A RARE CAUSE OF CERVICAL LIPOMAS: BANNAYAN-ZONANA SYNDROME

UNE CAUSE RARE DE LIPOMES CERVICAUX: SYNDROME DE BANNAYAN-ZONANA

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ABSTRACT

Introduction: Bannayan-Zonana syndrome (BZS) is a rare polymalformative condition characterized by macrocephaly, multiple lipomas, intestinal polyps, and vascular hamartomas. Cervical symptoms are extremely rare.
Observation: We report a case of a 25-year-old woman referred for compressive cervical swelling and inspiratory dyspnea. Previously diagnosed with BZS due to macrocephaly, diffuse lipomas, and intestinal polyps, her cervical MRI showed two large masses. Surgical excision of these lipomas resulted in a successful outcome with no recurrence after one year.
Conclusion: This case emphasizes the importance of recognizing BZS with compressive cervical lipomas, an atypical but critical manifestation, to prevent severe complications such as airway obstruction.
Key words: Bannayan-Zonana, Syndrome, Lipoma, Imaging

RÉSUMÉ

Introduction : Le syndrome de Bannayan-Zonana (SBZ) est une affection polymalformative rare caractérisée par une macrocéphalie, de multiples lipomes, des polypes intestinaux et des hamartomes vasculaires. Les symptômes cervicaux sont généralement asymptomatiques.
Observation: Nous rapportons le cas d'une femme de 25 ans adressée pour une tuméfaction cervicale compressive et une dyspnée inspiratoire. Précédemment diagnostiquée avec le SBZ en raison d'une macrocéphalie, de lipomes diffus et de polypes intestinaux, son IRM cervicale a révélé deux masses volumineuses. L'excision chirurgicale de ces lipomes a abouti à un résultat satisfaisant, sans récidive après un an.
Conclusion: Ce cas souligne l'importance de reconnaître le SBZ avec des lipomes cervicaux compressifs, une manifestation atypique mais critique, afin de prévenir des complications graves telles que l'obstruction des voies respiratoires.
Mots-clés: Syndrome, Bannayan-Zonana, lipome, imagerie

INTRODUCTION:

Bannayan-Zonana syndrome (BZS) is a polymalformative syndrome first described by Bannayan in 1971 [1], and was recognized as an autosomal dominant inherited disorder by Zonana in 1976 [2]. The main signs are macrocephaly, multiple lipomas, intestinal polyps, skin and vascular hamartomatous lesions [3]. Cervical symptoms are rarely discussed in the literature as they are very rare and mostly asymptomatic and non-life threatening.
We report a case of BZS in a 2,5-year-old girl, who was addressed to our Otolaryngology department for compressive cervical sweeling with dyspnea.

Patient presentation:

A 2,5-year-old girl was referred to our Otolaryngology department for cervical swelling with recent dyspnea. Six months previously, she was operated for intestinal obstruction in paediatric surgery department. Faced with the symptomatology (Macrocephaly, diffuse lipomas, and intestinal polyps) presented by this child, BZS was mentioned. The circular cervical swelling appeared at 4 months of age. The development of this mass was progressive, painless and without signs of respiratory or digestive compression. On clinical examination, bilateral laterocervical masses (12 cm in diameter each one) were renitent, painless, mobile in relation to the skin, and fixed to the deep plane. There was no "thrill" or murmur in the cervical area. The child had normal neuromotor development. A cervical MRI showed 2 voluminous basi-cervical fat-containing masses with T1 and T2 hypersignal. Theses masses dissapear after saturation of fat and are not enhanced after injection of gadolinium. They are measuring 73 x 45 mm on the left and 66 x 41 mm on the right with a bilateral axillary

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extension and in the left prescapularis. They push back and inside the jugulo-carotid vascular bundle which are permeable. At the bottom, they are prolonged by a mediastinal fat-containing formation moulding the pericardium without mass effect (Figure 1).

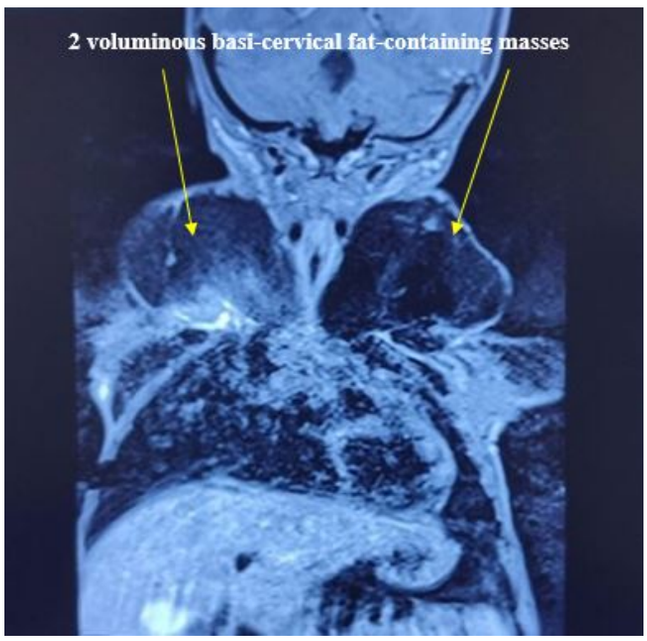

Figure 1: cervical MRI showing 2 voluminous basi-cervical fat-containing masses which dissapear after saturation of fat.

We performed a transverse cervicotomy through a bi-mastoid incision and we proceeded to the step-by-step dissection of the lipomas and the identification of the internal jugular vein, the carotid artery and the vagus nerve that have been preserved, as well as the spinal nerve (Figure 2).
The excision was complete for the entire two lipomas.

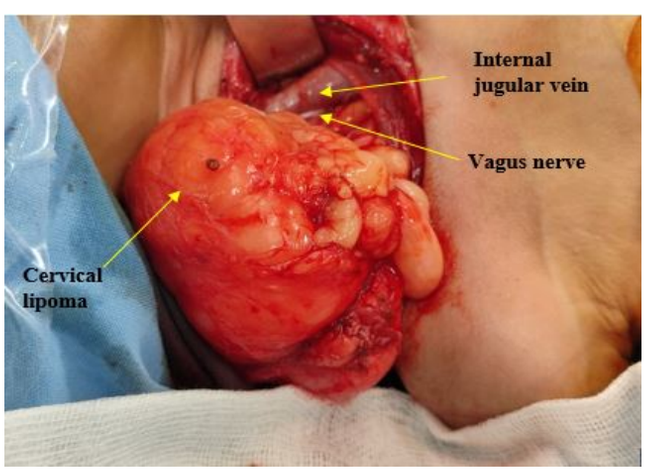

Figure 2: Intraoperative image, showing the surgical removal of cervical lipoma.

The postoperative courses were simples. The result of the anatomopathological examination revealed lipomas of the right and left latero-cervical regions. We are now at one year post operative, and no local recidivism is observed.

DISCUSSION:

The originality of the observation reported here lies in its occurrence in a girl, its sporadic nature, the rapidly increase in the volume of the lipomas and the unusual rise of inspiratory dyspnoea, and also comes from the complete clinical presentation of BZS.
The BZS is a rare poly-malformative syndrome characterized by macrocephaly and multiple soft tissue and visceral hamartomas and lipomas [4]. Inheritance is by autosomal dominant transmission with male predominance; however, occasional sporadic cases have been reported [5,6]. In 1992, Hayashi [3] reported 22 cases found in available literature. Gujrati [4] report 9 affected boys in 1998. In total, we found about 35 cases of BZS in the available literature [6-10]. To our knowledge, our patient is the first case of the literature of BZS with compressive cervical lipomas with dyspnoea.
The poly-malformative presentation of BZS may include lipomas, intestinal polyps, angiomas, lymphangiomas and vascular malformations [1,4,8,9]. Half of the patients develop moderate to severe psychomotor and mental retardation. This association of macrocephaly, cervical lipomas and intestinal polyps evoked the diagnosis in our patient who had normal neuromotor development.
In BZS, the lipomatous lesions are multiple and of variable localization [1,4,8,9]. The evolution of lipomas is gradual, causing functional and aesthetic complications. Deaths have been reported, caused by visceral lesions [6].
Imaging techniques are of interest for the diagnosis of this syndrome. Arteriovenous doppler and angio-MRI occupy a prominent place because of their performance in the diagnosis of the vascular nature of lesions and in their precise location [3]. Most BZS cutaneous vascular tumours are slow growing and easy to resection, while visceral and intracranial damage can cause symptomatic bleeding or mechanical compressions, especially in the spinal cord or spinal nerve roots. They may then require urgent surgical resection [3,4,6]. In some cases, the therapeutic possibilities are reduced, in particular because of the high risk of postoperative complications, especially haemorrhagic [3]. Therefore, abstaining from therapy and monitoring these patients remains the preferred attitude to adopt. In our patient, faced with the increase of the cervical lipomas volume and the functional gene, we proceeded to the excision of the entire two lipomas.
As reported by Bannayan [1] and Hayashi [3], 3 of the 22 patients died of diffuse infiltrating lipomatosis (1 case) and of arteriovenous malformations and lipomatosis (2 cases). These 3 patients were all females and were sporadic without family history similarly to our patient. The sporadic case seems to show worse prognosis than the familial case [3]. Thus, the severity of SBZ comes from arteriovenous malformations with a fatal outcome in most cases [3,4,6-8]. Mcheik [8] propose biannual clinical paediatric monitoring: depending on the clinical signs, a cervical or abdominal ultrasound

would be performed. He recommends a cerebral MRI and a digestive endoscopy every five years. In women, the breast examination is associated with an annual mammogram. Moreover, SBZ is considered as a cutaneous marker of malignant tumours. It also involves a greatly increased risk of breast, thyroid, endometrial, kidney and intracranial tumours. Monitoring should be continuous for early detection of these tumours which often occur in adulthood.

CONCLUSION:

This case highlights the rare presentation of Bannayan-Zonana syndrome with compressive cervical lipomas, emphasizing the need for early recognition and intervention to prevent life-threatening airway complications. Multidisciplinary management and regular monitoring are essential in managing the complexities of BZS.

Declaration of Conflicting Interests:

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Ethical approval:

The parents of our patient gave their approval for reporting the case.

REFERENCES:

1. Bannayan GA. Lipomatosis, angiomatosis, and macrencephalia. A previously undescribed congenital syndrome. Arch Pathol. 1971;92(1):1-5.
2. Miles JH, Zonana J, Mcfarlane J, Aleck KA, Bawle E. Macrocephaly with hamartomas: Bannayan-Zonana syndrome. Am J Med Genet. 1984; 19(2):225-34.
3. Hayashi Y, Ohi R, Tomita Y, Chiba T, Matsumoto Y, Chiba T. Bannayan-Zonana syndrome associated with lipomas, hemangiomas, and lymphangiomas. J Pediatr Surg. 1992;27(6):722-3.
4. Gujrati M, Thomas C, Zelby A, Jensen E, Lee JM. Bannayan-Zonana syndrome: a rare autosomal dominant syndrome with multiple lipomas and hemangiomas: a case report and review of literature. Surg Neurol. 1998; 50(2):164-8.
5. Zonana J, Rimoin DL, Davis DC. Macrocephaly with multiple lipomas and hemangiomas. J Pediatr. 1976; 89(4):600-3.
6. Khadir K, Eddaoui A, Chiheb S, Bourhnane D, Chaffik N, Azzouzi S, et al. Syndrome de Bannayan-Zonana d'évolution fatale. Arch Pédiatrie. 2009; 16(4):364-7.
7. Naidich JJ, Rofsky NM, Rosen R, Karp N. Arteriovenous malformation in a patient with Bannayan-Zonana syndrome. Clin Imaging. 2001;25(2):130-2.
8. Mcheik JN, Cordier MP, Longy M, Levard G. Macrocephaly and multiple hamartoma: a very variable entity. Arch Pediatr Organe Off Soc Francaise Pediatr. 2004;11(1):33-6.
9. Shimpuku G, Fujimoto K, Okazaki K. A case of Bannayan-Zonana syndrome. Masui. 2005;54(5):535-7.
10. Klifto MR, Balaratnasingam C, Weissman HH, Yannuzzi LA. Bilateral coats reaction in Bannayan-Zonana syndrome: a single case report. Retin Cases Brief Rep. 2017;11(3):286-9.