Congenital remnants as a cause of neonatal respiratory impairment

Filippo Carta1*, Riccardo Antonio Caria1*, Clara Gerosa2, Sara Sionis1, Melania Puddu3, Giovanni Ottonello3, Roberto Puxeddu1

1Department of Otorhinolaryngology, AOU Cagliari, University of Cagliari, Cagliari, Italy
2Department of Pathology, AOU Cagliari, University of Cagliari, Cagliari, Italy
3Neonatal Intensive Care Unit, Neonatal Pathology and Neonatal Section, AOU Cagliari, Cagliari, Italy
*These authors contributed equally to the manuscript

Abstract

Neonatal respiratory distress is a potentially life-threatening condition, representing a diagnostic and therapeutic challenge for physicians, especially when it is caused by rare pathologies. Head and neck remnants are benign congenital neoplasms rarely observed in newborns. Teratoma is the most common congenital tumor in childhood, while head and neck epithelial and mesenchymal hamartomas are uncommon. We report three cases of pharyngeal congenital remnants presenting with neonatal airway obstruction.

We observed a 9-month-old, 35-day-old, and 15-hour-old patients, who have been referred to our Department of Otorhinolaryngology with acute airway distress. All the patients showed a pharyngeal benign lesion, since teratomas originated from the left lateral wall of the pharynx in two cases and one “fibrovascular” hamartoma originated from the base of the tongue. Timely surgical excision through transoral CO2 laser microsurgery was curative in all the cases.

Dyspnoea in newborns is a challenging condition and must be managed, when possible, by a well-trained paediatric team. When clinicians face obstructive airway congenital remnants, a timely and radical surgical excision is necessary to avoid potentially lethal asphyxia.

Keywords

Congenital remnants, neonatal dyspnoea, teratoma, hamartoma, pharynx, CO2 laser.

Corresponding author

Filippo Carta, MD, Department of Otorhinolaryngology, University of Cagliari, Cagliari, Italy; email addresses: filippocarta@unica.it, pippocarta@tiscali.it.

How to cite

Introduction

Neonatal respiratory distress is a potentially life-threatening condition [1], representing a diagnostic and therapeutic challenge for physicians, especially when it is caused by rare pathologies. A wide variety of lesions may cause neonatal respiratory and feeding impairment but upper aero-digestive tract (UADT) anomalies are relatively uncommon [2]. Pharyngeal cysts, laryngomalacia, laryngeal stenosis, and vocal cord paralysis are the most common UADT anomalies reported in literature [3], while pharyngo-laryngeal congenital remnants are rarely reported. Teratoma is the most common congenital tumor observed in childhood [4], with an incidence approximately of 1/4,000 newborns [5], while head and neck hamartoma, a tumor-like mass composed of disorganized mature specialized cells and tissue elements that are indigenous to the location where it is found [6], is uncommon [7] with an incidence of 2-3/30,000 newborns [8]. Clinical presentation of UADT remnants is influenced by the size and the anatomical site from where they develop.

Authors report three cases of pharyngeal congenital remnants causing neonatal respiratory distress, aiming to improve the knowledge on these uncommon pathologies and to define their adequate management.

Materials and methods

Three patients were observed for congenital dyspnoea caused by pharyngeal congenital remnants at the Department of Otorhinolaryngology – University of Cagliari from December 2010 to June 2015. Age at diagnosis, clinical presentation, preoperative assessment, treatment and outcomes are detailed. Preoperative evaluation was performed using a paediatric 2.5 mm diameter flexible scope (Karl Storz®, Germany). Magnetic Resonance Imaging (MRI) was preoperatively performed in one patient only. A rigid 45° scope (Karl Storz®, Germany, diameter of 4 mm) connected to a HD-camera system (Karl Storz®, Germany) was added to the operating microscope (Carl Zeiss®, Germany – focal length of 250 mm) to improve the visualization of the lesion. Lesions were removed under general anaesthesia with oral intubation, with the aid of CO₂ laser surgery, with the aid of a Boyle Davis mouth gag, and paediatric laryngoscopes (Karl Storz®, Germany).

Case 1

A 9-month-old boy was referred to our Department for recurrent episodes of airway obstruction with oral breathing, snoring and dysphagia, after a previous standard evaluation performed by paediatrician, negative for the detection of UADT anomalies. Flexible scope and subsequent intraoperative examination of the UADT showed an oval and yellow-pinkish lesion with a diameter of about 2 cm originating from the left wall of the pharynx, at the junction between the nasopharynx and the oropharynx, extending behind the soft palate, almost completely obstructing the airway (Fig. 1).

A treatment was immediately planned and performed after intubation by transoral CO₂ laser surgery, with the aid of a Boyle Davis mouth gag. Histology showed a polypoid lesion (Fig. 2), composed by a chorion of fibro-adipose and fibro-connective tissue, pilosebaceous units and serous and mucous gland, covered by skin with a focal hyperkeratotic layer, and allowed the diagnosis of mature teratoma completely removed (Fig. 3).

Figure 1. Case 1: intraoperative endoscopic view of an oval and yellow-pinkish lesion with a diameter of about 2 cm originating from the left wall of the pharynx, at the junction between the nasopharynx and the oropharynx.

Congenital remnants causing neonatal dyspnoea

Case 2

A 35-day-old female was transferred to our Department from a Paediatric Department as an emergency for respiratory distress, cyanosis and bradycardia related to UADT obstruction. MRI of the head and neck region previously performed in the referring centre (axial T1- and T2-weighted scans with coronal and sagittal reconstructions) showed a soft tissue submucosal pharyngeal tumor (Fig. 4).

Intraoperative examination showed an oval pedicled lesion with the maximum diameter of 2 cm, originating from the left wall of the pharynx at the junction between the nasopharynx and the oropharynx, extending behind the soft palate.

The patient underwent transoral CO₂ laser excision with the aid of a Boyle Davis mouth gag. During the induction of general anaesthesia, the lesion firmly obstructed the laryngeal vestibule and required an urgent mechanical dislodging from the vestibule of the larynx. There were no complications and the postoperative course was uneventful. Histology allowed the diagnosis of mature teratoma completely removed as in the previous case.

Case 3

A 15-hour-old girl was referred to our Department from the Neonatal Intensive Care Unit for a severe congenital respiratory distress that required oro-tracheal intubation and umbilical vein catheterization as emergency procedures. The examination of the UADT showed the presence of a pinkish lesion with a diameter of about 2 cm originating from the midline of the dorsum/base of tongue and obstructing the airway. A timely transoral excision was performed under general anaesthesia through the paediatric laryngoscope, under microscopic view, with the aid of the CO₂ laser coupled with the bipolar diathermy at 15 Watt. The lingual polypoid mass appeared non-capsulated, with a broad base of implant, covered by normal
mucosa. Histology showed a mass (size of 12 x 11 x 9 mm) with brownish and darker-coloured areas at cut surface (Fig. 5).

The diagnosis of “fibrovascular” hamartoma was made on the basis of the prevalence of fibrous and vascular components (Fig. 6).

Follow-up

In all cases there were no complications related to the surgical procedure, the oro-tracheal tube was removed immediately after surgery, all patients left the hospital within 2-3 days, and at the present time all patients are free of disease: no recurrence has been observed after 30, 20 and 9 months respectively.

Discussion

Congenital remnants may develop deeply in the head and neck region or more superficially along the UADT, leading to anatomical or functional obstruction. Teratoma origins from a layer of pluripotent cells embedded during embryogenesis [9], generating tissue extraneous to the anatomical site from which it arises, with a variety of ectodermal, mesodermal and endodermal cell types [10]. Teratoma is generally classified into four types: dermoid, teratoid, true teratoma and
epignathi [3]. Dermoid type is the most common, frequently called “hairy polyp”, and contains tissue of ectodermal and mesodermal origin. Teratoid type contains poorly differentiated tissue from the three primary germ layers. The true teratoma is similar to the teratoid, but differentiated into histologically well recognisable tissues. The epignathi type is also tridermal in origin but differentiated into recognisable organs, sometimes with limbs or even a second foetus visible; it is very rare and generally incompatible with life [2, 11]. The highest incidence of teratomas is found in the neonatal period with a female/male ratio of 6:1 [12]. Teratoma is more frequently observed in the sacrococcygeal region, gonads and mediastium, while in the head and neck region it is uncommon (1-9% of all cases), generally in the deep neck areas and at the nasopharynx [4]. Teratoma can be associated with other pathologies (polyhydramnios, breech presentations, preterm birth, cleft lip and cleft palate, inguinal and abdominal hernia, hydrocele, chondrodystrophia, lobulated tongue, lingual hamartoma, pituitary duplication, soft palate insufficiency and cystic fibrosis) [2, 4, 11], but in our cases all lesion were isolated.

Hamartoma is classified as epithelial, mesenchymal, and mixed epithelial and mesenchymal types [13]. Fibrous hamartoma typically occurs in infants less than 2 years of age (average age of 10 months); onset is rare in older children and does not occur after adolescence [14]. UADT hamartoma can be associated to systemic manifestations or specific syndromes, such as the Cowden disease, characterized by macrocephaly, papilloedema and hamartomas, and the Peutz-Jeghers syndrome [15]. Teratoma is usually benign, but malignancy is possible (10% in newborns, but it significantly increases with age) [4], while hamartoma has no malignant potential since its proliferation is self-limiting for the excessive number of cells that reach maturity and cease the mitosis [9].

Clinical presentation of congenital remnants is related to the size and to the anatomical sites where they develop: naso-pharyngeal lesions may cause nasal obstruction, rhinolalia, snoring and the occurrence of sleep apnoea; oro- and hypopharyngeal lesions generally present with dysphagia, odynophagia and earache. The respiratory distress is generally related to the site of origin and the size of the lesion [2-4]. Dyspnoea can be severe: in literature mortality ranges of 9.7-17% despite surgery [4, 11] and two of the three cases we report required urgent care.

Since a wide variety of lesions may be responsible for neonatal respiratory and feeding impairment, newborns presenting with persistent airway symptoms must be endoscopically evaluated as to avoid the missing of lesions temporarily hidden by the soft palate or base of tongue.

Pharyngeal congenital remnants appear generally as sessile or short pedicled lesions [16]. Contrast-enhanced computed tomography or MRI should be performed to determine the exact location of the lesion for a correct staging of the disease and are useful to discriminate other expansive diseases or ectopic thyroid, as well as to exclude the intracranial extension of the lesion [4], but the degree of the respiratory distress drive the clinical management: patients with respiratory distress can be evaluated in emergency requiring immediate management like tracheal intubation or even tracheostomy [17], while patients with moderate respiratory or swallowing symptoms can be managed after imaging. Surgery is generally the treatment of choice [18]. Intraoperative rigid and flexible scopes coupled with the microscope allow for visualization of the exact origin of the lesion and its relationship with the surrounding structures.

Surgery has to be adequate to the benign nature of the lesion and to the fragility of newborns: the resection has to be performed in free margins with minimal removal of the surrounding healthy tissue. Main attention is required to reduce traumatism due to the endoscopic exposure and to achieve a precise haemostasis. CO₂ laser is an extremely safe and precise tool especially in the UADT of newborns: the limited thermal damage associated with a contextual hemostasis leads to minimal edema of soft tissues and/or bleeding that could cause asphyxia in the early postoperative period.

Histologically, the diagnosis of teratoma is relatively easy, while the wide variety of different cells of hamartomas requires, beyond routine staining, special stains and immunohistochemistry. In our case haematoxylin and eosin and Goldner’s stains showed abundant connective tissue; immunohistochemistry showed the expression of smooth muscle (SMA+), glandular tissue (CKAE1AE3+) and striated muscle bundles (Sarcomeric actin+), while CD34 was indicative of well-vascularised tumors [13]. The presence of all these well-differentiated components associated with a disorganized overgrowth, and a Ki67 (MIB1) reactivity of approximately 1% in the connective and gland tissue, allowed the definitive diagnosis of hamartoma.

Congenital remnants causing neonatal dyspnoea
Prognosis is excellent: in our series the postoperative course was uneventful in all cases. Although recurrences may be observed after incomplete surgical excision [19], in our series the microscopic view allowed a precise identification of the origin of the tumors with a consequent adequate resection with free margins, with no recurrences observed at the present time.

**Conclusion**

Newborns airway obstruction is a challenging condition and must be managed, when possible, by a well-trained team including the otorhinolaryngologist. The obstruction of the UADT must be examined endoscopically with extreme care as to avoid incorrect diagnosis, and, when congenital remnants are diagnosed, a minimally invasive radical excision is the treatment of choice.

**Declaration of interest**

The Authors declare that there is no conflict of interest.

**References**