Mental Health Disorders in 22q11 DS

Give yourself plenty of time to read this leaflet and do get in touch with us if you have any queries or concerns. The purpose of this leaflet is to give you a broad picture of the areas more frequently seen in people with 22q11DS but this should not prevent you from seeking specialist advice if the things you are thinking about are not covered here. The information has been drawn mostly from the Max Appeal Consensus Document of the Diagnosis and Management of 22q11DS. This document and references are available.

The thought that a member of your family or your friend could suffer with mental health problems is unpleasant and disturbing, unfortunately these issues are fairly common for people with 22q11DS; the brain is an organ and so problems can arise just like any other illness. It should also be remembered that psychiatric issues are common in the general population, especially for people with an intellectual disability or learning difficulty, which can cause emotional and behavioural issues too or people can have those as well.

What are mental health disorders?

Medicine is often about opinion and this is nowhere more so than in the field of mental health. Here's an example of a definition:

> "Abnormal and enduring thoughts, feelings, sensory impressions and behaviour that cause distress (to the individual or those around him/her) and interferes with personal functions." Or this can be expanded to: "Impairment of one or more important areas of functioning." (For 'functioning' read 'life'.)

Psychiatric issues and mental health problems are complex and often there aren't concrete facts or numbers, unlike a blood test. "Diagnoses" are simply a name given to certain symptoms and indicate the support needed and are the same as for people without 22q11DS of the same age and encountering the same issues. Try to avoid thinking in terms of 'labels'.

People with 22q11DS are more likely than others to encounter psychiatric problems throughout their life which is of concern and worry to families. So, whilst it is important to be aware that this problem may arise, try not to focus on it in daily life so that anxiety becomes a problem in itself.

It is, on the other hand, imprortant to act swiftly if you feel psychiatric issues are starting to appear because with timely recognition of problems and appropriate interventions (which might be changing things at home or work, psychological support, as well as medication in some situations) symptoms can usually be managed so that they are less distressing and do not limit a person's activities, achievements and relationships.

There are different categories of diagnoses, and sometimes people experience symptoms from a number of those. This is called co-morbidity. At other times a person might have just one or two symptoms, not fitting a particular diagnostic pattern, but still causing disruption to their day-to-day life and therefore need from support, which can be frustrating as there is no clear cut 'diagnosis'.

As children grow into adulthood there are the usual emotional and behavioural upheavals because of chemical and hormonal changes. People with 22q11DS and psychiatric issues have behaviours and symptoms that change, and consequently the diagnoses can change too.

People with 22q11DS have many differing behavioural and emotional symptoms, these are often unique to that person but this is more often the case than those without 22q11DS.

Is it possible to know whether a person is more or less likely to develop psychiatric problems?

Sadly, the answer to that is 'not really'.

However, young people who show behavioural and emotional problems at a younger age are more likely to have more symptoms throughout life. But this is not always the case - sometimes problems can be quite severe during childhood but then improve; for other individuals, major symptoms can appear later in adolescence or adulthood "out of the blue".

Importantly, no specific symptom or group of symptoms automatically mean that a person will develop problems later in life. There is no correlation between any physical features of 22q11DS and psychiatric problems or any degree of learning difficulty.

People with 22q11DS who are suffering psychiatric issues should, in addition to being treated by a psychiatrist, have their cardiac, calcium and other endocrine functions monitored and treated, as well as looking at diet and exercise.

What should trigger a referral for a mental health assessment for a child or adult with 22q11DS?

People who attend a specialist 22q clinic will be monitored regularly by a psychologist or psychiatrist but any doctor carrying out a general health review should be made aware that they need to ask some basic questions about emotional, behavioural and social well-being. This is an important part of the multi-disciplinary support needed for all people with 22q11DS.

If there are concerns about any distressing or disruptive behaviours, thoughts or emotions, then you should be encouraged to talk about it openly and reach out for help and support. Sometimes a simple chat with a GP or paediatrician may help to put an emotional symptom into perspective, and consider whether it is a problem or part of the patterns of behaviour that change as people move through their life, but if that doctor has an uncertainty then they should discuss the situation with a mental health professional before making a decision about a specialist referral

A key trigger is a significant change in behaviour, for example withdrawal from normal activities, seeming unhappy, afraid or disorientated, or acting in a way that is out of character. It should be remembered that people experiencing psychiatric symptoms often find it difficult to explain what they are feeling, and this may be especially true in 22q11DS.

People with 22q11DS are more likely to show signs of distress during times of change in their lives, and may require more support than other individuals in adjusting to change.

A drop in cognitive ability is noted as being a sign of the onset of psychiatric issues, this might be easier to spot in school aged children but, unfortunately, it is not frequently measured by schools.

The important thing to focus on is not knowing what the 'trigger' was or if there was one, but seeking advice and support at an early stage. Generally, problems treated earlier are more likely to respond effectively to intervention.

Are people with 22q11DS assessed differently to other people?

No, other than appropriate methods for age, development and communication abilities.

It is important for doctors to ask children, teenagers and adults with 22q11DS about their own symptoms (in a sensitive manner appropriate to their age and understanding), as well as seeking information from parents, carers and teachers. It is better to have the same person carrying out any assessments.

Are people with 22q11DS treated differently to other people?

Treatment of psychiatric symptoms in 22q11DS always requires an individually-tailored approach. However, there is no evidence that any particular treatments (medications or psychological approaches) are more effective for 22q11DS than for other individuals. Since there is a lack of research regarding the treatment of mental health problems in people with 22q11DS, often guidelines written for the general population, like the NICE (National Institute for Clinical and healthcare Excellence) guidelines, are being used.

Potential side-effects of drugs must be considered very carefully. Special attention should be paid to the use of medications that can lower seizure threshold or have potential cardiac side effects or affect blood counts. Particular reference is made to the antipsychotic drug clozapine and Ritalin, which is a stimulant medication (usually for ADHD), for which precautions and specialist advice should be considered.

People with 22q11DS may not respond as well as others to drug treatment, perhaps because they find it more difficult to say how they are feeling and that other medical conditions related to 22q11DS makes them more likely to suffer from side effects. There are NICE guidelines for the treatment of challenging behaviour in people with learning disabilities and/or autism. Also, people with 22q11DS are more vulnerable to psychiatric symptoms returning and may require longer term follow-up and on-going support.

It is advised that health care professionals who have experience of 22q11DS are consulted in order to maximise the benefit and reduce risks. However, it is probably more important to have easily-accessible local support (with specialist advice) than for treatment to be managed at a distance. These issues are potentially very debilitating and it is very beneficial for all professionals (including educators) to support the person and their family through these difficult situations.

Are people with 22q11DS more likely to suffer from mental health issues?

Unfortunately, yes. In the general population about 26% of people will suffer from mental health disorders, for people with 22q11DS this might be somewhere between 60% and 93%.

Compared to people with general intellectual disabilities, people with 22q11DS are more likely to have ADHD, anxiety disorder, mood disorder and psychotic disorders.

What are the specific problems that affect people with 22q11DS?

The sections below are not exclusive, but try to cover the main conditions and what treatments should be available.

1. Attention-Deficit/Hyperactivity Disorder (ADHD)

Just over 5 in 100 people in the general population suffer with ADHD as compared to about one third to almost a half of people with 22q11DS. Though it seems that inattention or poor concentration rather than hyperactivity is more common; and this is the case for both males and females.

What is ADHD?
ADHD is a pattern of inattention and/or hyperactivity-impulsivity seen for at least six months and for people with 22q11DS the symptoms must be more severe than for others and be seen in at least two settings causing interference on social interactions, a person's job or school work.

These are the sorts of symptoms a child might have:

- fail to give close attention to details and make careless mistakes in schoolwork
- not seem to listen when spoken to directly
- not follow through on instructions and fail to finish schoolwork or chores
- avoid, dislike, or are reluctant to engage in tasks that require sustained mental effort.

Children with 22q11DS and ADHD are also far more likely to suffer with other behavioural problems.

What is the treatment for ADHD?
There are NICE guidelines for the treatment of ADHD which combines a behavioural approach with medication. Ritalin is thought to be generally effective, though opinion on dosage varies and it is recommended that a comprehensive cardiac evaluation is made.

2. Autism Spectrum Disorders (ASD)

In the general population prevalence rates less than 2 people in 100 will have autism. But between 14 to 50 people in 100 with 22q11DS will have autism (that's very high). Though the social communication difficulties are slightly different in people with 22q11DS and sometimes not considered 'typically autistic'.

What is autism?
People with autism spectrum disorders have problems with social interaction and communication and have restricted repetitive and stereotyped pattern of behaviour, interests and activities. Usually social interactions, imaginative play and language are noticed to be poor before the age of 3 years. There is an impairment in functioning in either social interactions or language (the way it is used in social communication) or symbolic of fantasy play before the age of 3.

The symptoms listed in the DSM-IV (a psychiatrists' 'manual') are:

- impairment in using nonverbal behaviour
- not succeeding in making relationships with people of the same intellectual level
- lack of sharing pleasures and activities with others
- lack of social or emotional reciprocity
- delay or lack in the development of verbal language
- impairment in the ability to start or sustain a conversation
- stereotypical and repetitive use of language
- use of idiosyncratic language
- lack of varied, spontaneous fantasy play or social imitative play
- strong preoccupation with one of more stereotyped patterns of interest
- adherence to specific non-functional routines or rituals
- stereotyped and repetitive motor mannerisms
- preoccupation with parts of objects.

What are the treatments for Autism Spectrum disorders?
Specific guidance for people with 22q11DS has not yet been developed. There are therapies to improve symptoms in general population such as social-skills training, communication interventions. Also medication to address issues that often go hand in hand with autism such as attention deficit and anxiety. The specific pattern of social strengths and difficulties varies for each person with 22q11DS, and a supportive approach that builds social confidence is encouraged.

3. Generalised Anxiety Disorder (GAD)

About 3 in 100 people in the general population have GAD. Between 10 and 30 in 100 of people with 22q11DS will have GAD. Anxieties may be present at any age, and are often a persistent feature during late childhood and adolescence.

What is GAD?
People with GAD suffer from excessive anxiety and worries for more than half the time during a period of at least six months.

The symptoms are:

- a feeling of restlessness or of feeling keyed up or on edge
- being easily fatigued
- difficulties concentrating or mind going blank
- irritability
- muscle tension
- sleep disturbance.

In adults the worries and anxiety cause at least three of the above and in children one for a diagnosis to be made.

General anxiety disorder is different to conditions such as post-traumatic stress disorder, or anxiety caused by substance abuse or a general medical condition. GAD causes significant distress or reduced ability in social settings and work environments.

What is the treatment for GAD?
Treatment depends on a person's level of understanding. Communication strategies, such as a visual timetable can reduce uncertainty and Cognitive Behaviour Therapy may be effective. There is also emphasis on positive behavioural support. There isn't any advice on the use of medications specifically for 22q11DS people with GAD.

5. Specific phobia

In the general population this affects about 9 in 100 people but almost a quarter to over half of people with 22q11DS are thought to suffer with specific phobias.
A specific phobia is a marked and persistent fear towards a thing or event, caused by a specific thing or event. Adults with a specific phobia acknowledge that the fear is excessive or not reasonable; children do not recognise this.

There a five 'types' of phobias:

- animal
- natural environment
- blood-injection-injury
- situational
- 'other'

Most children with 22q11DS who had a specific phobia had fear of the dark, fear of lightning/thunder and/or animals.

What is the treatment for a specific phobia?
There is no treatment of a specific phobia just for people with 22d11DS. People with intellectual disabilities may respond to graded exposure to the phobia but the treatment for the general population is based on 'exposure' techniques.

6. Major Depression

Just under 6 in 100 generally suffer with major depression. In the 22q11DS population. Somewhere between 6 and 20 in 100 people with 22q11DS suffer from major depression, but it peaks during teenage years. Also people with 22q11DS have some of the symptoms of major depression with symptoms of other conditions.

What is major depression?
It is a period of at least two weeks in which a person experiences depressed mood and/or loss of interest or pleasure, and at least three of the following symptoms:

- weight loss or weight gain
- insomnia or hypersomnia
- psychomotor retardation or agitation
- fatigue or loss of energy
- feelings of worthlessness or guilt
- diminished ability to think or concentrate or indecisiveness
- recurrent thoughts of death or suicidal ideas.

In children and adolescents their mood can be irritable instead of depressed and children might fail to make weight gains.

What is the treatment for major depression?
In the general population the NICE guidelines for the general population advise anti-depressants and/or psychotherapy (counselling).

One report found find that there is considerable evidence that the medications called Selective Serotonin Reuptake inhibitors (SSRIs) may be useful for people with learning difficulties, starting with a low dose. However SSRIs and other medications may cause a person to become aggressive, destructive and self-harm.

On the other hand SSRIs may be beneficial for children and teenagers with learning difficulties. But there is the concern that antidepressants could increase suicidal feelings.

Like we said earlier; there is no clear cut answer.

7. Schizophrenia

Only 1 in 200 people in the general population will be affected by schizophrenia. For people with 22q11DS this figure is significantly higher with just under a quarter of young adults and more than 80 in 200 people aged over 25 years will suffer from schizophrenia spectrum disorders.

It is important to recognise that schizophrenia is a complex diagnosis, including many different symptoms which can affect each individual in a different way, with symptoms that can change over time, but it can be effectively managed with specialist support.

What is schizophrenia?

Schizophrenia is diagnosed when a person experiences two of the following symptoms for one month during a significant part of the time:

- delusions
- hallucinations
- disorganised speech
- grossly disorganised or catatonic behaviour
- negative symptoms like apathy or social withdrawal.

People with 22q11DS and schizophrenia are more impulsive, uncooperative and hostile than the general population.

Conditions such as hypoparathyroidism and hypothyroidism (thyroid hormone imbalances) could mimic psychotic features and this should be checked by an endocrinologist.

Risk factors for developing a psychotic disorder in 22q11DS continue to be investigated but might be related to lower verbal IQ scores, anxiety or symptoms of depression earlier in life or experiencing psychotic symptoms earlier in life.

What is the treatment for schizophrenia?

There are guidelines for the use of the new generation of antipsychotic drugs (not clozapine) for people with intellectual disabilities.

There is little research into the specific treatment of schizophrenia in people with 22q11DS. There is some research in to the use of clozapine but there appear to be significant side effects. It is also noted that some antipsychotic drugs cause arrhythmias (abnormal heart rhythms) in people with 22q11DS. Max Appeal recommends that specific advice should be taken from a psychiatrist who is familiar with the Consensus Document and is experienced in dealing with schizophrenia in people with 22q11DS.

8. Obsessive Compulsive Disorder (OCD)

Around 1 in 100 people generally suffer with OCD. For people with 22q11DS this may be about 5 or 6 in 100 of young people and adults, though various studies have put forward much higher numbers.

What is OCD?

Adults who have obsessions or compulsions recognise that they are not reasonable or that they are excessive. Symptoms of OCD in people with 22q11DS include contamination, aggressive obsessions, worries about somatic problems, hoarding, asking repetitive questions and cleaning. NB Somatoform disorders are characterised by physical symptoms that occur without an adequate physical cause. This is not malingering.

What is the treatment for OCD?

In the general population OCD is often treated with either medication (serotonin reuptake inhibitors - SSRIs) or with cognitive behavioural therapy. Specific treatment for people with 22q11DS is not known.

A final word!

This leaflet makes pretty grim reading for anyone who is concerned about a friend or loved one. BUT the 'take home' message is that mental disorders/psychiatric issues do not go away without help. The sooner a person recieves treatment the better the potential outcome in the long term.

It has been estimated that, for example, with schizophrenia (and let's be honest, this is the one that makes you feel very frightened) for every 5 people who develop schizophrenia, within five years of their first 'episode':

- 1 will get better.
- 3 will get better but will still have some symptoms.
- 1 will continue to have troublesome symptoms.

Getting help as early as possible has to make sense. A carer, friend or relative may well think of all the things they feel about mental health issues but this is about the person that they would like to help and doing nothing isn't often the best course of action. Call Max Appeal if you want someone to talk to in confidence about these very sensitive issues.