

22q11.2 Deletion Syndrome Fact Sheet - Treatable Psychiatric Illnesses in Adults

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Treatable psychiatric illnesses in adults with 22q11.2 Deletion Syndrome

Treatable psychiatric illnesses are one of the most common features of 22q11.2 Deletion Syndrome (22q11.2DS) in adults. About 6 of every 10 adults with 22q11.2DS have a treatable psychiatric illness. Like several other features associated with 22q11.2DS such as thyroid disease, psychiatric illnesses are very similar to those found in the general population. Individuals with 22q11.2DS just have a greater overall tendency to develop these illnesses. Also like thyroid diseases, psychiatric illnesses are not observable at or before birth. They are important later onset features of the syndrome. Adults with 22q11.2 who develop these illnesses do so at fairly similar ages to others with the same illness who do not have 22q11.2DS, and are treated in similar ways. Appropriate monitoring for these conditions and optimal care once diagnosed will give the best outcomes, as for any illness.

Common psychiatric illnesses in adults with 22q11.2 Deletion Syndrome

Mood and anxiety disorders: The most common psychiatric illnesses in adults with

22q11.2DS are also very common in the general population: anxiety and depression. These illnesses have the same signs and symptoms (e.g., anxiety, depression, irritability and usually several physical symptoms), and are diagnosed the same way as they are for anyone from the general population who does not have a 22q11.2 deletion. The likelihood of developing a treatable anxiety disorder though is higher for someone with 22q11.2DS than for individuals in the general population. Anxiety disorders include generalized anxiety disorder, obsessive compulsive disorder and panic disorder. These may begin in childhood and persist into adult years or may begin later on. All anxiety disorders are treatable with standard medications and/or cognitive-behavioural approaches. Similarly, standard treatments for major depression appear to work just as well in adults with 22q11.2DS as in anyone else.

Schizophrenia and related disorders: The next most common group of psychiatric illnesses in adults with 22q11.2DS are schizophrenia and related disorders, sometimes termed psychotic illnesses. “Psychotic” is often understood to mean that the person is out of touch with reality. This usually involves changes in thinking like delusions (beliefs that are held that are not true) and/or hallucinations (changes in the perception of senses like hearing or vision). Examples of these would include falsely believing that people are following you (a delusion) or hearing voices when there is no one in the room (a hallucination). These are due to changes in the brain.

Schizophrenia and related illnesses like schizoaffective disorder are serious but treatable psychiatric illnesses. Their association with 22q11.2DS has been known for about 20 years, since speech pathologist Robert Shprintzen published a letter in 1992 about the first few individuals from his cohort who had developed psychotic illness. Since then, it has become clear that about one in every four to five individuals with 22q11.2DS will develop schizophrenia or a related psychotic illness. This is about a 20 to 25 times greater chance than someone without the 22q11.2 deletion. It is important to note that the majority of individuals with 22q11.2DS will not develop schizophrenia. However, because the association of 22q11.2DS with schizophrenia is a great source of

concern for individuals, their families and their clinicians, we will highlight some key points that may be helpful.

Monitoring for common psychiatric illnesses in adults with 22q11.2 Deletion Syndrome

Why do individuals with 22q11.2DS, their families and their clinicians need to be aware of and monitor for changes that may indicate the presence of a psychiatric illness? Like almost any medical condition, early detection and effective treatment of psychiatric illness will lessen the long term effects of the illness. These illnesses can occur at any time from childhood through adult years but perhaps the most common time is in the teen years through the 20s. What are the changes to be on the lookout for? Psychiatric illnesses are brain illnesses so many changes to look for involve common brain functions: thinking, emotions and behaviour. Changes in thinking include changes in memory, concentration and attention as well as delusions and hallucinations (changes in perception of reality). Changes in thinking also include the loss of previous abilities to understand other people, like misperceiving or misunderstanding what others are saying, or taking things “the wrong way” more than before. Changes in emotions include greater levels of fear, worry, anxiety or nervousness or a deepening sadness or apathy - just not enjoying things anymore. Changes in behaviour include changes in the individual’s ability to function - at home, in social situations, at school or at work. The other changes that often occur in psychiatric illnesses are physical, like changes in sleep, appetite, weight, and/or energy level. Changes in functioning are also important because they can indicate the severity of the other symptoms; that is, the effect the changes in thinking, emotions, behaviour and sleep or other physical changes are having.

Often, others are better able to see these changes than the person who is experiencing them. The lack of ability to recognize that one has an illness, or loss of insight, is a common feature of psychiatric illnesses. Over-concern and over-monitoring of course is

not desirable. The key is to recognize that important changes have occurred or are occurring and to consult a doctor, ideally a specialist such as a psychiatrist, about these changes to see whether they may indicate a treatable illness.

Diagnosing common psychiatric illnesses in adults with 22q11.2 Deletion Syndrome

Diagnosis of a psychiatric illness in someone with 22q11.2DS is the same as for anyone else. There are no blood tests or x-rays that are diagnostic - the key is an accurate history of how the person is now and how they were before. The other important things for the doctor to consider when evaluating the signs and symptoms that are present are the other features of 22q11.2DS. This would include the endocrine disorders such as hypothyroidism or hypocalcemia/hypoparathyroidism as well as seizure disorders, for example. These other conditions may or may not have been previously recognized and treated. Possible effects of any treatments the person may already be receiving for other medical conditions should also be considered when determining whether the adult with 22q11.2DS has or is developing a treatable psychiatric illness.

Other illnesses that are common in the general population may also be present in individuals with 22q11.2DS. These would include alcohol and drug abuse. Also, individuals can have more than one psychiatric illness, such as major depression and alcohol abuse. Less common psychiatric illnesses are also found in adults with 22q11.2DS, as they are in the general population. In all cases, the illnesses would be assessed, diagnosed and treated in a standard manner.

Predicting and preventing psychiatric illnesses in adults with 22q11.2 Deletion Syndrome

There are no known predictors of psychiatric illness, including schizophrenia, in someone with 22q11.2DS. For example, the intellectual level of the person is not a

predictor. Whether the person had attention deficit disorder as a child does not predict later onset of schizophrenia. Indeed, there are no clinically proven predictors of psychiatric illness in anyone without a 22q11.2 deletion either. This is however a great source of current research interest.

There are also no proven prevention strategies for psychiatric illness. Good advice though is to avoid marijuana and other drug and alcohol abuse. Marijuana use, especially heavy early marijuana use, for example, in the general population shows an increased association with schizophrenia. Good nutrition and exercise are also recommended as general health measures, as for anyone.

Knowledge is Hope

Treating psychiatric illnesses in adults with 22q11.2 Deletion Syndrome

Psychiatric illnesses in general are treatable conditions. Schizophrenia in particular is often a challenging illness to manage, whether or not one happens to have a 22q11.2 deletion. However, by carefully following standard clinical practice guidelines for schizophrenia, and paying close attention to the individual's physical health issues, most individuals with 22q11.2DS who have illnesses like schizophrenia will improve. This is based on many years of experience of psychiatric specialists who follow patients with 22q11.2DS and schizophrenia.

Psychiatric illnesses are no different from the other conditions associated with 22q11.2DS, including the heart and palatal abnormalities and the thyroid problems. As for almost all medical conditions, there is currently no cure but there are effective treatments and management strategies. All treatments have the potential to have some side effects. However, standard management for the individual condition is generally effective. The key in 22q11.2DS is to be aware of the individual's other associated conditions and important general issues in adults with the syndrome. One example would be the lowered seizure threshold in 22q11.2DS. This means that the doctor would ensure that hypocalcemia is monitored and treated and would more often use

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anti-seizure medications in addition to anti-psychotic medications. In this way, the doctor can help to prevent the person from having a seizure.

While clinics specializing in adults with 22q11.2DS may be a preferred option for diagnosis and treatment of later onset conditions, there are currently very few such clinics available. The key ingredients to the successful treatment of psychiatric illnesses in individuals with 22q11.2DS are the willingness of the person to have the necessary assessments and to follow the recommended treatments, and the help and support of caregivers and a doctor who is interested in the person and in the many facets of 22q11.2DS.

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