Laryngeal Manifestations of Common Genetic Syndromes

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ABSTRACT

Stenosis in the region is a common cause of respiratory distress in the newborn. Although aspiration, feeding, and feeding difficulties are associated with congenital laryngeal anomalies, hypopharyngeal or high epiglottic, laryngeal, and anophthalmic dysgenesis are associated with developmental and respiratory failure. In addition, unilateral and bilateral defects of various syndromes are associated with congenital laryngeal anomalies. In this study, we reviewed the clinical features associated with the laryngeal anomalies and described the clinical features associated with these syndromes.

Patient-Hall Syndrome

- Extremely rare syndrome with less than 100 cases clinically recognized before the age of 20
- Autosomal or X-linked transmission
- Diagnosed clinically
- Most common abnormalities include:
  - Anterior laryngeal webs (65%)
  - Laryngeal clefts (40%)
  - Epiglottis (30%)
  - Airway obstruction
- Additional less common craniofacial abnormalities include:
  - Hypertelorism
  - Cleft palate
- Pathophysiology of Bilateral Epiglottitis:
  - Epiglottis develops as a result of the fourth brachial arch
  - Normally, these swellings fuse completely leading to a common epiglottis
  - Bifid epiglottis results when the mesenchyme fails to fuse properly
- Signs & Symptoms of Bilateral Epiglottitis:
  - Dysphagia
  - Epiglottitis
  - Neonatal lethargy
  - Cough
  - Difficulty breathing
  - Oxygen saturation

Recommended Evaluation for Bilateral Epiglottitis:

- Early diagnosis of bilateral epiglottitis syndrome will prevent significant mortality and morbidity for these patients including:
  - Hypoxic respiratory distress
  - Severe airway obstruction

- Both sides of the epiglottis should undergo evaluation for other congenital anomalies, particularly those associated with the bilateral epiglottitis syndrome
  - Chromosome analysis
  - Genetic testing
  - Endoscopic evaluation, serially to monitor hypopharyngeal, tracheobronchial, and lung involvement
- In conclusion, bilateral epiglottitis syndrome is associated with neonatal laryngeal anomalies and at-risk evaluation for associated craniofacial abnormalities

Posterior Laryngeal Clefts:

- Most infants present with airway obstruction and respiratory distress
- Type 1: cleft extends below the cords causing a partial cleft of the vocal cords
- Type 2: extend below the cords causing a partial cleft of the cricothyroid joint
- Type 3: laryngeal arrest occurs after week 9; characterized by partial airway obstruction

Anterior Laryngeal Clefts:

- Commonest cause of anterior laryngeal cleft is incomplete resorption of the 4th arch
- Most clefts are incomplete (Type 1)
- Type 2: extend below the cords causing a partial cleft of the vocal cords
- Type 3: laryngeal arrest occurs after week 9; characterized by partial airway obstruction

CONCLUSIONS

- Although rare, laryngeal anomalies are often found with Pallister-Hall Syndrome, Opitz-Frias Syndrome, and 22q11 Deletion Syndrome. Pallister-Hall Syndrome is associated with bifid epiglottis. Opitz-Frias Syndrome can present with laryngeal findings. 22q11 Deletion Syndrome is connected with anterior glottic webs.

REFERENCES