Turner syndrome

SUBJECT:- MEDICAL BIOLOGY

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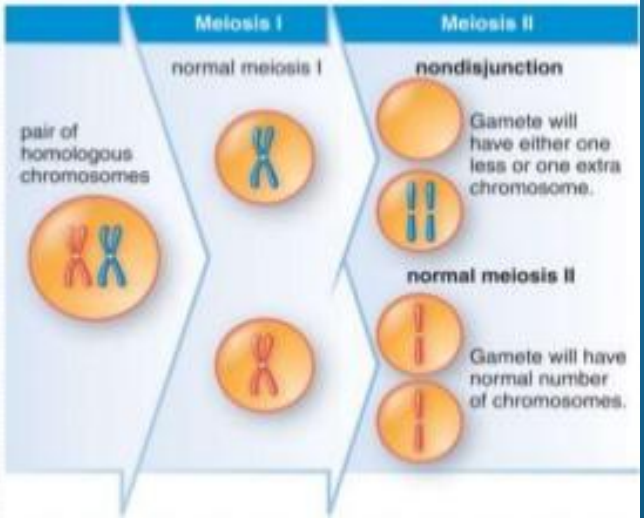
Turner Syndrome

Overview; What Is It?

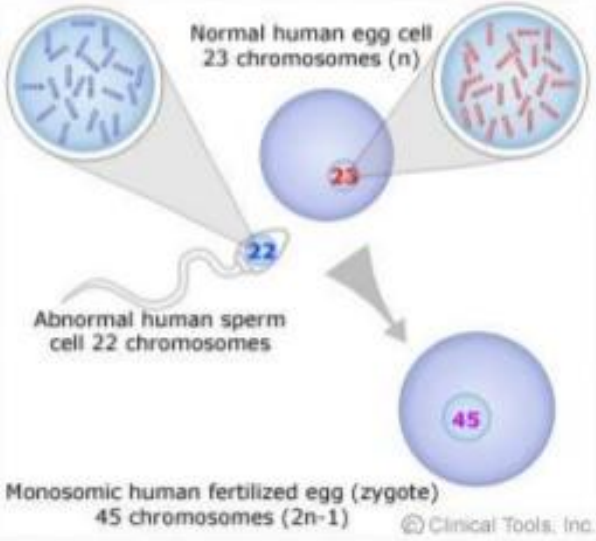
- Turner syndrome (TS) is a chromosomal condition that describes girls and women with common features that are caused by complete or partial absence of the second sex chromosome.[1][2][3] ...
- The missing genetic material affects development before and after birth.[7]
- TS occurs in approximately 1 of every 2000-2500 live female births and approximately 10% of all miscarriages.[1][2][3]

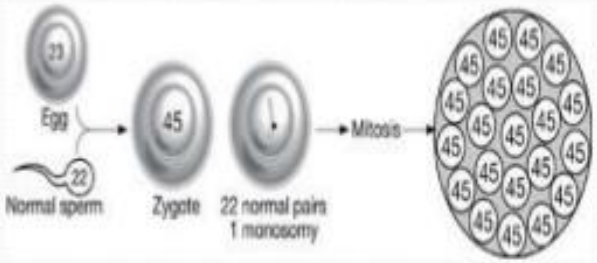
What causes Turner syndrome ?

- Normally, a girl inherits one X chromosome from her mother and one X chromosome from her father. But girls who have Turner syndrome are missing one of their X chromosomes.[1]
- Turner syndrome is typically caused by what is called **nondisjunction.**[1] [4]
- Nondisjunction happens when a pair of sex chromosomes fails to separate during the formation of a sperm or egg.[1]



- When sperm with no X chromosome unites with a normal egg to form an embryo, that embryo will have just one X chromosome (X rather than XX). [1] [2]
- As the embryo grows and the cells divide, the X chromosome will be missing from every cell of the baby's body. [1]



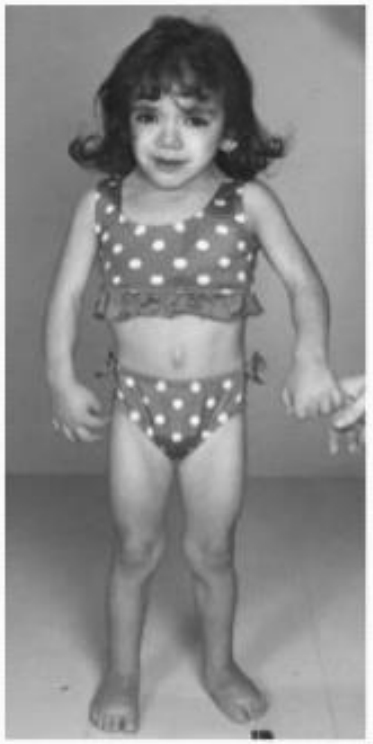


- Researchers have not determined which genes on the X chromosome are associated with most of the features of Turner syndrome. [2]
- They have, however, identified one gene called **SHOX**[1] that is important for bone development and growth. [1] [2]
- The loss of one copy of this gene likely causes short stature and skeletal abnormalities in women with Turner syndrome. [1] [2]

[1] SHOX: The official name of this gene is "short stature homeobox."
Read more about the SHOX gene and the X chromosome.

Other names for Turner syndrome [2]

- 45,X
- Monosomy X
- TS
- Turner's syndrome
- Ulrich-Turner syndrome

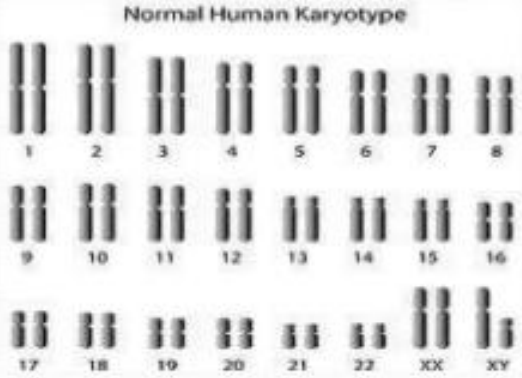


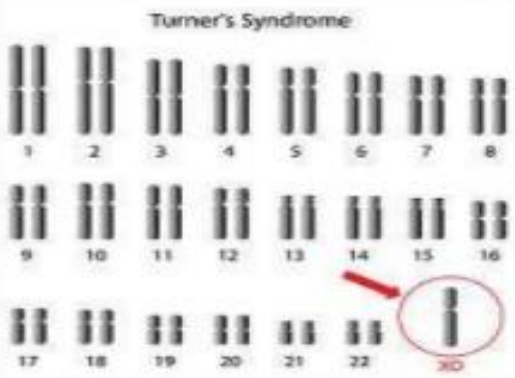
Types of Turner syndrome

1. X-Chromosome Monosomy
2. X-Chromosome Mosaicism
3. X-Chromosome Defects

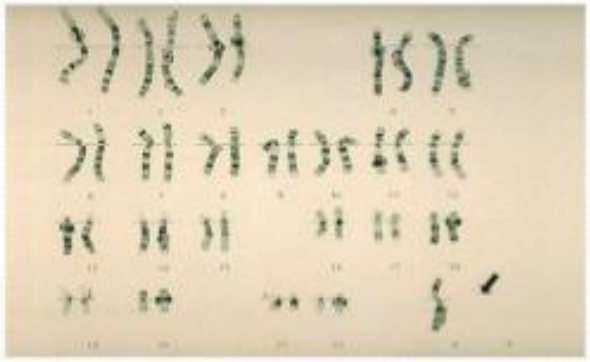
X-Chromosome Monosomy

- About 50 percent of individuals with Turner syndrome have Monosomy X, which means each cell in the individual's body has only one copy of the X chromosome instead of the usual two sex chromosomes. [2] [4]



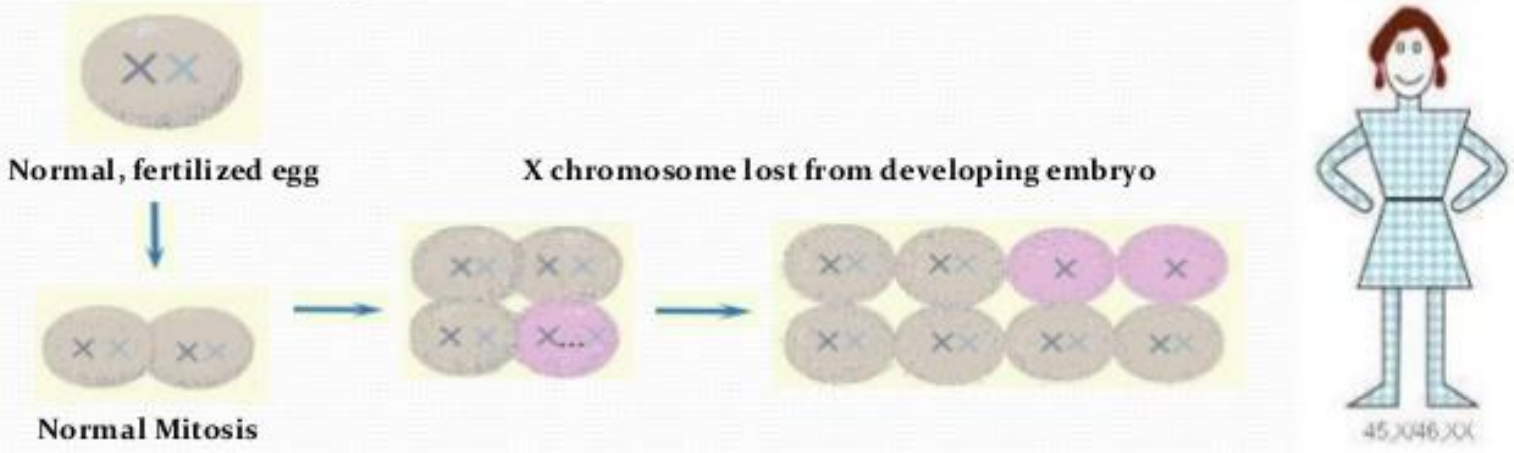


- During the process in which oocytes (eggs) or sperm are formed, one of the sex chromosomes is sometimes "lost".
- An embryo receiving only a Y-chromosome can not survive, but an embryo receiving only a X-chromosome may survive and develop as a female with Turner syndrome.
- [45XO] is known as classic TS.[5]



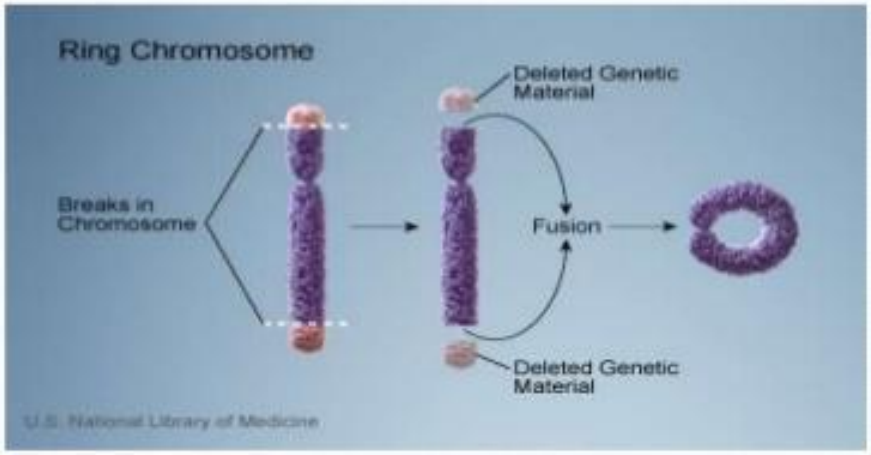
X-Chromosome Mosaicism

- About 30 percent of girls with the disorder are missing the X chromosome in just some of their cells. [1]
- This mixed chromosome pattern is known as Turner mosaic. [46XX/45XO]
- Girls with this pattern may have fewer symptoms because they still have some normal (XX) cells. [1] [4]



X-Chromosome Defects

- about 20 percent of Turner syndrome cases, both X chromosomes are present, but one is abnormal.
- It may be shaped like a ring or missing some genetic material. [1] [4]



Symptoms of Turner syndrome

- The term 'syndrome' is used to describe a collection or combination of symptoms which result from a single cause, it does not necessarily mean that all are present in the person who has a syndrome.[5]
- Girls with TS may have only a few or several of the features associated with TS, but **short stature** and **infertility** are *nearly* always present. [3] [6]
- Turner syndrome affects growth and sexual development.[1]
- The most common feature of Turner syndrome is: [3] [5][7]

short stature

- **The most common feature** of Turner syndrome is short stature, which becomes evident by about age 5.
- Girls who have Turner syndrome are shorter than average. They often have normal height for the first five years of life, but then have a slow growth rate.
- At puberty they do not have the usual growth spurt.

Non-functioning ovaries

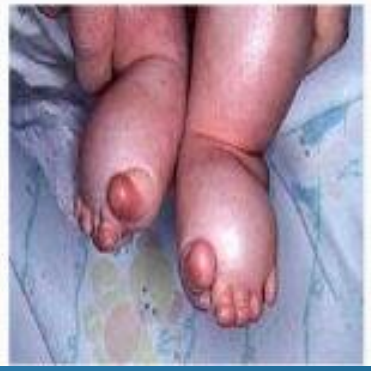
- Normally a girl's ovaries begin to produce sex hormones (estrogen and progesterone) at puberty. This does not happen in most girls who have Turner syndrome.
- However, most girls with Turner syndrome will not produce these sex hormones which means that:
- unlike other girls, they may not start their monthly periods naturally.
- they may not fully develop breasts.
- they may be infertile.

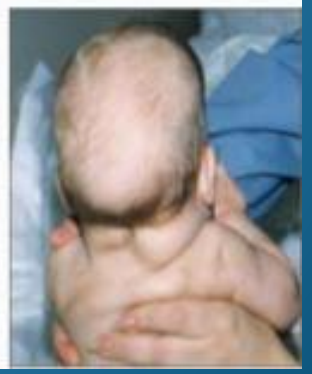
intelligence

- Most girls and women with Turner Syndrome are usually of normal intelligence with good verbal skills and reading skills .[1] [2]
- Some girls, however, have problems with math, memory skills and fine-finger movements. [2]

Other symptoms of Turner syndrome: [3]

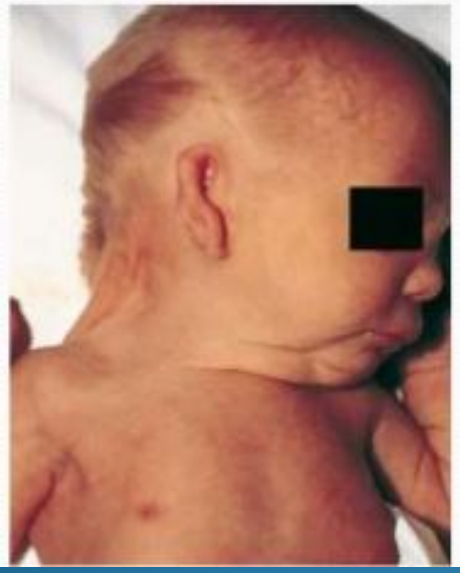
- About 30 percent of females with TS have extra folds of skin on the neck (webbed neck). [2]
- A low hairline at the back of the neck.
- A broad chest and widely spaced nipples.
- Puffiness or swelling (lymph edema) of the hands and feet.
- Small and narrow fingernails, toenails that turn up. [7]
- Skeletal abnormalities.
- Kidney problems.

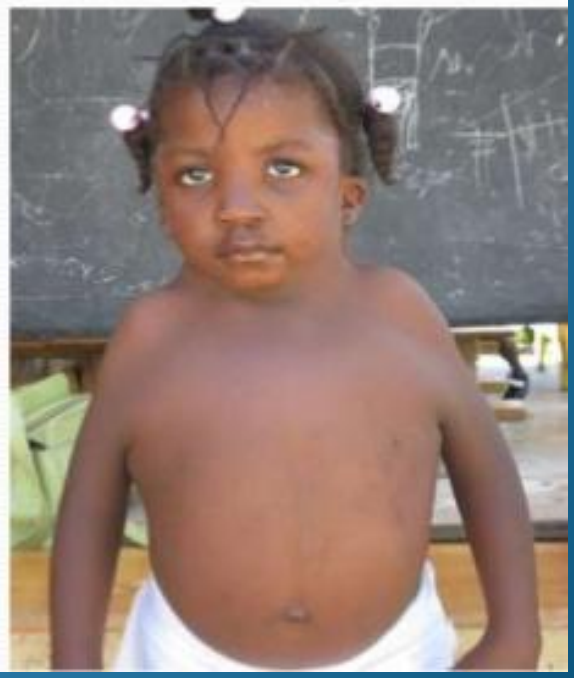




- A heart murmur, sometimes associated with narrowing of the aorta (blood vessel exiting the heart).
- A tendency to develop high blood pressure (so this should be checked regularly).
- Minor eye problems that are corrected by glasses.
- Scoliosis (deformity of the spine) occurs in 10 percent of adolescent girls who have TS.
- The thyroid gland becomes under-active in about 10 percent of women who have TS.

- Osteoporosis can develop because of a lack of estrogen.
- Arms that turn out slightly at the elbow.



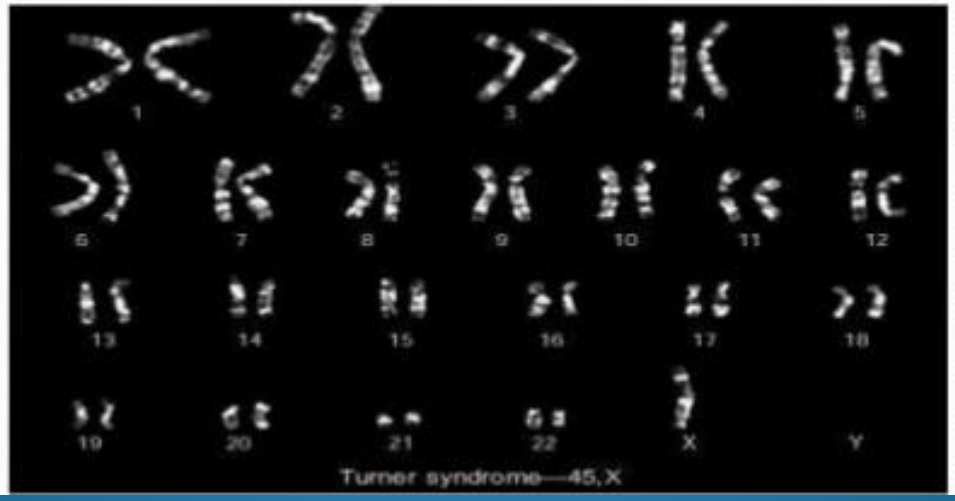


Diagnose of turner syndrome

- 1. before the baby is born: Turner syndrome may be diagnosed during pregnancy with chorionic villus sampling (CVS) or amniocentesis. Alternatively, an ultrasound (a machine that uses sound waves to look inside a mother's uterus) can identify the disorder by its physical symptoms.[1]

- 2. About half of the cases are diagnosed within the first few months of a girl's life by the characteristic symptoms (swelling of the hands and feet, heart defect).[1] [3]

- 2. Other patients are diagnosed in adolescence because they do not grow normally or go through puberty. [1] [3]
- 3. A blood sample to make a karyotype (a chromosome analysis), because the diagnosis can be confirmed.[1] [3]



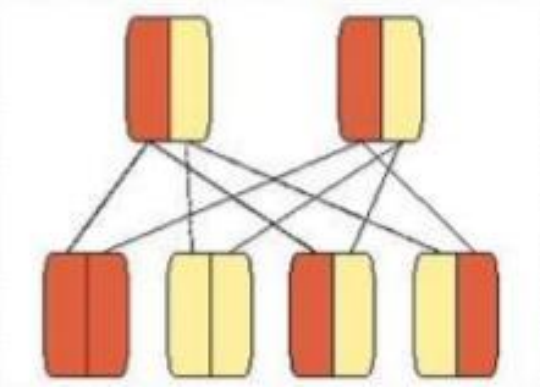
Treatment of Turner syndrome

- The best way to treat this disorder is:
  Hormone replacement therapy [1]
- Hormone replacement therapy is usually started at the time of normal puberty, around 12 years.[3]
- Teenagers are treated with growth hormone to help them reach a normal height. [1]
- They may also be given low doses of androgens (male hormones that females also produce in small quantities) to increase height and encourage hair and muscle growth. [1]
- Some patients may take the female hormone estrogen to promote sexual development. [1]

- Babies born with a heart murmur or narrowing of the aorta may need surgery to correct the problem.[3]
- Blood pressure should be checked routinely and, if necessary, treated with medication. [3]
- Almost all women are infertile, but pregnancy with donor embryos may be possible. [3]
- Having appropriate medical treatment and support allows a woman with Turner syndrome to lead a normal, healthy and happy life. [3]

Is Turner syndrome inherited?

- Most cases of Turner syndrome are not inherited.[2]
- While Turner syndrome is genetic in that it involves the loss or abnormal expression of X-chromosome genes, it is not usually hereditary.[4]
- An error in cell division called nondisjunction can result in reproductive cells with an abnormal number of chromosomes.[2]



- Turner syndrome affects all races, nationalities and regions of the world equally, and parents who have produced many unaffected children may still have a child with Turner syndrome.[4]
- There are no known toxins or environmental hazards that increase the chances of Turner syndrome.[4]

Interesting facts about Turner syndrome

- Turner Syndrome affects 60,000 females in the United States. This disorder is seen in 1 of every 2000 to 2500 baby girls, with about 800 new cases diagnosed each year. [1]
- In 75-80% of cases, the single X chromosome comes from the mother's egg; the father's sperm that fertilizes the egg is missing its sex chromosome. [1]
- Turner syndrome is named for Dr. Henry Turner, who in 1938 published a report describing the disorder. [1]

- The average height of an untreated woman with Turner syndrome is 4 feet 8 inches.[1]
- A female fetus (normally XX) that is missing one of its X chromosomes can survive, but a male fetus (normally XY) cannot. The X chromosome is a long DNA molecule with many genes that are needed for cells to function; it is essential for life. In contrast, the Y chromosome carries few genes and is not essential for life.[1]

QUESTIONS AND ANSWER

1. What is Turner Syndrome?
2. What causes Turner Syndrome?
3. What are the other names for Turner Syndrome?
4. How many types of Turner Syndrome are there?
5. What are the Symptoms of Turner Syndrome?
6. How we can Diagnose Turner Syndrome?
7. What are the Treatment of Turner Syndrome?
8. Is Turner Syndrome inherited?
9. What is x-chromosome Monosomy?
10. What is x-chromosome Mosaicism?
11. What is x-chromosome Defects?

THANK YOU MAM