Thymic Aplasia (DiGeorge Syndrome)

Thymic aplasia, also called DiGeorge syndrome, is caused by a 22q11 deletion on chromosome 22. It is thought that this deletion causes defects in migration of neural crest cells, particularly affecting the development of the third and fourth pharyngeal pouches. This affects the development of the thymus gland, which is an organ in the mediastinum that plays a role in the differentiation and induction of tolerance in T-cells as well as the parathyroid glands, which is responsible for the regulation of blood calcium levels. Therefore, individuals with DiGeorge syndrome have a T cell deficiency, making them prone to recurrent viral and fungus infections. Due to defective of parathyroid glands, individuals also suffer from hypocalcemia, often presenting as tetany. Congenital heart defects and abnormalities of the aortic arch are also extremely common in these patients. Some patients display palatal abnormalities, particularly velopharyngeal incompetence, as well as cleft lip and cleft palate. On chest X-ray, there will be an absent thymic shadow due to thymic aplasia.



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Pathophysiology

DiGeorge Syndrome
Inquisitive-George-monkey
Thymic aplasia is also called DiGeorge syndrome, named after the pediatric endocrinologist Angelo DiGeorge.

22q11 Deletion
Two dolls in (22) tutus and (q) queen with (11) double wand
This disease is caused by a 22q11 deletion on chromosome 22, which is thought to cause defects in the migration of neural crest cells.

Failure to Develop Third and Fourth Pharyngeal Pouches
(3) Tree and (4) Fork in Pharaoh pouches
The 22q11 deletion is thought to affect the migration of neural crest cells, particularly affecting the development of the third and fourth pharyngeal pouches. The third pouch develops into the inferior parathyroids and thymus, while the fourth pouch develops into the superior parathyroids.

Signs and Symptoms

Undeveloped Thymus
Thigh
Due to abnormal development of the third and fourth pharyngeal pouches, the thymus fails to develop properly. The thymus gland is an organ in the mediastinum that plays an important role in the differentiation and induction of tolerance in T cells.

T Cell Deficiency
Deflated Tennis-balls
Because the thymus plays an important role in the differentiation and induction of tolerance in T cells, thymic aplasia causes a severe T cell deficiency.

Recurrent Viral and Fungal Infections
Virus and Fun-guys are holding the plate
T cell deficiency caused by thymic aplasia makes individuals susceptible to recurrent viral and fungal infections due to poor cellular immunity.

Undeveloped Parathyroids
Parathyroid-parachute
The third pharyngeal pouch develops into the inferior parathyroids while the fourth develops into the superior parathyroids. Failure of these pouches to develop properly leads to improper development of the parathyroids, leading to hypocalcemia.

Hypocalcemia
Hippo-calcified-cow
Hypocalcemia is characterized by low calcium levels in the serum and is caused by hypoparathyroidism, which plays an essential role in calcium homeostasis.

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Tetany

Titanic

This is a medical sign consisting of the involuntary contraction of muscles, typically seen in the hands. Low blood calcium levels increase the permeability of neuronal membranes to sodium ions that cause a progressive depolarization, which leads to increased possibility of action potentials.

Aortic Defects

A-orca

Improper neural crest migration also causes aortic arch defects in many individuals, because neural crest migration is responsible for the truncal and bulbar ridges that spiral and fuse to form the aorticopulmonary septum. Common defects include an interrupted tetratology of Fallot, interrupted aortic arch, and persistent truncus arteriosus.

Congenital Heart Defects

Heart

Improper neural crest migration also causes congenital heart disease in about 40% of individuals with DiGeorge syndrome. Common defects include tetralogy of Fallot and ventricular septal defects.

Diagnosis

Absent Thymic Shadow on CXR

Absent Thigh on plate

On chest X-ray, patients with DiGeorge syndrome will have an absent thymic shadow due to thymic aplasia.

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