What are microdeletions and microduplications?

To the point- Microdeletion and microduplication syndromes occur when an individual has a small piece of a chromosome that is missing (deleted) or extra (duplicated). Microdeletions and microduplications include the term “micro” because the missing or additional genetic material is too small to be seen with a microscope or with a conventional karyotype (picture of the chromosomes) but is big enough that it typically leads to the absence of several genes in the area of the deletion or duplication. The health and developmental effects associated with microdeletions and microduplications can vary tremendously and depend on where in the genome the deletion/duplication is and how many genes it involves. The effects can be so mild that no significant health or developmental concerns are seen, more moderate, or in some cases quite severe.

What does it mean for me or my baby if a microdeletion or microduplication is diagnosed or suspected?

The answer to that question really depends on where the deletion or duplication is located – what genes and how many genes are included in the duplication or deletion. Some microduplications and microdeletions are very rare and have only ever been reported in one or a few individuals. Others are seen more commonly (although, even the more “common” microdeletions” are still pretty rare). The associated effects of any specific microdeletion or microduplication depend on what genes are involved, but can also be variable even among people who have the same deletion or duplication. For example, a parent could have a microdeletion or microduplication and have virtually no symptoms, but their child may have more symptoms, even though it is the same microdeletion or microduplication.

Genetics Review of Microdeletions and Microduplications

As we reviewed in Genetics 101 each gene basically could be thought of as an instruction book to build a specific protein that has some role in how our body functions. Our genes are inherited in pairs, one from each parent, and are passed on through our chromosomes. Humans typically have 23 pairs of chromosomes for a total of 46, with one set passed on from their biological mother and one set form their biological father. You could think of a chromosome as being a bookshelf that holds a certain set of instruction books with each instruction book telling the body how to make a certain protein. Each instruction book is a gene.
There are several types of genetic conditions. Some conditions are caused by a small change, known as a mutation, in a single gene. This could be thought of as a typo in one instruction book that does not allow the protein to be made properly.

Aneuploidy conditions are caused by entire extra or missing copies of whole chromosomes. For instance, Down syndrome is caused by three copies of the 21st chromosome.

A microdeletion could be thought of as a situation in which several instruction books or genes are removed from one of the bookshelves (chromosomes).

A microduplication could be thought of as a situation in which several duplicate copies of specific instruction books (genes) are added to the bookshelf (chromosomes).

**What causes a microdeletion or microduplication?**

There is nothing currently known that a parent could do to cause or prevent a microduplication or microdeletion from occurring.

Microdeletions and duplications can be passed on from parent to child in some cases or occur due to a chance event in the chromosome in the formation of the egg or the sperm or very early in fetal development. The chance that a microduplication or deletion is inherited vs brand new (de novo) in a baby depends on the specific location of the duplication/deletion.

**Information about Specific Microdeletion and Microduplication Syndromes**

Below you will find links to some more specific information regarding some microdeletion and microduplication syndromes. Testing for some of these conditions is done using cell-free DNA (cfDNA).

For some points to consider about pursuing cfDNA for screening for microdeletions, clinic here: What should I know about cfDNA screening for microdeletions? (hyperlink)

[22q11.2 Deletion syndrome](#)

[1p36 Deletion syndrome](#)
15q Angelman syndrome
15q Prader-Willi syndrome
11q - Jacobsen syndrome
8q - Langer-Giedion syndrome
5p - Cri-du-chat syndrome
4p - Wolf-Hirschhorn syndrome