

CHROMOSOMAL DISORDERS

A person's cells can sometimes have extra or missing chromosomes (or pieces of chromosomes), referred to as a chromosomal disorder. If there is an extra chromosome or piece of a chromosome, there are three copies of the genes on that chromosome, called trisomy. If there is a missing piece of a chromosome, there is only one copy of these genes, and this is called monosomy. Since there are hundreds to thousands of genes on each chromosome, there may be many genes affected by a chromosome abnormality. Having this difference in copy number of some genes in each cell can cause a person to have problems in the way that their organs form, or to experience growth and/or developmental delay. Each specific chromosome abnormality can cause different symptoms, because different genes are affected. However, specific developmental or medical complications can be seen in children with similar chromosomal disorders, giving us the ability to provide these individuals information about what the future will hold. There are many chromosomal disorders which have not yet been described in the medical literature, thus we still have much to learn about the way that they might affect the body's growth and function. However, we are committed to providing and coordinating the best clinical care for your child, regardless of their chromosome disorder.

CLINIC PROVIDERS

Ada Hamosh, MD- Clinical Geneticist

Julie Hoover-Fong, MD- Clinical Geneticist

Celide Koerner, RN, MS- Nutritionist

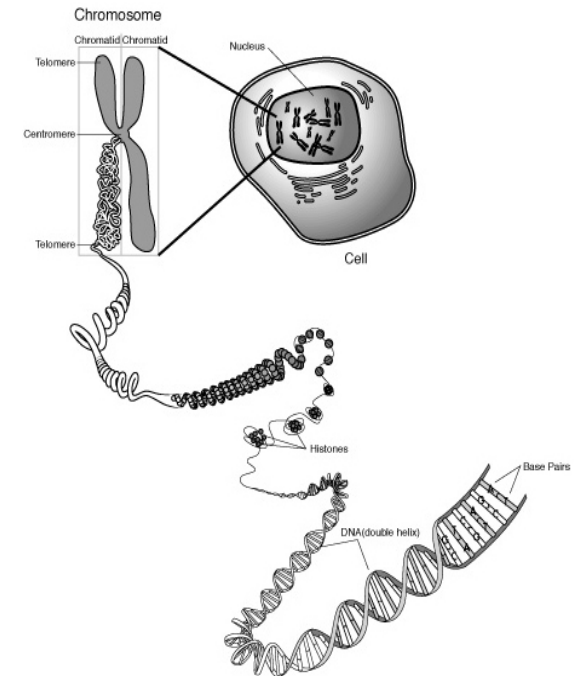
Emily Lisi, MS- Genetic Counselor and Clinic Coordinator



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RARE CHROMOSOMAL DISORDER CLINIC



WHAT ARE CHROMOSOMES?

The human body is made up of trillions of cells. DNA, which codes for our genetic information, is present in each cell. This information tells our bodies how to grow and develop and determines our unique characteristics, much like a blueprint. The DNA code is divided into individual segments, called genes. Each gene codes for a protein which has a specific function in the body. There are approximately 20,000 different genes in each cell of our body! The genes are packaged together on chromosomes.

We typically have 23 pairs of chromosomes in each cell. One copy of each pair comes from our mother in her egg cell and the other comes from our father in his sperm cell, making 46 chromosomes in all. We each have 22 pairs of “numbered” chromosomes (autosomes) and two sex chromosomes. Females have two X chromosomes while males have an X and a Y chromosome. It is important to have two complete copies of each autosome and two sex chromosomes for normal growth and development. A picture of a person’s complete set of chromosomes is known as a karyotype, as shown below.



Normal Male Karyotype

RARE CHROMOSOMAL DISORDER CLINIC

Johns Hopkins Medicine now offers a specialized clinic for individuals with rare chromosomal disorders. You and your family will have the opportunity to meet with experts in the field who will be seeing only patients with similar diagnoses that day. This clinic will enable you to coordinate appointments with other specialists at Johns Hopkins and learn what to expect in the future. In addition, you may be able to make contact with other families who are affected with similar conditions. These contacts may offer you support in a way that even the best health care providers may not.

What disorders would be appropriate for this clinic?

22q11 deletion syndrome (VCF)

Smith-Magenis syndrome

Williams Syndrome

Turner Syndrome

Microdeletion/duplication syndromes

Unbalanced translocations

Other rare chromosome abnormalities

Those individuals with Trisomy 21 (Down syndrome) are also welcome to visit our clinic but will be referred to a specialized Down syndrome clinic at Kennedy Krieger Institute for long-term care.

How do I make an appointment?

Please call the Johns Hopkins Genetics Office at 410-955-3071 to schedule an appointment. Emily Lisi, a certified genetic counselor, will discuss your child’s particular chromosome abnormality and his/her health and development. If possible, please have a copy of the chromosome test results available when you call. You may be asked to have an echocardiogram, a renal ultrasound, a brain MRI, or other testing prior to your clinic visit, so that we can provide the most comprehensive care for your child. More studies or specialty referrals may be indicated following your genetics clinic visit and we will help coordinate these studies. A referral may be required from your pediatrician/primary care physician, depending on your insurance plan.

CONTACT INFORMATION

For more information, please contact:

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Please visit www.rarechromo.org to learn more about rare chromosomal disorders.