

**22q11.2
Deletion
Syndrome:
What does it mean
for mental health?**



Who is this booklet for?

This booklet was developed as a resource about 22q11.2 deletion syndrome (22qDS). It contains information about how 22qDS can impact mental health and provides practical strategies and recommendations to protect and improve mental health.

This is not a comprehensive guide to treating mental illness and you should contact your local mental health providers for further guidance specific to your situation.

This booklet was developed to be a reliable source of information for families. It was developed by genetic counselors and families who live with 22qDS working together.

This booklet is intended to be a supplement to genetic counseling. You can find a genetic counselor in your area using the National Society of Genetic Counselors' Find a Genetic Counselor tool at nsgc.org.



What is 22qDS?

22qDS (22q11.2 deletion syndrome) is also called Velocardiofacial syndrome (VCFS) and DiGeorge syndrome (DGS). About 1 in 4000 people have 22qDS. It is a condition caused by a small missing (deleted) piece of chromosome 22. This missing piece of genetic material can contribute to developmental and/or health issues. There is a lot of variation between people with 22qDS in terms of whether they have health issues, and how serious these are.

Some of the more common health issues that an individual with 22qDS may develop are listed below. Not everyone with 22qDS will have all of the health issues listed.

- Heart defects
- Cleft palate (an opening at the roof of the mouth)
- Learning difficulties
- Immune system problems (being more vulnerable to infections)
- Differences in facial appearance

Additional health issues that may occur are:

- Growth delays
- Feeding difficulties
- Kidney problems
- Low levels of calcium
- Hearing loss
- Developmental delays
- Speech delay
- Seizures
- Bone differences
- Eye problems
- Gastrointestinal problems
- Mental illness*

*This booklet will focus on mental illness.



What is mental illness?

Mental illness refers to a wide range of conditions that affect how people think, feel and/or behave. Mental illness can cause distress but there are a variety of treatments available, and people can recover. Even if a person does not experience mental illness, it is important for everyone to take care of their mental health. A selection of mental illnesses are described below:

Anxiety Disorders

Anxiety disorders involve excessive fear or anxiety beyond that of normal nervousness or anxiousness, and may include specific phobias or panic disorders.

Schizophrenia

Schizophrenia involves delusions (beliefs that are not based in reality), hallucinations (e.g. seeing or hearing things that are not there), and trouble with thinking and concentration.

See page 15 for more information about schizophrenia.

Mood Disorders

“Mood disorders” is a term that covers both depression (low mood) and bipolar disorders (periods of low mood and periods of “high” or irritable mood).

Schizoaffective Disorder

Schizoaffective disorder involves a combination of symptoms of schizophrenia and symptoms of mood disorders.

Obsessive Compulsive Disorder (OCD)

OCD involves symptoms that include uncontrollable, reoccurring thoughts (obsessions) and behaviors (compulsions) that people engage in to try to cope with that obsession.

What causes mental illness?

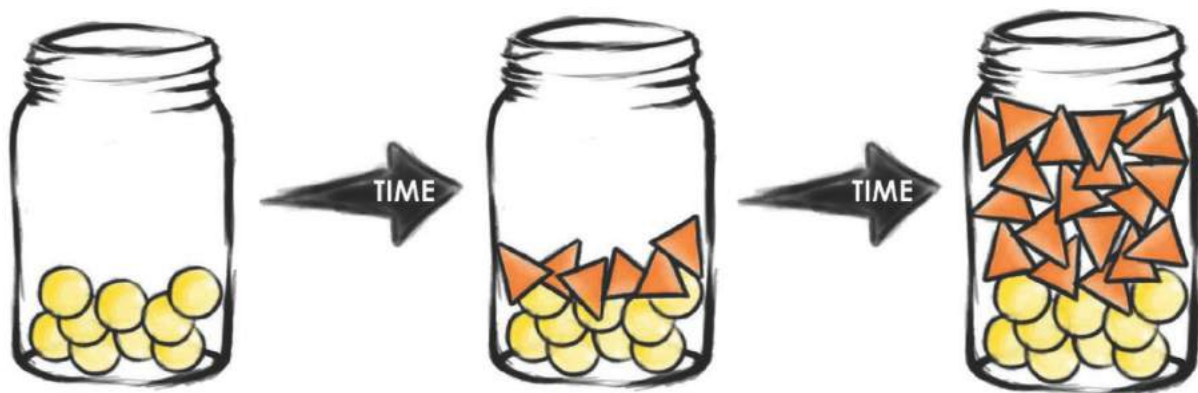
The Mental Illness Jar

Most of the time mental illness does not have one single cause. Mental illness is typically caused by a combination of genetic and environmental factors (“nature” and “nurture”).

The following pictures use a jar to represent how **genetic (inherited) factors** and **environmental factors** work together to cause mental illness. Everyone has a mental illness jar, which can be filled with both genetic and environmental factors.



If the mental illness jar becomes full, a person experiences an episode of mental illness. If the jar is not full, a person is not experiencing an episode of mental illness. The closer the jar becomes to being full, the more vulnerable to mental illness the person is.



Genetic Factors

Genetic factors are represented in the jar as yellow balls. Research has shown that likely everyone has some genetic factors for mental illness. There are different types of genetic factors for mental illness. The most common type of genetic factor that people can have is represented by a small yellow ball. Some people have a small amount of them and some have a large amount, but most people have a medium amount. Each one of this type of genetic factor makes only a very small difference to whether a person develops mental illness.



22qDS - The Big Yellow Ball

Missing or extra pieces of chromosomes - like the one that causes 22qDS - are a different type of genetic factor for mental illness. These are less common in the general population, but they can make a bigger difference to whether or not a person develops mental illness.



We use a large yellow ball in the mental illness jar to represent 22qDS because studies have shown that this genetic factor makes a bit of a bigger difference to whether or not someone develops mental illness.

People with 22qDS likely have other smaller yellow balls too (just like everyone else does) in their mental illness jar.



Environmental Factors

Environmental factors can be thought of as “life experiences”. We don’t know what all of the environmental factors for mental illness are, but some of the ones we do know about are listed below.

We are all different in how sensitive we are to environmental factors, like stress. Stress can include both positive and negative experiences. People who are more sensitive may be more likely to develop mental illness after a stressful experience. The differences in how sensitive people are might be due to having different amounts of genetic factors in one’s mental illness jar.

Environmental factors that make a person more likely to have either **schizophrenia** or **schizoaffective disorder** or **bipolar disorder**:

- Being born in winter months
- Experiencing stressful life events
- Taking certain illegal drugs (like cannabis or methamphetamine)

Environmental factors that make a person more likely to have **schizophrenia**:

- Being born after a difficult pregnancy or delivery
- Being brought up in a large city
- Being an immigrant
- Having a childhood head injury

Most of these experiences make it only slightly more likely that a person will develop mental illness. For example, for the average person in the general population, the chance to develop schizophrenia is about 1% (1 out of 100 people). But, someone whose birth was difficult is twice as likely to have a mental illness, like schizophrenia, than someone whose birth was not difficult. This means that a person whose birth was difficult has a chance to develop schizophrenia of 2% rather than 1%.



Environmental Factors

22qDS

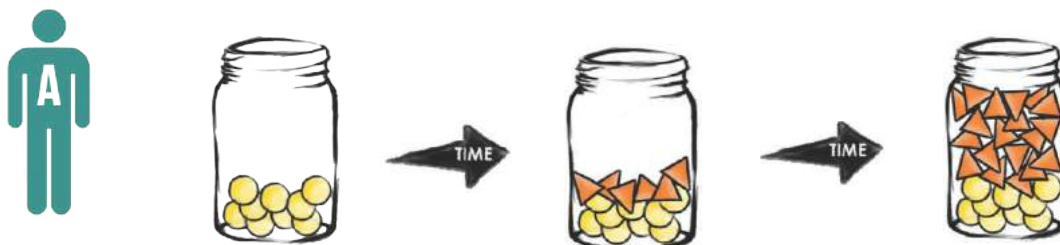
Below are some environmental factors that a person with 22qDS may be more likely to experience than a person without 22qDS.

- Frequent medical appointments or visits to the hospital
- Difficulties making friends
- Feeling different than their peers or siblings, or experiencing bullying
- Finding school stressful or difficult

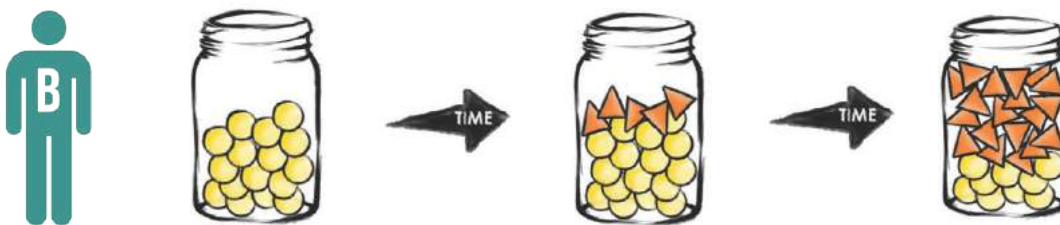
Putting It All Together

Mental illness can be caused by different combinations of genetic and environmental factors. It is not anyone's fault when someone develops a mental illness.

Person A has a small amount of genetic factors, and has experienced a large amount of environmental factors that have filled up their mental illness jar.



Person B has a larger amount of genetic factors and a smaller amount of environmental factors which has resulted in their mental illness jar being filled.



Person C has 22qDS and has some other, more common, genetic factors as well. They have also experienced environmental factors, which filled up their mental illness jar.



A person who has a large amount of genetic factors – including individuals with 22qDS – may be more likely to develop mental illness than someone who has only a small amount.

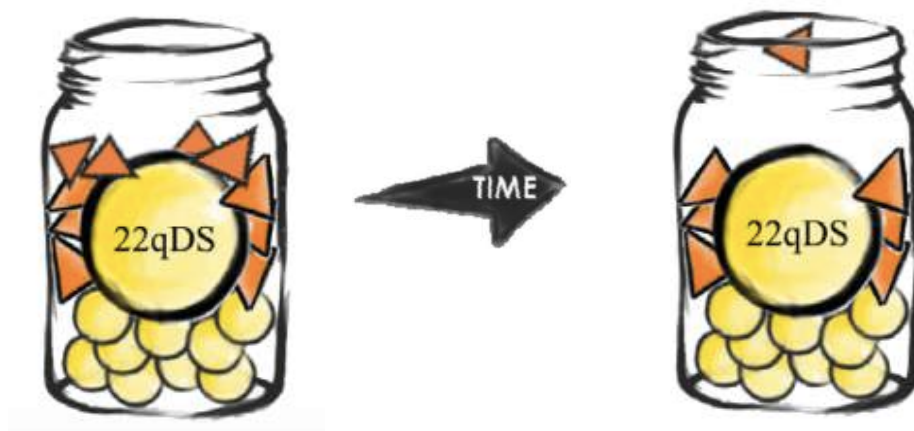
But, a person with 22qDS may not develop mental illness.

Protecting Mental Health in People with 22qDS

People with 22qDS can do things to protect their mental health.

The rest of this booklet will focus specifically on how to take care of mental health. Most of the concepts apply for people without 22qDS as well as those that have 22qDS.

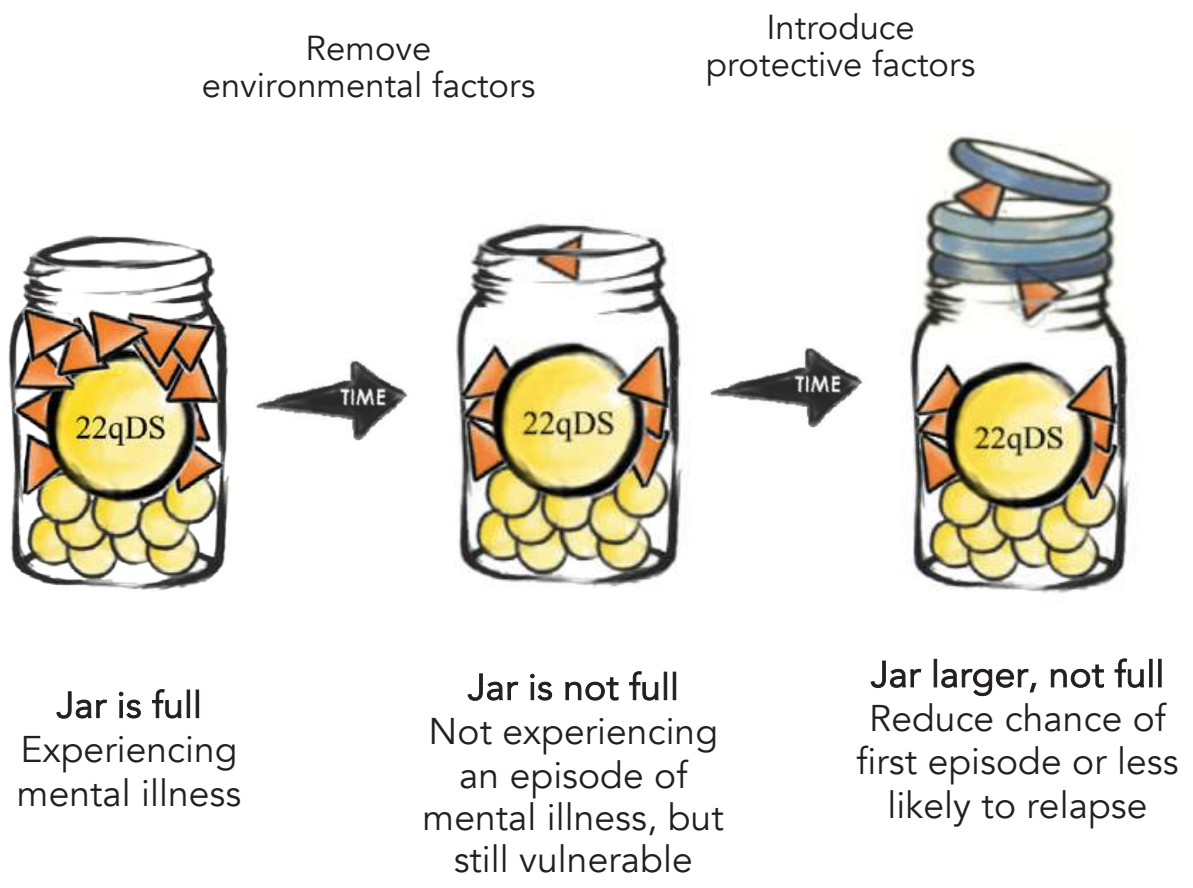
People cannot change their genetic factors; however, some environmental factors can be avoided, changed or removed from the mental illness jar. Someone might remove environmental factors by:



- Finding ways to reduce stress
 - See page 12 for some examples of activities to reduce stress
- Not taking drugs like cannabis and methamphetamine

Protecting Mental Health in People with 22qDS

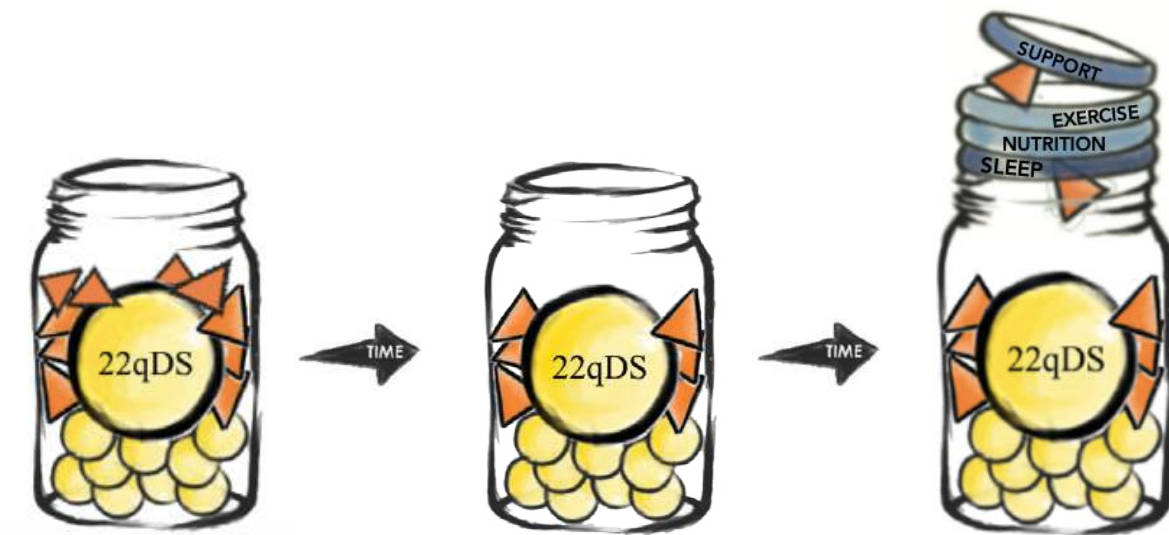
People with 22qDS can also protect their mental health by adding protective factors onto their jars. Protective factors can be thought of as rings that stack on top of the jar to make it bigger. This makes it less likely for the jar to fill to the top. Protective factors can make more room if someone's jar is already full and help with recovery.



Protecting Mental Health in People with 22qDS

Research has shown that the following things are protective of mental health in everyone:

- **Exercise:** some form of regular exercise or physical activity
- **Nutrition:** regular meals and a healthy balanced diet
- **Sleep:** adequate and consistent sleep
- A good **support system** of people you trust



People with 22qDS are also recommended to have **regular check-ins with a mental health professional** (psychiatrist or psychologist) **starting in early childhood** (i.e. before age 5). These visits can help monitor the individual for any symptoms and can connect parents with appropriate treatments and resources. Early intervention is one of the best ways of improving outcomes for people with mental illness. This professional may be part of a 22qDS clinic or care team, or may be a separate part of your or your child's care.



Protecting Mental Health in People with 22qDS

As well as the protective factors that work for all of us, there are others that different people may find helpful. Some examples are listed below:

- Stretching
- Yoga
- A hot bath
- Meditation
- Spending time with a pet
- Talking to someone
- Taking a break from the stressor
- Doing something that makes you laugh
- Listening to your favorite music
- Looking through old family photos
- Deep breathing
- Reading a book
- Working on a coloring book
- Painting
- Gardening
- Going to the park
- Dancing
- Making a craft
- Playing a game
- Asking for help
- Work through a mental health workbook
- Make a mental health plan

It is important for individuals to find what works best for them and their family. Encouraging healthy habits and having a safe space to discuss feelings can be very helpful for protecting a person's mental health.

What is the chance for a person with 22qDS to develop mental illness?

Mental illness is common; the chance for someone without 22qDS to develop any mental illness is ~20%. The most common mental illnesses are depression and anxiety, and 1% of people develop schizophrenia.



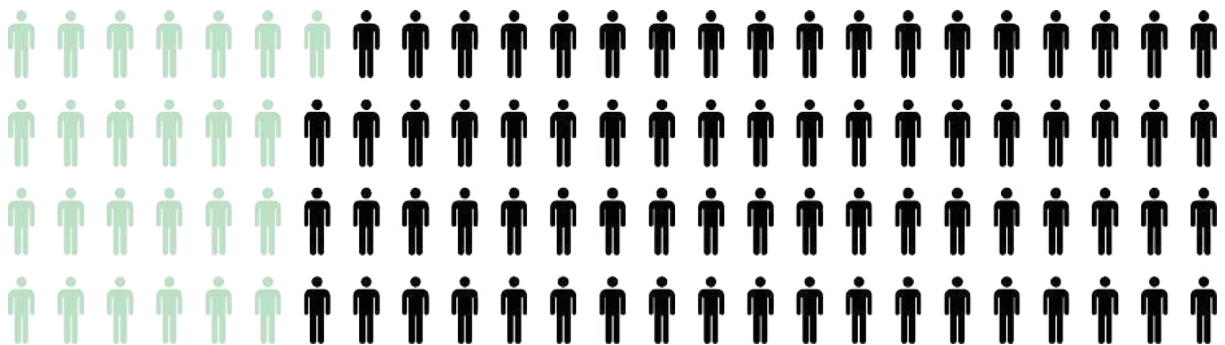
A person with 22qDS is more likely to experience mental illness than a person without 22qDS, but the chance of developing a mental health problem is difficult to estimate. The most common mental illnesses, like for someone without 22qDS, are depression and anxiety.



More studies have been done about schizophrenia and 22qDS:

About 25% (1 in 4) people with 22qDS will develop schizophrenia. This means that most people (75% or 3 out of 4 people) with 22qDS do **not** develop schizophrenia.

*See page 15 for information about schizophrenia.



Out of 100 people with 22qDS, approximately 25 will develop schizophrenia in their lifetime and 75 will not develop schizophrenia.



22qDS and Mental Illness Facts

When mental illness happens, the symptoms most often start during the **late teens to early 20's**. This is similar between people with and without 22qDS.

Early diagnosis and treatment for mental illness leads to the best outcomes for people over the long term. This is why regular mental health monitoring is important for children with 22qDS.

*See page 16-17 for how to recognize emerging symptoms of mental illness and pages 11-12 for protective and wellness factors.

Researchers are trying to determine whether there are treatments for mental illness that are specific to 22qDS. Currently, mental illness treatment options are similar between people who do and do not have 22qDS.

Individualized treatment plans for mental illness can be developed between mental health professionals and the family. Possible treatment options include:

- Medication
- Different types of talk therapy (psychotherapy), including Cognitive Behavioral Therapy (CBT)
- Psychoeducation
- An in-patient stay in the hospital

Treatment often uses a combination of therapy and medication.



Schizophrenia

Schizophrenia can cause a change in the way a person thinks, feels, and behaves. During an active episode, a person with schizophrenia may be unable to distinguish between what is real and what is not real. Types of symptoms that may occur in schizophrenia are listed below.

Positive symptoms are changes in thoughts and behavior that are “added” to a person’s experiences. These symptoms are often referred to **psychosis** and represent a loss of touch with reality. Some examples are:

- Hallucinations (hearing, seeing, feeling, tasting or smelling something that is not there)
- Delusions (false or mistaken beliefs that are firmly held)
- Paranoia


Negative symptoms are thoughts or behaviors that exist before a person gets sick, but are not present once a person is ill.

- Less ability to plan, reduced speech or difficulty expressing emotion

Cognitive symptoms

- Problems with attention, concentration or memory
- Confused or disordered (“disorganized”) thinking and speech

Symptoms of schizophrenia typically begin between the ages of 16 and 30. People with schizophrenia may not be aware that they are developing symptoms of mental illness, so family or friends may be the first to recognize changes and seek help.



Recognizing Emerging Symptoms of Mental Illness


Early intervention (treatment) has been shown to improve long-term outcomes for people with mental illness. Recognizing emerging symptoms of mental illness can help with getting early treatment. Examples of emerging symptoms are listed below:

Anxiety:

- Constantly worrying or having negative thoughts
- Sleep or appetite changes
- Complaining of physical ailments (e.g. stomach aches and headaches)
- Feelings of panic (e.g. heart racing or difficulty breathing)
- Problems thinking or troubles concentrating
- Drop in functioning (for example in grades or school performance)
- Avoiding specific situations (e.g. school or work)
- Irritability

Mood Disorders:

- Lack of interest or enthusiasm for things that used to be interesting or enjoyable
- Mood changes/extreme shifts between highs and lows
- Feeling disconnected
- Sleep or appetite changes
- Irritability
- Drop in functioning (for example in grades or school performance)
- Difficulty Concentrating
- Low energy
- Withdrawal/spending more time alone than usual



Recognizing Emerging Symptoms of Mental Illness

Psychosis:

In addition to the symptoms listed on the previous page, emerging symptoms of psychosis (schizophrenia) may also include:

- Increased sensitivity to smell, sound, touch, etc.
- Illogical thinking
- Difficulty telling what is real from what is not
- Nervousness or suspiciousness beyond what is typical for them
- Unusual behavior
- Loss of personal hygiene or self-care

A person who experiences some of these symptoms does not necessarily have an emerging mental illness. But if you are concerned about how frequent or severe these symptoms are, or about a sudden change in behavior, it can be helpful to check with a mental health professional, like a psychiatrist or psychologist.





Things To Remember

1. Mental illness is caused by BOTH genes and environment. Usually, inherited (genetic) factors and environmental (lifestyle) factors contribute together to cause mental illness.
2. 22qDS is one genetic factor that contributes to someone's susceptibility to mental illness, but does not by itself cause mental illness.
3. A person who has a larger amount of genetic factors (e.g. a person with 22qDS) may be more likely to develop mental illness than someone who has a smaller amount. But, not everyone with 22qDS will develop a mental illness.
4. People with 22qDS who have had an episode of mental illness can recover. There are treatments for mental illness.
5. Environmental factors can sometimes be removed from the mental illness jar, but we cannot control or prevent all environmental factors.
6. There are things to protect mental health for the future. Some suggestions are listed on pages 11 and 12. Protective factors can reduce the chance of experiencing a first episode of mental illness or make it less likely that another episode occurs.
7. If you are worried that someone might have symptoms of mental illness, get help quickly. Options listed on pages 19 might help.
8. Regular mental health check-ins should be a routine part of care for people with 22qDS.



Mental Health Resources

Emergency mental health resources:

- For anything urgent or life-threatening call 911 or your local emergency number immediately.
- Find a Crisis Line in your area. For help finding a Crisis Line visit yourlifecounts.org.

Routine, ongoing mental health resources:

- See a psychologist or psychiatrist regularly, starting in early childhood if possible.
- Seek help from your primary care provider, 22qDS team, or other health care provider.

Taking care of your own mental health:

- Reach out to close friends or loved ones.
- Contact a minister, spiritual leader or someone in your faith community (if applicable).
- Check out some of the resources on the next page which might be helpful for you.



Mental Health Resources

Organizations that you might find helpful include:

- Canadian Mental Health Association (CMHA)
- Anxiety Canada
- National Alliance on Mental Illness (NAMI)
- Anxiety & Depression Association of America (ADAA)
- The International 22q11.2 Foundation
- Rare Disease Foundation

Meditation or mindfulness websites/apps such as:

- Headspace
- Calm
- Stop, Breath & Think

Kid-friendly books and resources include:

- Mighty Moe: An Anxiety Workbook for Children
- Taming Worry Dragons
- What To Do When You Worry Too Much



Thank You

This resource was created by Stephanie Chieffo, Emily Morris, and Caitlin Slomp, with input from parents of individuals with 22qDS, and designed by Vanessa Macdonald.

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