Welcome to the neighborhood!

Chromosome 1q21.1

Why does location matter so much? Why all the fuss about deletion vs. duplications?

How many chromosomes do we have? Most people have 23 pairs of chromosomes, for a total of 46.

Typically you get one chromosome from mom and one from dad. This means that most people have 2 copies of chromosome 1.

The location of a deletion or duplication determines what types of features are shown. Changes within the ‘typical’ region of 1q cause different features than changes within the TAR region.

Sometimes, a child is born with differences in his or her chromosomes. A person might have extra copies or fewer copies of a whole chromosome or just part of one. This is called a copy number variant or CNV.

A 1q21.1 deletion means that there is genetic material missing. A 1q21.1 duplication means that there is extra genetic material. Having a deletion versus duplication affects what types of features people have.

If you would like to learn more, join our community, or contribute to the ongoing research, please visit simonsvipconnect.org

What types of CNVs happen to chromosome 1q21.1?

Chromosomes have arms. Each chromosome in your body has a short (or p) arm and a long (or q) arm.

Chromosomes have stripes. The arms have different sections and regions.

Chromosomes have addresses. Every chromosome contains different sections and regions.

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We are still learning about 1q21.1. Scientists have discovered a lot about 1q21.1 with the help of families. Being said, we still do not know everything. **Want to help?**

**deletion**

Missing information from ~146.6-147.4 Mb:

Features: Mood and anxiety disorders, short stature, smaller head size (microcephaly), low muscle tone (hypotonia), cataracts, tremors, over active reflexes, heart problems

**duplication**

Extra information from ~146.6-147.4kb:

Features: Autism Spectrum Disorder (ASD), Attention Deficit Hyperactivity Disorder (ADHD), mild intellectual disability, larger head (macrocephaly), low muscle tone (hypotonia), gastric ulcers, scoliosis, impairment of phonological processing and articulation, impairment of fine and overall motor skills

**What is the TAR region?**

The ‘TAR’ region of 1q21.1 is next to the ‘typical’ region. For some individuals, the 1q21.1 CNV may include both the typical region and the TAR (Thrombocytopenia Absent Radius) region. When the TAR region is deleted on one chromosome and there is another genetic variant in the same region on the other chromosome 1, individuals may have TAR syndrome. Individuals with TAR syndrome have problems with poor blood clotting and underdevelopment or malformation of bones in the arms or legs.

**Everyone Is Unique**

Not everyone with a 1q21.1 deletion or duplication exhibits the same features. Even individuals in the same family may show a wide range of features and how mild or severe these are.

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