World Down Syndrome Day

World Down Syndrome Day (WDSD) is celebrated annually on the **21st of March** and is a global awareness day that has been officially observed by the United Nations since 2012.

The date for WDSD is set on the 21st day of the 3rd month as a representation of the extra copy of the triplication (trisomy) of the 21st chromosome that people with Down syndrome are born with.

On this day, people with Down syndrome and those who live and work with them throughout the world organize and participate in activities and events to raise public awareness and create a single global voice advocating for the rights, inclusion and well-being of people with Down syndrome.

Source: News18.com

Our Students of Department of Zoology, aimed to boost up the level of knowledge and of awareness about World Down Syndrome Day (WDSD) by submitting short articles to know more about Down Syndrome.

GENETICS BEHIND DOWN SYNDROME

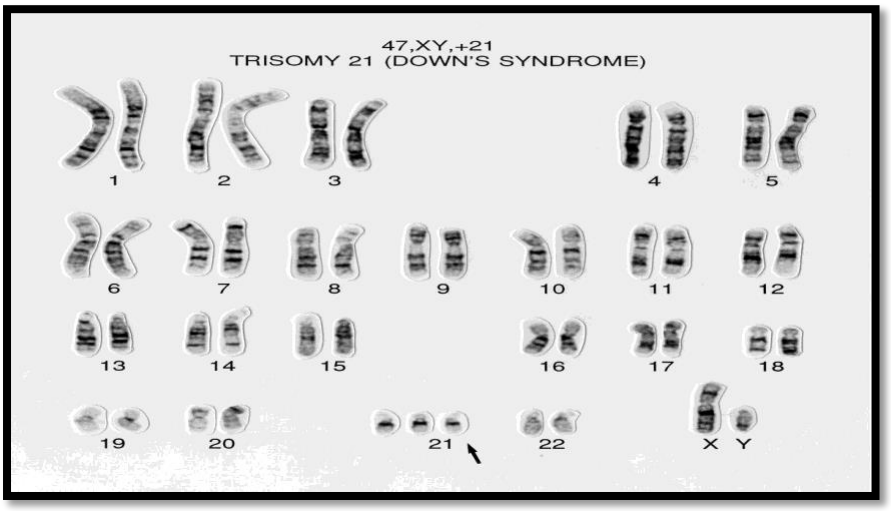
By Anushka Ghosh , 4th semester (2020-2023) , UG dept of zoology , Barasat Govt. College

Down syndrome is also referred as **TRISOMY 21** . It is a genetic disorder , that can't be cured. Specifically disorder of **chromosome 21** , due to development of extra genetic material. People with down syndrome , generally looks like **MONGOLIANS**, so also reffered as **MONGOLISM** or **MONGOLOID IDIOCY**, includes flattened face, Mongolian types of **Epicanthal fold** on eyelid, short heighted, small ears, small neck and many more abnormalities.

The followings are the causes of down syndrome :

TRISOMY 21:

The main reason behind down syndrome is the trisomy**(2n+1)** of chromosome 21, that is why , there is an extra chromosome 21 occures along with normal diploid **(2n)** chromosome. It means there are three copies of chromosome 21 instead of normal two. People with down syndrome having 47 chromosomes in their cells instead of 46. The **karyotype** of down syndrome is **46,XX+21** or **46,XY+21 .** About 95 percent of the time, the reason is trisomy .



Karyotype of down syndrome

To create the trisomy condition of chromosome 21, the **NON-DISJUNCTION** of chromosome 21 occurs during the development of normal gamet with normal chromosome. The **NON-**

DISJUNCTION occurs during **MEIOSIS I** or **MEIOSIS II** cell division , so , the one gamet contain two chromosome 21 and another gamet contain no chromosome 21.

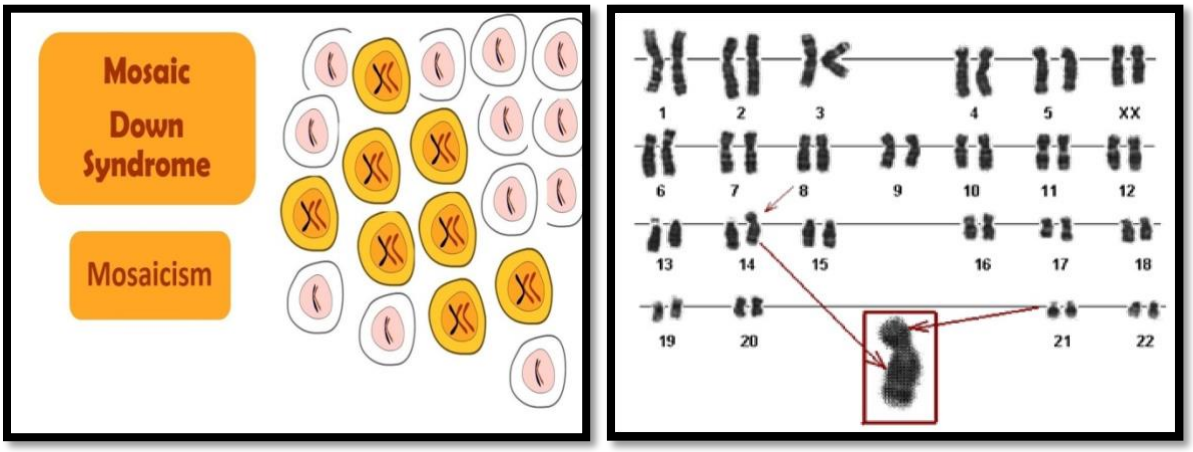
Now the gamet with abnormal and exceptional two chromosome 21 fertilizes with the gamet of normal chromosome 21, resulting three copies of chromosome 21, this is called as TRISOMY of chromosome 21 .The result is , the birth of baby with down syndrome.

ROBERTSONIAN TRANSLOCATION:

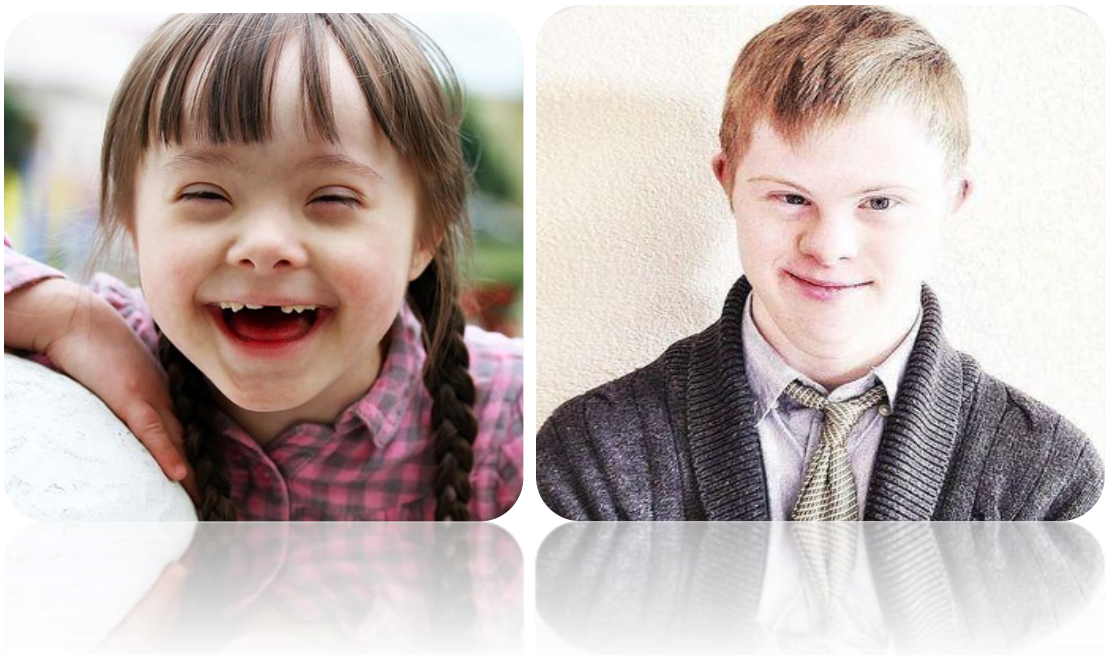
In this rare case, the chromosome 21 attaches with chromosome 14 through translocation. Now during the development of gamet , one 14-21 chromosome and one free chromosome 21 are unitedly included into single gamet. Due to the fertilization of this abnormal gamet with normal one , three copies of chromosome 21 will occur in that zygote and from that zygote, the down baby will born. This type of down syndrome often occurs in family , that is why it is known as **FAMILIAL DOWN SYNDROME.** About 4 percent of the cases are this type.

MOSAIC DOWN SYNDROME:

In this form of down syndrome , a person has only some cells with an extra copy of chromosome 21. This mosaic of normal and abnormal cells is caused by abnormal cell division after fertilization. Mosaicism is the rare and least common form of down syndrome and accounts for only about 1 percent of all cases of down syndrome.



Mosaic down syndrome and translocation down syndrome



Girl and boy with down syndrome

WEBLINK

https://en.wikipedia.org/wiki/Down_syndrome

IMAGE SOURCE

- cdc.gov
- en.wikipedia.org

WHAT IS DOWN SYNDROME ?

BASUSRI DANDAPAT, 4TH SEM (2020-2023) , DEPARTMENT OF ZOOLOGY, BARASAT GOVERNMENT COLLEGE.

All the living beings are are made of cells- basic functional and structural unit of life. Each cell contains several organelles among which the most important one is the NUCLEUS. We all know the nucleus contains the genetic material in the form of genes which are codes for replicating the cell and to carryout functions of a cell properly. These are responsible for our inherited traits and are arranged in structures called chromosomes. Human cell contains 23 pairs of chromosomes or 46 chromosomes.

Down syndrome is a consequence of genetic variation. The full or partial extra copy of chromosome 21 in an individual results in down syndrome. This extra amount of genetic material varies the developmental process and it is associated with certain traits.

DISCOVERY:

British physician, John Langdon Down in 1866 first described Down syndrome and called it “Mongolism”. Afterwards it came to known as Down syndrome after him. The term came to use during the early 1970s. In 1959 French Pediatrician/Geneticist Professor Jerome Lejeune discovered that individuals with Down syndrome have an extra chromosome. Very soon chromosome studies were developed to know about the proper diagnosis of this syndrome.

SYMPTOMS:

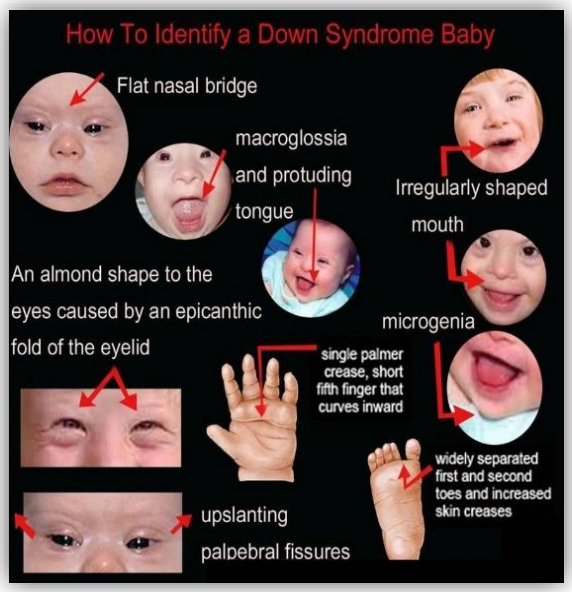
The symptoms include intellectual disability and characteristic facial profile. Early symptoms are:

- Loss of interest in being sociable, conversing or expressing thoughts and declined ability to pay attention.
- Sadness, fearfulness or anxiety, irritability, uncooperativeness or aggression.
- Restlessness or sleep disturbances. Unable to coordinate.
- Seizures occuring in adulthood.

CHARACTERISTIC TRAITS:

The characteristic traits of this syndrome are:

- Small sized skull
- Upward slant of eyes and epicanthic folds
- Small nose with the flat nasal bridge
- Mouth has a narrow short palate with small teeth and furrowed protruding tongue
- Single crease on the hand (simian crease) at birt
- Delayed development and behavioural problems. And Cognitive disability



Other complications like heart defects, vision problems, gut problems etc. are also associated.

The only prevention can be prenatal diagnosis. The families with members suffering from this syndrome must seek consultation from mental health professionals. The child or adult must be treated with care and love.

WEBLINKS:

https://i1.wp.com/sport4you.net/wp-content/uploads/2021/07/boy-in-green-shirt-with-downs-syndrome-1.jpg?w=675&ssl=1

https://www.ptckids.com/wp-content/uploads/2019/02/down_syndrome.jpg

DOWN SYNDROME: POPULATION & AGE

-MEGHNA DAS (UG SEM- IV, ZOO DEPT)

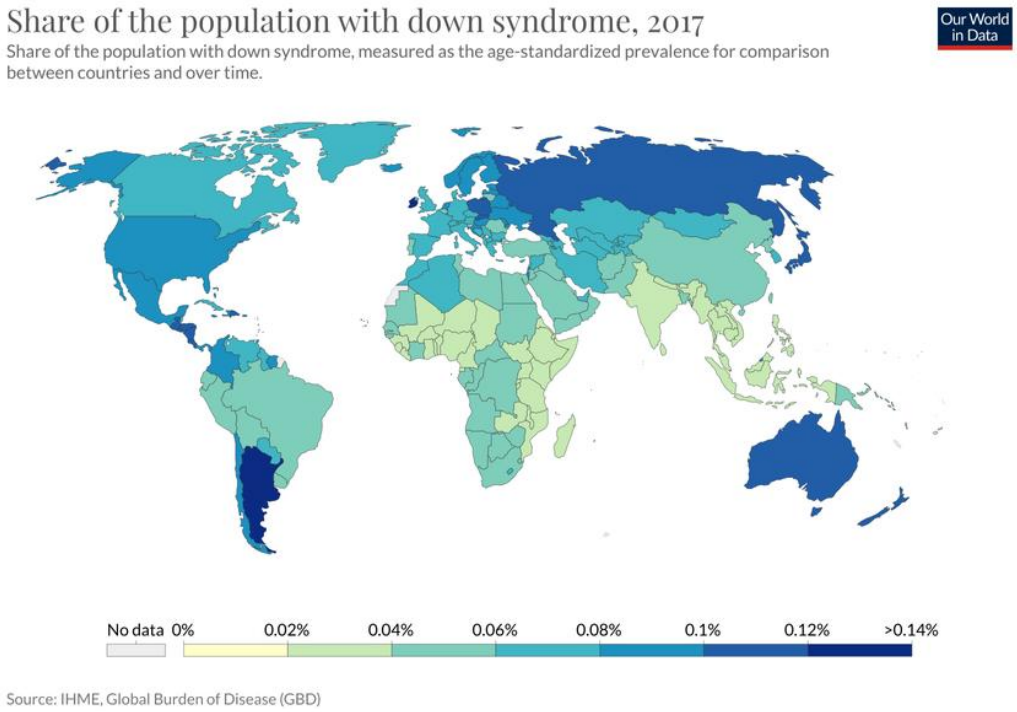
WORLD POPULATION FOR DOWN SYNDROME

Down syndrome is said to be occurred when there is an extra copy of 21st chromosome during pregnancy. This leads to some distinctive signs and symptoms such as recognizable facial features in addition to developmental & intellectual difficulties. Though Down syndrome is the most commonly occurring genetic disorder, the way the condition presents itself in each person will differ.

The estimated occurrence of Down syndrome is between 1 in 1,000 to 1 in 1,100 babies worldwide, according to the World Health Organization. Down syndrome is the most common chromosomal disorder in USA. About 6,000 babies are born with Down syndrome each year.

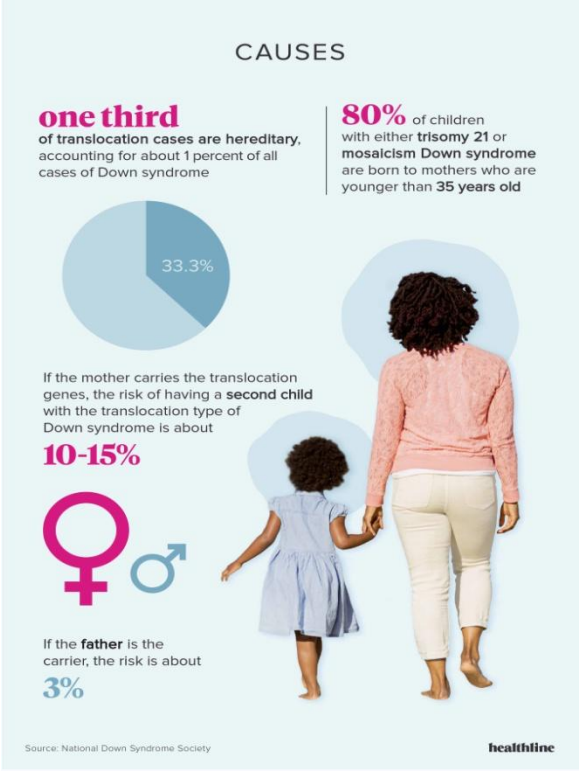
Between 1979 and 2003, the number of babies with Down syndrome has increased by 30%. Older mothers are more likely to have a baby affected by Down syndrome than younger mothers. In other words, the occurence of Down syndrome increases as the mother's age increases. For estimation of the occurence of Down syndrome, the number of pregnancies affected by Down syndrome is compared to the total number of live births.

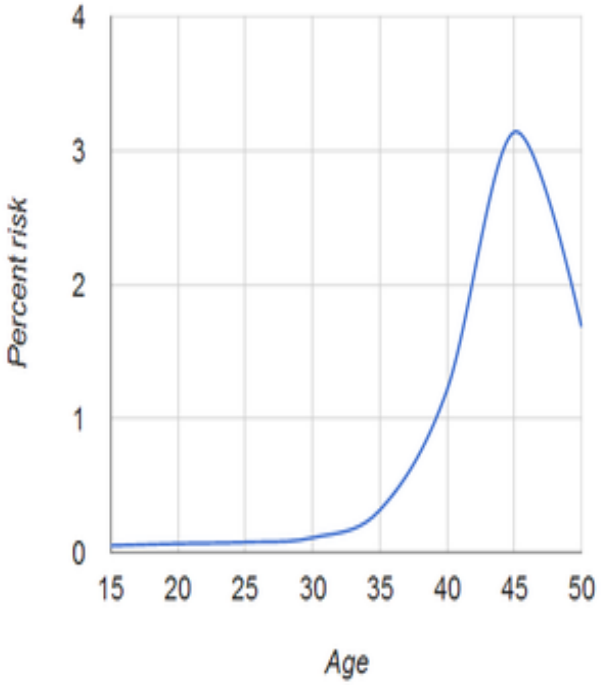
WEST BENGAL: in the Genetics Department of West Bengal there 85 cases which are diagnosed as Down syndrome. Ramakrishna Mission Seva Pratishthan, Kolkata, India has taken control of 30 individual. More than 90% of Down syndrome patients have flat faces, low life span, and abnormal eye problems. 56.5% patients of such conditions suffer from Congenital Heart Diseases and 41.2% of them suffer from jaundice at birth. These factors are significantly low by the help of Health Drive.



MATERNAL AGE FACTOR IN DOWN SYNDROME

Almost 80% of the newborns are born with trisomy 21 or mosaicism Down syndrome to the younger mothers below the age of 35. Younger mothers tend to have babies more frequently so the number of babies with Down syndrome increases subsequently. However mothers of age more than 35 have the possibility to have a baby with this condition is naturally more.





According to the National Down Syndrome Society, a 35 year old mom has a chance of having a child with Down syndrome is 1 in 350. This chance gradually increases to 1in 100 at the age of 40 an approximately 1 in 30 by the age of 45 and above. About one-third of total translocation cases are hereditary accounting for about 1% of all the cases of Down syndrome. Both parents can be carriers of the translocation genes without showing any symptoms of Down syndrome in their body. Mothers who have a child with Down syndrome have an increased chance of having another baby with same condition. When a woman has a child with Down syndrome, than the risk of having another child with Down syndrome is about 1 in 100 up to the age of 40.

The risk of having a second child with the translocation gene of own syndrome is about 10 to15 5 if the mother carries the genes. But is about 3% if the father carries the gene.

Reference:

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3. https://www.cdc.gov
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How to treat with Down syndrome patient?:

-Tista Das (B.Sc SEM 4)

Down syndrome is a genetic disorder causing developmental and intellectual delays. People with Down syndrome are in a great risk for a number of health problems. We can identify such patients with many symptoms like flattened face, short neck, almond shaped eyes that slant up, small hands and feet, tiny white spots on the Irish of the eye etc. Down syndrome cannot be cured. Early treatment programs like speech, physical, occupational, educational therapy and mental support is the essential ways that help them to lead a happy, productive life.

1. **Physical therapy** includes activities and exercises that help to build up skills, increase muscle strength. A physical therapist might help a child to establish an efficient walking pattern.
2. **Speech language therapy** can help children with Down syndrome improve their communication skills and use language effectively before they can speak. A speech therapist can help children to learn many other ways of communication such as sign languages and pictures etc.
3. **Occupational therapy** helps find ways to maintain daily tasks and conditions to match a person's need and ability. This type of therapy teaches self dependable skills such as eating, getting dressed, writing and using a computer. At the high school level, an occupational therapist could help teenagers to identify jobs, careers, or skills that match their personalities.
4. **Emotional and behavioral therapies** work to find useful responses to both pleasant and unpleasant behaviors.

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2. Source of image: pinterest.com

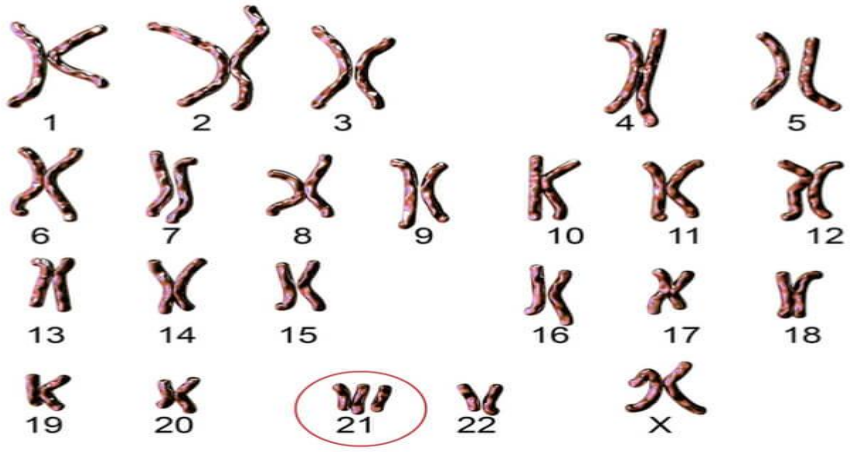
DOWN SYNDROME AWARENESS

Prsented By- ***Sahin Ali*** *[PG/ZOOLOGY/SEM-II/Roll-06] -* ***BARASAT GOVT. COLLEGE***

❖ What Is Down Syndrome?

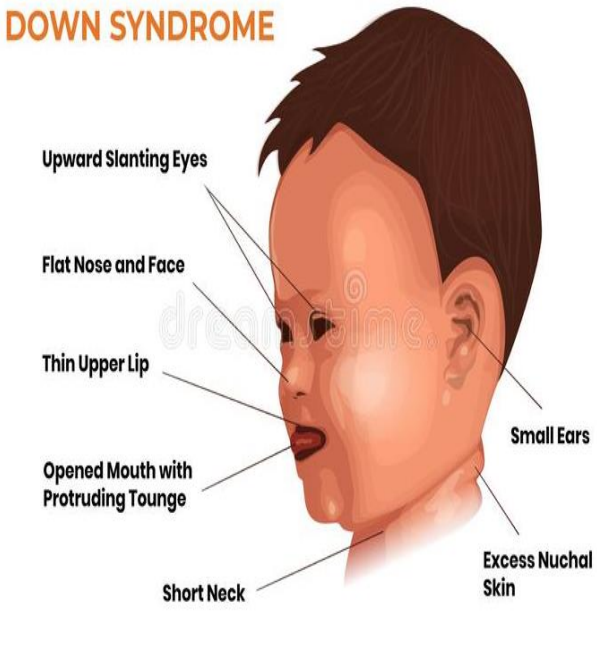
Down syndrome is a genetic disorder caused when abnormal cell division results in an extra full or partial copy of chromosome 21. This extra genetic material causes the developmental changes and physical features of Down syndrome.

A medical term for having an extra copy of a chromosome is 'trisomy.' Down syndrome is also referred to as **Trisomy 21**.



❖ Physical Features of Down Syndrome

- Flattened face
- Small head
- Short neck
- Protruding tongue
- Upward slanting eye lids (palpebral fissures)
- Unusually shaped or small ears
- 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
- Short height



❖ Causes and Risk Factors

Advancing maternal age. A woman's chances of giving birth to a child with Down syndrome increase with age because older eggs have a greater risk of improper chromosome division. A woman's risk of conceiving a child with Down syndrome increases after 35 years of age. However, most children with Down syndrome are born to women under age 35 because younger women have far more babies.

Being carriers of the genetic translocation for Down syndrome. Both men and women can pass the genetic translocation for Down syndrome on to their children.

Having had one child with Down syndrome. Parents who have one child with Down syndrome and parents who have a translocation themselves are at an increased risk of having another child with Down syndrome. A genetic counselor can help parents assess the risk of having a second child with Down syndrome.

❖ Diagnosis

There are two basic types of tests available to detect Down syndrome during pregnancy: screening tests and diagnostic tests.

Screening Tests- Screening tests often include a combination of a blood test, which measures the amount of various substances in the mother's blood and an ultrasound, which creates a picture of the baby. During an ultrasound, one of the things the technician looks at is the fluid behind the baby's neck. Extra fluid in this region could indicate a genetic problem. These screening tests can help determine the baby's risk of Down syndrome.

Diagnostic Tests- Diagnostic tests are usually performed after a positive screening test in order to confirm a Down syndrome diagnosis. Types of diagnostic tests include:

Chorionic villus sampling (CVS)—examines material from the placenta

Amniocentesis—examines the amniotic fluid (the fluid from the sac surrounding the baby)

Percutaneous umbilical blood sampling (PUBS)—examines blood from the umbilical cord

These tests look for changes in the chromosomes that would indicate a Down syndrome diagnosis.

REFERENCES

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- https://www.mayoclinic.org
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PICTURE SOURCE:-

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- https://www.google.com/url?sa=i&url=https%3A%2F%2Fwww.dreamstime.com%2Fillustration%2Ftrisomy.html&psig=AOvVaw0MYniWBRzkutQ5BB7SJb3j&ust=1648818827214000&source=images&cd=vfe&ved=0CAgQjRxqFwoTCNC8x_i28PYCFQAAAAAdAAAAABAJ

AWARNESS ON DOWN SYNDROME

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Down syndrome is one of the most common chromosome abnormalities. It caused due to trisomy of chromosome number 21.due to this extra copy of chromosome it changes the baby's appearance and their mental growth.

TYPES

There are 3 types of down syndrome

Trisomy 21: in this case each cell in the body has 3 separate copies of chromosome 21 instead of the usual 2 copies.

Translocation Down syndrome: This occurs when an extra part or a whole extra chromosome 21 is present, but it is attached or "trans-located" to a different chromosome rather than being a separate chromosome 21.

Mosaic Down syndrome: some of their cells have 3 copies of chromosome 21, but other cells have the typical two copies of chromosome 21.

RISK FACTORS

1.sometimes the advanced maternal age increases the chance of down syndrome of the baby. A woman's risk of conceiving a child with Down syndrome increases after 35 years of age. But according to statistics most children with Down syndrome are born to women under age 35 because younger women have far more babies.

2. Both father and mother can pass the genetic translocation for Down syndrome on to their children.

3.parents having one child with down syndrome have an increased risk of another child also with down syndrome.

Case ID	Age (years)	Gender	Birth order	Mother's age at birth	Diagnosis/ method	Level of ID
1	6	Male	First	20	Trisomy/lab confirmed	Moderate
2	8	Male	First	18	Clinical observation	Moderate
3	14	Female	Third	24	Clinical observation	Severe
4	3	Male	First	22	Trisomy/lab confirmed	Mild

ID: Intellectual disability, DS: Down syndrome

Table- Profile of children with DS

Ref-Down syndrome in tribal population in India: A field observation Ram Lakhan and Madhavaram Thomas Kishore J Neurosci Rural Pract. 2016 Jan-Mar; 7(1): 40–43. doi: 10.4103/0976-3147.172167

Though the increased maternal age is told to be a major risk factor but from the above survey we can see the age of the mother during the child birth with DS is not above 23 also.so there may be some more hidden reasons that we have to identify.

Picture – A child in India having DS

SCREENING OF DS DURING PREGNANCY

Screening should be done to avoid the unfortune birth of a abnormal child not only for DS but for various other syndromes and diseases so that if necessary within the given time (according to the law and medical condition) the fetus can be aborted.

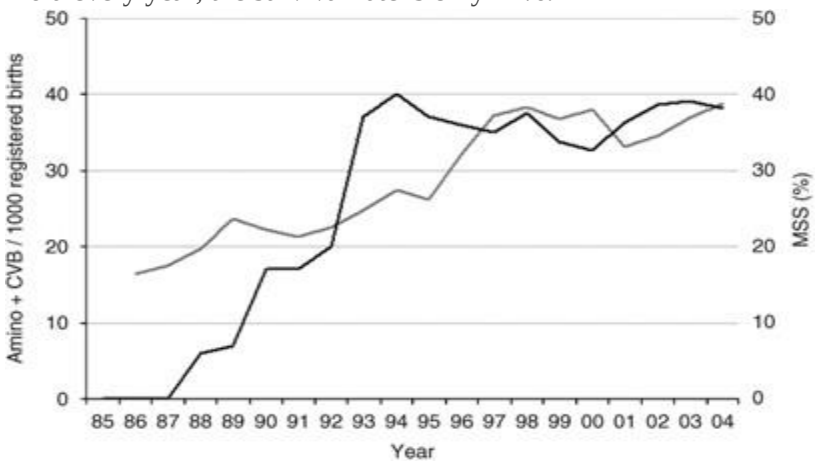
1.Quadrupel blood screening test (14-20 weeks)

2.20 weeks screening test

3. multiple marker serum screening test

STATUS OF INDIA REGARDING DS

India has a high number of people suffering with DS .Down Syndrome affects approximately 23,000-29,000 children born in India every year. In compare to other countries this condition is fatal in India due to negligence. People with DS also have several parallel serious health issues like congenital heart diseases, pulmonary hypertension etc. Out of the 23,000 to 29,000 kids born with Down Syndrome in India every year, the survival rate is only 44%.



20 years trend of prevalence and survival of DS (European journal of Human Genetics)

Ref-Twenty-year trends in prevalence and survival of Down syndrome Claire Irving, Anna Basu, Sam Richmond, John Burn & Christopher Wren *European Journal of Human Genetics* 02 July 2008

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