DiGeorge-anomaly

GENERAL INFORMATION

Description:

DiGeorge anomaly is part of a rare congenital abnormality that is the result of defects during early fetal developmental. These defects occur in areas known as the 3rd and 4th pharyngeal pouches, which later develop into the thymus and parathyroid glands, heart and other important structures. These defects are associated with a chromosome abnormality called "22q11 chromosome deletion".

Alternative names:

- Hypoplasia of thymus and parathyroids
- Third and fourth pharyngeal pouch syndrome
- DiGeorge syndrome chromosome region; DGCR
- DiGeorge syndrome
- Takao VCF syndrome
- Conotruncal anomaly face syndrome
- CATCH-22

Classification:

- Other well-defined immunodeficiency syndromes

Inheritance:

Autosomal dominant

OMIM:

- #188400 DiGeorge syndrome; DGS
- #192430 Velocardiofacial syndrome
- *602054 T-BOX 1; TBX1

Incidence:

1: 4000-5000 live births.

CLINICAL INFORMATION

Description:

There are various clinical features of this anomaly. Patients can have: heart defects, abnormal thyroid function, hypocalcemia due to abnormal parathyroid function, poor numbers of T lymphocytes which normally mature in the thymus. The cardiac abnormalities typically include tetralogy of Fallot, truncus arteriosus, septal defects. The severity of the cardiac abnormalities often determine the outcome. There is often a dysmorphic face with cleft palate, low-set ears, and fish-shaped mouth. There is highly variable immunodeficiency, associated with absence or reduction of thymic size. Severe forms may present as SCID with absent T cells. Learning difficulties and abnormal behaviour are also common.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Microdeletion 22q11, ORHANET
- DiGeorge Syndrome, eMedicine

Genetic:

- EDDNAL
- 22q11.2 Deletion Syndrome, GeneTest

Therapeutic options:

- Surgical treatment of the cardiac abnormalities define prognosis and there are on the first place. If there is evidence for significant humoral deficiency then intravenous immunoglobulins is requiered. Prophylactic antibiotic therapy in case of mild immune defects. Severe defects, with absent T cells, should be considered for BMT. Thymic transplants have been tried but are of uncertain value.
- DiGeorge Syndrome, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies
- Immunologic Evaluation in Patients With DiGeorge Syndrome or Velocardiofacial Syndrome, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name: DGCR
Alias(es): CAFS 6, CATCH22 2, CTHM 6, DGS 2, DORV 6, TBX1 6, TGA 6, VCF 2, VCFS 6, DGS, VCF, DiGeorge syndrome chromosome region

Localization:

Chromosomal Location:

22q11

Maps:

DGCR (Map View)

Other gene-based resources:

Ensembl: ENSG00000184058, GENATLAS: DGCR, GeneCard: DGCR, UniGene: 474233, Entrez Gene: 1714, euGenes: 1714, GDB: 119843

PROTEIN INFORMATION

Description:

Other features:

Expression pattern for human:

Tissue	Exp. (%)	Clones
breast cancer	71.15	1:68
normal squamous epithelium, floor of mouth	23.83	1:203
esophagus	3.28	2:2949
human skeletal muscle	0.45	1:10746
cervix	0.38	2:25325
uterus, pooled	0.31	1:15533
pancreas, exocrine	0.23	1:21418
germ cell, pooled	0.13	1:35870
pool, lung+testis+B-cell	0.09	1:55714
colon	0.06	1:85835

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- European Society for Immunodeficiencies
- NIH/National Institute of Allergy and Infectious Diseases

Disease specific:

- The 22q11 Group

Other information sources:

- DiGeorge Syndrome
- DiGeorge Syndrome