Genetic Education Materials for School Success

22q Deletion Syndrome

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Acknowledgment:

Genetic Education Materials for School Success (GEMSS) was created and maintained by an interdisciplinary team of genetic counselors, physicians, families, and students working with New England Regional Genetics Network (NERGN) and the Institute on Disability at the University of NH. Thank you to these amazing partners. This work was supported by HRSA grant UH7MC30778.

Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

22q Deletion Syndrome

At a Glance

22q Deletion syndrome (also known as Velocardiofacial Syndrome or DiGeorge Syndrome) is a genetic condition caused by a missing piece of chromosome 22. This condition is highly variable in its severity and in the number of body systems that are affected. 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. This syndrome almost always presents as a new case within a family, i.e., there is no family history or way to predict it occurring.

Meet Dani on page 11.

Common Features of 22q Deletion Syndrome:

- Distinctive facial features
  - Underdeveloped chin, low-set ears, wide-set eyes
- Heart defects - may require surgery
- Poor immune system function = frequent infections
- Cleft palate (abnormality in the roof of the mouth)
  - Could affect feeding or speaking
- Speech difficulties
- Delayed development
  - Delayed language development
  - Behavioral and emotional challenges

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In Depth

Medical and Dietary Considerations

What you need to know

Medical issues are typically most problematic during infancy. In preschool and beyond, management switches to focus more on 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. *Be aware that carbonated drinks may worsen hypocalcemia. Caffeine may contribute to or worsen anxiety.*

Physical characteristics and/or symptoms:

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

- Heart defects are common in affected individuals. These heart defects can range from very mild to very severe. Some children may have already had heart surgery and some may require surgeries in the future.
- Palate (roof of the mouth) abnormalities are found in common in affected individuals. These can include clefts (holes) in the palate that require surgery or less significant abnormalities that may not require surgery. These abnormalities may affect speech and a high percentage of children with this condition require speech therapy.
- Facial features of **c**hildren with 22q deletion syndrome may 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 about 3/4 of individuals with 22q deletion syndrome. School age children will not usually require active management for their immune deficiency.

- Hypocalcemia (low calcium levels) may be serious in infancy but tends to normalize in school age children. Be aware that low calcium levels can lead to seizures.
- 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) abnormalities
    - Hearing loss
    - Growth hormone deficiency and possible skeletal abnormalities
    - Autoimmune disorders may occur at a higher frequency, including these possibilities:
        - Juvenile rheumatoid arthritis
        - 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)
        - Celiac disease
        - Certain blood conditions (hemolytic or aplastic anemia, autoimmune neutropenia, idiopathic thrombocytopenia)

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, is not eating, or shows signs of infection.
- It is important to be aware of any academic or behavior changes. Contact parents when any differences are noticed.

Education Supports

It is important to have high learning expectations for children who have 22q Deletion utilizing their strengths and interests as a foundation. Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

What you need to know

Most children (90%) with 22q deletion experience some degree of developmental disability with delayed speech and language development the most consistent feature. 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. 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). Typically have

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
- May struggle to remember, process, and organize information efficiently.
- Executive function difficulties affect planning, thinking flexibly and understanding abstract ideas.
    - Can cause problems in more complex math or in reading comprehension.
    - Difficulty in planning and executing plans may affect social interactions

Math:

- Math learning difficulties in 22q deletion include difficulties in understanding and representing quantities and in accessing the numerical meaning from symbolic digits.
- Word problems may be a significant area of weakness due to their procedural nature and difficulty in reading comprehension.

Motor and sensory:

- 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 text is difficult, requiring coordination and the ability to hold information in memory for the short term.

Communication:

- Hypernasal speech is common (75%).

  - Likely due to velopharyngeal incompetence (VPI) related to cleft palate and similar abnormalities
  - Some children with VPI experience nasal regurgitation in which food and drink comes out of the nose. May requires referral to a specialist from the 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

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

An IEP or a 504 plan may be needed to address the educational challenges more individually. Structure and routine can help reduce anxiety. Consider interventions like those described below.

Interventions for attention and memory:

- Frequently repeat verbal instructions.
- Break instructions down into clear steps.
- Use a voice recorder.
- 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. (Try graph paper.)
- 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.
- Visual instruction may work better than verbal.
- Limit written homework.
- Use modified notetaking, i.e., fill-in-the-blank notes.

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 because there are many different factors.
- A pediatric speech pathologist may be helpful.
- For more information on speech-language disorders, see: http://www.asha.org/public/speech/disorders/.

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 and autism spectrum disorders can also be present and contribute to social withdrawal.
- Individuals may have poor social judgement and decision-making.
- May have executive function challenges, may wander.
- Psychiatric illness is more common than in the general population and may include bipolar disorder, schizophrenia, anxiety, perseveration and depression.

What you can do

- Consider counseling. Discuss involving behavioral or mental health professionals with the child's parents, if needed.
- Be proactive with behavioral supports.
  - Firm directions, rules, and clear expectations are helpful
  - The child may benefit from positive behavioral interventions
- It is important to help individuals set goals that match their desires and abilities.
- 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.

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.

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.

School Absences and Fatigue

What you need to know

- Additional surgeries may cause increased absences.
- In rare cases immune deficiency can cause an increased susceptibility to infections that could cause absences.
- Anxiety or depression may cause increased absences and school phobias.

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.
- If a child has sleep disturbances at night, he or she may need a rest break during the day.

Emergency Planning

Many children with 22q deletion syndrome may not need a specific emergency plan. Emergency plans will be individually determined, based on behaviors and medical issues. Develop this with input from the parent and physician. Review the plan yearly. It is important to mention new signs, symptoms, or pain to the child's parents.

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Resources

Medline Plus

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide a good medical overview of the condition without being too technical.
https://medlineplus.gov/genetics/condition/22q112-deletion-syndrome/

International 22q11.2 Foundation

The International 22q11.2 Foundation works with top medical experts from around the world to build understanding of 22q, improve treatment options for families, and conduct the necessary research for longer, healthier lives for patients. Click on the Resources tab, then click "Family Resources." You will see a list on the right including "School and Learning."
https://22q.org/

22q Family Foundation

The 22q Foundation is dedicated to raising awareness for 22q11.2 deletion syndrome while connecting, supporting, and offering accurate information to families affected by the disorder. Click on the 22Q Education tab and scroll down to "School Success – A Handbook for Parents and Educators" for another great resource.
https://22qfamilyfoundation.org/

Supporting Children with Genetic Syndromes in the Classroom: The Example of 22Q Deletion

This is an interesting article describing the unique learning strengths and needs of this disorder and recommended classroom interventions.
https://nasenjournals.onlinelibrary.wiley.com/doi/10.1111/1467-9604.12029

Info for School Nurse and Primary Care Staff

22q Deletion Syndrome is a complex condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team. Work with the parents to get signed releases to share information at the school nurse's office and the primary care office.

Gene Reviews is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The entry for 22q Deletion Syndrome provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues. It is a great resource for medical providers. https://www.ncbi.nlm.nih.gov/books/NBK1523/

HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, remind the parents to sign a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

Example letter for physician to provide to school

"My patient_____ has been diagnosed with 22q Deletion Syndrome. This is a rare genetic condition that is characterized by cleft palate, heart defects, developmental delays, speech and learning difficulties, and immune suppression. This condition necessitates an Individualized Education Program (IEP) or 504 plan.

Medical complications of 22q Deletion Syndrome may cause more frequent absences than the typical student. This student would benefit from evaluation by speech therapy and special education professionals to define appropriate modifications and accommodations."

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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 affected 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 is in control works 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."

Thank you so much to Dani and family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.

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