International 22q11.2 Foundation

Newsletter June 2023

Philadelphia, PA, USA

22q at the Zoo – Thank You!

The International 22q11.2 Foundation would like to thank every one of our 22q at the Zoo collaborators including 22q organizations, healthcare providers, basic scientists, industry partners, and especially families for your selfless service to our 22q community – with a total of 22 international locations and 50 US sites participating in 22q at the Zoo Worldwide Awareness Day in 2023!

As you have witnessed over the past few weeks, a social function intended as a friend raiser can have an enormous global positive impact. We know, based on previous feedback, that 22q at the Zoo can help carry families through struggles emerging between zoo events. This includes our local Philadelphia-area participants, those coming together at events large and small on the 3rd Sunday in May or any other day around the world, as well as those who cannot attend in person but witness the magic virtually as Facebook posts begin to emerge from New Zealand and Australia and they no longer feel alone.

It certainly takes a village to coordinate one of these events, let alone 13 including during a pandemic, but it is with the amazing support of our International 22q11.2 Foundation volunteers, led by **Carol Cavana**, **Debbie DeLoach**, **Lauren Lairson**, and **Donna McDonald-McGinn**, as well as our extraordinarily hard working and committed **global volunteers**, that another incredible moment happened for the international 22q community. So, thank you again from the bottom of our collective hearts.

We hope to see even more of you gathering next year for our 14th Annual 22q at the Zoo Worldwide Awareness Day to be held on **May 19, 2024** - because there is no question that 22q at the Zoo Changes You!

22q at the ZOO WORLDWIDE AWARENESS day®

22q at the Zoo - Worldwide Awareness Day

Photos from May 21, 2023

Our annual 22q at the Zoo event aims to raise awareness about chromosome 22q11.2 differences while providing families, friends, and professionals a chance to socialize.

Here are some photos from our 2023 event**.**

Visit our social media for more photos.

Atlanta, GA, USA

Copenhagen, Denmark

Some of the 22q at the Zoo Locations 2023

Argentina
- Tigre

Australia
- Gold Coast
- Ipswich
- Melbourne
- Perth
- Sydney

Belgium
- Mechlen

Canada
- Toronto

Denmark
- Copenhagen

England
- Birmingham

Ireland
- Dublin

Italy
- Florence

Japan (Virtual)

Netherlands
- Emmen

New Zealand
- Hamilton

Northern Ireland
- Newtownards

Poland
- Warsaw

Serbia
- Subotica

Spain
- (multiple locations)

United States
- Montgomery, AL
- Phoenix, AZ
- Fresno, CA
- Oakland, CA
- Orange, CA
- Sacramento, CA
- Denver, CO
- Bridgeport, CT
- Boca Raton, FL
- Melbourne, FL
- Tampa, FL
- Atlanta, GA
- Boise, ID
- Baton Rouge, LA
- Shreveport, LA
- Grand Rapids, MI
- St. Paul, MN
- Kansas City, MO
- St. Louis, MO
- Asheboro, NC
- Lincoln, NE
- West Orange, NJ
- Bronx, NY
- Niagara Falls, NY
- Syracuse, NY
- Cincinnati, OH
- Cleveland, OH
- Portland, OR
- Columbus, OH
- Halifax, PA
- Philadelphia, PA
- Pittsburgh, PA
- Columbia, SC
- Sioux Falls, SD
- Abilene, TX
- Austin, TX
- Beaumont, TX
- Dallas / Fort Worth, TX
- Houston, TX
- North Centra, TX
- Rio Grande Valley, TX
- San Antonio, TX
- Tyler, TX
- Salt Lake City, UT
- Richmond, VA
- Seattle, WA
- Northern WV

If there were no 22q at the Zoo events in your area this year, **you can host one in 2024**! This extraordinary event grows every year because of dedicated volunteers like you.

To ensure maximum success and awareness, the International 22q11.2 Foundation has developed guidelines for 22q at the Zoo Worldwide Awareness Day. Please visit the **Host a 22q at the Zoo Event** section on our website for more information. We even have an event planner letter ready for you! No zoo? No problem. Consider gathering at a park, a farm, or even an aquarium.

If you have any further questions or concerns, please contact us at **info@22q.org**. We are here to help you every step of the way. On behalf of The International 22q11.2 Foundation, Inc. we thank you for joining us as we expand awareness of the 22q11.2 differences.

See you at the zoo on May 19, 2024!

Recommended Check Ups for Individuals with 22q11.2 Deletion Syndrome

Individuals with 22q11.2DS often need to deal with multiple medical problems. From time to time, it is important to have health checks, as these may reveal conditions that need attention. Earlier this year, international 22q11.2DS experts published updated clinical recommendations for managing children and adults with the syndrome. Each of these documents contains a useful table that lists the recommended periodic assessments. We have now made these two tables into checklists in the format of a fillable savable pdf. Whenever you or your loved one completes an assessment, just put a check mark directly on the checklist on your computer.

To use these check lists on your computer:

1. **DOWNLOAD** these files onto your hard drive or your cloud first before entering check marks digitally, or else your check marks will not be saved.

Children/Youth **Adult**

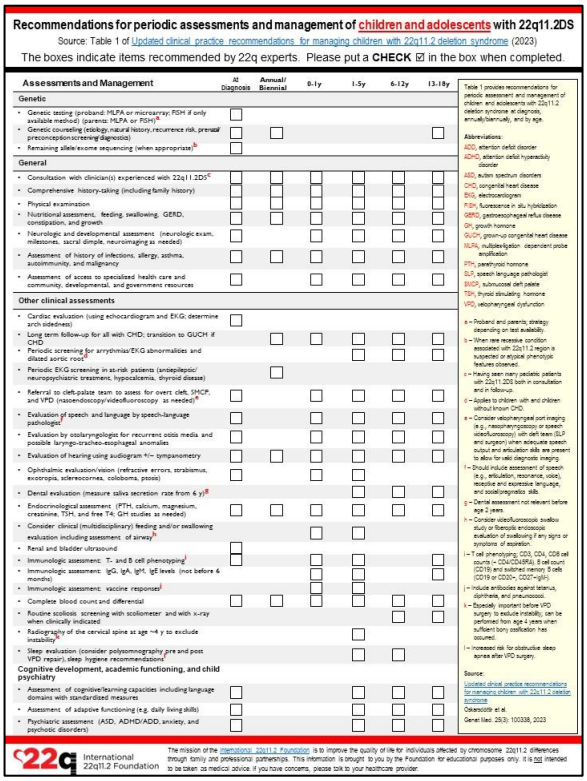
2. Open these files from your **system viewer** (i.e. **Finder** on Macs or **File Explorer** on Windows), not a web browser
3. These files are fillable and savable PDFs, so you can type directly into the forms and save your responses.

If you are still unable to enter check marks or save your responses, please consider updating your Adobe Reader. Alternatively, you can print the forms and fill them out on paper.

Here are the links to the clinical recommendations:

- Updated clinical practice recommendations for managing **children** with 22q11.2 deletion syndrome
- Updated clinical practice recommendations for managing **adults** with 22q11.2 deletion syndrome

This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have any concerns, please talk to your healthcare provider.



Recommendations for periodic assessments and management of children and adolescents with 22q11.2DS

Source: Table 1 of Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome (2023)

The boxes indicate items recommended by 22q experts. Please put a **CHECK** ☑ in the box when completed.

Assessments and Management	At Diagnosis	Annual/ Biennial	0-1y	1-5y	6-12y	13-18y
Genetic						
• Genetic testing (proband: MLPA or microarray; FISH if only available method) (parents: MLPA or FISH)[a]	☐					
• Genetic counseling (etiology, natural history, recurrence risk, prenatal/ preconception screening/diagnosis)	☐	☐				☐
• Remaining allele/exome sequencing (when appropriate)[b]	☐					
General						
• Consultation with clinician(s) experienced with 22q11.2DS[c]	☐	☐	☐	☐	☐	☐
• Comprehensive history-taking (including family history)	☐	☐	☐	☐	☐	☐
• Physical examination	☐	☐	☐	☐	☐	☐
• Nutritional assessment, feeding, swallowing, GERD, constipation, and growth	☐	☐	☐	☐	☐	☐
• Neurologic and developmental assessment, (neurologic exam, milestones, sacral dimple, neuroimaging as needed)	☐		☐	☐	☐	☐
• Assessment of history of infections, allergy, asthma, autoimmunity, and malignancy	☐	☐	☐	☐	☐	☐
• Assessment of access to specialized health care and community, developmental, and government resources	☐		☐	☐	☐	☐
Other clinical assessments						
• Cardiac evaluation (using echocardiogram and EKG; determine arch sidedness)	☐					
• Long term follow-up for all with CHD; transition to GUCH if CHD			☐	☐	☐	☐
• Periodic screening for arrythmias/EKG abnormalities and dilated aortic root[d]				☐	☐	☐
• Periodic EKG screening in at-risk patients (antiepileptic/ neuropsychiatric treatment, hypocalcemia, thyroid disease)		☐				
• Referral to cleft-palate team to assess for overt cleft, SMCP, and VPD (nasoendoscopy/videofluoroscopy as needed)[e]	☐		☐	☐	☐	☐
• Evaluation of speech and language by speech-language pathologist[f]	☐		☐	☐	☐	☐
• Evaluation by otolaryngologists for recurrent otitis media and possible laryngo-tracheo-esophageal anomalies	☐		☐	☐	☐	☐
• Evaluation of hearing using audiogram +/– tympanometry	☐	☐	☐	☐	☐	☐
• Ophthalmic evaluation/vision (refractive errors, strabismus, exotropia, sclerocornea, coloboma, ptosis)	☐		☐	☐		
• Dental evaluation (measure saliva secretion rate from 4-y)[g]				☐	☐	☐
• Endocrinological assessment (PTH, calcium, magnesium, creatinine, TSH, and free T4; GH studies as needed)	☐	☐	☐	☐	☐	☐
• Consider clinical (multidisciplinary) feeding and/or swallowing evaluation including assessment of airway[h]			☐	☐		
• Renal and bladder ultrasound	☐					
• Immunologic assessment: T- and B cell phenotyping[i]	☐		☐	☐		☐
• Immunologic assessment: IgG, IgA, IgM, IgE levels (not before 6 months)			☐	☐		☐
• Immunologic assessment: vaccine responses[j]			☐	☐		
• Complete blood count and differential	☐	☐	☐	☐	☐	☐
• Routine scoliosis screening with scoliometer and with x-ray when clinically indicated					☐	☐
• Radiography of the cervical spine at age ~4 y to exclude instability[k]				☐		
• Sleep evaluation (consider polysomnography pre and post VPD repair), sleep hygiene recommendations[l]				☐	☐	
Cognitive development, academic functioning, and child psychiatry						
• Assessment of cognitive/learning capacities including language domains with standardized measures	☐			☐	☐	☐
• Assessment of adaptive functioning (e.g. daily living skills)	☐			☐	☐	☐
• Psychiatric assessment (ASD, ADHD/ADD, anxiety, and psychotic disorders)	☐			☐	☐	☐

Table 1 provides recommendations for periodic assessment and management of children and adolescents with 22q11.2 deletion syndrome at diagnosis, annually/biannually, and by age.

Abbreviations:
ASD, autism spectrum disorders
CHD, congenital heart disease
EKG, electrocardiogram
FISH, fluorescence in situ hybridization
GERD, gastroesophageal reflux disease
GH, growth hormone
GUCH, grown-up congenital heart disease
MLPA, multiplex ligation -dependent probe amplification
PTH, parathyroid hormone
SLP, speech-language pathologist
SMCP, submucosal cleft palate
TSH, thyroid stimulating hormone
VPD, velopharyngeal dysfunction

a – Proband and parents; strategy depending on test availability.
b – When rare recessive condition associated with 22q11.2 region is suspected or atypical phenotypic features observed.
c – Having seen many pediatric patients with 22q11.2DS both in consultation and in follow-up.
d – Applies to children with and children without known CHD.
e – Consider velopharyngeal port imaging (e.g., nasopharyngoscopy or speech videofluoroscopy) with cleft team (SLP and surgeon) when adequate speech output and articulation skills are present to allow for valid diagnostic imaging.
f – Should include assessment of speech (e.g., articulation, resonance, voice), receptive and expressive language, and social/pragmatics skills.
g – Dental assessment not relevant before age 2 years.
h – Consider videofluoroscopic swallow study or fiberoptic endoscopic evaluation of swallowing if any signs or symptoms of aspiration.
i – T cell phenotyping: CD3, CD4, CD8 cell counts (= CD4/CD8MRA). B cell count (CD19) and switched memory B cells (CD19 or CD20+, CD27+IgD–).
j – Include antibodies against tetanus, diphtheria, and pneumococci.
k – Especially important before VPD surgery to exclude instability; can be performed from age 4 years when sufficient bony ossification has occurred.
l – Increased risk for obstructive sleep apnea after VPD surgery.

Source:
Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome
Óskarsdóttir et al.
Genet Med. 25(3): 100338, 2023

22q International 22q11.2 Foundation

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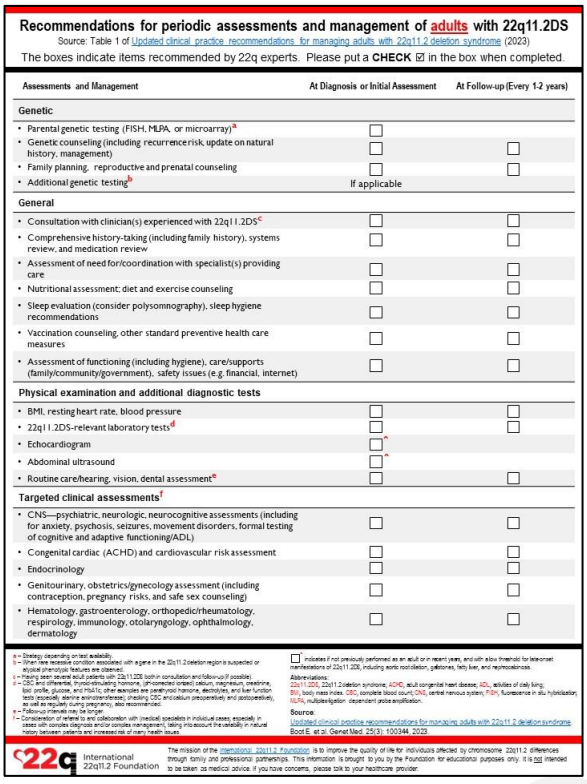
↑ Checklist for Children and Youth

↓ Checklist for Adults



Recommendations for periodic assessments and management of adults with 22q11.2DS

Source: Table 1 of Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome (2023)

The boxes indicate items recommended by 22q experts. Please put a **CHECK** ☑ in the box when completed.

Assessments and Management	At Diagnosis or Initial Assessment	At Follow-up (Every 1-2 years)
Genetic		
• Parental genetic testing (FISH, MLPA, or microarray)[a]	☐	
• Genetic counseling (including recurrence risk, update on natural history, management)	☐	☐
• Family planning, reproductive and prenatal counseling	☐	☐
• Additional genetic testing[b]	If applicable	
General		
• Consultation with clinician(s) experienced with 22q11.2DS[c]	☐	☐
• Comprehensive history-taking (including family history), systems review, and medication review	☐	☐
• Assessment of need for/coordination with specialist(s) providing care	☐	☐
• Nutritional assessment; diet and exercise counseling	☐	☐
• Sleep evaluation (consider polysomnography), sleep hygiene recommendations	☐	☐
• Vaccination counseling, other standard preventive health care measures	☐	☐
• Assessment of functioning (including hygiene), care/supports (family/community/government), safety issues (e.g. financial, internet)	☐	☐
Physical examination and additional diagnostic tests		
• BMI, resting heart rate, blood pressure	☐	☐
• 22q11.2DS-relevant laboratory tests[d]	☐	☐
• Echocardiogram	☐^	
• Abdominal ultrasound	☐^	
• Routine care/hearing, vision, dental assessment[e]	☐	☐
Targeted clinical assessments[f]		
• CNS—psychiatric, neurologic, neurocognitive assessments (including for anxiety, psychosis, seizures, movement disorders, formal testing of cognitive and adaptive functioning/ADL)	☐	☐
• Congenital cardiac (ACHD) and cardiovascular risk assessment	☐	☐
• Endocrinology	☐	☐
• Genitourinary, obstetrics/gynecology assessment (including contraception, pregnancy risks, and safe sex counseling)	☐	☐
• Hematology, gastroenterology, orthopedic/rheumatology, respirology, immunology, otolaryngology, ophthalmology, dermatology	☐	☐

a – Strategy depending on test availability.
b – When rare recessive condition associated with a gene in the 22q11.2 deletion region is suspected or atypical phenotypic features are observed.
c – Having seen several adult patients with 22q11.2DS both in consultation and follow-up (if possible).
d – CBC and differential; thyroid-stimulating hormone, (phi)corrected (ortho) calcium, magnesium, creatinine, lipid profile, glucose, and HbA1c; other examples are parathyroid hormone, electrolytes, and liver function tests (especially alanine aminotransferase); checking CBC and calcium preoperatively and postoperatively, as well as regularly during pregnancy, also recommended.
e – Follow-up intervals may be longer.
f – Consideration of referral to and collaboration with (medical) specialists in individual cases, especially in cases with complex diagnosis and/or complex management, taking into account the variability in natural history between patients and increased risk of many health issues.

☐ indicates if not previously performed as an adult or in recent years, and with a low threshold for late-onset manifestations of 22q11.2DS, including aortic root dilatation, gallstones, fatty liver, and nephrocalcinosis.

Abbreviations:
22q11.2DS, 22q11.2deletion syndrome; ADHD, adult congenital heart disease; ADL, activities of daily living; BMI, body mass index; CBC, complete blood count; CNS, central nervous system; FISH, fluorescence in situ hybridization; MLPA, multiplex ligation -dependent probe amplification.

Source:
Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome
Bird E. et al. Genet Med. 25(3): 100344, 2023.

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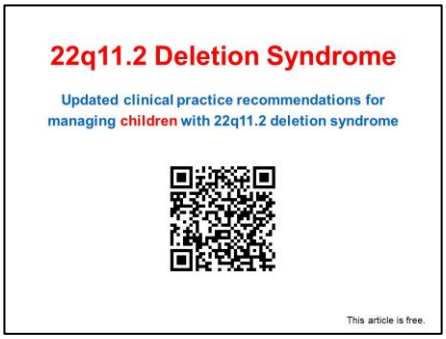
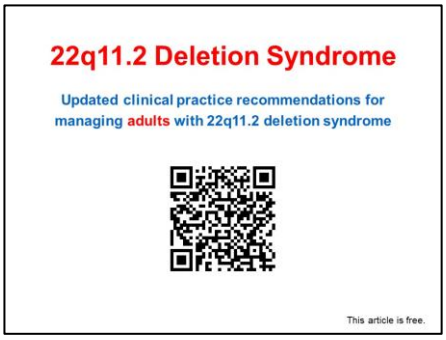
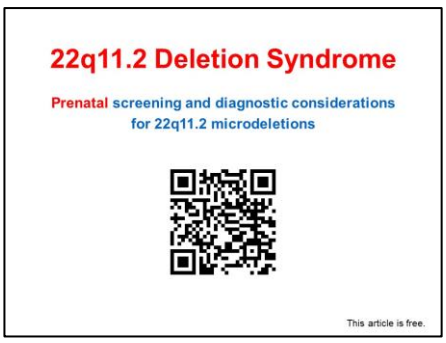
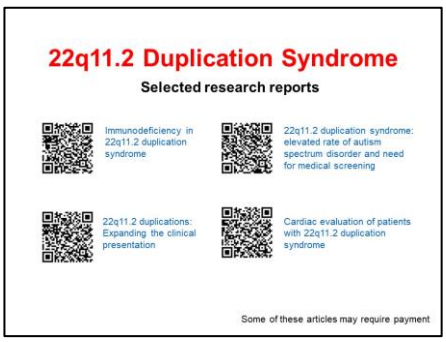
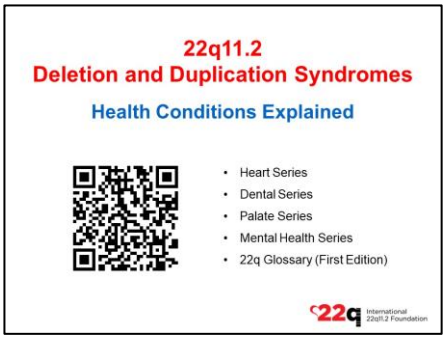
Sharing 22q Resources Using QR Codes











Toronto, ON, Canada

Multiple resources are available for families affected by 22q differences and the medical professionals who take care of them. Let's use technology to get the right resources to the people who need them!

At 22q at the Zoo in Toronto this year, we shared multiple important documents using QR codes (see the images on the left and on top). A QR code is an image that stores information – in our case, the information stored in each QR code is the website address of a 22q resource.

Zoo day participants in Toronto simply scanned the code using their smart phones and accessed the webpages directly. With this easy-to-use method, we shared:

- 3 sets of updated clinical recommendations for 22q11.2 deletion syndrome
- 4 research articles for 22q11.2 duplication syndrome
- The **Health Conditions Explained** section from the website of our Foundation

Feel free to enlarge this newsletter on your screen, focus on one of the QR codes on the left, and use a second phone to scan it. You'll reach the resource!

For your next event, you can make your own QR codes for 22q-related resources using a free online code generator. Or email **info@22q.org** if you would like to get them from us.

TIP: Share the clinical recommendation documents with healthcare providers who are less familiar with 22q11.2 deletion or duplication syndrome.

Mental Health Series

Information sheets now available on our website

https://22q.org/symptoms-care/health-conditions-explained/

Medical terms can be overwhelming, especially for families who are affected by multiple health conditions. To help our patients and families navigate through their medical journeys, we started a section called "**Health Conditions Explained**" on our website. The information sheets are generated by individuals who are highly involved in 22q-related education and communication, and the content is carefully reviewed by 22q experts.

The topics published previously are:

HEART SERIES

- The Heart and Normal Blood Flow
- Ventricular Septal Defect (VSD)
- Tetralogy of Fallot (ToF)
- Truncus Arteriosus (TA)
- Interrupted Aortic Arch (IAA)

DENTAL SERIES

- Let's Talk 22q Teeth – Info for Families
- Dental Health in Children with 22q – Info for Dentists

PALATE SERIES

- The Velopharynx
- Cleft Palate & Submucous Cleft Palate
- Velopharyngeal Dysfunction: Introduction and Causes
- Velopharyngeal Dysfunction: Diagnosis
- Velopharyngeal Dysfunction: Surgery
- Velopharyngeal Dysfunction: Speech Issues

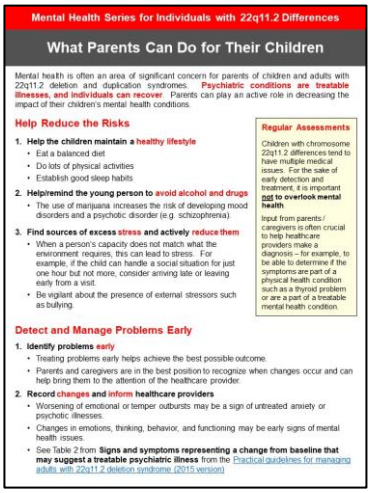
22q GLOSSARY

- First Edition (87 entries)

We will add more contents on various topics as they become available.

Mental health is often an area of significant concern for parents of children and adults with 22q11.2 deletion and duplication syndromes. Psychiatric conditions are **treatable** illnesses, and individuals can recover. Parents can play an active role in decreasing the impact of their children's mental health conditions.

The latest addition to our "**Health Conditions Explained**" series focusses on mental health. The 8 information sheets and the Q&A provide background information about specific conditions and some tips on possible management. Each person is different – Please consult your healthcare provider about the best approach to assessment and management for you or your loved one with 22q11.2DS or 22q11.2DupS.


Mental Health Series for Individuals with 22q11.2 Differences

What Parents Can Do for Their Children

Mental health is often an area of significant concern for parents of children and adults with 22q11.2 deletion and duplication syndromes. **Psychiatric conditions are treatable illnesses, and individuals can recover**. Parents can play an active role in decreasing the impact of their children's mental health conditions.

Help Reduce the Risks

1. **Help the children maintain a healthy lifestyle**
   - Eat a balanced diet
   - Do lots of physical activities
   - Establish good sleep habits
2. **Help/remind the young person to avoid alcohol and drugs**
   - The use of marijuana increases the risk of developing mood disorders and a psychotic disorder (e.g. schizophrenia).
3. **Find sources of excess stress and actively reduce them**
   - When a person's capacity does not match what the environment requires, this can lead to stress. For example, if the child can handle a social situation for just one hour but not more, consider arriving late or leaving early from a visit.
   - Be vigilant about the presence of external stressors such as bullying.

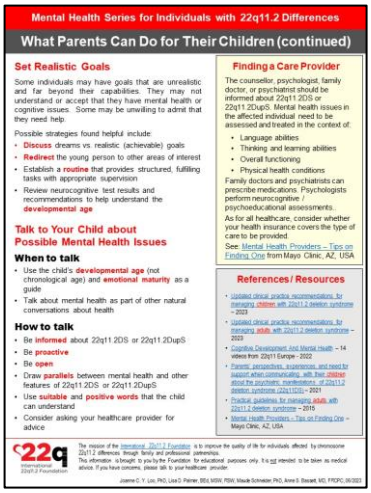
Regular Assessments

Children with chromosome 22q11.2 differences tend to have multiple medical issues. For the sake of early detection and treatment, it is important **not to overlook mental health**.

Input from parents / caregivers is often crucial to help healthcare providers make a diagnosis – for example, to be able to determine if the symptoms are part of a physical health condition such as a thyroid problem or are a part of a treatable mental health condition.

Detect and Manage Problems Early

1. **Identify problems early**
   - Treating problems early helps achieve the best possible outcome.
   - Parents and caregivers are in the best position to recognize when changes occur and can help bring them to the attention of the healthcare provider.
2. **Record changes and inform healthcare providers**
   - Worsening of emotional or temper outbursts may be a sign of untreated anxiety or psychotic illnesses.
   - Changes in emotions, thinking, behavior, and functioning may be early signs of mental health issues.
   - See Table 2 from **Signs and symptoms representing a change from baseline that may suggest a treatable psychiatric illness** from the Practical guidelines for managing adults with 22q11.2 deletion syndrome (2015 version)


Mental Health Series for Individuals with 22q11.2 Differences

What Parents Can Do for Their Children (continued)

Set Realistic Goals

Some individuals may have goals that are unrealistic and far beyond their capabilities. They may not understand or accept that they have mental health or cognitive issues. Some may be unwilling to admit that they need help.

Possible strategies found helpful include:

- **Discuss** dreams vs. realistic (achievable) goals
- **Redirect** the young person to other areas of interest
- Establish a **routine** that provides structured, fulfilling tasks with appropriate supervision
- Review neurocognitive test results and recommendations to help understand the **developmental age**

Talk to Your Child about Possible Mental Health Issues

When to talk

- Use the child's **developmental age** (not chronological age) and **emotional maturity** as a guide
- Talk about mental health as part of other natural conversations about health

How to talk

- Be **informed** about 22q11.2DS or 22q11.2DupS
- Be **proactive**
- Be **open**
- Draw **parallels** between mental health and other features of 22q11.2DS or 22q11.2DupS
- Use **suitable** and **positive words** that the child can understand
- Consider asking your healthcare provider for advice

Finding a Care Provider

The counsellor, psychologist, family doctor, or psychiatrist should be informed about 22q11.2DS or 22q11.2DupS. Mental health issues in the affected individual need to be assessed and treated in the context of:

- Language abilities
- Thinking and learning abilities
- Overall functioning
- Physical health conditions

Family doctors and psychiatrists can prescribe medications. Psychologists perform neurocognitive / psychoeducational assessments.

As for all healthcare, consider whether your health insurance covers the type of care to be provided.

See: Mental Health Providers – Tips on Finding One from Mayo Clinic, AZ, USA

References/ Resources

- Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome – 2023
- Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome – 2023
- Cognitive Development And Mental Health – 14 videos from 22q11 Europe – 2022
- Parents' perspectives, experiences, and need for support when communicating with their children about the psychiatric manifestations of 22q11.2 deletion syndrome (22q11DS) – 2021
- Practical guidelines for managing adults with 22q11.2 deletion syndrome – 2015
- Mental Health Providers – Tips on Finding One – Mayo Clinic, AZ, USA

MENTAL HEALTH SERIES

- Mental Health and 22q11.2 Deletion Syndrome
- Mental Health and 22q11.2 Duplication Syndrome
- What Parents Can Do for Their Children
- Attention Deficit Hyperactive Disorder
- Autism Spectrum Disorder
- Anxiety Disorders
- Psychotic Disorders [Including schizophrenia]
- Mood Disorders
- Mental Health Q&A (Spring 2023)

Congratulations to Our Graduates!

Children and youth with 22q11.2 differences often face multiple challenges – speech and language delays, intellectual disabilities, and learning disabilities. In addition, they often need to take time off school to deal with their medical problems. To be able to graduate is a significant accomplishment!

Graduates – We admire your perseverance. Congratulations on your success!

Parents, caregivers, teachers, therapists, and everyone who have been part of the journey – For the care and encouragement, we salute you!

Here are some of the 2023 graduates from our 22q community.

Congratulations Saralyn!

Celebrating Saralyn for her school graduation! We wanted to share encouraging words from Saralyn's mom… for our 22q families…

"From the time I got pregnant with Saralyn, doctors didn't give her much of chance of even surviving to birth. After birth, it still seemed to be one challenge after another and we were told she would never walk, run, talk, or learn. The brick and mortar school in our area basically suggested life skills-only classes. But we are so happy to share June 5th, Saralyn graduated kindergarten and though she receives special education class work, she has excelled and surprised everyone with the potential she holds inside!!! A church friend created a slideshow for her to commemorate all her life and all her important milestones of graduating kindergarten."

So proud of you, Saralyn—keep up the great work! Saralyn is featured in the International 22q Faces of Sunshine calendar for the month of September.

Congratulations Adam!

Meet 11-year-old Adam, born with 22q11.2 Deletion syndrome. Adam is graduating from Grade 6 and moving on to middle school in September. Adam is nervous and will miss his teacher, who he has been lucky to have for the past 4 years. School has been very challenging for Adam, but we try our best to keep it positive. We are from Kitchener, Ontario, Canada.

Congratulations Maren!

Maren is graduating from Access Academy in Little Rock, Arkansas. She was diagnosed with 22q11.2 deletion syndrome at 15 months. Maren loves horses, books, and tik tok. She plans to volunteer in her community, take art classes, and seek employment after graduation. We are so grateful for the support of Maren's teachers, therapists, and her healthcare team. We love you, Maren, and we are proud of you!

Congratulations Elijah!

Elijah is graduating from Central East High School in Fresno, California! Elijah was diagnosed with 22q11.2 Duplication Syndrome at age 11, after years of visiting countless specialists and therapists. While it helped to have a diagnosis, we know there is much advocacy and research to be done to help this population! We are so proud of Elijah and all that he has overcome to finish high school on time. After graduation, he will be participating in a day program to help with his executive functioning and independent living skills. His future is bright!

Congratulations Madeleine!

My name is Madeleine Dawley and I have 22q. After years of hard work, I finally reached my goal of receiving my Associate of Arts degree.

I was diagnosed with 22q when I was very young. Through velo-pharyngeal surgery, dental rehab surgery, 2 spinal surgeries and many other complications I was able to still focus on my education. I am very proud to say that I am a part of the college graduating class of 2023. Although I couldn't have done it alone. I wanted to give a very special thank you to my mom and dad, for everything they have done for me. Without them me graduating and enjoying my best life wouldn't have been possible. I also wanted to shout out my grandparents for celebrating every accomplishment with me and my parents and for supporting me through everything. Lastly, I wanted to thank everyone else for their support and love through everything. Anything is possible.

Congratulations Jackson!

My name is Jackson Cabrera, I am 23 years old, I was born with 22q11.2 deletion syndrome and I have undergone 8 heart surgeries because of being born with Tetralogy of Fallot. I just graduated from Palomar College with an AA degree in Cinema Studies. I will be transferring to JP Catholic University in the fall to achieve my BFA in the Game Development Program with an emphasis in Game Development and Design.

Congratulations Kathryn!

My name is Kathryn Gainor and I have 22q. I finally got to walk the stage for graduation on May 20, 2023! I was a student from 2010 to 2014. I dropped out in 2014 to take care of my health. I worked for 6 and a half years. I did work full time for 4 of those years.

I decided to go back to college in 2019. I found out McDaniel College was dropping my major and I really wanted to finish it. At one point, I was working part time and going to school part time. During covid I was able to take more classes virtually, so it worked for me!

In December 2021, I was 2 weeks away from finishing my senior seminar. That was when I got diagnosed with triple negative breast cancer, one of the most aggressive breast cancers. I had to start chemo within weeks, so I had to defer that semester. In March of 2022, I finally graduated after passing my senior seminar. I was going to walk in May of 2022, but was too sick on chemo. It was literally my last week of chemo, and I could barely stand for long periods of time. Instead, I got my diploma and hung it up.

Fast forward, I finally beat cancer and finished treatment in October. I decided this is the year that I will walk, and so I finally walked and rang the bell out of college which is McDaniel's tradition. It is tradition to ring the bell in and ring it out.

So yes, I am very proud! I hope that my story inspires others to never give up! I will always love learning! I also learned that health is most important and if you take care of yourself, you can succeed in anything! I am so proud to officially have a Bachelor of Arts in religious studies from McDaniel College!

22q and me – Finnegan's Story

Finnegan ("Finn") was born in December of 2015 at UPMC Mercy Hospital of Pittsburgh, Pennsylvania. He is enjoying school, his friends, and annoying his older sister. He is turning 7 this December!

Finn's 22q11.2 deletion was diagnosed when he was 1 month old. His parents knew from an anatomical ultrasound during pregnancy that he would be born with bilateral club feet, but otherwise had a normal, full-term pregnancy. He was delivered via scheduled C-Section and happily got to go back to the hospital room with Mom and Dad. The neonatologist came into the hospital room to examine Finn more closely and noticed a few midline defects (bifid uvula, submucosal cleft palate) in addition to the club feet and wanted him to be monitored more closely, so he went to the NICU at UPMC Mercy. That night, he had an apnec episode, so the next day he was transferred to UPMC Children's Hospital of Pittsburgh to undergo many tests and needed to get over several more hurdles before he could go home more than three weeks later. Finnegan's first Christmas and New Year were spent in the NICU.

One of the many tests and examinations that Finn underwent right after birth was a genetic panel. About 30 days later, Finn's doctors were able to explain all of Finn's irregularities as actually being related to his genetic diagnosis of DiGeorge Syndrome or 22q11.2 deletion. Shortly thereafter, Finn's schedule became filled with more tests and doctors' appointments to monitor his syndrome, and to understand which body systems had been affected: Cardiology, Nephrology, Endocrinology, Immunology, ENT, Orthopedics, Pediatric Surgery, Ophthalmology, Cleft-Craniofacial Team and Genetics.

At 10 months old, Finn suffered his first apnec seizure – the first of countless others in a 4-month period. Regular hospital stays and Neurology appointments entered Finn's life until the doctors were able to control his seizures

with medication. Finn had difficulty swallowing and could not nurse at birth, so a G-Tube was placed for nutrition. While Finn no longer uses the tube for feedings, it has become quite handy for administering medications.

This December, Finn will be undergoing palate surgery to help with speech. His family cannot wait to hear all the stories Finn has to tell them when he has words. Until then, he will continue to use the sounds and gestures that he must get his point across.

Finn is a happy and very silly first grader. He spends about 85% of his day in the classroom with his schoolmates. The rest of the day Finn spends with school interventionists and therapists (Occupational, Physical, and Speech). Finn is learning how to read, sign, and use his talker at school.

Finn has participated in several activities such as swimming lessons, gymnastics, and some martial arts. This Fall was Finn's first soccer season! He loves to play with his vehicles (mostly trucks and buses), ride his bike, jump on the trampoline, swim, and play video games (he is very competitive!).

Finn's family is so proud of everything that he has accomplished so far and cannot wait to see what is next for him. Finn wants to grow up to be a firefighter and school bus driver.

For more inspirational stories, please visit:
https://22q.org/inspirational-stories/

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Our mission: to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.

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- Create Your Own Event or Fundraiser Contact us at **info@22q.org**

Thank you!

The mission of the **International 22q11.2 Foundation** is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

International 22q11.2 Foundation, Inc.
PO Box 532, Matawan, NJ 07747
www.22q.org (877) 739-1849 **info@22q.org**