مدينة الملك فهد الطبية
King Fahad Medical City

CHROMOSOMAL SYNDROMES

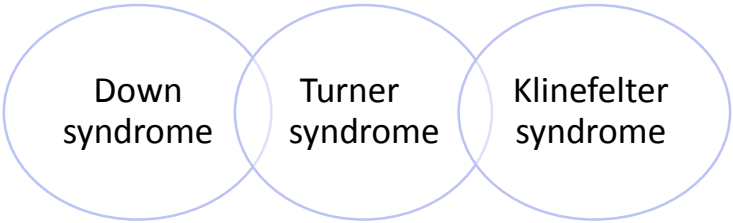
What is syndrome ?

Syndrome is a group of signs and symptoms that occur together and characterize a particular abnormality or condition due to presence of chromosomal abnormalities.

Types of chromosomal anomalies:

There are many types of chromosome anomalies that can be organized into two basic groups:

1. **Numerical such as:**



2. Structural such as:

Down syndrome

Down syndrome:

Also known as (trisomy 21), is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21.

Causes of trisomy 21:

1. 92%-94% trisomy 21 due to non-disjunction during fertilization.

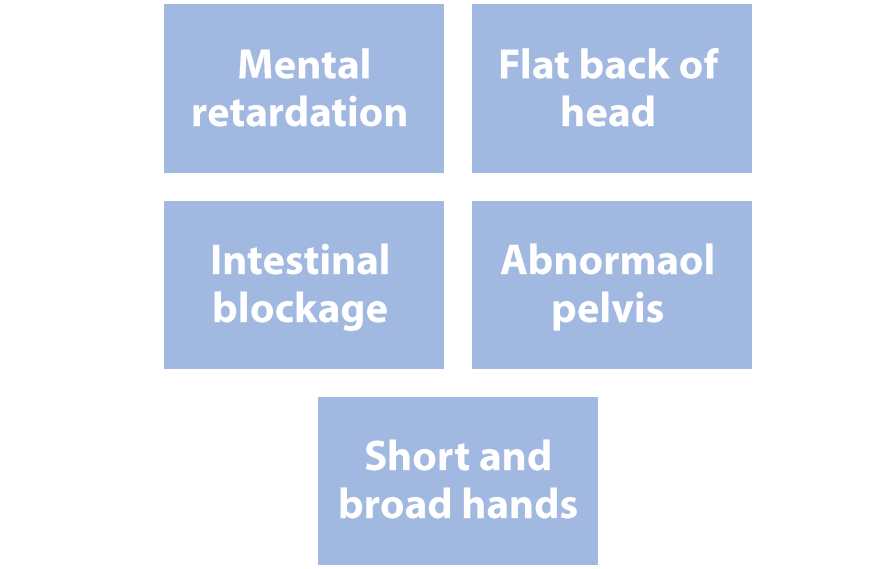
2. 3-4% translocation of chromosome 21 breaking and attaching to other chromosome during gametogenesis.
3. 2-4% mosaicism error in cell division after fertilization.

Identification of Down syndrome:

Down syndrome can be identified either by:

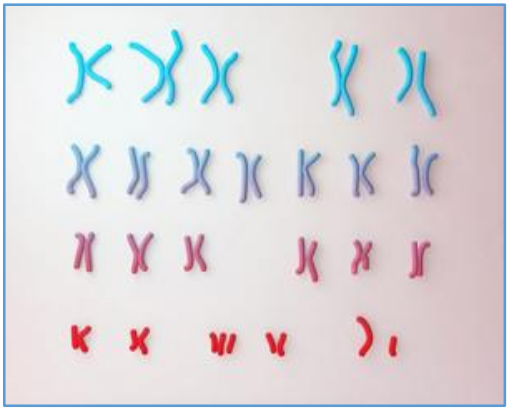
1. During pregnancy by ultrasound which initiate prenatal test by amniocentesis or chorionic villi sampling.

2. After birth by characteristic clinical features and chromosome karyotyping such as:



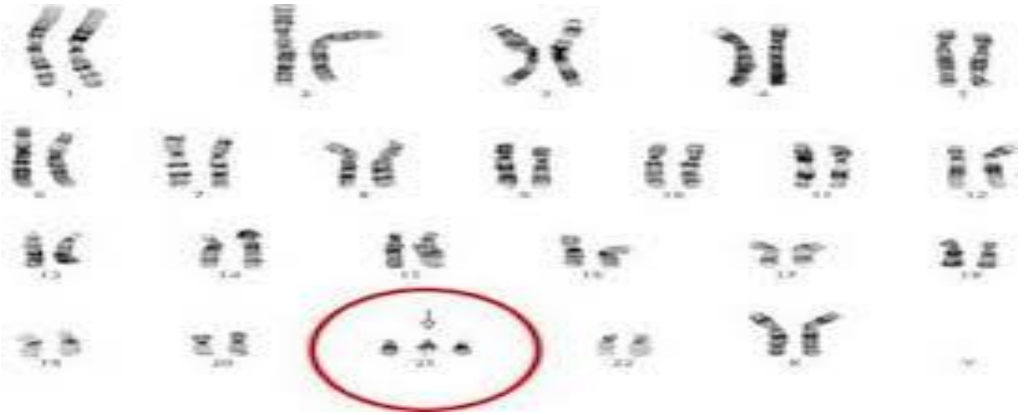
- **Down syndrome is one of the most common chromosome numerical abnormalities in human, It occurs in about one per 1000 babies born each year.**

1. before birth by prenatal test (amniocentesis)



Trisomy 21 (also known by the karyotype 47,XX,+21 for females and 47,XY,+21 for males)

2 .Confirmed by chromosome karyotyping



Turner syndrome (TS):

Also known as (45, X or 45,XO), is a condition in which a female is partly or

completely missing one of the X chromosomes.

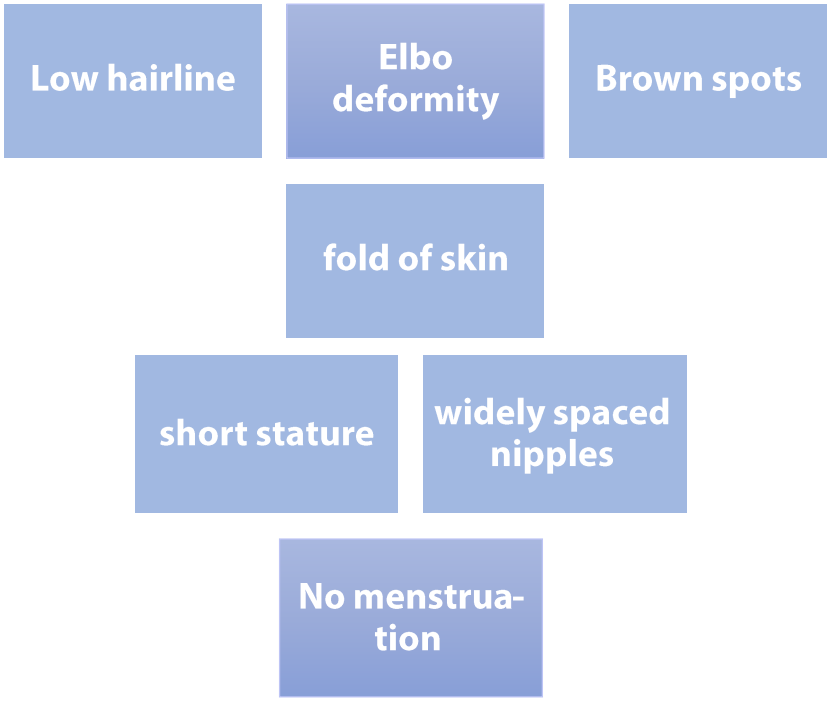
Causes of Turner syndrome:

1. 48% monosomy X due to error in sperm or ova non-disjunction before fertilization.
2. 18% in the form of iso-chromosome of the q arm of the X chromosome (mirror image of the q arm).
3. 27% is due to structural rearrangement of the X chromosome.
4. 7% of Turner's syndrome is in the form of mosaicism error in cell division after

fertilization: 45 X with one or more additional cell lineages.

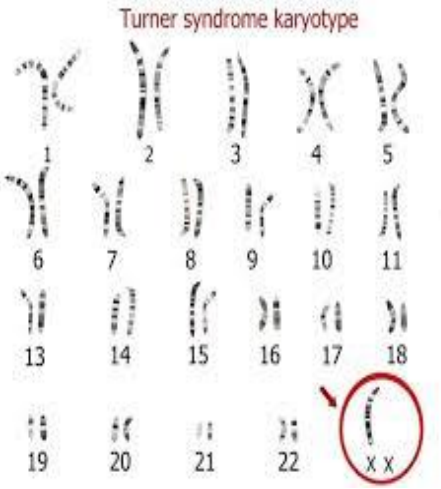
Common symptoms of Turner syndrome:

An individual may have any combination symptoms and is unlikely to have all symptoms:

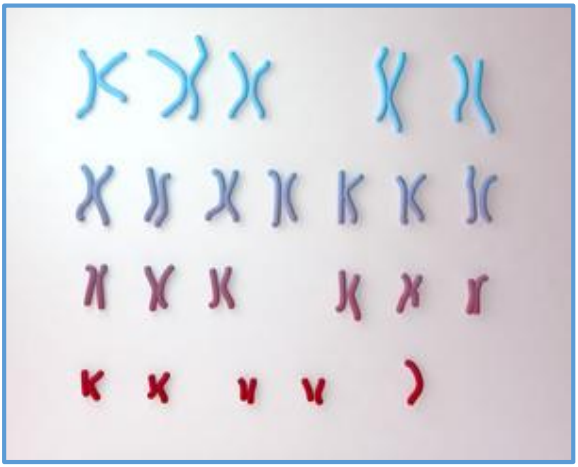

Low hairline
Elbo deformity
Brown spots
fold of skin
short stature
widely spaced nipples
No menstrua-tion

- **Turner syndrome is a chromosome abnormalities, It occurs in about one per 3000 live-born girl.**

Confirmed by chromosomal karyotyping:



Also known by the karyotype 45,X for females



Klinefelter's syndrome (KS):

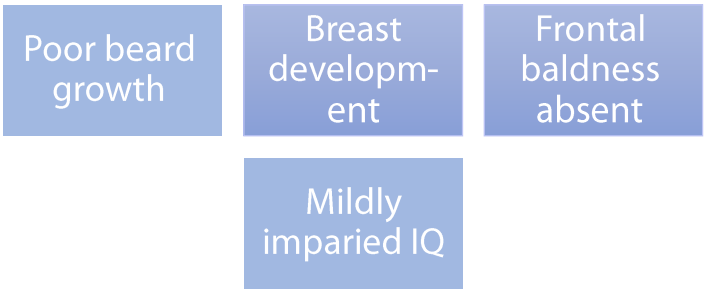
Also known as (47 XXY), is the set of symptoms that result from the present of two or more X chromosomes in males giving a total of 47 or more chromosomes rather than the usual 46.

Symptoms are typically more severe if there are additional copies of the X chromosomes (three or more).

Causes of Klinefelter's syndrome:

1. **(XXY) due to non-disjunction during fertilization where 40% of the additional X chromosome is of a maternal origin.**
2. **Mosaicism error in cell division after fertilization.**

Common symptoms of Klinefelter's syndrome:



Tall Stature

Slightly feminized physique

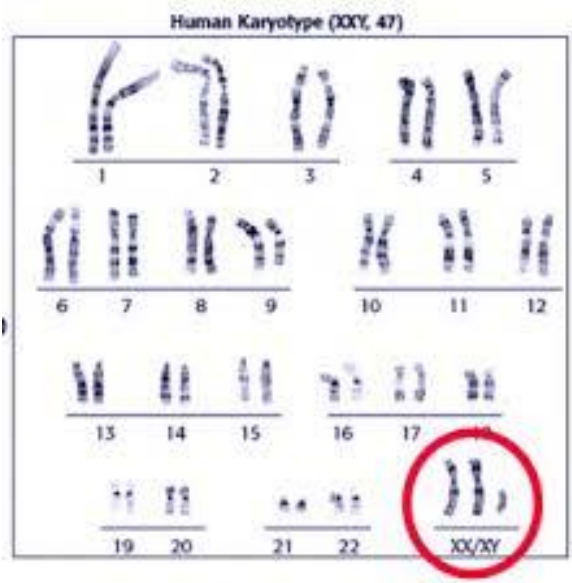
Tendency to lose chest hair

Osteoporo-sis

Klinefelter's syndrome is a chromosome abnormalities, It occurs in about one per 1000 live-born boy.

This figure shows a karyotype from a KS syndrome patient.

Confirmed by chromosome karyotyping



Also known by the karyotype 47,XXY

✗ Digeorge syndrome:

Also known as (Velocardial facial syndrome "cleft lip and palate") or (22q11.2deletion syndrome), it is a common genetic disorder resulting in medical complications, cognitive impairment, and brain morphologic changes, caused by the deletion of a small segment of chromosome 22 near the middle of the chromosome at a location designated *22q11.2*—signifying its location on the long arm of one of the pair of chromosomes. This syndrome is inherited in an autosomal dominant pattern.

Common symptoms of Digeorge

- Congenita-l heart disease
- Learning difficulties
- Hearing loss
- Growth hormone deficiency
- Skeletal defects
- Congenit-al defects of the kidney
- Defects in the larynx, trachea and esophagus
- The lack of activity in the parathyroid glands
- Bouts of convulsions
- Autoimm-une disorders

syndrome:

Digeorge syndrome is a chromosome abnormalities, It occurs in about one per 4000 live-born.

لأن الوعي وقاية ..

إدارة التثقيف الصحي

قسم علم الأمراض الطبية

HEM1.17.000571

Written by Health Promotion Department
Designed by CPAV-PRAMA