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ADULT PRESENTATION OF BILATERAL BRANCHIAL SINUS WITH AUTOSOMAL DOMINANT TRANSMISSION IN FOUR MEMBERS OF THE FAMILY: A RARE CASE STUDY

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ABSTRACT

This report describes a case of a bilateral second branchial fistula that presented as a persistent discharging sinus in the lower neck associated with pain and swelling in the neck, in a 36-years-old female with autosomal dominant transmission in her all the three daughters. Diagnostic-imaging modalities to aid in diagnosis and treatment plan are discussed. The surgical technique of removing the fistula through a stepladder incision anterior to the sternocleidomastoid muscle and an upward dissection is described along with a discussion on other surgical methods. Four cases were encountered in the family, with no otologic or kidney symptoms, which is quite different from the classical branchio-oto-renal syndrome which associates severe inner ear and kidney congenital anomalies.

KEYWORDS

Branchial Fistulae, Imaging Modalities, Surgical Techniques, Bilateral Branchial Sinus, Branchial Apparatus, Branchial.

INTRODUCTION

Structures of head and neck during foetal life are derived from the branchial apparatus which begins to emerge from the 4th week of intra-uterine life. It consists of six pairs of arches which are mesodermal in origin. Between the arches, sits internal pouches originated from endodermal tissue and externally it invaginates into clefts that are ectodermal in origin. Each branchial arch is supplied by an artery and a nerve and develops into well-defined muscles, bones, and cartilage. The branchial arches usually involute by the 7th week of gestation but failure to do so, may lead to various developmental anomalies such as branchial cysts, sinuses and fistulae.

In utero, the 2nd arch grows externally in a caudal manner to cover 3rd and 4th arch and gets fused with the 5th arch eventually resulting in obliteration of the clefts. Failure of this obliteration of clefts results in branchial cysts lined by ectoderm. On the other hand, if the 2nd arch fails to fuse with the 5th arch, a thin tract connects the clefts to the skin and this is termed as branchial sinus and they mostly ends as a blind tubular or saccular anomaly within mesenchymal tissue. Branchial fistula suggests complete communication from the ectodermal surface to the endodermal surface i.e., the tract communicates internally and externally and presumably relate to an incompletely closed or ruptured branchial plate. [1-6]

We here report a rare case of bilateral branchial sinus fistula in a young female with family history of same presentation occurring in all 3 of her daughters presuming autosomal dominant transmission of the disease. A discussion follows about the genetic transmission of this pathology in the family.

Case Study

A 36-year-old female presented in E.N.T. OPD with a history of discharge from right and left sides of lower part of her neck since birth. The discharge was whitish-yellow, sticky in nature and was on and off in pattern at the time of presentation. The patient also gave history of having bilateral neck swelling over the same site, which got visible around 1 year of her age, and then burst open spontaneously later in life followed by discharge from the swelling. The discharge use to be different in consistency since then; watery or whitish viscous sometimes or yellowish sticky or bloody in nature sometimes. She also complained of episodes of fever and pain at the site of discharge which were associated with increase in the amount of discharge at that time. She was consulting local practitioners and in-spite of taking different medications, she was not getting relieved and was eventually advised surgical treatment.

There was no history of recurrent upper respiratory tract infections, dental or jaw infections. There was no history suggestive of Tuberculosis or Any Malignancy related to bones or soft tissues. But she mentioned that all her 3 daughters are suffering from the same condition since their birth. Her eldest daughter is 13yrs of age and have same presentation i.e., bilateral branchial sinus in neck. The 2nd daughter has a unilateral branchial sinus with watery discharge and is 7yrs of age and the last one, again have bilateral branchial sinus associated with discharge and is of 1 ½yrs of age. None of them have any signs and symptoms showing association with Branchio-oto-renal syndrome or BOR syndrome.

General physical examination and systemic examination of the patient is within normal limits. Local examination revealed sinus opening bilaterally 2-3cm away from midline. The sinus opening on right side located just above the mid clavicle between two heads of right sternocleidomastoid muscle. On the left side the sinus opens above the right mid clavicle on the anterior border of sternocleidomastoid muscle more medially placed. Active whitish yellow, viscous, non-foul-smelling discharge was present from both the sinus openings. Sinus tract was palpable on both sides just lateral to sternocleidomastoid muscle. It moved with deglutition and protrusion of tongue.

Oral cavity examination was within normal limits. Routine blood investigations and chest X-ray were within normal limits. The patient was planned to undergo exploration and excision of the sinus and fistulous tract.

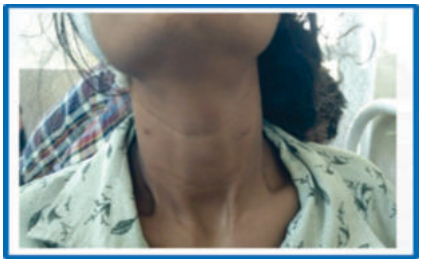

Image 1: Patient with B/L sinus presentation.

Image 2: Patient With Her 3 Daughters With Same Presentation.

Surgical Procedure

Under general anaesthesia, the patient was intubated and placed in Rose 's position. The external opening of the tract was dilated with a probe to delineate the tract. Methylene blue dye was injected to see the extent of the tract and for proper identification while exploration. Excision of both sinus tract was done which turned out to be fistulous tract. Dissection was done with stepladder pattern incision with lower incision being half elliptical around the fistula.

It was done by deepening the incision and elevating sub-platysmal plane. Blunt dissection is done by separating the underlying fat, fascia, muscles and soft tissue where needed and extended up to the angle of mandible. Fistulous tract was identified lying superficial to sternocleidomastoid muscle, facial vein, greater auricular nerves, Ansa-cervicalis and was passing between external and internal branches of carotid arteries on both the sides. Fistula was cut at the upper end of dissection and probing was done with a guide wire. Here the tract was obliterated so was ligated there using vicryl suture material. A subplatysmal drain was placed and the subcutaneous tissue, platysma and skin were approximated with vicryl 3-0 and silk 3-0 respectively. The same procedure was done on the other side also.

The patient was extubated uneventfully. Movement of the vocal cords were bilaterally equal. The patient was shifted to post-operative ward in a stable condition. The patient was then put on parenteral antibiotics for 7 days. The drain was removed at postop day 2 confirming no collection at the operated site. The post-operative period was uneventful and the patient recovered completely. The patient was discharged on the 8th post-operative day.

A histopathological examination of the specimen confirmed the presence of a tract lined by pseudostratified columnar epithelium with abundant subepithelial lymphoid tissue. A final diagnosis of bilateral branchial fistula was made with autosomal dominant transmission to her daughters. None of her daughter showed complete symptoms of BOR syndrome suggesting variable penetration and mutation of the genes.

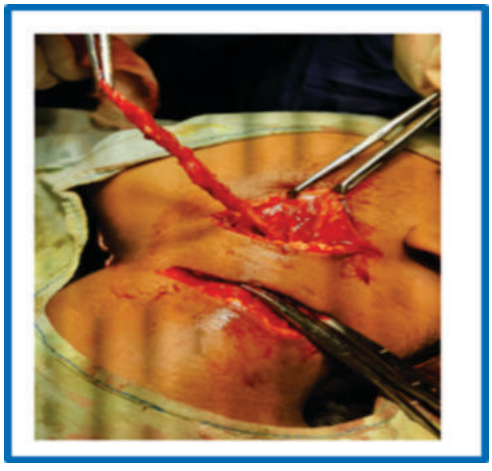

Image 3: Surgical excision using step ladder incision.

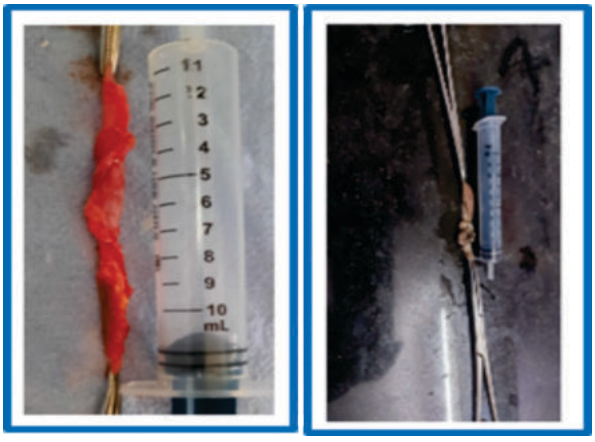

Image 4: B/L fistulous tract dissected and removed.

Image 4: Post-op picture.

DISCUSSION

Branchial apparatus anomalies are the second most common congenital anomalies of the head and neck region mostly present during childhood. However, they may get sometimes ignored and discovered in adulthood.

Congenital anomalies of the branchial arches can lead to absence or malformation of the head and neck structures arising from the branchial apparatus. This may result in microtia, hearing loss and facial asymmetry. This can occur in isolation or as part of a syndrome e.g., BOR syndrome etc. Developmental anomalies of the branchial clefts lead to cysts, sinuses or fistulae formation that are found in the neck.

First branchial cleft anomalies are caused by embryonic duplication of the first cleft or first arch and cleft; which is classified by Work in 1972 into type I and type II respectively. These are rare, occurring in approximately 5% of branchial cleft anomalies. First branchial cleft anomalies present as an external fistula tract opening located anterior and inferior to the tragus (type I) or as a fistula tract between an anterior opening close to the angle of the mandible with a posterior opening found in the ear canal or conchal bowl (type II). The fistulae tracts are present from birth and can present with recurrent skin or ear discharge or may be asymptomatic. Treatment is with complete surgical excision of the tract, taking care not to injure the facial nerve; particularly relevant with type II lesions which have an inconstant relationship to the facial nerve which may run either deep or superficial to the fistula tract.

Second branchial cleft abnormalities are the most commonly encountered, representing approximately 95% of all branchial apparatus abnormalities. The second branchial pouch differentiates into the palatine tonsil and supratonsillar fossa. The second branchial cleft however, do not contribute to a definitive structure, instead, they fused with the third and fourth clefts to form a cavity which is temporary called as the Cervical Sinus of His. The second branchial fistula occurs when the second branchial arch fails to grow caudally over the third and fourth arches leaving remnants of the second, third and fourth branchial clefts in contact with their surface by a narrow canal. The anatomic relationship of the second branchial apparatus and cervical sinus is such that, the anomalies of the second branchial cleft may occur anywhere from the tonsillar fossa to the supraclavicular region in the neck.

Second cleft sinus or fistulae tracts are found along the anterior border of the sternocleidomastoid muscle. The tract extends cranially along the carotid sheath which can pass between the internal and external carotid arteries and may run deep to the hypoglossal nerve. The internal opening of a second branchial cleft fistula is mostly found in the tonsillar fossa. Management is with complete surgical excision of the tract, taking care to avoid injury to the carotid arteries or hypoglossal nerve.

Third and fourth branchial cleft abnormalities are rare and are difficult to differentiate clinically. Third cleft fistulae have a skin opening anterior to the sternocleidomastoid muscle in the lower third of the neck, with an internal opening in the piriform fossa, above the level of the superior laryngeal nerve and on the other side fourth cleft fistulae have similarly located skin openings but the internal opening in the piriform fossa is located below the level of the superior laryngeal nerve. Clinical features are recurrent skin infections and abscesses. Recurrent thyroiditis is a potential feature of fourth cleft abnormalities

as the fistula tract may be closely related to the thyroid gland. [1-6]

Most often the branchial fistula arising from the second branchial cleft are unilateral. Bilateral branchial fistulae are seen in less than 4% cases. It is not uncommon to find these anomalies to run in families especially in case of bilateral branchial fistula in the form of Branchio-oto-renal syndrome or BOR syndrome. BOR syndrome shows autosomal dominant inheritance with presence of ear malformations, pre-auricular pits, renal anomalies and the branchial apparatus anomalies. BOR syndrome is associated with mutations of EYA1 gene. If there is a suspicion of BOR syndrome then the patient should be examined thoroughly and abdominal sonography should be done to rule out renal anomalies. Branchial fistulae are often not true fistula since a true internal opening is usually not present is often covered with a thin membrane.

Clinical examination alone could provide sufficient information to make a diagnosis of branchial anomalies in the majority of cases. However, exceptions to this will be in the case of infected lesions or abnormal fusion of branchial clefts. Imaging is necessary to assist in making the definitive diagnosis as well as in the surgical planning. Various imaging modalities are available such as fistulography, ultrasonography, computed tomography (CT scan) and magnetic resonance imaging (MRI) have been used to help confirm the diagnosis. An alternative method of diagnosing a fistula is through injecting methylene blue into the external opening, in which case if the fistula is complete, the blue dye may appear in the throat. Occasionally, this test may be negative pre-operatively but positive at the time of operation due to the relaxation of neck muscles under general anaesthesia.

The standard treatment of branchial sinus and fistula is total surgical excision with meticulous dissection. Care should be taken to excise the tract in-toto to avoid recurrence which can be challenging to deal with. An infection of the tract should be dealt with antibiotics and surgery may be delayed until such an event subsides. Histological examination of the fistula tract often shows the presence of squamous epithelium or columnar epithelium. Rarely muscle fibres or lymphoid aggregates may also be seen.

Surgical Techniques

Surgical techniques described in the literature are unique to the course of the fistula. The stepladder method, which involves multiple stab incisions and meticulous dissection was described by Bailey in 1933. Taylor (1977) advocated stripping of the fistulous tract using wire stillettes, vein strippers or arterial intimal strippers that are passed into the tract.[7] This method enables removal of the tract without extensive dissection and incisions, resulting in lesser scar. The disadvantage of this blind method is the possibility of injuring the adjacent vital structures.

We have opted for a direct vision using step ladder incision method by making an elliptical incision around the neck sinus along the anterior border of the sternocleidomastoid muscle during the dissection in order to avoid such injuries. The fistulae were located along the carotid sheath, passing between the internal and external carotid arteries. The tract ascended superficially, anterior to the sternocleidomastoid muscle and then got obliterated.

There was no necessity to perform the tonsillar incision in the present case, as the tapering shape of the fistula at its superior end enabled its delivery through the neck incision. The Oro-cervical or pull through fistulectomy that was described by Talaat (1992) gives a better cosmetic result, however rough handling of the fistulous tract may result in incomplete removal and recurrence. [8] In retrospect, a lazy 'S' shaped incision along the anterior border of sternocleidomastoid would leave a more pleasing scar rather than the vertical incision that was performed.

Among the surgical complications reported are local infection, seroma or haematoma, neurological injuries to the hypoglossal and upper laryngeal nerves, unsightly scars, persistent fistulas and injury to the internal jugular vein.[9]

Healing was uneventful in the present case with no complications and the case got satisfactorily discharged on 8th postop day.

Patient's daughters are under examination and will be promptly plan for surgical exploration and excision of the fistulae, but signs and symptoms related to BOR syndrome has not been found yet in the children, but further studies are required for confirming the exclusion. Till now it seems that the gene might showing variable penetration among the family members.

CONCLUSIONS

The present case illustrates the classical features of bilateral branchial fistulae. With careful dissection under direct vision, the fistulae were completely removed in one piece and surgical morbidity was minimal with uneventful discharge of a stable patient. Similar cases of branchial fistulae with no otologic or kidney symptoms were present in the family, with a dominant autosomal transmission pattern.

REFERENCES:

1. Morgan AG. Benign disease of neck, Scott Brown's otolaryngology, 6th edn. Butterworth Neineman: 1997. p. 5\16\4-5\16\5.
2. Moore KL, Persaud TVN, Torchia MG. The Developing Human: Clinically Oriented Embryology. 10th ed. Philadelphia, PA: Elsevier; 2016.
3. Schoenwolf GC, Bleyl SB, Brauer PR, Francis-West PH. Larsen's Human Embryology. 5th ed. Philadelphia, PA: Churchill Livingstone; 2015.
4. Shekhar C, Kumar R, Kumar R, Mishra SK, Roy M, Bhavana K The Complete Branchial Fistula: A Case Report. Indian J Otolaryngol Head Neck Surg 2005;57:320-22.
5. Sharma P, Marwah S, Nityasha et al. Bilateral branchial sinus: a case study. Int J Health Sci Res. 2017; 7(6):358-361.
6. Laura Warner, Christopher Jennings and John C. watkinson surgical anatomy of the neck, Scott Brown's otolaryngology, 8th edn. 2019. p. 541-543.
7. TAYLOR PH, BICKNELL PG. Stripping of branchial fistula – a new technique. J Laryngol Otol 1977; 91: 141-149.
8. TALAAT M. Pull through branchial fistulectomy: A technique for the Otolaryngologist. Ann Otol Rhinol Laryngol 1992; 101: 501-502.
9. AGATON-BONILLA FC, GAY-ESCODA C. Diagnosis and treatment of branchial cleft cysts and fistulas. A retrospective study of 183 patients. Int J of Oral Maxillofac Surg 1996; 25: 449-452.