Frequently Asked Questions

1. If no members of my family have a chromosomal anomaly can I still have a child with the condition?

Yes. There are two ways chromosomal anomalies occur in an individual, through inheritance from a parent or through a new change happening in the individual himself. Sometimes, parents may be carriers of a chromosomal anomaly without them being affected themselves. However when it is passed on to a child, symptoms may manifest due to imbalances of the chromosomes.

2. What are some examples of chromosomal anomalies?

Name	Description
Down Syndrome	Trisomy 21
Patau Syndrome	Trisomy 13
Edward Syndrome	Trisomy 18
Turner Syndrome	Monosomy X in females
Klinefelter Syndrome	Additional X in males
Cri du Chat Syndrome	Deletion on chromosome 5
Wolf Hirschhorn Syndrome	Deletion on chromosome 4
DiGeorge Syndrome	Deletion on chromosome 22

3. If I have a child with a chromosomal anomaly, what does this mean for subsequent pregnancies?

As discussed earlier, there are two ways chromosomal anomalies occur. If the change had occurred for the first time in your child (new mutation), there is a very low risk that subsequent pregnancies will be affected as well. On the other hand, if the change was inherited from a parent, there is a significantly higher risk of recurrence. A geneticist or genetic counsellor will be able to give you more details about this.

References

Mutations and Health. Genetic Home Reference. U.S. National Library of Medicine [online] Available from: http://ghr.nlm.nih.gov/handbook/mutationsanddisorders [Accessed June 2010]

Fact Sheet 6. Changes to Chromosomes – Number, Size and Structure [online] Available from: http://www.genetics.com.au/pdf/factsheets/fs06.pdf [Accessed June 2010]

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MALAYSIAN RARE DISORDERS SOCIETY

Rare Disorders Series:

Chromosomal Anomalies

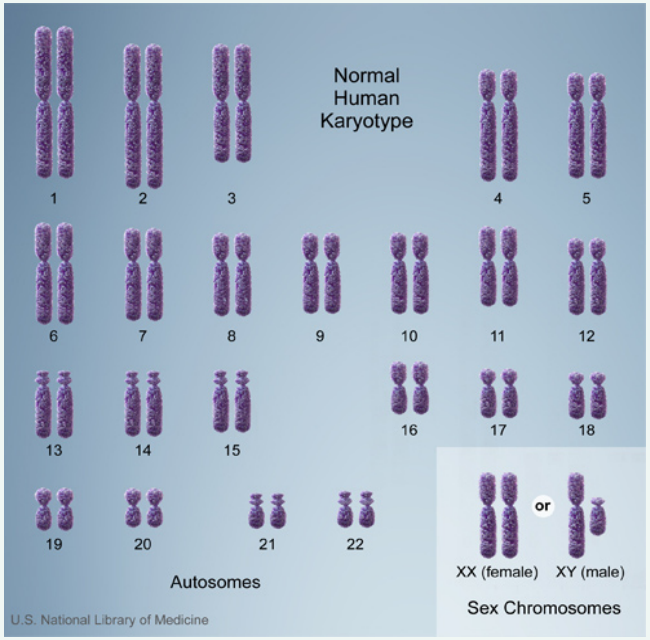
What are chromosomes?

Chromosomes are structures located in our cells which contain our genetic information.

Human beings have 23 pairs of chromosomes. One chromosome from each pair comes from the father and the other from the mother.

22 of the chromosome pairs are called autosomes.
These are numbered from one to 22, roughly according to their sizes from largest to smallest.
The 23rd pair is also known as the sex chromosomes.
Females have two X chromosomes while males have one X and one Y chromosome.

Each chromosome carries thousands of genes. They contain vital information which determine the various characteristics, growth and development of an individual.



Karyotype – A pictorial representation of the human chromosomes

Chromosomal anomalies

Chromosomal anomalies refer to changes in the number or structure of the chromosomes.

This may be due to a chromosome with the change being inherited from a parent, or the change arising for the first time in an individual.

These changes occur randomly and is not due to anything done or not done by either parent before or during pregnancy.

Chromosomal anomalies are individually rare but can be grouped into several general categories:

Structural changes

a. **Deletion** – A part of a chromosome breaks off resulting in a loss of genetic material.

b. **Duplication** – A part of a chromosome is copied too many times resulting in a gain of genetic material.

c. **Translocation** – Parts on two different chromosomes break off and swap places. May result in the genetic material remaining the same, or a loss or gain depending on the type of translocation.

d. **Inversion** – A part of a chromosome breaks off, reverses and reattaches.

Numerical changes

a. **Monosomy** – Loss of one chromosome.

b. **Trisomy** – Presence of an extra chromosome.

The effects of these chromosomal changes vary from one person to another. They are dependant on the number and function of genes lost or gained by the individual, as well as several other factors.

Testing

Genetic counselling by qualified personnel is advised if there is a family history of any genetic condition and before any genetic testing is done.

Gross changes in chromosomes such as large structural changes, monosomies and trisomies can be detected from a karyotype analysis.

The analysis is performed by looking at the picture of an individual's chromosomes (karyotype) and checking for changes by comparing it to a standard set of chromosomes.

Most chromosomal changes however, involve very subtle differences that require analysis at the DNA level.

Both types of analyses require a blood sample to be drawn from the patient and in many cases from the parents as well.

Treatment and management

There is currently no known cure for any kind of chromosomal anomaly.

Treatment is administered based on symptoms shown by the individual.

Many chromosomal anomalies result in developmental delay in the individual.
Early intervention and continuous education are key factors in helping those affected achieve their maximum potential.

Patients are advised to consult a geneticist to obtain further information on the best treatment and management plans according to symptoms.