22q Deletion Velocardiofacial
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (**Error! Hyperlink reference not valid.**).

Physical characteristics and/or symptoms

- Heart defects
- Speech difficulties
  - Oral motor apraxia
  - Hypernasal speech
- Palate abnormalities
  - Submucous or frank cleft palate
  - Velopharyngeal insufficiency (poorly closing palate with speech or feeding)
  - Bifid uvula
- Distinct facial features:
  - Small-appearing eyes
  - Small mouth
  - Straight narrow nose
- Learning difficulties
- Attention deficit disorder
- Psychiatric illness
- Immune deficiency
  - Poorly functioning T-cells
  - Reduced numbers of T-cells
  - Improves over time
- Hypocalcemia

Other less common findings may include:

- Significant feeding problems
- Renal anomalies
- Hearing loss
- Differences in the larynx, trachea, and/or esophagus
- Growth hormone deficiency
- Autoimmune disorders
- Seizures may occur due to hypocalcemia
- Skeletal abnormalities

Recommended Routine Surveillance

- Routine Blood work:
  - Serum ionized calcium and thyroid studies
  - Reevaluation of immunologic status prior to any live virus vaccines
  - Annual CBC and differential
- Ophthalmologic and auditory evaluation prior to school
- Surveillance for scoliosis
- Regular speech/language/developmental evaluations if appropriate
- Routine dental care

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful: https://www.epilepsy.com/learn/managing-your-epilepsy/seizure-action-plans

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist:
  - Cardiac defects
- Craniofacial clinic/Plastic surgery:
  - Palate abnormalities
- Developmental pediatrics:
  - Developmental delays
  - Autism
  - ADHD
- Developmental evaluation:
  - Speech therapist
  - Physical therapist
  - Occupational therapist
- Ear, Nose and Throat:
  - Hearing loss
  - Evaluate for velopharyngeal insufficiency (VPI)
- Endocrinologist:
  - Hypocalcaemia
  - Thyroid and growth hormone deficiency
- Gastroenterologist:
  - Significant feeding problems
- Geneticist / Genetic Counselor:

    - Diagnosis
    - Coordination of care
    - Genetic risk for family
    - Clinical trials
- Immunologist:
    - Immune deficiency requires aggressive treatment of infections
- Nephrologist:
    - Renal anomalies
- Neurology:
    - If seizures present
- Orthopedist:
    - Skeletal abnormalities
- Psychiatry:
    - Psychiatric illness
- Ophthalmologist:
    - Surveillance
- Rheumatologist:
    - Autoimmune disorders

Sample Forms

Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient_____ has been diagnosed with 22q11.2 deletion syndrome. 22q11.2 deletion syndrome is characterized by congenital heart defects, palatal anomalies, characteristic features, an immune deficiency, and learning difficulties. Medical complications with this condition include management of immune deficiencies, gastrointestinal reflux, palate abnormalities, hypocalcaemia, and congenital heart disease. Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

"22q Deletion Velocardiofacial at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

Practical Guidelines for Managing Adults with 22q11.2 deletion syndrome
https://www.nature.com/articles/gim2014175.pdf

Gene Reviews: National Center for Biotechnology Information (NCBI) Bookshelf - 22q11.2Deletion Syndrome
https://www.ncbi.nlm.nih.gov/books/NBK1523/?report=printable

Medline Plus
https://medlineplus.gov/genetics/condition/22q112-deletion-syndrome/

Additional resources including support group information can be found on the main website.