

## Congenital neck anomalies- A clinical and anatomy study

Rovena Bode<sup>1</sup>

<sup>1</sup>Department of General Surgery, Medicine University, "Mother Tereza" University Hospital Center, Tirana, Albania

Address for correspondence: Dr. R. Bode, Surgery Department, University of Medicine, Tirana, "Mother Tereza" University Hospital Center, Rruga e Dibrës, 1025 Tirana, Albania.

e-mail: [rovenabode@gmail.com](mailto:rovenabode@gmail.com)

---

**Abstract:** *Background:* Congenital neck anomalies in the adult are a rare pathology encountered in clinical practice. They are related to disturbances in the development of branchial apparatus of fetus. Although some diagnoses may be evident on physical examination, others may present a diagnostic dilemma. They include branchial cysts and sinuses, thyroid gland abnormalities, and especially thymic and parathyroid gland ectopies. Some of them are inherited, however most of them are caused by environmental factors.

*Methods:* We are presenting our experience with these anomalies, a retrospective study undertaken in the General Surgery Service, "Mother Teresa" University Hospital Center, Tirana, Albania. Because they have varied manifestations and may present at any age they should always be considered. Usually they represent as midline neck masses or lateral neck swellings (fistulae).

*Resultats:* Patient age varies from 12-53 years old. Infrahyoid and suprahyoid thyroglossal duct cysts, median and lateral colli fistulae, thyroid and parathyroid gland ectopies were some the cases treated by us. Parathyroid ectopies are discovered during surgical treatment for primary hyperparathyroidism.

*Conclusion:* Congenital neck anomalies may present at any age, because of this they should be considered. Surgical excision is the only curative treatment.

**Keywords:** *branchial arches, developmental anomaly, fistulae colli laterale, thyroglossal cysts*

---

### I. INTRODUCTION

Congenital malformations of the neck are a wide and heterogeneous group because this region contains parts of almost all organ systems [1-4]. They represent structural or functional anomalies which are present at the time of birth. Approximately 50% of them cannot be assigned a specific cause, but genetic factors and environmental factors are accused. An intimate knowledge of the anatomy and embryology are necessary to better understand, diagnose and treat these anomalies [1-4, 6].

The most common neck anomalies are thyroglossal duct cysts. They are most seen in childhood but also during adolescence and adulthood. They are caused by the persistence of thyroglossal duct during the descent of the thyroid from the foramen caecum to its prelaryngeal position in the neck [6, 8]. They represent as cysts, fistulae or sinuses. Infrahyoid cysts accounts 65% of cases and the suprahyoid type only 20%.

Branchial anomalies are a consequence of disturbances in the development of branchial apparatus of fetus. They are classified in 4 types according to the branchial pouch origin and their anatomic relationships. Second arch anomalies are the most common and represent 90-95% of branchial anomalies. Usually they manifest as cystic lesions than fistulae, located anterior and deep to the sternocleidomastoideus muscle [1-5].

Another possible anomaly is ectopic thyroid tissue. They are attributed to the descent of thyroid tissue through the foramen caecum to its final position. The most common site of ectopic tissue is lingual position. They can manifest with dysphagia, dyspnoea, stridor and hemorrhage [7, 8].

In this group enter also parathyroid and thymus disorders. Especially parathyroid ectopies related to the defects in migration of parathyroid tissue. Inferior parathyroids are more variable in their position than superior parathyroid [8, 9].

### II. MATERIAL AND METHODS

We have carried out a retrospective study for patients admitted for surgical treatment, between January 2009 – December 2011 for congenital neck anomalies in the General Surgery Department, "Mother Tereza" University Hospital Center of Tirana, Albania. The data on patient's histories, diagnostic procedures, treatment and complications were acquired from medical reports. The diagnose was established by physical examination, neck ultrasonography and Ct- Scan. Thyroid hormone levels were measured also.

### III. RESULTS

#### ***Thyroglossal duct cysts***

There were 6 patients referred to the Service of General Surgery nr1, "Mother Tereza" Hospital Universitary Center, Tirana, Albania, from January 2008 to December 2010. We found 4 woman and 2 men with thyroglossal duct cysts, with age varying from 12 to 33 years old. Clinically, 4 patients had a cyst and two a fistula. The cysts were represented as midline neck masses, varying in size from 1.5 cm to 4 cm. Three of them were infrahyoid thyroglossal cysts and one was a submental cyst. The cysts had inflammatory changes in all cases, with a purulent fluid in its content, with skin changes. From the external button of the fistulas a purulent discharge was noted in all two patients. All patients were treated with antibiotics preoperatively and underwent surgical exploration with a Sistrunk procedure. Two of them required a single reoperation for recurrence. The only complication observed was a seroma in two cases.

#### ***Branchial anomalies***

A patient of 16 years old resulted with a right lateral neck fistula, along the sternocleidomastoideus muscle. He had a history of 3 years since noticed a lateral neck mass that began to discharge a purulent fluid - from the external button. During the examination it was noticed a fistulous tract along the lateral border of sternocleidomastoideus muscle. During the operation a lateral neck fistulous tract was noticed beginning from the external, cutaneous button toward the carotid sheath, in close contact with it. No communication with thyroid gland was noticed. A branchial fistula was diagnosed and treated.

#### ***Thyroid abnormalities***

##### ***Ectopic lingual thyroid***

A 33 years old female patient, was referred to the department of General Surgery nr1, from Endocrinology department with the diagnosis of "lingual thyroid". The patient was hospitalized in this service for a swelling at the base of the tongue, noticed 6 years ago, during the first pregnancy. The mass during these years had been grown, and the patient complained of the sensation of foreign body in the mouth, dysphagia, dispnea, cough and bleeding during the end of pregnancy.

On physical examination it was noticed a well localized lesion measuring 5-6 cm in size, smooth, rubbery and reddish mass at the base of the tongue (Fig. 1). No signs of ulceration, bleeding or pus discharge were identified on the surface of the mass. Neck examination revealed no palpable thyroid gland or neck mass but a cervicotomy scar.

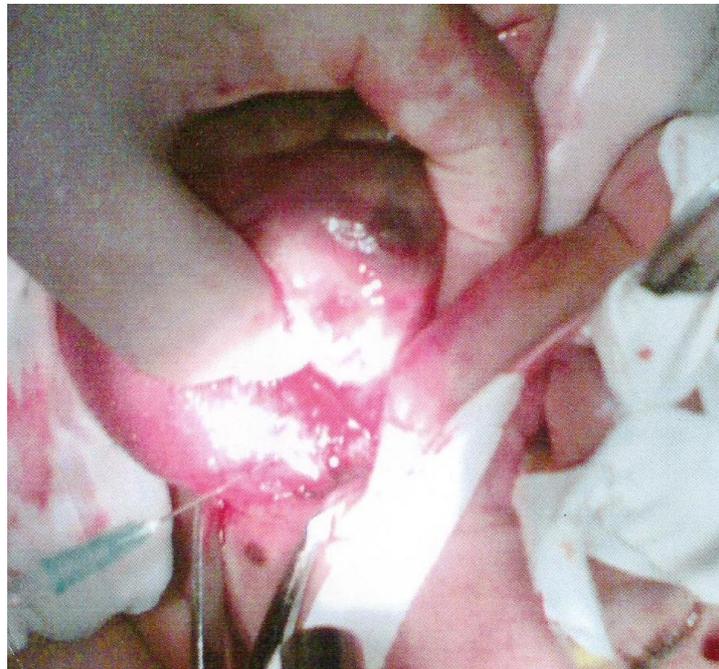


Fig1.Lingual Thyroid

Thyroid tests were as follow: TSH 15, 5 uIU/ML, T3 3, 08 pg/ml, T4F 0, 506 ng/dl.

The patient was healthy and in her medical history revealed of an operation done during childhood for a Thyroid ultrasound revealed no thyroid gland in its normal position. Scintigraphy revealed increased isotope uptake in

the lingual region and absence of isotope uptake in the neck region. The final diagnosis was ectopic lingual thyroid gland.

Treatment began with the administration of L-thyroxine for 3 months and after that with I-131.

Elective surgical resection of the thyroid gland was performed through oral approach under general anesthesia. The surgery and postoperative period were uneventful. The specimen was sent for histopathology analysis and the result was follicular goiter Fig.2.

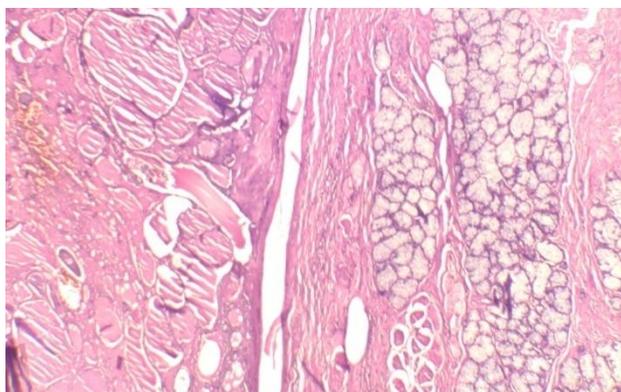


Fig. 2 Hystopathology specimen of a follicular goiter

#### ***Parathyroid gland anomalies***

These cases are discovered during surgical treatment for primary hyperparathyroidism, the same period. In a group study of 8 patients, two ectopic inferior parathyroid glands are found. One patient resulted with an inferior parathyroid gland carcinoma localized in the mediastinum. The other patient resulted with an inferior parathyroid adenoma localized in the thyrothymic ligament (Fig. 3).



Fig. 3 Ectopic parathyroid adenoma

#### **IV. CONCLUSION**

Congenital neck anomalies may present at any age, because of this they should be considered. Surgical excision is the only curative treatment.

#### **REFERENCES**

- [1] Whetstone J, Branstetter BF, Hirsch BE. Fluoroscopic and CT fistulography of the first branchial cleft. *AJNR Am J Neuroradiol*. 2006;27 (9): 1817-9. [AJNR Am J Neuroradiol \(full text\)](#) - [Pubmed citation](#)
- [2] Al-ghamdi S, Freedman A, Just N et-al. Fourth branchial cleft cyst. *J Otolaryngol*. 1992;21 (6): 447-9. - [Pubmed citation](#)
- [3] Joshi MJ, Provenzano MJ, Smith RJ et-al. The rare third branchial cleft cyst. *AJNR Am J Neuroradiol*. 2009;30 (9): 1804-6. [doi:10.3174/ajnr.A1627](#) - [Pubmed citation](#)
- [4] Benson MT, Dalen K, Mancuso AA et-al. Congenital anomalies of the branchial apparatus: *embryology and pathologic anatomy*. *Radiographics*. 1992;12 (5): 943-60. [Radiographics \(abstract\)](#) - [Pubmed citation](#)

- [5] Gold BM. Second branchial cleft cyst and fistula. *AJR Am J Roentgenol.* 1980;134 (5): 1067-9. [AJR Am J Roentgenol \(citation\) - Pubmed citation](#)
- [6] Noyek AM, Friedberg J. Thyroglossal duct and ectopic thyroid disorders. *Otolaryngol Clin North AM* 1981, 14:187-201
- [7] Peters P, Stark P, Essig G Jr, et al. Lingual thyroid: an unusual and surgically curable cause of sleep apnoea in a male. *Sleep and Breathing* 2010; 14: 377-380
- [8] Di Benedetto V. Ectopic thyroid gland in the submandibular region simulating a thyroglossal duct cyst: a case report. *Journal of Pediatric Surgery* 1997; 321: 745-1746
- [9] Wermers RA, Khosla S, Atkinson EJ, et al. Incidence of primary hyperparathyroidism in Rochester, Minnesota, 1993-2001: an update on the changing epidemiology of the disease. *J Bone Miner Res* 2006; 21:171-177