Welcome to the neighborhood!

Chromosome 15q11.2–13.1

Did you know?

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

Chromosomes have addresses
Every chromosome contains different sections and regions.

15q11.2–13.1
Chromosome 15 Arm q Region 11.2–13.1

Chromosomes have stripes
The arms have different and unique regions, which can look like stripes (bands) under a microscope.

Typically you get **one** chromosome from mom and **one** from dad
This means that most people have 2 copies of chromosome 15.

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.

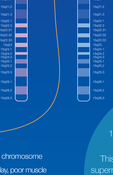
Why all the fuss about mom & dad?
Unlike most chromosomes, genes from the 15q11.2-13.1 region behave differently depending on whether they are inherited from mom (maternal) or from dad (paternal).

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

Does the number of copies matter?
Yes, having too few or too many maternal or paternal copies of genes from 15q11.2-13.1 can impact a child's development.

Chromosomes come from your mom & dad.

23 chromosomes from mom

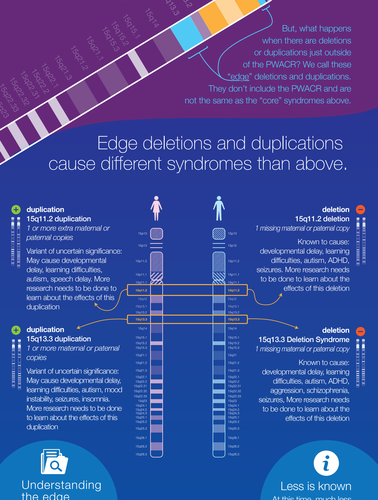


23 chromosomes from dad

Children typically have 46 chromosomes but sometimes they have extra or missing chromosomal material.

This is called a Copy Number Variant or CNV.

What types of CNVs happen to chromosome 15q11.2–13.1?



deletion
Angelman Syndrome (AS)
Missing maternal information

Features: Developmental delay, seizures, motor and language impairments, intellectual disability, happy personality with unprovoked laughter

deletion
Prader-Willi Syndrome (PWS)
Missing paternal information

Features: developmental delay, poor muscle tone, intellectual disability, insatiable appetite, and behavior problems

duplication
Dup15q Syndrome
Extra maternal information

Maternal interstitial (within chromosome) duplication

Features: developmental delay, poor muscle tone, seizures, motor and language impairments, intellectual disability, autism and other behavioral differences

duplication
15q11.2-13.1 duplication
Extra paternal information

Paternal interstitial (within chromosome) duplication

Known to cause: developmental delay, learning difficulties, autism, ADHD seizures. More research needs to be done to learn about the effects of this duplication

duplication
Dup15q Syndrome
Extra maternal information

Maternal extra isodicentric chromosome

Features: developmental delay, poor muscle tone, seizures, motor and language impairments, intellectual disability, autism and other behavioral differences

What's in a name?
Sometimes, extra copies of 15q11.2-13.1 connect and form their own, extra chromosome.

This may have different names, like supernumerary 15q, isodicentric 15q, inverted duplication 15q, and others.

Although technically these names may mean something slightly different, they all refer to an extra chromosome made from 15q11.2-13.1

All of the syndromes above have one thing in common: something's up with 15q11.2-13.1

We call these "core" deletions and duplications. This region contains important genes, like *UBE3A, SNRPN*, and others. We refer to this as the Prader-Willi/Angelman Critical Region (PWACR)

But, what happens when there are deletions or duplications just outside of the PWACR? We call these "edge" deletions and duplications. They don't include the PWACR and are not the same as the "core" syndromes above.

Edge deletions and duplications cause different syndromes than above.

duplication
15q11.2 duplication
1 or more extra maternal or paternal copies

Variant of uncertain significance: May cause developmental delay, learning difficulties, autism, speech delay. More research needs to be done to learn about the effects of this duplication

deletion
15q11.2 deletion
1 missing maternal or paternal copy

Known to cause: developmental delay, learning difficulties, autism, ADHD, seizures. More research needs to be done to learn about the effects of this deletion

duplication
15q13.3 duplication
1 or more maternal or paternal copies

Variant of uncertain significance: May cause developmental delay, learning difficulties, autism, mood instability, seizures, insomnia. More research needs to be done to learn about the effects of this duplication

deletion
15q13.3 Deletion Syndrome
1 missing maternal or paternal copy

Known to cause: developmental delay, learning difficulties, autism, ADHD, aggression, schizophrenia, seizures. More research needs to be done to learn about the effects of this deletion

Understanding the edge
Scientists are working to identify and understand important genes in the edge region.

Compared to differences involving the PWACR, changes in these "edge" areas have much more variable outcomes.

Less is known
At this time, much less is known about these "edge" deletions and duplications.

how?

- May cause milder or altogether different symptoms
- They have been studied in fewer people
- May not cause any symptoms in some people
- May not be as affected by parent of origin (from mother or from father)
- Are much more likely to be inherited, or passed down from parents

Since scientists are still unsure about the effects of most "edge" duplications and deletions, they are considered "variants of uncertain significance" (VUS).

Research continues in an effort to figure out which VUS cause symptoms and to fully describe their impact on behavior and learning.

Learn more and support the efforts of the Dup15q Alliance at www.dup15q.org

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dup15q.org