SNS COLLEGE OF ALLIED HEALTH SCIENCES

SNS Kalvi Nagar, Coimbatore - 35

Affiliated to Dr MGR Medical University, Chennai

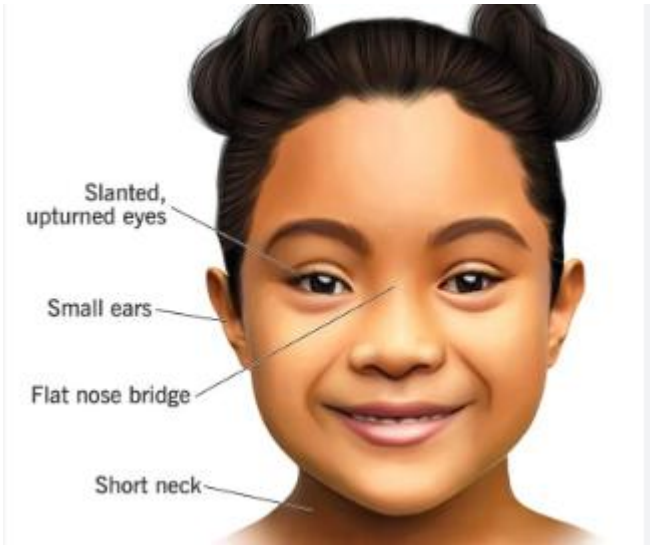
DEPARTMENT: ALLIED HEALTH SCIENCES

COURSE NAME: PAEDIATRICS

Topic:Downs syndrome

Definition

- A genetic chromosome 21 disorder causing developmental and intellectual delays.
- Down's syndrome is a genetic disorder caused when abnormal cell division results in extra genetic material from chromosome 21.(or)
- Down syndrome is a genetic condition where a person is born with an extra copy of chromosome 21. This means that they have a total of 47 chromosomes instead of 46. This can affect how their brain and body develop. People diagnosed with Down syndrome have happy and healthy lives with supportive care.

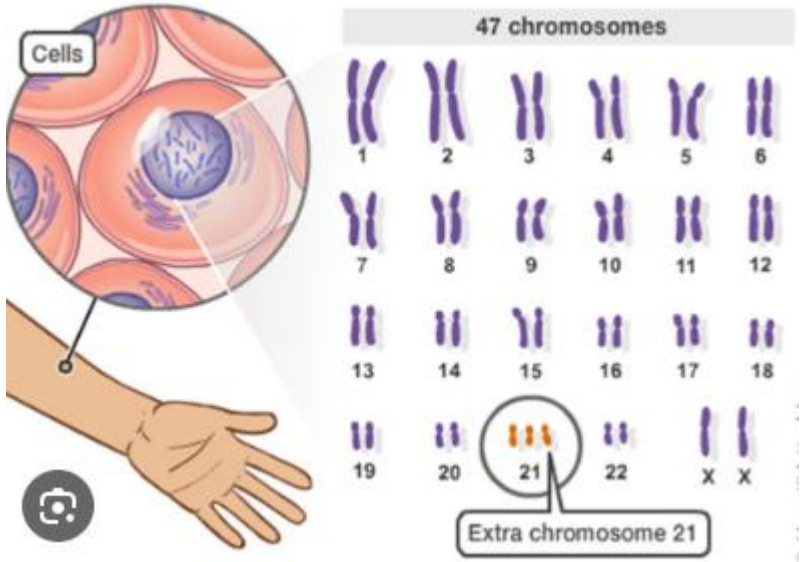


Etiology

- genetic condition
- autosomal dominant

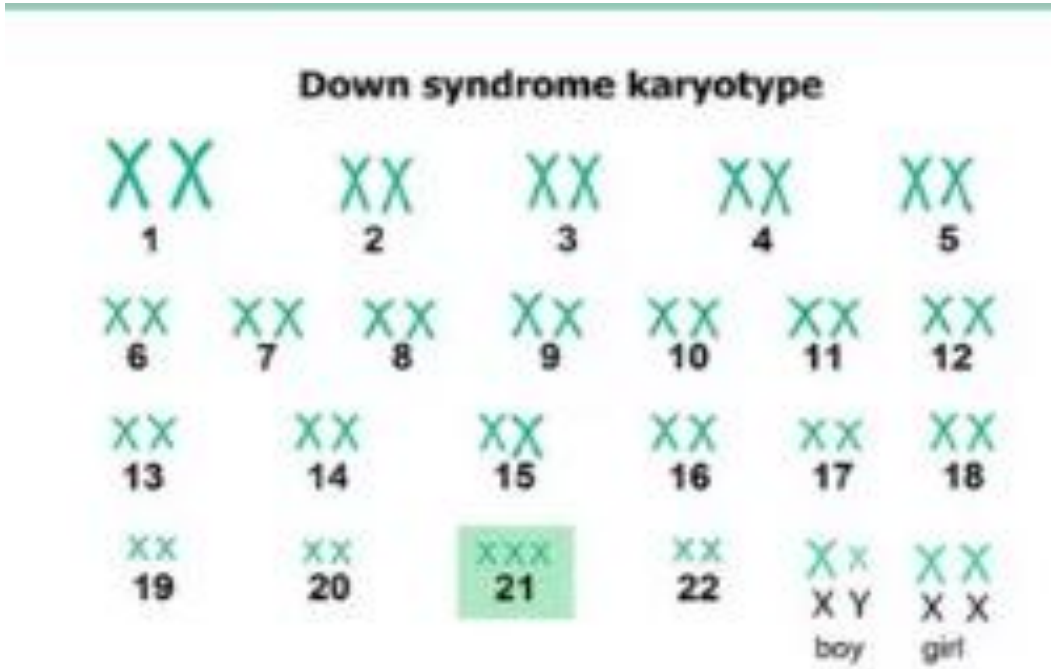
There are **three types of Down syndrome** with different causes, including:

- Trisomy 21-Trisomy 21 is the most common type of Down syndrome. The term “trisomy” means having an extra copy of a chromosome. Trisomy 21 occurs when a developing fetus has three copies of chromosome 21 in every cell instead of the typical two copies. This type makes up 95% of all cases of Down syndrome.
- Translocation-Translocation is a type of Down syndrome where there’s a partial or full amount of chromosome 21 attached to another chromosome. Unlike trisomy 21, translocation occurs when chromosome 21 isn’t separate, but it relocates to another numbered chromosome. This type of Down syndrome accounts for less than 4% of all cases.
- Mosaicism-Mosaic Down syndrome is the rarest type of Down syndrome and accounts for less than 1% of all cases. Mosaicism occurs when only some cells contain the usual 46 chromosomes and some cells contain 47. The extra chromosome in some cells is chromosome 21.



Pathophysiology

- About 95 percent of the time, Down syndrome is caused by trisomy 21 — the person has three copies of chromosome 21, instead of the usual two copies, in all cells. This is caused by abnormal cell division during the development of the sperm cell or the egg cell. Mosaic Down syndrome.



Diagnosis

- **The first trimester combined test**
- The first trimester combined test, which is done in two steps, includes:
- **Blood test.** This blood test measures the levels of pregnancy-associated plasma protein-A (PAPP-A) and the pregnancy hormone known as human chorionic gonadotropin (HCG). Abnormal levels of PAPP-A and HCG may indicate a problem with the baby.
- **Nuchal translucency test.** During this test, an ultrasound is used to measure a specific area on the back of your baby's neck. This is known as a nuchal translucency screening test. When abnormalities are present, more fluid than usual tends to collect in this neck tissue.
-

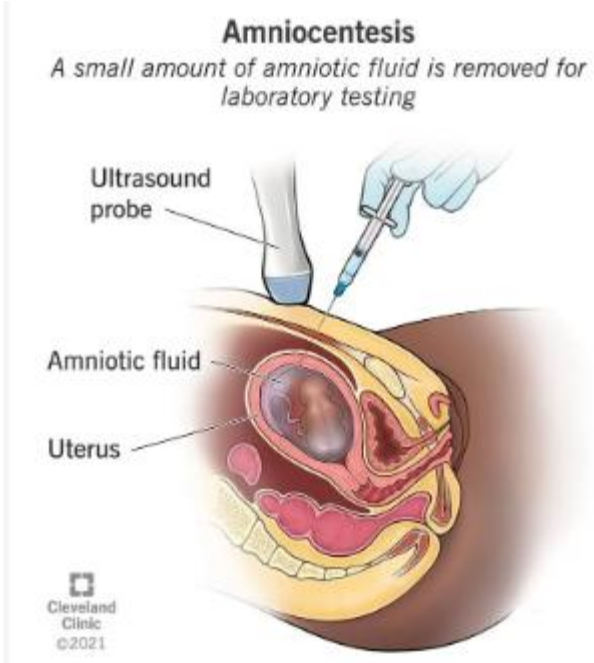
- **Integrated screening test**
- The integrated screening test is done in two parts during the first and second trimesters of pregnancy. The results are combined to estimate the risk that your baby has Down syndrome.
- **First trimester.** Part one includes a blood test to measure PAPP-A and an ultrasound to measure nuchal translucency.
- **Second trimester.** The quad screen measures your blood level of four pregnancy-associated substances: alpha fetoprotein, estriol, HCG and inhibin A.

Diagnosis

- Prenatal screening tests
- **Amniocentesis-**Amniocentesis is a prenatal testing procedure usually performed during the second or third trimester of pregnancy. It can diagnose certain chromosomal conditions (such as Down syndrome) or genetic conditions (such as cystic fibrosis).

- **Chorionic villus sampling** (CVS)-Chorionic villus sampling (CVS) is a type of prenatal testing. CVS testing takes a small sample of cells from the placenta, the organ that forms during pregnancy to deliver nourishment to the fetus.

- **Percutaneous umbilical blood sampling** (PUBS)



Diagnosis

Diagnostic tests for newborns

- After birth, the initial diagnosis of Down syndrome is often based on the baby's appearance. But the features associated with Down syndrome can be found in babies without Down syndrome, so your health care provider will likely order a test called a chromosomal karyotype to confirm diagnosis. Using a sample of blood, this test analyzes your child's chromosomes. If there's an extra chromosome 21 in all or some cells, the diagnosis is Down syndrome.

Signs & Symptoms

- Flattened face
- Small head
- Short neck
- Protruding tongue
- Upward slanting eye lids (palpebral fissures)
- Unusually shaped or small ears
- Poor muscle tone
- Broad, short hands with a single crease in the palm
- Relatively short fingers and small hands and feet
- Excessive flexibility
- Tiny white spots on the colored part (iris) of the eye called Brushfield's spots
- Short height

Risk factors

- **Advancing maternal age**
- **Being carriers of the genetic translocation for Down syndrome.**
- **Having had one child with Down syndrome**

- **Heart defects.**
- **Gastrointestinal (GI) defects.**
- **Immune disorders.**
- **Sleep apnea.**
- **Obesity.**
- **Spinal problems.**
- **Leukemia.**
- **Dementia.**
- **Other problems.** Down syndrome may also be associated with other health conditions, including endocrine problems, dental problems, seizures, ear infections, and hearing and vision problems.

Treatment

- There is no cure for Down syndrome, but early intervention and treatment can improve the overall quality of life for individuals with the condition. Treatment may include:
- Early childhood intervention programs, including speech and language therapy, occupational therapy, and physical therapy
- Special education services
- Medical treatment for any associated health conditions, such as heart defects or hearing loss
- Behavioral and mental health counseling for individuals and families