22q Deletion Velocardiofacial *At a Glance*

22q Deletion Velocardiofacial syndrome is a genetic condition caused by a very tiny missing piece on chromosome 22. This condition is highly variable in its severity and in the number of body systems that are affeced. There is a difference in severity even between affected individuals in the same family. The most commonly affected areas are the heart, the palate (roof of the mouth) and speech, learning and behavior.

About one in every 4 - 6,000 babies is born with 22q deletion syndrome. Children with 22q deletion may have distinct facial features.

Other names for 22q deletion syndrome include:

- 22q11.2 deletion syndrome
- DiGeorge syndrome
- Velocardiofacial syndrome
- Shprintzen syndrome
- Conotruncal anomaly face syndrome
- Caylor cardiofacial syndrome
- Autosomal Dominant Opitz G/BBB syndrome.

(Learn more about physical characteristics and/or symptoms of 22q deletion syndrome at the end of this document.)

Things to Think About

1. Medical / Dietary Needs

What you need to know

- Medical problems such as heart defects, immune deficiency, feeding problems, low muscle tone, and cleft palate typically occupy the management in infancy. In preschool and beyond, management switches to cognitive, behavioral and learning disorders.
    - However, some children with 22q deletion syndrome will have ongoing medical issues, such as repeated surgeries to repair a complex heart defect.

    - Ongoing treatment of speech difficulties may require additional surgeries in school age children. Speech therapy commonly continues throughout elementary school.

- Sleep disturbances may occur in 22q deletion syndrome due to structural and functional abnormalities. These can lead to behavior and/or learning difficulties.
- No special diet is required for 22q deletion, although a well-balanced diet is important.

What you can do

- It is important to meet with the parents to learn about the child's individual medical needs. Although many of the early medical needs may have resolved, each individual child is different and may have ongoing issues that are being addressed and may impact that child's success in the classroom.
- Up to date immunizations are very important. A yearly check-up and studies as needed should occur in the child's Medical Home. Yearly vision screening can be done at school. Notify parents if the child seems tired.
- It is important to be aware of any academic or behavior changes. Contact parents when any differences are noticed.

2. Education Supports

It is important to have HIGH LEARNING EXPECTATIONS for children who have 22q Deletion. Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

What you need to know

- Most children (90%) with 22q deletion experience some degree of developmental disability with delayed speech and language development as the most consistent feature.
- In formal standardized testing, most school aged children have a full scale IQ in the category of borderline intellectual disability (full scale IQ of 71-85).
- A school aged child with 22q deletion will typically have an unusual neuropsychological profile with a significantly higher verbal IQ than performance IQ with strengths and weaknesses suggestive of a nonverbal learning disorder.
- Common strengths
    - Rote verbal learning and memory
    - Reading, decoding and spelling
- Common difficulties
    - Nonverbal processing

- Visual-spatial skills
- Complex verbal memory
- Attention
- Working memory
- Visual-spatial memory
- Mathematics

Attention and memory

- Diagnosis of attention deficit disorder is common in 22q deletion.
- Attention to details but not the whole
- Problems with concentration on tasks
- Executive function difficulties affect planning, thinking flexibly and understanding abstract ideas.
- This may cause children to struggle to remember, process, and organize information efficiently.
- Executive function difficulties can cause problems in more complex math or in reading comprehension.
- May also affect social interactions because of the difficulty in planning and executing plans

Math

Individuals with 22q deletion syndrome may have significant visuospatial dysfunction, diminished math attainment, and executive dysfunction.

- Deficits may be seen in areas of nonverbal processing, visual-spatial skills, complex verbal memory, attention, working memory, visual-spatial memory, and mathematics.

Math learning difficulties in 22q deletion include difficulties in understanding and representing quantities and in accessing the numerical meaning from symbolic digits.

- Individuals may show adequate fact retrieval while development of procedural strategies appears to be delayed.
- Word problems may be a significant area of weakness. They are challenging due to their procedural nature and difficulty in reading comprehension.

Motor and sensory

Motor and sensory abilities in children with 22q deletion syndrome may be delayed.

- Poor muscle development in children with 22q deletion syndrome may lead to delayed motor milestones.

- This can lead to coordination problems that can persist into adolescence. Children may find it difficult to perform tasks that require dexterity and control of movements.
- Children may struggle with visual information to guide their actions. They may find it difficult to perform tasks requiring spatial awareness.

- Copying down text is difficult, as it requires coordination and the ability to hold information in memory for the short term.

Communication

Speech and language development is delayed in the majority of children with 22q deletion syndrome. This may be due in part to structural differences such as a cleft palate (a hole in the roof of the mouth) or to functional difficulties (either VPI or oral apraxia).

- Hypernasal speech is common (75%).
    - Excessively nasal speech due to velopharyngeal incompetence (VPI)
    - Some children with VPI experience nasal regurgitation in which food and drink comes out the nose.
    - Remediation of VPI requires specific referral to a specialist from your primary care physician
- Articulation disorders
    - May be compensatory
    - May be due to oral apraxia (inability to coordinate facial and lip movements) or dysarthria (weakness of oral muscles)
- Slow vocabulary growth and difficulty in forming complex sentences is also common.

What you can do

Consider an IEP or 504 plan in order to address the educational challenges in a more individualized manner. Structure and routine can help reduce anxiety. Visual reminders may help with verbal/auditory learning and reduce frustration.

Be alert for warning signs of problems:

- Late or missing assignments
- Unfinished work
- Work attempted, but done incorrectly
- Quietness in class – lack of questions
- Difficulty retelling a story
- Social or behavioral problems

Interventions for attention and memory

- Repeatedly using verbal instructions
- Break down instructions into clear steps
- Use a tape recorder while reading to the class
- Allow student to use a word bank on a test to help with recall
- Teach a system of remembering assignments using a chart and/or an assignment book

Interventions for learning math

- Provide a template for complex or multistep problems; break down the steps
- Help teach the concepts of numeracy and the associations between numbers and quantities. For example using a board game in which the playing pieces are moved around a board.
- Line up the numbers for calculations
- Use active learning to teach concepts, such as baking or cooking to teach fractions
- Help the child learn to apply the information in new circumstances

Interventions for motor and sensory development

- Occupational, speech and physical therapy may be helpful for motor development, feeding and swallowing, etc.
- Visual instruction may work better than verbal.
- Limit written homework

Interventions for communication

- It is important to rule in/out developmental motor speech disorder. This includes:
    - Childhood apraxia of speech which is a motor planning problem
    - Developmental dysarthria, which is a motor execution problem.
- The diagnosis and the treatment of speech and language problems are challenging. Many different factors may be involved. However, remediation has led to excellent prognosis in a large majority of cases.
- As a result of nasal regurgitation, the child may experience more nasal infections. This gap between the velum and pharynx (area in the back of the throat) may also lead to difficulty swallowing, or dysphagia. It is important to remember that feeding may be a challenge for these children, and they may tire easily during mealtimes. A pediatric speech pathologist will be helpful if feeding is a concern. A speech language pathologist will work with the family to ensure swallow safety, implement feeding techniques, and reduce the risk of pulmonary complications.
- For more information on speech-language disorders, see: http://www.asha.org/public/speech/disorders/.

3. Behavior & Sensory Support

What you need to know

- Social withdrawal is common and may be in part due to speech problems.
- Attention deficit is common and may make the behaviors in a classroom challenging.
- Anxiety, perseveration and autism spectrum disorders can also be present and contribute to social withdrawal.
- Individuals may have poor social judgement and decision-making.
- It is important to help individuals set goals that match their desires and abilities.

What you can do

- Consider treatments:
    - Behavioral supports
    - Counseling
    - Medication
- Be proactive with behavioral supports. Discuss involving behavioral or mental health professionals, or medication with the child's parents, if needed.
    - Firm directions, rules, and clear expectations are helpful
    - The child may benefit from positive behavioral interventions
- Advocate for continued speech therapy to address speech differences
- Be alert for signs of autism and advocate for appropriate support
- Monitor for the need for additional support services for anxiety or depression
- Structure and routine can help reduce anxiety.

4. Physical Activity, Trips, Events

What you need to know

- Hypotonia (low muscle tone) is sometimes still an issue in the school years and may impact the child's ability to participate in a physical education program.
- Special accommodations needed for individuals who have 22q deletion syndrome are dependent on the individual child.
- A child with 22q deletion with ADHD or executive function challenges, may wander or be confused on a field trip.
- Speech difficulties may make field trips and special events more challenging.
- If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports Programming (www.nepassage.org).

What you can do

- Involving the parent in the planning is important so that the special needs for special events or field trips can be addressed.
- Assist in developing an adaptive physical education program if needed.

5. School Absences and Fatigue

What you need to know

- Additional surgeries may cause increased absences.
- In rare cases the immune deficiency can cause an increased susceptibility to infections.
- Anxiety or depression may cause increased absences and school phobias.
- If a child has sleep disturbances at night, he or she may need a rest break during the day.

What you can do

- Work with the student and parents to communicate about absences for medical reasons and help provide the extra help needed.
- Be alert for signs of anxiety or depression and contact the parents.

6. Emergency Planning

What you need to know

Emergency plans will be very individually determined and many children with 22q deletion syndrome will not have a need for a specific emergency plan.

7. Resources

The International 22q11.2 Deletion Syndrome Foundation

http://www.22q.org/

Perhaps you've found your way to our website because you or a loved one was recently diagnosed with the 22q11.2 deletion. Maybe you're a physician, therapist, or educator looking for information about a new patient or student. However you came to find us, you've come to

the right place. Our growing Foundation is committed to improving the lives of people with the 22q11.2 deletion syndrome and their families; with your help, we can realize our goals of detection, care, and cure.

National Center for Biotechnology Information (NCBI) Bookshelf - 22q11.2 Deletion Syndrome

http://www.ncbi.nlm.nih.gov/books/NBK1523/?report=printable
Learn more about the genetics of 22q Deletion Syndrome

Educating Children with Velo-cardio-facial Syndrome

https://www.cutlerlandsman.com/

http://www.amazon.com/Educating-Velo-Cardio-Facial-Syndromes-Communication-Disorders/dp/1597564923

Purchase this book from respected author, Donna Cutler-Landsman.

Cutler-Landsman, Donna. San Diego: Plural Publishing, 2007. Print.
ISBN10: 1-59756-109-6
ISBN13: 978-1-59756-109-9

The Dempster Family Foundation

www.dempsterfamilyfoundation.org

The Dempster Family Foundation is dedicated to improving the quality of life for the growing community of those affected by 22q11.2 Deletion Syndrome. We do this through partnerships that positively impact greater awareness, education, and advocacy

Educational Issues for Children with Chromosome 22q.11.2 deletion

http://www.ucdmc.ucdavis.edu/mindinstitute/research/cabil/presentations/dultz_education-nov05.pdf

This educational slide presentation was prepared for families by Cheryl Dultz, San Juan Unified School District, CA. It includes an overview of current research at the time, strategies for learning in the classroom, and how to help your child at home. Knowledge is Hope.

The 22 Crew

www.22crew.org

The 22 Crew is a website from the UK developed by families of children with 22q. Their motto is "22Q CAN-DO". Their mission is to fund research and provide social, educational and medical support for all those affected by 22q Deletion Syndrome.

Supporting children with genetic syndromes in the classroom: the example of 22q deletion

http://onlinelibrary.wiley.com/doi/10.1111/1467-9604.12029/epdf
by Colin Reilly and Lindsey Stedman, the British Journal of Learning Support

Practical guidelines for managing adults with 22q11.2 deletion syndrome

http://www.nature.com/gim/journal/vaop/ncurrent/abs/gim2014175a.html

This article was published online in 2014, in Genetics in Medicine, the official publication of the American College of Medical Genetics.

Note: *This printable version does not include the information found under the green button marked "Transitions" on the website. Those general pages may be printed separately.*

Meet a child with 22q – *Dynamic Dani!*

Dani lives in a small New England college town with her parents. Having just turned three, she is transitioning from Early Supports and Service (ESS or Early Intervention) to educational and other services in her community. She will be in a combination of a local private Preschool/Child Care and the Preschool run by her town's public school system.

Her Mother Kim describes Dani as an active little girl who is mildly involved with 22q. She likes to be around people she likes and asks to see her twin cousins every single day! They are 3 months younger and one of them, who is quiet like Dani, is Dani's best friend in the world.
Dani is "really fun," very active, and loves to be outside. She might be shy when she meets someone but warms up as she gets to know the person.
Dani was first diagnosed after she was late on some developmental milestones and was referred at about 18 months because she wasn't speaking. She was referred to a neurologist who made the diagnosis after doing some testing.

Dani started talking near her third birthday and now signs as well as speaks (about 75% talking and 25% signing, but often does both at the same time). She has made great gains with her OT, PT and Speech therapies in ESS. Dani has some challenges with low muscle tone in her core and hands especially. She has no cardiac issues but does have some difficulty with her immune system. For example, if she gets a cold, she may have it for months. She is active all the time

and “never sits still” according to her mother. Dani has sensory sensitivities, especially to noises and water. Bath time is especially challenging. Kim has found that giving her choices and helping her feel like she in in control work very well in helping Dani feel like she is in charge. And that eliminates her getting upset. For instance, instead of telling her to eat her sandwich, she may give her a choice of two healthy items, such as sandwich or apple. “Most often she chooses what I wanted her to choose anyway!” says Kim!

She advises parents to be very positive and to “Keep some time for yourself. Step back to look at what you have instead of what you don’t have.” Kim has found the Dempster Foundation’s Website extremely helpful and it even has a “Family Locator” for families wishing to connect to other families.

Kim also advises teachers to be patient, and to try to be open to different techniques. She states, “Dani can understand everything but sometimes just doesn’t want to talk. Every child with this (condition) is extremely different.”

Physical characteristics and/or symptoms

Not all people with 22q deletion have all of these characteristics.

- **Heart** defects are found in 74% of affected individuals. These heart defects can range from very mild to very severe and may require no surgery or many surgeries.

- **Palate** (roof of the mouth) abnormalities are found in 69% of affected individuals. These can include clefts (holes) in the palate that require surgery; submucosal (involving the muscular layer beneath) clefts that may or may not require surgery but may affect speech; and velopharyngeal insufficiency, a condition in which the palate closes improperly during speech. These abnormalities contribute to the high percentage of children with this condition who require speech therapy.

- **Face:** Children and adults with 22q deletion syndrome may have distinct facial features that include a nose that is broad at the top and narrow at the bottom, smaller appearing eyes and a small mouth. These facial features are usually not distinct enough to be recognized by the untrained eye.

- **Learning difficulties** are found in 70-90% of individuals with 22q deletion syndrome. The most common delays in early childhood are in the area of motor development, often related to low muscle tone, and language development.

- **Attention** difficulties are the most commonly found behavioral difference and it is estimated that 30-50% of children with 22q deletion fit the diagnostic criteria for ADHD. Autism spectrum disorders are common – found in about 20% of individuals with 22q deletion syndrome.

- **Psychiatric illness** is more common than in the general population and may include bipolar disorder, schizophrenia, anxiety, perseveration and depression.

- **Immune deficiency**, primarily in the numbers of T-cells (immune functioning blood cells) is present in 77% of individuals with 22q deletion syndrome. Despite this, very few school age children require active management for their immune deficiency.

- **Hypocalcemia** (low calcium levels) may occur and may be serious in infancy, and can cause seizures. The levels tend to normalize, and this is no longer commonly an issue in school age children.

- **Other less common findings may include:**
    - Significant feeding problems, including severe difficulty swallowing requiring nasogastric tube (NG-tube) feedings and/or gastrostomy tube (G-tube) placement.
    - Kidney (renal) anomalies (31%)
    - Hearing loss (both conductive and sensorineural)
    - Differences in the larynx, trachea, and/or esophagus, including vascular ring and laryngeal webs
    - Growth hormone deficiency
    - Autoimmune disorders may occur at a higher frequency, including these possibilities:
        - Juvenile rheumatoid arthritis
        - Idiopathic thrombocytopenia (blood condition)
        - Hyperthyroidism or hypothyroidism
        - Vitiligo (a skin condition in which there is a loss of brown color from areas of skin, resulting in irregular white patches that feel like normal skin)
        - Hemolytic anemia
        - Autoimmune neutropenia (low levels of white blood cells)
        - Aplastic anemia
        - Celiac disease
    - Seizures may occur due to hypocalcemia (low levels of calcium) but are rare in the absence of low calcium levels.
    - Skeletal abnormalities including upper and lower extremities anomalies, vertebral, and rib anomalies.