

Part of the curriculum: Genetics and evolution [1A1 – Chromosomal abnormalities]

A Look at Translocation, Trisomy and Mosaic Down Syndrome

The 3 different types of Down syndrome explained

Adapted from www.verywell.com

By Kathleen Fergus

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Translocation Down syndrome is one of the three primary types of Down syndrome. It's due to reciprocal translocation, a special type of chromosome abnormality. Any chromosome can be involved in a reciprocal translocation. For example, a reciprocal translocation between the long arm of chromosome 14 and the short arm of chromosome 21, results in two new fused chromosomes called derivative chromosomes.

Instead of having three independent, separate number 21 chromosomes, a person with translocation Down syndrome has two independent #21 chromosomes and a #21 chromosome that is attached to another chromosome. The attached chromosomes are called a derivative chromosome and can occur for the first time in the person with translocation Down syndrome, or the derivative chromosome can be inherited from a parent. Therefore when a child is diagnosed with translocation Down syndrome, it is very important that the parents of the child also have karyotype testing.

Translocation Down syndrome is one of the three forms of Down syndrome (the others are trisomy and mosaicism). It accounts for 4 to 5% of Down syndrome cases. Normally human beings inherit 23 chromosomes from both the mother and the father for a grand total of 46 chromosomes. However, people with trisomy Down syndrome get an extra copy of chromosome 21, for a total of 47 chromosomes. Mosaicism Down syndrome is different from other types of Down syndrome because not all the cells in the body have 46 chromosomes. Instead, a percentage of cells can have 47 chromosomes (with an extra copy of chromosome 21)...

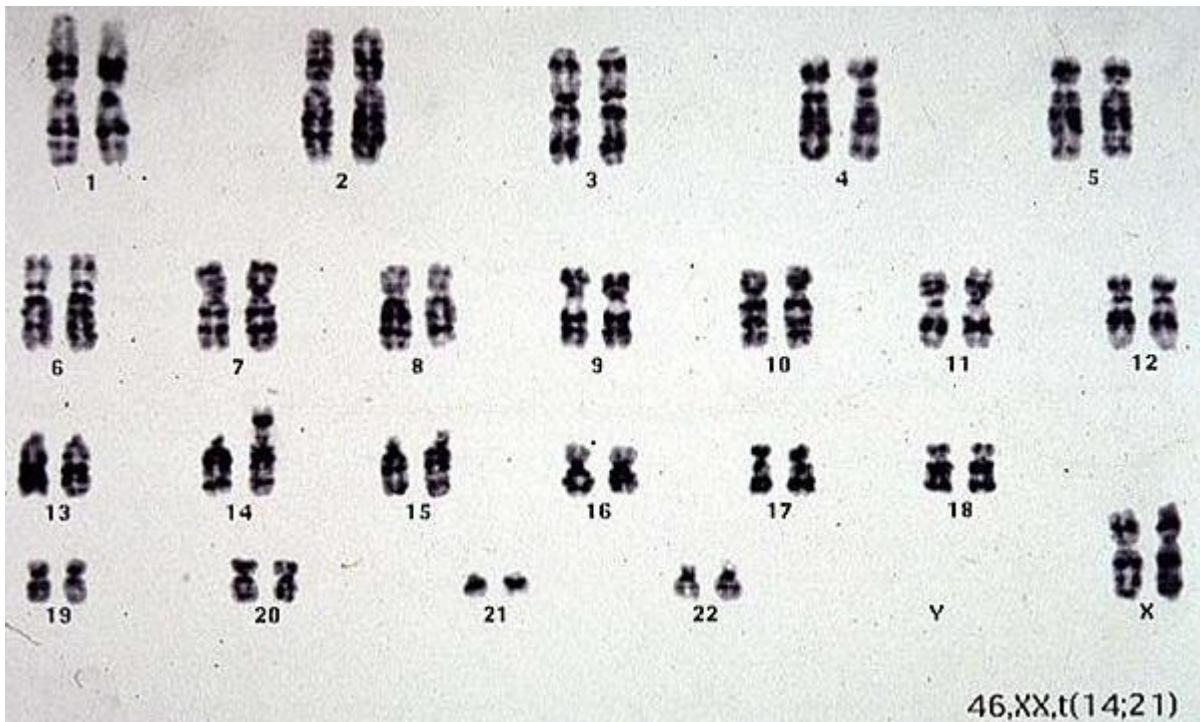
Use the document as well as your scientific knowledge to :

- 1. Explain how a translocation can lead to Down syndrome, and why the parents of an affected child must have a karyotype testing.**
- 2. Explain the origin of trisomy Down syndrome, using sketches and the additional document.**

You may use the following key words:

Non-disjunction – Meiosis – Gamete

Down Syndrome caused by unbalanced translocation between chromosomes 14 and 21



**Balanced translocation carrier between chromosomes 14 and 21 in male
Karyotype 45, XY, t (14;21)**

