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DiGeorge Syndrome

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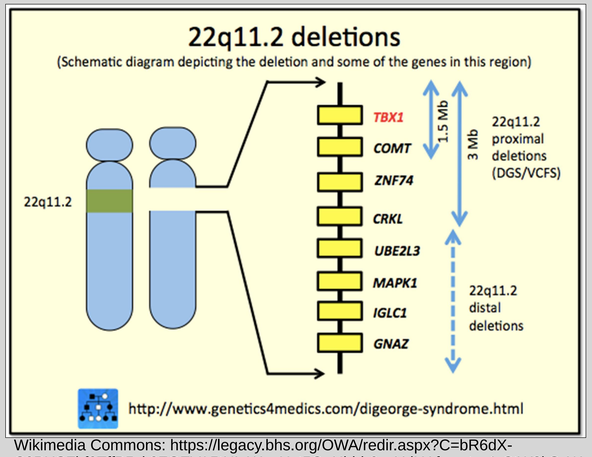
CASE DESCRIPTION

We present a case of DiGeorge syndrome, a rare chromosomal disorder due to the deletion of the q 11 region of chromosome 22. It is a part of CATCH 22 syndrome and also referred to as Velo-Cardio-Facial Syndrome.

An 18 year old male with this syndrome, with a BMI of 36.7, with a history of generalized anxiety disorder and developmental delay presented for dental rehabilitation.

The diagnosis of this syndrome was done in infancy with genetic testing when he presented with neonatal hypocalcemia. Following inhalational induction, nasal intubation was performed with direct laryngoscopy and the perioperative course was uneventful.



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DiGeorge Syndrome

DiGeorge Syndrome (1)
- CHD
- Palatal abnormalities
- Dysmorphic face
- Thymic hypoplasia
- Immune deficiency
- Hypoparathyroidism
- Neonatal hypocalcemia
- Speech and learning disability

Craniofacial Abnormalities (3)
- Micrognathia/retrognathia
- Small mouth
- Low set malformed ears
- Prominent nose
- Hypoplastic nares
- Velopharyngeal anomalies

CATCH -22 (2)
(Mnemonic CATCH-22 summarizes salient features and identifies the location of the chromosomal abnormality)
- Cardiac defects
- Abnormal facies
- Thymic hypoplasia
- Cleft palate
- Hypocalcemia/Hypoparathyroidism

Airway Anamolies (4)
- Micrognathia/retrognathia
- Small mouth
- Velopharyngeal deficiency
- Short trachea
- Tracheomalacia
- Bronchomalacia

CHD in DiGeorge Syndrome(3)
- Tetralogy of Fallot,
- Pulmonary atresia with VSD
- Truncus arteriosus
- Interrupted aortic arch
- Isolated anomalies of the aortic arch
- Isolated VSD

Anesthetic Concerns
- Pre-op anxiety
- Difficult airway
- CHD
- Hypocalcemia and tetany
- Immune deficiency - prone to infection
- Avoid respiratory alkalosis
- Rapid blood transfusion: Hypocalcemia

DISCUSSION

Patients diagnosed with DiGeorge syndrome typically exhibit retrognathia/micrognathia, long face, high and broad nasal bridge, narrow palpebral fissures, as well as small teeth. Additionally patients may develop asymmetrical crying face, downturned mouth, short philtrum low-set, malformed ears, and hypertelorism. These patients are also subject to congenital heart defects, either a cleft palate or incompetence of the soft palate, immune deficiencies, and growth hormone deficiency. Renal, pulmonary, gastrointestinal (GI), skeletal, and ophthalmologic are common as well.

Since these patients exhibit craniofacial dysmorphism, they may have a difficult airway due to cleft palate and short trachea. Hyperventilation and respiratory alkalosis during intraoperative and recovery period results in hypocalcemia and may lead to seizures.

General anesthesia with muscle relaxants masks seizures and whenever possible regional anesthesia is preferred. If General anesthesia is planned anesthesia should be maintained with volatile anesthetic agents or intravous anesthesic agents without muscle relaxants. Hyperventilation during intraoperative or recovery period should be avoided. Serum calcium levels should be measured in the perioperative period and calium supplimentation should be done if the levels are low.

Rapid blood transfusion can lead to chelation of calcium by citrate and decrease serum ionized calcium levels.

CONCLUSION

Patients with DiGeorge syndrome may present with difficult airway and are at risk for hypocalcemic tetany during perioperative period. Careful attention to serum calcium level is essential and factors predisposing to hypocalcemia should be avoided.

References

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