

Facilitating discussions about the psychiatric phenotypes of 22q11.2 deletion syndrome

Virginia Commonwealth University

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Our Study:

The research team wants to know what conversations about a diagnosis of 22q11.2 deletion syndrome look like from the point of view of people with 22q and/or the parents or legal guardians of someone with 22q. We want to know what information was or was not given about the mental health problems associated with 22q:

- What information was helpful?
- What information was not helpful?
- What information did they want to know?
- What information did they not want to know?
- When is the best time to talk about the chance to have mental health problems as a part of 22q?

We want to use what we learn to create a pamphlet about the mental health problems that are part of 22q11.2 deletion syndrome, but we need help from study participants. If you 1) have 22q11.2 deletion syndrome AND/OR 2) are the parent or legal guardian of someone with 22q, we want to hear what this was like for you. We hope a pamphlet about the mental health problems of 22q will be helpful for people, their family and friends, and the doctors and other healthcare providers who care for people with 22q11.2 deletion syndrome.

If you are interested in participating, you may access the study survey online using the following website:

<https://redcap.vcu.edu/surveys/?s=LT98YYD7MF>

Study Contact Information:

If you have any questions or concerns, you can contact the student researcher:

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What is 22q11.2 deletion syndrome?

The 22q11.2 deletion syndrome is a genetic condition. This means a change in our DNA, or the information that tells our body how to work, causes the condition. There are many symptoms of 22q, and these symptoms are different among people. Some people may not even know they have the condition. Symptoms can even be different among people in the same family. The symptoms can affect any part of our body.

Some common symptoms of 22q11.2 deletion syndrome include:

Heart:	A person's heart may beat too fast or too slow. The heart may have formed differently and may need surgery to fix it.
Lip and/or palate:	A person may have a split in their lip or in the roof of their mouth. They may get a lot of ear infections and have trouble talking.

Immune system:	A person may get sick a lot.
Development:	A person may need extra help in school. He/she may not learn things like other people. He/she may walk or talk later than others.
Mental health problems:	A person may be told they have schizophrenia, depression, bipolar disorder, autism spectrum disorders, oppositional defiant disorder, ADHD, anxiety and mood disorders, and others.
Feeding, swallowing, nutrition:	A person may have a hard time eating. He/she may have a hard time gaining weight.
Facial features:	A person may look like other people who have 22q11.2 deletion syndrome.

Other symptoms of the condition include:

Endocrine:	A person may have problems with thyroid or low calcium levels.
Kidney and urinary tract:	A person may wet the bed a lot or get lots of bladder infections. He/she may have problems with his/her kidneys.
Growth:	A person may be short. He/she may have a hard time growing.
Brain:	A person may have seizures or episodes where they cannot stop his/her body from shaking.
Skeletal:	A person may have a curve in their spine. The bones in their head may form differently.
Blood:	A person may bleed a lot or get bruises easily.

For most people, there is a low risk of having a mental health problem, such as schizophrenia. For people with 22q11.2 deletion syndrome, the chance of having a mental health problem is higher. Unfortunately, the mental health problems of 22q are not talked about as much as the other symptoms, and the research team wants to change that for the better.

We thank you for taking the time to learn about our study. We hope to hear from you soon.