





Defect in the development of the **third** and **fourth pharyngeal pouches**, which results in **abnormal thymic development**, **cardiac defects**, and **parathyroid developmental abnormalities**. It follows an **autosomal dominant** inheritance pattern, though most cases are **sporadic**.

AD
Autosomal dominant

CLINICAL FEATURES

CATCH 22

- **Cardiac defects:** conotruncal defects (tetralogy of Fallot, interrupted aortic arch, truncus arteriosus), VSD 
- **Abnormal facies:** bulbous nose, micrognathia, ear anomalies, prominent nasal bridge, asymmetric crying facies, craniosynostosis
- **Ophthalmologic:** cataract, coloboma, strabismus 
- **Thymic hypoplasia:** immunodeficiency
- **Cleft palate**, hypernasal speech, bifid uvula
- **Hypocalcaemia** and **hypoparathyroidism**

22q11.2 deletion

CNS:

- Hypotonia
- Microcephaly
- Seizures



Development:

- **Intellectual disability**
- Learning difficulties

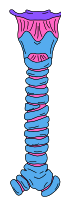
Psychiatric:

- Autism
- **Schizophrenia**
- ADHD
- Anxiety



Laryngotracheoesophageal:

- Vascular rings
- Laryngeal web
- Subglottic stenosis
- Esophageal atresia



Gastrointestinal:

- Diaphragmatic hernia
- **Umbilical and inguinal hernia**
- Constipation

Genitourinary:

- Renal anomalies
- Cryptorchidism
- Hypospadias



MSK:

- Scoliosis
- Clubfoot
- Rib and vertebral anomalies



DIAGNOSIS

Confirmatory Genetic Testing:

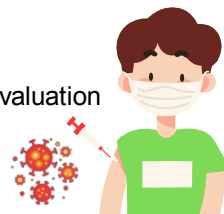
- Karyotype
- Fluorescence in situ hybridization (FISH) to assess for **22q11.2 deletion**
- TBX1 gene sequencing



INVESTIGATIONS / SURVEILLANCE

At diagnosis, then annually:

- **CBC with differential:**
 - Cytopenias (hemolytic anemia, neutropenia, thrombocytopenia)
- **TSH, free T3, T4**
- **Immunodeficiency work-up:**
 - Lymphocyte subsets, T cells evaluation
 - Immunoglobulin levels
 - Lymphocyte proliferation
 - Vaccine titers



- **Calcium, PTH:** *q1–3 months (until stable), then annually*
- **Growth hormone:** *q3–6 months x2 years, then annually*

MANAGEMENT

- **Calcium supplementation** for hypocalcemia.
- **Developmental therapies** (physical, occupational, speech, special education, etc).
- **Endocrine monitoring** for growth and thyroid issues.
- **Cardiac surgery** for congenital heart defects.
- **Immune support** with antibiotics, immunoglobulin, or thymus/stem cell transplants if needed.
- **Mental health support** for anxiety, ADHD, and/or other psychiatric conditions.



February 2025