DIGEORGE SYNDROME

What is it?

DiGeorge Syndrome (DGS) is a particular group of clinical features that frequently occur together as a result 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. The features of the syndrome widely vary and can affect almost any part of the body. 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 more mild cases, diagnosis may be made after development of autoimmune disease, feeding difficulties, or speech delay. Today, genetic testing is more widely used to confirm a diagnosis of DGS.

What are the characteristics or complications?

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

- Frequent infections including recurrent ear infection, respiratory infection, and autoimmune disease
- 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 GERD, constipation
- Kidney problems including missing or malformed kidney
- Oral motor problems including cleft palate, feeding disorders, dysphagia, speech impairment
- Delayed growth and development
- Learning disabilities
- Higher rates of behavioral, psychiatric, and communication disorders including AD/ADHD, anxiety, ASD, affective disorders, OCD

The unique facial features associated with DGC include:

- Abnormal distance between body parts and most often seen with eyes
- Low set or malformed ears
- Underdeveloped mouth, chin, teeth
- Heavy or hooded eyelids
- Overbite
- Bulbous nose tip

What is the treatment?

There is no cure for DGS and treatment is therapy focused to manage systems and symptoms. This could include antibiotic medication, calcium supplements, hormone replacement, cardiac and/or oral surgery, ear tubes or hearing aids, and PT/OT/SLP services.

Kennedy Krieger Institute

The **Specialized Health Needs Interagency Collaboration (SHNIC)** program is a collaborative partnership between the Kennedy Krieger Institute and the Maryland State Department of Education.

Suggested school accommodations

Most children with DGS experience some degree of development disability with delayed speech and language development. Common strengths include word processing speed, spelling, grammar, computer skills, and 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 as well social and emotional functioning are also common and require planning in the educational setting. Supporting students with this condition in the school require educators and parents/guardian to work as a team. Some accommodations to consider for a 504/IEP could include:

- PT/OT/SLP evaluations
- Functional Behavior Plan
- Use of assistive technology
- Adaptive PE if necessary
- Student location in classroom/preferential seating related to hearing and vision
- Clear, concise directions
- Visual versus verbal instruction
- Extended time for response
- Copies of notes made available
- Visual/spatial strategies such as adjusted spacing on papers
- Assignment book for memory, graphic organizers
- Use of active learning (ie. baking or cooking to teach math)
- Monitor for fatigue, offer rest breaks as appropriate
- Emotional support, peer groups
- Staff education/training as appropriate
- Emergency Evacuation Plan (EEP)

Specific health issues for Individualized Healthcare Plan

- Diagnosis including baseline assessments for any immune, cardiac, respiratory, kidney, GI, feeding, and behavioral or psychiatric concerns
- Current medication list including PRN medications
- Nutrition orders and feeding protocol if identified feeding disorder
- Note concerns for hearing loss, use of assistive devices
- Location for rest periods if needed due to cardiac condition and fatigue
- Communicate with school staff, parents/guardian, and provider any changes or concerns about the disease
- Emergency Care Plan(s) (ECP) related to medical needs in the school setting and staff education/training as appropriate for each

Resources & Manuals

Immune Deficiency Foundation
https://primaryimmune.org/

The 22q Family Foundation
https://22qfamilyfoundation.org/

The International 22q11.2 Foundation Inc.
https://www.22q.org/