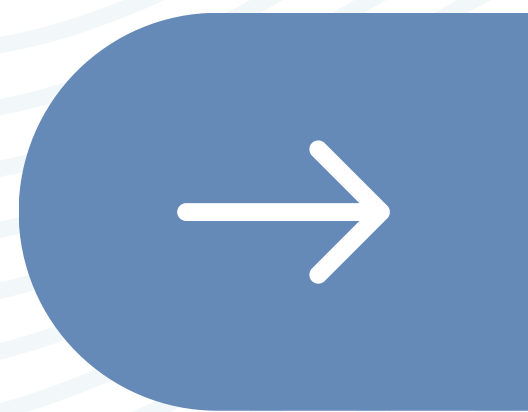


#DIBS BY NEXTILLO

DAILY INFORMATION BULLETIN SERVICE

DIGEORGE SYNDROME





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DIGEORGE SYNDROME

It is also known as [Velocardiofacial Syndrome](#). Heart defects may include a hole between the lower chambers of the heart (ventricular septal defect); only one large vessel, rather than two vessels, leading out of the heart (truncus arteriosus); or a combination of four abnormal heart structures (tetralogy of Fallot).



IMAGE DESCRIPTION



- *The image shows abnormal facies of Digeorge Syndrome And Tetrology Of Fallot (Boot Shaped Heart).*
- *Hypertelorism*
- *Antimongoloid slant*
- *Short philtrum*
- *Mandibular hypoplasia*



IMPORTANT INFORMATION

- ***Chromosome 22q11 microdeletion - Failure of development of 3rd & 4th pharyngeal pouches.***

Triad:

- ***A. Hypoplasia of Thymus: Decreased T cells.***
- ***B. Parathyroid Hypoplasia: Hypocalcemia***
- ***C. Ultimo-brachial body: Defect in heart & great vessels***



CLINICAL FEATURES

CATCH

- ***C - Cardiac defects like TOF***
- ***A - Abnormal Facies***
- ***T - Thymic aplasia***
- ***C - Cleft palate***
- ***H - Hypocalcemia***



DIAGNOSIS

- ***CXR: Absent thymic shadow***
- ***Blood: Decreased CD 3 T Cells, increased B cells, decreased IgA, increased IgE.***
- ***Lymph node: Para cortical area depletion***
- ***It is diagnosed at birth or in infancy based on clinical observation of multiple symptoms with various organs.***
- ***A genetic test is done to confirm the diagnosis for Chromosome 22q11 microdeletion.***



MCQ

Question:

A 3-year-old child presented to the OPD with failure to thrive and a developmental defect of the third and fourth pharyngeal pouches and was referred for further evaluation. The Fluorescence In Situ Hybridization was done. What is the most likely diagnosis?

- A.) *Williams syndrome*
- B.) *Pitt syndrome*
- C.) *DiGeorge syndrome*
- D.) *Cri-du-chat syndrome*

Ans - C.) DiGeorge syndrome