DiGeorge Syndrome

Background

DiGeorge syndrome (DGS) is a particular group of clinical features that frequently occur together because of a chromosomal 22 defect. The term “22q11.2 deletion syndrome” is commonly used. DGS is the most common micro-deletion syndrome causing poor development of several body systems. DGS is primarily a disease of immunodeficiency caused during fetal development.

A DGS diagnosis is made based on signs and symptoms present at birth or shortly after. The classic features include congenital heart defects, susceptibility to infection, unique facial characteristics, and low blood calcium levels. For other milder cases, diagnosis may be made after feeding difficulties, speech delay, or diagnosis of an autoimmune disease.

The features of the syndrome widely vary, even among family members. Most children experience only some of the symptoms of DGS although at least 30 have been identified. The most common symptoms include:

- Frequent infections including recurrent ear infections, respiratory infections
- Cardiac defects including heart murmur, congenital heart defects
- Bone and muscle problems including poor muscle tone, scoliosis, arthritis, and growth hormone deficiency
- Gastrointestinal problems including reflux, constipation
- Kidney problems including missing or malformed kidneys
- Oral motor problems including cleft palate, feeding disorders, dysphagia, speech impairment
- Delayed growth and development
- Learning disabilities and higher rates of behavioral, psychiatric, and communication disorders including anxiety, attention-deficit/hyperactivity disorder (ADHD), autism spectrum disorder (ASD), and affective disorders

Unique facial features are also characteristically associated with DGS including abnormal distance between the eyes; low set or malformed ears; underdeveloped mouth, chin, or teeth; heavy or hooded eyelids; overbite; and bulbous nose tip.

There is no cure for DGS. Treatment is focused on managing symptoms and can include antibiotic medication for recurrent infections. Calcium supplements, hormone replacement, cardiac and/or oral surgery, ear tubes or hearing aids, and PT/OT/SLP services may also be utilized.

Top Takeaways for School

Most children with DGS experience some degree of developmental disability with delayed speech and language development.

Common strengths include word processing speed, spelling, grammar, computer skills, rhythm and musical talent.

A neuropsychological profile could suggest nonverbal learning disorder with common difficulties that include receptive and expressive language, non-verbal processing, executive function, and abstract reasoning.

Students may have significant visuospatial dysfunction and be challenged in math application and reading comprehension.

Difficulties with fine and gross motor skills and social and emotional functioning are also common and require planning in the educational setting.

Considerations for the Individualized Healthcare Plan (IHP)

- Nursing diagnoses: Risk for infection, imbalanced nutrition: less than body requirements, risk for decreased cardiac output, risk for injury, impaired growth and development
- Nutrition interventions and equipment (consider brand/size of feeding tube, tube replacement, water flushes, fluid intake goal and supplements); note school district policy on tube replacement and consider keeping backup feeding tube kit at school if applicable
- Use of specialized equipment, adaptive equipment, and orthotics
- Equipment troubleshooting (consider equipment/device user manual, battery, charger)
- Consider emergency action plans (EAPs) and emergency evacuation plans (EEPs) related to special health care needs, including staff education/training

Discussion Starters for the Educational Team

1. Would the student benefit from evaluations or assessments in any of the following areas: physical therapy, occupational therapy, speech and language therapy, assistive technology, adapted physical education, functional behavior, psychology, hearing and vision?
2. Would the student benefit from additional academic support and/or modified education (e.g., copies of notes, extra time, reduced workload, simplified instructions, alternative formats for presentation of material, 504/IEP)?
3. Does the student need additional adult support to access the academic curriculum in the least restrictive environment?
4. Does the student need support with gross or fine motor skills in the classroom?
5. Does the classroom environment support the student's needs and/or equipment (e.g., flash pass for bathroom or nurse)?

Resources

Kennedy Krieger Institute: Neurology and Neurogenetics Clinics
kennedykrieger.org/patient-care/centers-and-programs/neurology-and-neurogenetics-clinics

International 22q11.2 Foundation
22q.org/

The 22q Family Foundation
22qfamilyfoundation.org/

The Immune Deficiency Foundation
primaryimmune.org/

For more information, please scan the QR code or visit: KennedyKrieger.org/SHNIC

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