How to Conquer a Chromosome Abnormality—What is a chromosome abnormality?

University of Texas Health Science Center at San Antonio, School of Medicine
The Chromosome 18 Registry and Research Society

Special points of interest:

* Genetic material—genes, chromosomes, DNA
* Chromosome anatomy
* Chromosome analysis
* Common chromosome 18 syndromes

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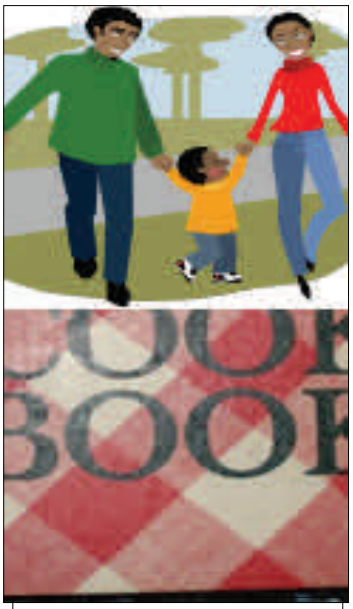
What is a chromosome 18 abnormality?

When confronted with a chromosome abnormality diagnosis, parents often have an avalanche of questions; the first of which is "What is a chromosome abnormality?" In order to understand what a chromosome abnormality is, we first need to understand what a chromosome is and some background about genetics.

You have surely noticed that children tend to look like their parents, often having some features like their mom and some like their dad. This is because they received half of their genetic material from each parent.

Your genetic material contains the instructions for every part of your body. These instructions are for virtually everything from how your ears are shaped to how you digest your foods to how susceptible you are to infections. These units of instruction within your body are essentially the cookbook of life. The individual units are called genes. Genes are organized within cells into packages called chromosomes much like recipes are organized in chapters in cookbooks.

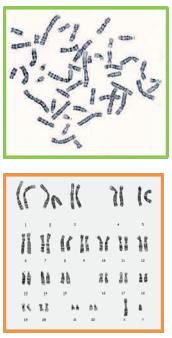
You may have heard of the human genome. Think of this as the cookbook of life. It contains all the recipes you will ever need. And every cell in the body contains the complete cookbook with all the recipes. As the cookbook is divided into chapters, our genome is divided into chromosomes. And as each chapter has many recipes, each chromosome contains many genes. While the cookbook is made of paper, your genes and chromosomes are made of DNA.



Genome = Cookbook
Chromosomes = Chapters
Genes = Recipes

Chromosome anatomy—important to understand

The top picture is of chromosomes from a white blood cell viewed under a light microscope. They have been treated with stain that makes them appear to have black and white bands. However, this random assortment of chromosomes is difficult to analyze for differences from normal.



The bottom picture shows the chromosomes paired up and in numerical order. This makes analysis easier. Chromosomes are found in pairs, one from mom and one from dad. Males have one X and one Y chromosome while females have two X chromosomes. Each chromosome has a "waistband" called a centromere, and two arms—the short arm is the p arm and the long arm is the q arm.

Chromosome Analysis

The analysis of chromosomes involves looking for missing pieces, duplicated pieces, rearrangements or losses or gains of entire chromosomes. Quite literally, anything you could do to a piece of paper with scissors, glue and a copy machine, could really happen and does really happen to chromosomes. However, the majority of these

changes are not compatible with life. The majority of babies conceived who have chromosome changes are miscarried early in pregnancy. Babies with any type of chromosome changes that survive are said to have chromosome abnormalities.

So, a chromosome abnormality is a change in the content or structure of chromsomes.

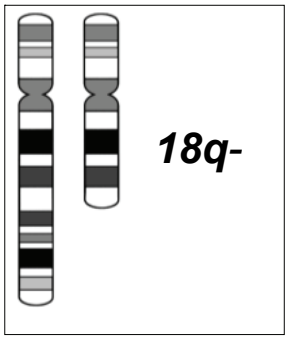
Because chromosomes are made up of genetic material (genes) the results can cause abnormal development or function.

"A chromosome abnormality is a change in the content or structure of chromosomes."

The Most Common Syndromes of Chromosome 18

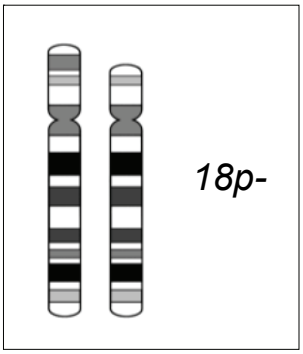
18q-

This picture shows 18q-, a deletion of any portion of the long arm, also called the q arm. Someone could have a deletion that is smaller or larger than the one pictured. The deletion could be from the end of the chromosome like in the picture and thus would be called a terminal deletion. The deletion could also be from somewhere in the middle of the long arm and as such would be called an interstitial deletion. It is also important to appreciate that one chromosome is normal and only one chromosome of the pair has the deletion



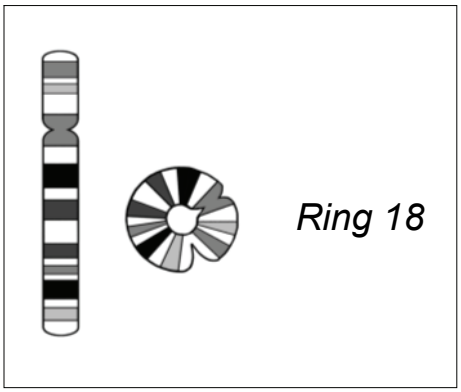
18p-

Shown here is 18p-. Like 18q-, the 18p deletions can be large or small, terminal or interstitial. 18p– involves the short or p arm of one of the two copies of chromosome 18.



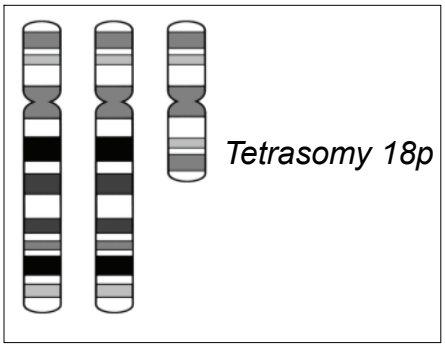
Ring 18

Shown here is Ring 18. In this condition one of the two chromosomes forms a ring. This means that by definition, the protective cap called the telomere is missing from each end of the chromosome. However that does not mean that genes must be missing from both ends of the chromosome—but they usually are. Again like the other two conditions, the size of the deletion from either end of the chromosome can be large or small. There could be a big q arm deletion and a small p arm deletion or two small deletions or any combination. It is important to note that the deletion of genes in ring chromosomes always involves the ends of the chromosome arms.



Tetrasomy 18p

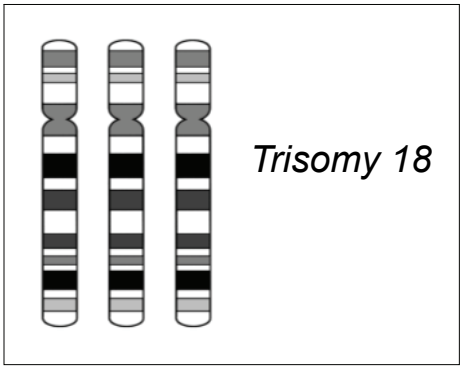
Shown to the left is Tetrasomy 18p. In this condition there are two normal copies of chromosome 18 along with an extra chromosome composed of two p arms of chromosome 18. This means there are a total of 4 copies of the chromosome 18 p arm, which is why it is called Tetrasomy 18p.



"Because chromosomes are made up of genetic material (genes), the results can cause abnormal development or function."

Trisomy 18

The last major syndrome of chromosome 18 is Trisomy 18, in which there are simply 3 copies of chromosome 18. This is, by far, the most common of the chromosome 18 syndromes. However, this condition is different from the other conditions because children with Trisomy 18 usually have shortened lifespans and many die before their first birthday.



SCHOOL OF MEDICINE
UT HEALTH SCIENCE CENTER
THE CHROMOSOME 18 CLINICAL RESEARCH CENTER

UTHSCSA—Department of Pediatrics
The Chromosome 18 Clinical Research Center
MSC 7820
7703 Floyd Curl Drive
San Antonio, TX 78229-3900

Phone: 210-567-5321
Fax: 210-567-0919
E-mail: hilla3@uthscsa.edu

For more information, you may contact the authors and principal investigators of the Chromosome 18 Clinical Research Center at the phone numbers or email shown to the left.

Authors & Principal Investigators:
Jannine D. Cody, PhD and Daniel E. Hale, MD

Our Motto
To provide individuals and families affected by chromosome 18 abnormalities with comprehensive medical and educational information with a focus on treatment options.

We are on the web!

http://pediatrics.uthscsa.edu/centers/chromosome18/

Summary

To summarize, this is what you have learned so far:

* Your genetic material is packaged in chromosomes.
* The chromosomes contain genes.
* Genes are the instructions for all body development and function.
* There is a variety of chromosome copy number changes that can occur.
* When they do occur, normal development and function is often altered.

Information provided by The Chromosome 18 Clinical Research Center to:

The Chromosome 18 Registry & Research Society

http://www.chromosome18.org/
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