***التشوهات او الاعتلالات الكروموسومية*** ***Chromosome Aberration***

ويقصد بها الطفرات او الاعتلالات الغير طبيعية التي تحدث على مستوى الكروموسوم وهي على نوعين:

1. تشوهات او اعتلالات تركيبية Structural Abnormalities : وتعني التغيرات في تركيب الكروموسومات وهي على عدة انواع:

* حذف قطعه من الكروموسومDeletion :
* إضافة قطعه من الكروموسومInsertion :
* استبدال قطعه من الكروموسومTranslocation :
* عكس قطعه من الكروموسومInversion :
* مضاعفة قطعه من الكروموسومDuplication :

1. تشوهات او اعتلالات عدديه Numerical Abnormalities : وتعني التغيرات في عدد الكروموسومات ففي الحالة الطبيعية هنالك 23 زوج كروموسومي وتسمى بـ Disomy اما اذا حدث تغير في عدد الكروموسومات ضمن الزوج الواحد فمثلا إضافة كرموسومي فيصبح هنالك ثلاث كروموسومات بدلا من اثنين وتسمى هذه الحالة Trisomy وعلى العكس لو فقد كروموسوم يبقى كروموسوم واحد وتسمى هذه الحالة Monosomy.

***المتلازمات الوراثية Genetic Syndrome :*** *ويقصد بها الظواهر او الأمراض الوراثية الناتجة عن الاعتلالات الكروموسومية التركيبية والعددية وأهمها:*

**1-Numerical Abnormalities:**

When an individual is missing either a chromosome from a pair (monosomy) or has more than two chromosomes of a pair (trisomy). An example of a condition caused by numerical abnormalities is Down Syndrome, also known as Trisomy 21 (an individual with Down Syndrome has three copies of chromosome 21, rather than two). Turner Syndrome is an example of monosomy, where the individual - in this case a female - is born with only one sex chromosome, an X.

**2-Structural Abnormalities:**

When the chromosome's structure is altered. This can take several forms:

* **Deletions:** A portion of the chromosome is missing or deleted.
* **Duplications:** A portion of the chromosome is duplicated, resulting in extra genetic material.
* **Translocations:** When a portion of one chromosome is transferred to another chromosome. There are two main types of translocations. In a **reciprocal translocation**, segments from two different chromosomes have been exchanged. In a **Robertsonian translocation**, an entire chromosome has attached to another at the centromere.
* **Inversions:** A portion of the chromosome has broken off, turned upside down and reattached, therefore the genetic material is inverted.
* **Rings:** A portion of a chromosome has broken off and formed a circle or ring. This can happen with or without loss of genetic material.

Most chromosome abnormalities occur as an accident in the egg or sperm. Therefore, the abnormality is present in every cell of the body. Some abnormalities, however, can happen after conception, resulting in mosaicism, where some cells have the abnormality and some do not.

Chromosome abnormalities can be inherited from a parent (such as a translocation) or be "de novo" (new to the individual). This is why chromosome studies are often performed on parents when a child is found to have an abnormality.

**How do chromosome abnormalities happen?**

Chromosome abnormalities usually occur when there is an error in cell division. There are two kinds of cell division.

* Mitosis results in two cells that are duplicates of the original cell. In other words, one cell with 46 chromosomes becomes two cells with 46 chromosomes each. This kind of cell division occurs throughout the body, except in the reproductive organs. This is how most of the cells that make up our body are made and replaced.
* Meiosis results in cells with half the number of chromosomes, 23 instead of the normal 46. These are the eggs and sperm.

In both processes, the correct number of chromosomes is supposed to end up in the resulting cells. However, errors in cell division can result in cells with too few or too many copies of a chromosome. Errors can also occur when the chromosomes are being duplicated.

Glossary of Terms

**Acrocentric chromosomes** - those chromosomes, specifically numbers 13, 14, 15, 21 and 22, that are able to take part in Robertsonian translocations.

**de novo** - a chromosome abnormality that occurred in the individual and was not inherited from the parents.

**Mosaicism** - abnormal chromosome division resulting in two or more kinds of cells, each containing different numbers of chromosomes (chromosome mosaicism).

**Reciprocal translocation** - when segments from two different chromosomes have been exchanged.

**Ring chromosome** - a portion of a chromosome has broken off and formed a circle or ring. This can happen with or without loss of genetic material.

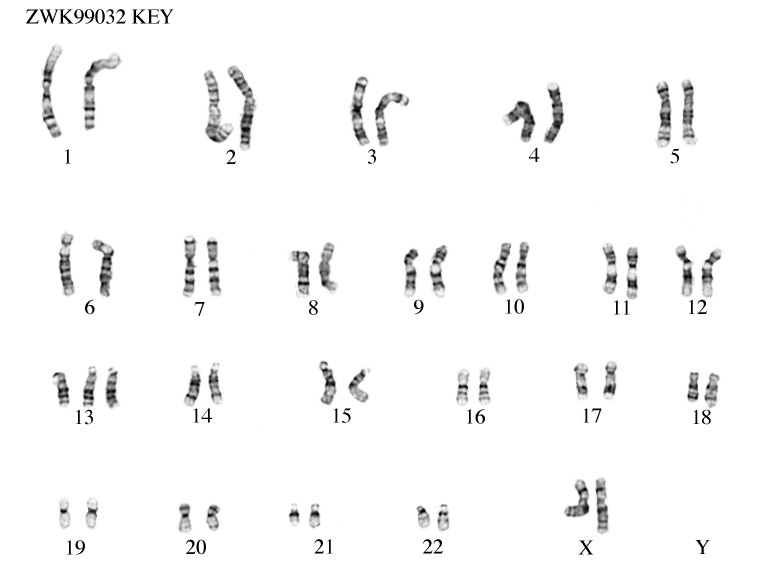
**Robertsonian translocation** - when two chromosomes fuse, usually at the centromere, creating a translocation. Only certain chromosomes, called acrocentric chromosomes, are capable of participating in this kind of translocation.

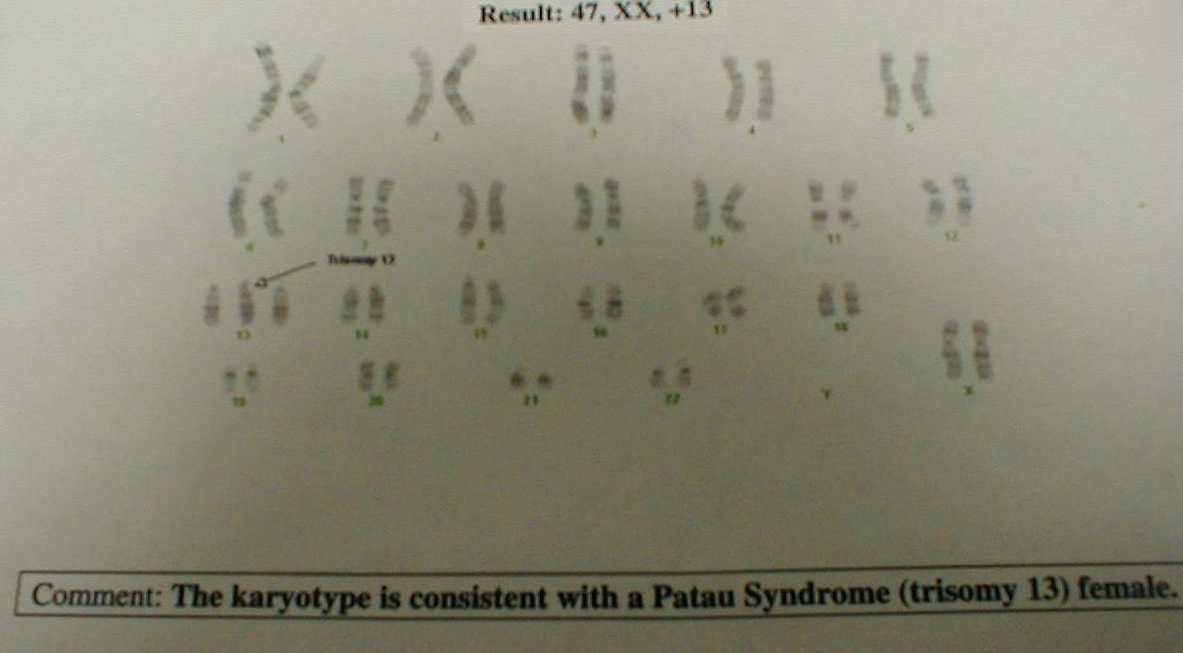
Examples of Genetic Disorders Resulted From Chromosomal Abnormalities:

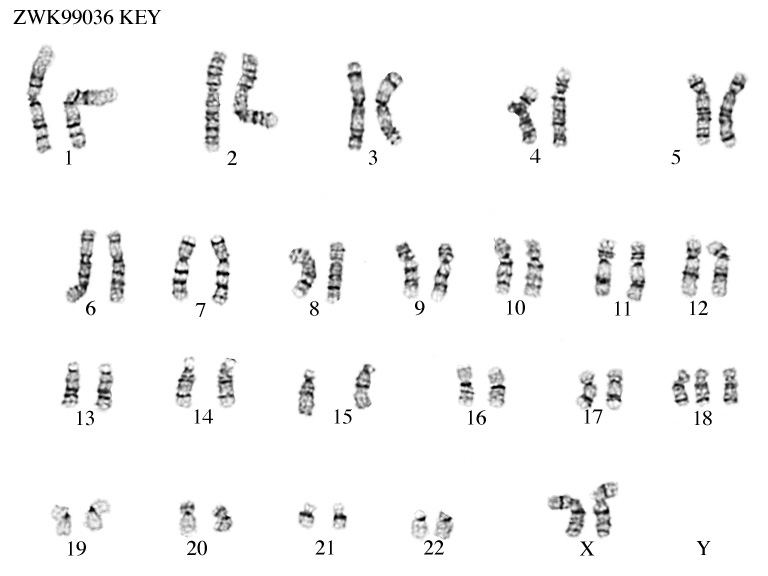
**1-Numerical Chromosomal Abnormalities:** (Change in the No. of Chromosomes set not in structure) includes:

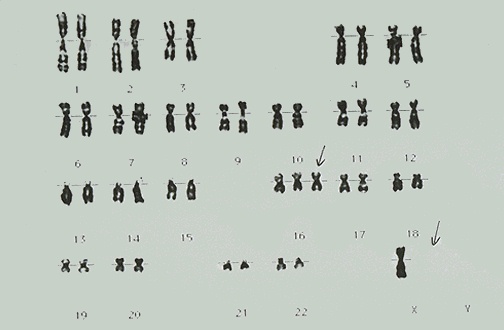
1. Autosomal Chromosome abnormalities:

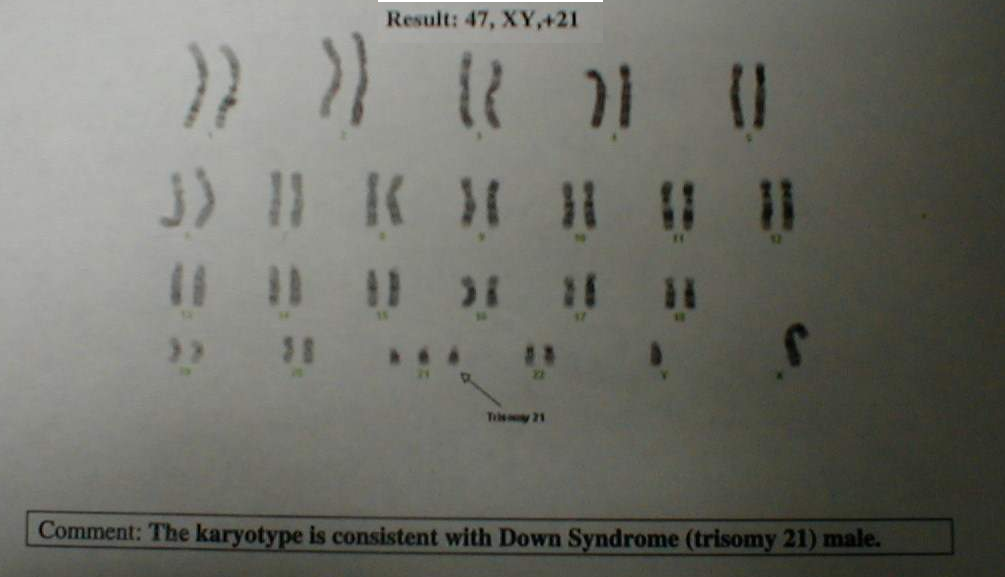
* [**Patau Syndrome**](http://en.wikipedia.org/wiki/Patau_Syndrome): also called D-Syndrome or trisomy13



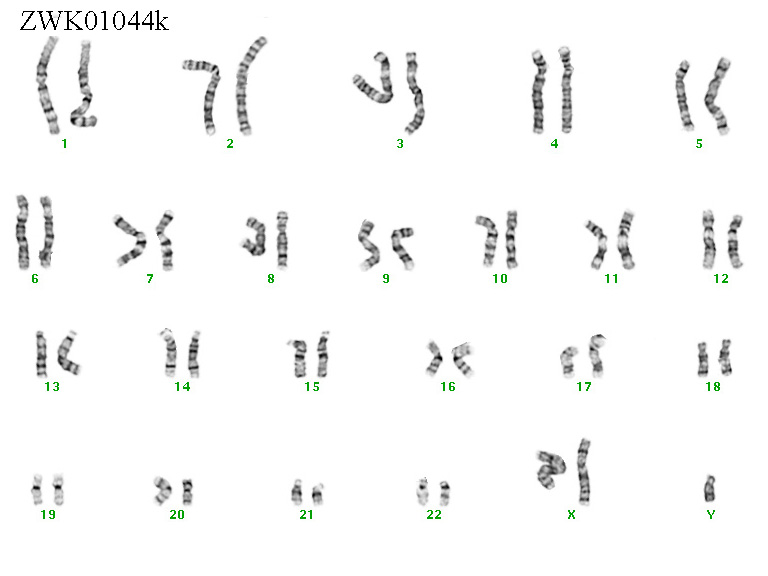
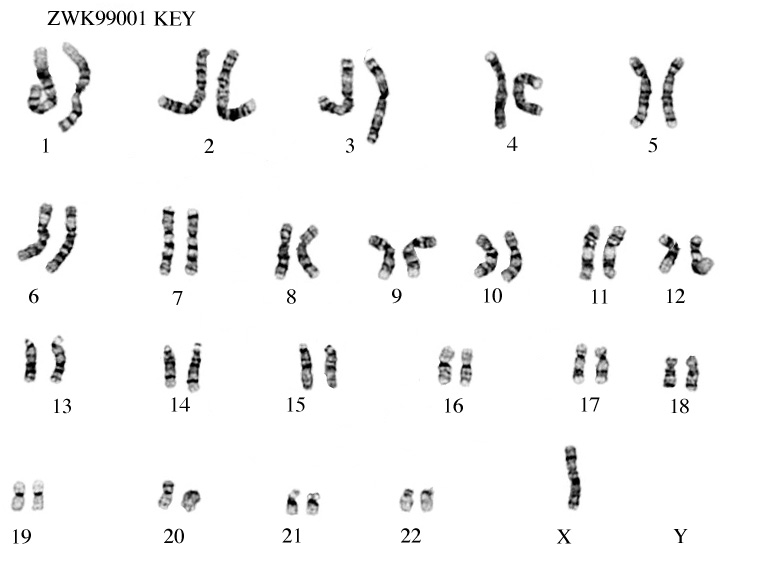


* ****[**Edwards syndrome**](http://en.wikipedia.org/wiki/Edwards_syndrome), or trisomy18
* **Trisomy 16** is a [chromosomal abnormality](http://en.wikipedia.org/wiki/Chromosomal_abnormality) in which there are 3 copies of [chromosome 16](http://en.wikipedia.org/wiki/Chromosome_16_%28human%29) rather than two.[[1]](http://en.wikipedia.org/wiki/Trisomy_16#cite_note-1) It is the most common trisomy leading to [miscarriage](http://en.wikipedia.org/wiki/Miscarriage) and the second most common chromosomal cause of it, closely following [X-chromosome monosomy](http://en.wikipedia.org/wiki/Turner_syndrome). Like most chromosomal abnormalities, trisomy 16 usually causes miscarriage in the first trimester of [pregnancy](http://en.wikipedia.org/wiki/Pregnancy).



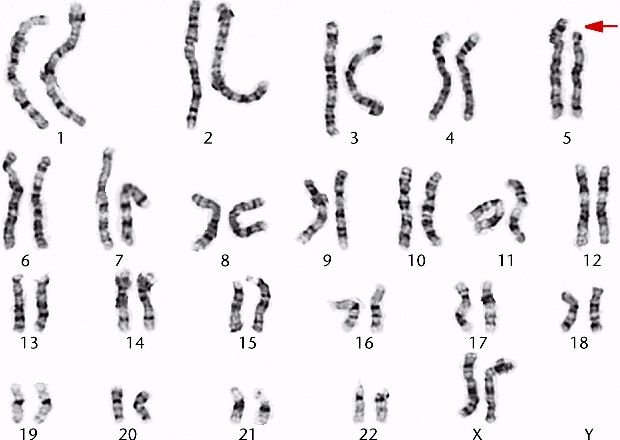
* [**Down syndrome**](http://en.wikipedia.org/wiki/Down_syndrome): or [trisomy 21](http://en.wikipedia.org/wiki/Trisomy_21)

1. Sex Chromosome abnormalities:incudes

* **Klinefelter’s syndrome**: trisomy XXY (supermale)
* **Turner’s syndrome:** monosomy absence of Y chromosome (Female).

**2-Sructural Chromosomal Abnormalities:** (Change in the structure of Chromosomes not in the No. of Chromosome) includes:

* **Cri-du-chat**: result from deletion of about half of the short arm of chromosome 5.



**Karyotyping:**

