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Fact Sheet about Chromosomes and Syndromes

All human beings have **23 pairs of chromosomes**, and each chromosome has many genes on it. Genes provide a roadmap for how a person develops, and might be thought of as the "building plan" or a blueprint for our development. Though the scientists can now read all of the codes in a human genome, or set of genes, they still don't know what every gene's job is in the building of a human. It is complicated to make a person! But we have learned a lot about how humans develop.

There are several ways that errors in chromosomes can cause a genetic syndrome. First, some syndromes are caused by abnormalities in chromosomes, meaning a person has one or more chromosomes that are "atypical." Parts of one may be missing or they may have an extra chromosome than most people. For example, Down syndrome, also known as Trisomy 21, is caused by too many copies of a chromosome.

Another way that chromosomes can cause a syndrome is when genetic material is missing. These are known as "microdeletion syndromes." This means small amounts of the gene broke off or are missing. This missing genetic material is then often found to have been critical to normal or "typical" development. Some microdeletion syndromes commonly associated with IDD are: Prader Willi Syndrome, Williams syndrome and 2q11 Deletion Syndrome.

One last way chromosomes can cause a syndrome is having too much genetic material or too many repeats of a gene sequence. Having too much genetic material can also disrupt physical and especially, brain development. An example of this is Fragile X Syndrome.