International Journal of Pediatrics and Adolescent Medicine 9 (2022) 182–184

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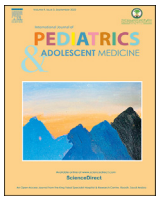
International Journal of Pediatrics and Adolescent Medicine

journal homepage: http://www.elsevier.com/locate/ijpam



Images in pediatrics

Laryngeal web with 22q11.2 deletion syndrome

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ARTICLE INFO

Article history:
Received 3 November 2021
Received in revised form
1 February 2022
Accepted 20 February 2022
Available online 10 March 2022

Keywords:
Congenital heart disease
22q11.2 deletion syndrome
Laryngeal web
Polyhydramnios

ABSTRACT

Laryngeal web is a rare congenital or acquired disease that results in airway stenosis. Depending on the severity of atresia, patients with laryngeal web show a wide variety of symptoms ranging from asymptomatic to life-threatening respiratory dysfunction that may require emergency tracheostomy immediately after birth. We report a neonatal case of laryngeal web with 22q11.2 deletion syndrome. Post-delivery, the infant showed dysphonia and had a ventricular septal defect with characteristic craniofacial features. The infant underwent an endoscopic incision of the web and cardiac surgery. Among patients with laryngeal web, 30% have 22q11.2 deletion syndrome. 22q11.2 deletion syndrome is the most common chromosomal microdeletion syndrome and the second most common chromosomal abnormality associated with congenital heart disease. Therefore, if an infant has laryngeal web with comorbidities such as congenital heart disease, 22q11.2 deletion syndrome should be considered in differential diagnosis.

1. Images in pediatrics

A female infant was delivered at 38 weeks of gestation via cesarean section because of non-reassuring fetal status. Polyhydramnios was detected at 25 weeks of gestation. Post-delivery, the infant showed dysphonia without respiratory distress. She also had a ventricular septal defect and characteristic craniofacial features including hooded eyelids, ear anomalies, and micrognathia. Since laryngeal endoscopy revealed partial membranous atresia of the larynx (Fig. 1A), laryngeal web was diagnosed. Based on this diagnosis and comorbidities, 22q11.2 deletion syndrome was suspected. A definitive diagnosis of 22q11.2 deletion syndrome was made by fluorescence *in situ* hybridization.

The infant underwent an endoscopic incision under general anesthesia following intubation with a 2.5-mm endotracheal tube on day 7 (Fig. 1B). After extubation, the dysphonia disappeared and re-stenosis was not observed (Fig. 1C).

Laryngeal web is a rare congenital or acquired disease of partial laryngeal atresia [1]. Congenital laryngeal web is caused by abnormal development of the larynx by 10 weeks of gestation and often relates to syndromes such as 22q11.2 deletion syndrome. On the other hand, acquired laryngeal web is induced by prolonged intubation, trauma, and comorbidities of surgery. Depending on the severity of atresia, patients with laryngeal web show a wide variety of symptoms ranging from asymptomatic to life-threatening respiratory dysfunction that may require emergency tracheostomy immediately after birth. Among patients with congenital laryngeal web, 30% have 22q11.2 deletion syndrome [1].

22q11.2 deletion syndrome is the most common chromosomal microdeletion syndrome and second most common chromosomal abnormality associated with congenital heart disease [2]. Airway abnormalities including submucosal cleft palate, laryngomalacia, and laryngeal web are also common in 22q11.2 deletion syndrome. Among patients with 22q11.2 deletion syndrome who have airway

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Peer review under responsibility of King Faisal Specialist Hospital & Research Centre (General Organization), Saudi Arabia.

https://doi.org/10.1016/j.ijpam.2022.02.001

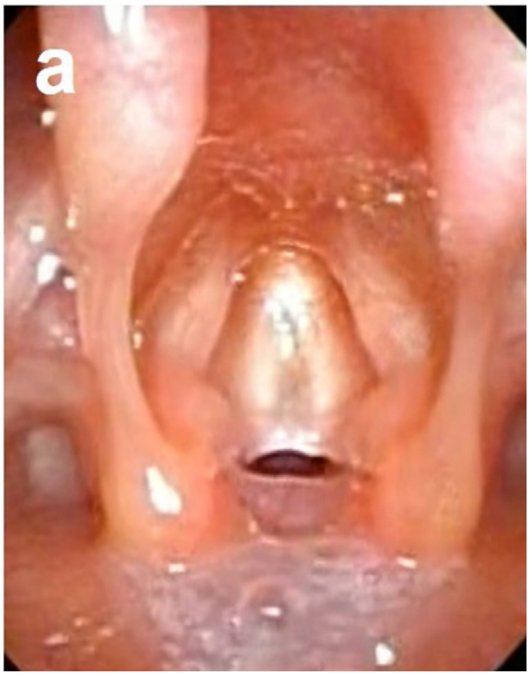

Fig. 1A. Laryngeal endoscopy shows partial membranous atresia of the larynx.

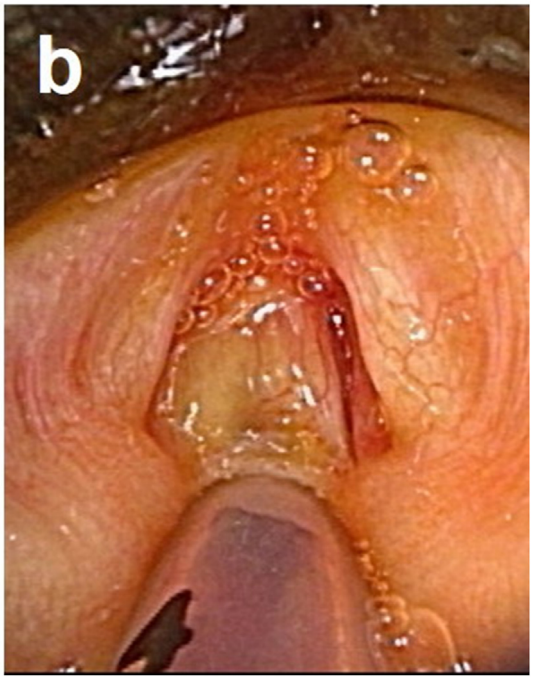

Fig. 1B. The infant was intubated with a 2.5-mm endotracheal tube before endoscopic incision.

abnormalities, 20% have laryngeal web [1–3]. Therefore, if an infant has laryngeal web with comorbidities such as congenital heart disease, 22q11.2 deletion syndrome should be considered in differential diagnosis.

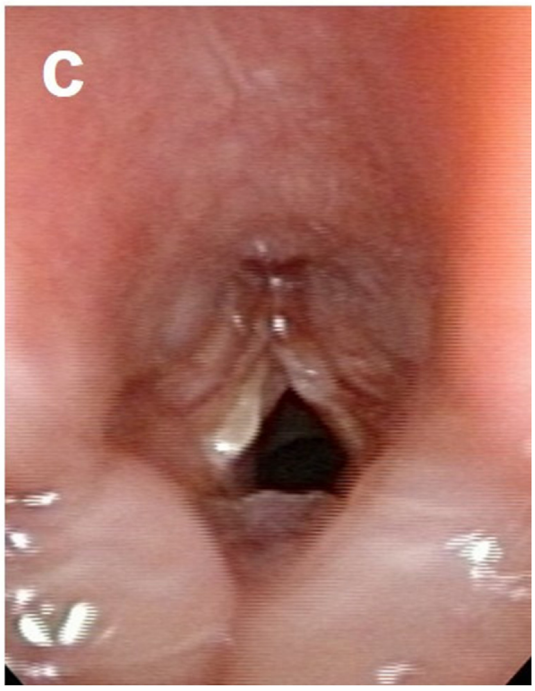

Fig. 1C. Laryngeal endoscopy shows recanalization of the larynx after extubation.

Funding sources

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

Ethical statement

All authors have no conflicts of interest to declare. Written informed consent was obtained from the patient's parents for publication.

Author statement

This manuscript has not been published or presented elsewhere in part or in entirety and is not under consideration by another journal. All study participants provided informed consent, and the study design was approved by the appropriate ethics review board. We have read and understood your journal's policies, and we believe that neither the manuscript nor the study violates any of these. There are no conflicts of interest to declare.

Declaration of competing interest

None.

Acknowledgements

The authors are grateful to Mitsuhiro Kimura and Shinpei Urabe (Department of Otorhinolaryngology, Shimane Prefectural Central Hospital, Shimane, Japan) for their cooperation in the treatment of the patient.

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