Disability Awareness Begins With You

Chromosome Disorders

What are Chromosomes?

Simply put, chromosomes are the structures that hold our genes. Genes are the individual instructions that tell our bodies how to develop and keep our bodies running healthy. In every cell of our body there are 50,000 to 100,000 genes that are located on 46 chromosomes. These 46 chromosomes occur as 23 pairs. We get one of each pair from our mother in the egg, and one of each pair from our father in the sperm. The first 22 pairs are labeled longest to shortest. The last pair are called the sex chromosomes labeled X or Y. Females have two X chromosomes (XX), and males have an X and a Y chromosome (XY). Therefore everyone should have 46 chromosomes in every cell of their body. If a chromosome or piece of a chromosome is missing or duplicated, there are missing or extra genes respectively. When a person has missing or extra information (genes) problems can develop for that individual's health and development.

Each chromosomes has a p and q arm; p (petit) is the short arm and q (next letter in the alphabet) is the long arm. Some of the chromosomes like 13, 14, and 15 have very small p arms. When a karyotype is made (see below) the q arm is always put on the bottom and the p on the top. The arms are separated by a region known as the centromere which is a pinched area of the chromosome. The chromosomes need to be stained in order to see them with a microscope. When stained the chromosomes look like strings with light and dark 'bands'. Each chromosome arm is defined further by numbering the bands, the higher the number, the further that area is from the centromere.

What is a Karyotype?

A karyotype is an actual photograph of the chromosomes from one cell. The cells analyzed are usually white blood cells from a regular blood draw or from a prenatal speciman. After staining, the chromosomes can be seen as banded strings under 1,000 x magnification. They are analyzed by specially trained cytogenetic technologists, Ph.D cytogeneticists, or medical geneticists. 'Cytogenetics' is a word for the study of chromosomes. After analysis under the microscope a picture (karyotype) is printed. In a karyotype the chromosomes can appear bent or twisted. This is normal and is simply reflecting how they are sitting on the slide. Chromosomes are flexible structures made up of DNA. The coding order of that DNA makes up the genes. Chromosomes are analyzed during a time in the cell cycle when they are compact. During other times in the cell cycle the chromosomes unwind into long strands of DNA. At that time we would not be able to see them under the microscope. If you were to pull out all the chromosomes into long strands of DNA there would be over 7 feet of DNA in each cell! That's about 80 billion miles of DNA in the average human adult!

Down Syndrome

Children with Down Syndrome (DS) account for one of every 800 births. The risk of chromosome disorders like DS, trisomy 13 and trisomy 18 increases with maternal age. The incidence of DS at birth is lower at age 20 (1/1600) than at age 35 (1/370), but many more younger women have children than older women. So most (75-80%) DS children are born to younger women. If a couple has a child with DS, there is usually an increased risk for a second affected child. An extra whole chromosome 21 in all cells examined is found in about 92 per cent of all DS individuals. DS is common enough that it may appear that there is an excess cluster or hot spot when several DS children are born in the same area, but this is just chance and statistical variation. As far as we know, there is no relationship between DS and diet, drugs, economic status, or life style. Some evidence suggests that it is a little more common in families with Alzheimer's disease in one or more older family members.

Non-disjunction results from unequal chromosome division, usually in the mother's egg production. This is the form of DS that increases in incidence with increased maternal age. But DS is so common, it is not rare in young parents. If a couple has a child with DS, the risk is higher for the next pregnancy (1/100). Obviously, this means that the risk is 99% that the next child will NOT have DS. If the risk is already 1/220 at birth, the risk at age 37, the risk is usually estimated as twice the risk for age. Risks for amniocentesis results are higher because half to three-quarters of DS fetuses die before birth of natural causes. The risk of DS does not appear increased in siblings of trisomy 21 individuals. Prenatal testing is always recommended for couples who are worried about a second affected child. But prenatal testing procedures may not be for everyone, since they are expensive and many parents may not want to know or act on the information. The testing is more than 99 per cent reliable in most genetic centers, only rarely is there a sampling of maternal cells, failure of cells to grow or bacterial or yeast contamination.

Rare Chromosome Disorders

Rare chromosome disorders encompass extra, missing or re-arranged chromosome material but do not include the more common conditions such as Down's Syndrome. Using the latest technology, it is now possible for smaller and more complex chromosome defects to be identified. The amount of chromosome material duplicated, missing or rearranged can vary a great deal. This means that it may be difficult to identify two people who have exactly the same chromosomal disorder. The clinical problems of those affected can also vary enormously even when the chromosome diagnoses are similar.

It is immediately following diagnosis that these families have the greatest need for emotional and practical support and above all, for information. But even among the more common "rare" disorders, it is likely that the professionals in the local community - the GP, Social Worker or even hospital specialists - will have never before come across anyone with the same disorders. The usual sources of support are not available to affected families, yet the effects of the disorders can be devastating. The vast majority of families have a desperate feeling of isolation.

Most rare genetic disorders are viewed by the Medical Profession as "Orphan Diseases." Such terminology indicates that too few people are affected to interest the investment of capital in research for cures and therapies. Thus, the progress of such research proceeds at a snail's pace, while the affected children have little hope for breakthroughs in their lifetimes.

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