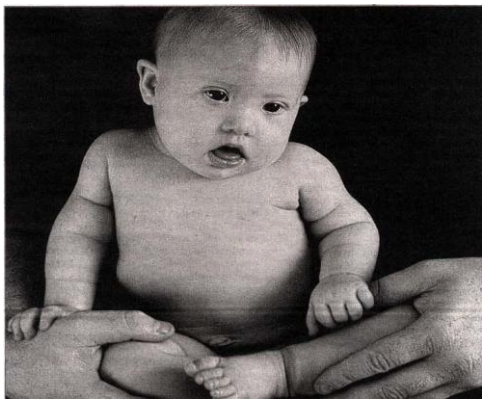
WORLD DOWN SYNDROME DAY

21 March 2015





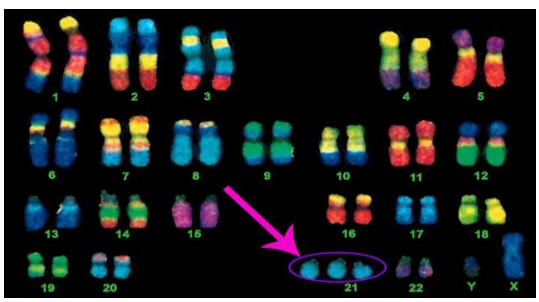
"HEALTH AND WELLBEING:

ACCESS AND EQUALITY FOR ALL"

WHAT TO KNOW ABOUT DOWN SYNDROME?

1. What is Down Syndrome?

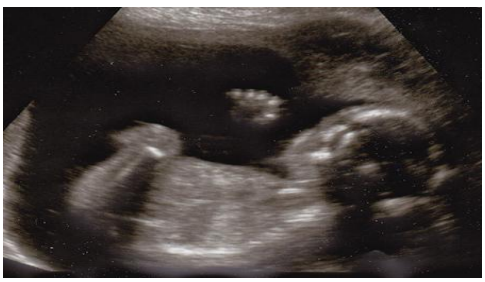
Down syndrome is a genetic condition that causes delays in physical and intellectual development. It occurs in one in every 691 live births. Individuals with Down syndrome have 47 chromosomes instead of the usual 46. It is the most frequently occurring chromosomal disorder.



2. What causes Down Syndrome?

Down syndrome is usually caused by an error in cell division called nondisjunction, which leaves a sperm or egg cell with an extra copy of chromosome 21 at conception. The error is not related to anything the mother did during pregnancy.

The probability that an egg will contain an extra copy of chromosome 21 increases significantly with advancing maternal age. Therefore, woman over 35 are more likely to give birth to a baby with Down Syndrome.



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3. Down Syndrome diagnosis

Down syndrome can be diagnosed before birth (during pregnancy) or shortly after birth.

Prenatal Diagnosis for Down Syndrome:

❑ **Screening Tests:** Done between 14 and 20 weeks of gestation.

- "The Triple Screen" is a combination of three tests that measure quantities of various hormones and proteins in the blood.
- Ultrasound Scan can show some physical traits that are helpful in calculating the risk of Down syndrome.

❑ **Diagnostic Tests:**

- **Amniocentesis** - a process in which amniotic fluid is sampled using a hollow needle inserted in the uterus. It is performed between 12 and 20 weeks gestation.
- **Chorionic Villus Sampling** (CVS) is a process in which a sample of the chorionic villi cells are taken from the placenta through the cervix or the umbilicus, guided by ultrasound. It is conducted between 9 and 13 weeks.
- **Percutaneous Umbilical Blood Sampling** (PUBS) is a process in which a sample of the fetus blood is taken directly from the umbilical cord. It is performed at 18 weeks gestation.

Postnatal Diagnosis for Down Syndrome:

The diagnosis of Down syndrome is usually made soon after birth because of some subtle differences in the baby's appearance. The diagnosis is based on the presence of an extra number 21 chromosome on a **karyotype** (visual display of the chromosomes grouped by their size, number and shape). Chromosomes may be studied by examining blood or tissue cells.

4. Common characteristics of people with Down Syndrome

A. Facial Features:

- Almond shaped eyes with epicanthal folds
- White spots on the iris
- Flat nasal bridge with small nose
- Small mouth with protruding tongue
- Flat facial profile with small ear

B. Small stature and short neck

C. Hands and feet:

- Single crease in the palm of the hand
- Short stubby fingers
- Small hands with short fingers
- Single flexion furrow of the fifth finger
- Small feet
- Large space between the big and second toes

D. Hypotonia (low muscle tones and weak muscles)

- Delayed motor skills

E. Medical problems

- Congenital heart defects
- Vision and hearing problems
- Respiratory problems and infections
- Thyroid conditions

E. Cognitive problems

- Mild to moderate mental retardation
- Speech delay
- Developmental delays

It is important to remember that while children and adults with Down syndrome experience developmental delays, they also have many **talents and gifts** and should be given the **opportunity and encouragement** to develop them.

Early intervention services including **physical, speech and developmental therapies** can be very helpful. Most children can attend schools, some in regular classes and others in special education classes.

Many adults with Down syndrome are capable of **working in the community**, but some require a more structured environment.

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