

Prenatally Diagnosed *Chromosome Rearrangements*

INTRODUCTION

Finding out that you or your baby carries a chromosome rearrangement can be confusing. We hope that this pamphlet will help to explain the chromosome findings and answer some questions you may be having.

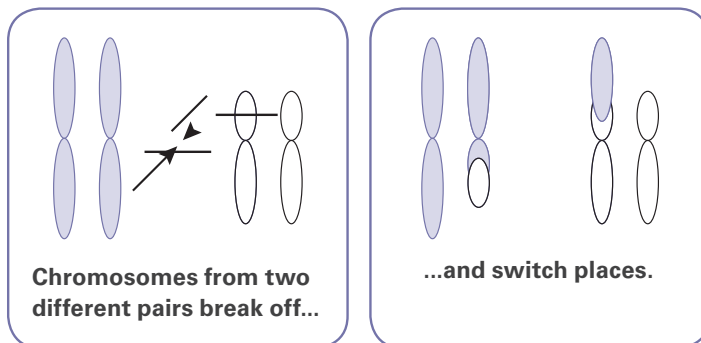
Chromosomes and genes

To begin, we would like to refresh your memory of chromosomes and genes. You may remember from science class that our bodies are made up of cells. Inside each cell, there are structures called chromosomes. The chromosomes are inherited from our parents through the egg and sperm cells. There are 46 chromosomes in each cell that can be arranged into 23 pairs. One of each pair comes from the mother and one from the father.

Each chromosome contains hundreds of genes. The genes are the instructions that tell our bodies how to function and develop.

Chromosome translocations

A translocation is a rearrangement of chromosome material. Sometimes pieces of chromosomes from two separate pairs break off and switch places with one another as illustrated below.



Although the chromosome material is rearranged, all of the genes are still there. Therefore, the translocation is said to be balanced and does not cause mental or physical handicaps.

Unbalanced translocations

Occasionally, a small piece of chromosome material is lost during the formation of a translocation. If this happens, the missing genes can cause intellectual disabilities and/or birth defects. This type of translocation is called an unbalanced translocation. Unfortunately, the missing material may be so small that it can't be seen through a microscope. So, we can't always tell if a translocation identified through prenatal diagnosis will affect the baby or not.

Parents' chromosomes

Many translocations diagnosed prenatally are inherited. This means that the chromosome rearrangement has been passed through the egg or sperm cell to the fetus. In this case, one parent can be found to have an identical translocation. If the parent has no physical or mental handicaps caused by the translocation, the baby will usually not have related health problems either.

Balanced translocation carriers

Carriers of balanced translocations are usually able to have healthy children, but may have some reproductive problems. This is because a person with a balanced translocation can make egg or sperm cells with too much of one chromosome and too little of the other. This can cause infertility, miscarriage or the birth of a child with intellectual disabilities and/or birth defects. A geneticist or genetics counselor should be able to tell you the approximate risk for the above mentioned problems given your specific rearrangement and history. Prenatal diagnosis, such as amniocentesis or chorionic villus sampling, is routinely offered to translocation carriers in future pregnancies.

Since the balanced translocation can be inherited, the carrier parent may also have inherited the rearrangement. As such, other family members such as parents, brothers, sisters or children of a balanced translocation carrier may also be at risk for these reproductive problems. relatives at risk may wish to read this pamphlet and consider having chromosome testing as well.

If parents' chromosomes are normal

If neither parent has the rearrangement, the laboratory may use special stains or perform additional tests on the specimen. In some cases these tests may determine whether the fetus has extra or missing genetic material.

An ultrasound evaluation of the fetus between 18 and 22 weeks of pregnancy may also be useful. Many fetuses with unbalanced translocations have associated birth defects. In some cases, these birth defects can be detected with a detailed ultrasound evaluation. Not all birth defects can be identified in this manner, however.

Sometimes, it is impossible to tell whether the identified chromosome abnormality will cause problems for the baby.



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