CLINICAL CASE

TRANSNASAL ENDOSCOPIC APPROACH ON A RARE CASE OF CRANIAL JUVENILE FIBROMATOSIS

Cristina Ghiţă[1], B. Mocanu[2]
[1]The University of Medicine and Pharmacy "Carol Davila", Bucharest, Romania
[2]BRAIN Institute, Monza Hospital, Bucharest, Romania

Corresponding author: Cristina Ghiţă
Phone no. 0040721900265
E-mail: cristina.ghita8@yahoo.com

Abstract

Juvenile fibromatotsis is very uncommon and determins locally agressive fibroblastic proliferation. The location in the paranasal sinus is even more uncommon and only 6 more cases have been reported in the world until now, this being the first one in our country. The election treatment for this pathology is surgery with large resection margins, radio and chemiotherapy being used only for unresectable tumours or agressive reccurences. Our aim is to present an exceptionally rare case of intracranial juvenile fibromatosis on a 2 year old pacient treated surgically in a transnasal endoscopic manner using high precision equipment.

Keywords*: juvenile, fibromatosis, intracranial*

Introduction

Infantile fibromatosis represents a benign pathology of the fibrous tissue, characterized by local infiltration and proliferation of mature fibroblasts and collagen. However, there is no celular anaplasia present [1]. A specific attribute of this condition would be its capacity to invade surrounding structures, but without creating distant metastasi [2]. The cause of infantile fibromatosis is yet to be discovered, but numerous factors such as genetic, viral agents, estrogen, trauma or irradiation are thought to be related [3].

On macroscopic examination, infantile fibromatosis appears as a white mass with poor margins which is very invasive into surround tissues. Microscopically, its structure is formed of fibroblasts and fibrocytes along with myofibroblasts situated in a collagenous to myxoid stroma [4]. In order to establish the exact size of the tumour for further surgical approach and the follow-up treatment, radiological evaluation is performed [5]. The treatment consists mainly of surgery and radiotherapy [6]. Gold standard surgical approach is en-bloc resection, only if the tumour is located in the head and neck and its size does not increase the chance of morbidity after excising it [7].

Case presentation

We present the case of a 2 year old female child who was known with right nasal cavity tumour with extension to the right orbit, when she presented to our clinic. She was diagnosed in another clinic with juvenile hyaline fibromatosis based on histopathology and imunohistochemistry tests. The patient first experienced obstruction of the right nasal cavity followed by swelling of the internal angle of the

right eye. The evolution of this pathology led to right eye proptosis (Figure 1).

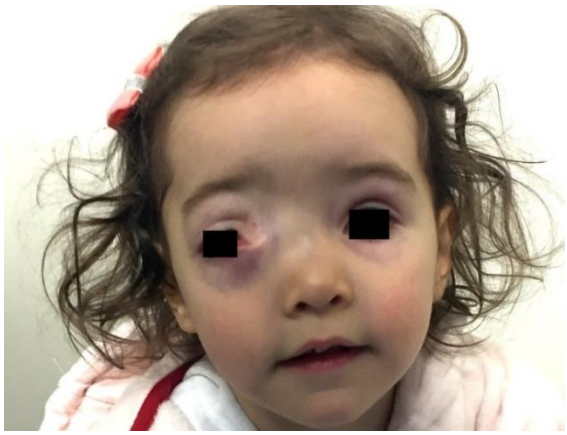

Figure 1 - Preoperatory aspect of the patient

Further investigations revealed that the tumour had protuded significantly in the right orbit and had also extended to the ethmoid, sphenoid sinus and maxillary sinus (Figures. 2, 3, 4). Bearing in mind the proximity of the tumour towards the optic nerve and the ethmoid, immediate surgical approach was considered.

Transnasal macroscopic ablation of the tumour was performed, under general anestesia, using a neuronavigation system with a 0,3 mm error. The procedure was started by placing the patient in supine position with orotracheal intubation and antibiotic treatment.

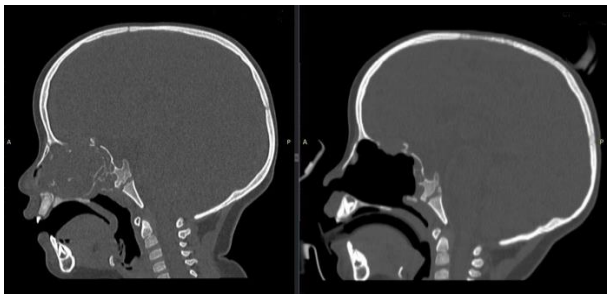

Figure 2 - Comparative sagital CT preoperatory (left) and postoperatory (right)

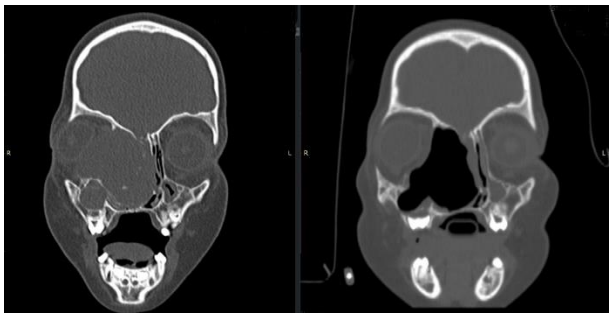

Figure 3 - Comparative coronar CT preoperatory (left) and postoperatory(right)

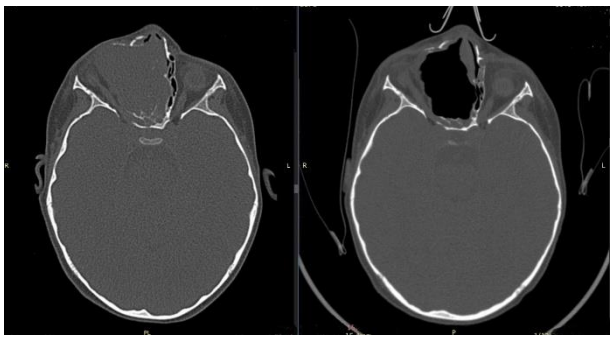

Figure 4 - Comparative axial CT, preoperatory (left) si postoperatory (right)

On endoscopic inspection of the nasal cavity a large erythematous tumour with firm capsulae and friable, hemorrhaging content. Moreover, we noticed that the tumour was comprising the the right inferior nasal turbinate, while also producing the laterodeviation of the nasal septum to the left, blocking almost completely the access to the left nostril. In addition to this, the right eye was pushed laterally. The tumour extended up to the ethmoid bone, occupied the right maxillary sinus entirely and the right sphenoethmoidal recess. We also noticed that the tumoral capsulae had infiltrated in the right lamina papyracea and partially in the orbiatry periosteum.

In order to perform the ablation of the tumour we used the „peace-meal" technique. There was an abundant bleeding at the site of the maxillary artery which was overcome with bipolar hemostasis (3OW), one unit of blood and one unit of plasma were transfused. At the site of tumoral resection hemostastis was performed using monopolar cauterisation (14W) was applied, followed by HHC cream, lidocaine, tetracyclin and rifampicin. Consequently, nasal lavage with Axentine and Cathejell is applied.

During the procedure were performed two biopsies, one from the capsulae and one from the content of the tumour, which were sent to histopathology and immunohistochemistry for further evaluation. The hemostasis was efficient, with no need for nasal packing.

Postoperatory, while the patient was still under general anesthesia a contrast CT of the paranasl synuses was performed revealing entire tumour resection with clear margins (Figures. 2, 3, 4). In this way, we managed to preserve nerves and blood vassels using a minimally

invasive approach. In the postoperatory picture the complete absence of swelling at the inner corner of the right eye can be noticed (Figure 5).

Figure 5. Postoperatory aspect of the patient

The postoperatory evolution was favourable, and the patient was discharged 3 days after the intervention.

Figure 6. Aspect of the patient 6 months after the surgery

The final histological report confirmed the presence of hyaline juvenile fibromatosis. Microscopically, the tisular fragments which were biopsied consisted of proliferated fusiform cells with no abnormal nucleii, placed in short bundles or isolated in hyaline mass. The fragments contained also respiratory mucosa with oedema and inflammatory infiltrate. Bony trabecular patterns were also found in the proximity of the tisular fragments.

Immunohistochemistry tests were positive for antibodies such as: Ki67, Vimentin (found in fusiform cells), SMA, CD34 (found in vascular structures) and S100 (in rare cells).

The patient was discharged with antibiotic, antiinflammatory treatment, as well as local treatment for the regeneration of the nasal cavity mucosa. On her 6 months follow-up we noticed that the patient was healing nicely with no complications (Figure 6).

Discussions

The group of fibromatoses has been defined as consisting of non-metastasising fibrous tumours that invade locally and recur after surgical excision [8,9]. On occasion, they may regress spontaneously [10-14].

Juvenile hyaline fibromatosis is a disorder with autosomal-recessive transmission. Its etiology has not yet been established, but it is known that a gene located on 4q21 is responsible [15]. Research has shown that JHF is a pathology that affects the connective tissue in which fibroblasts produce aberrant quantities of glycosaminoglycans [16]. The tumour connsists mostly of dermatan sulfate, but also of condroitin sulfate and hyaluronan, whereas normal skin is defined by high quantities of hyaluronan [17]. Normally it affects one or more siblings, more frequently in children aged 2 to 5 [15].

Literature shows that common locations of this tumour are the abdominal wall, the shoulders and upper arms, where the connective tissue that overlies the fascia or aponeurosis starts to proliferate. The location of IF in the head and neck has not been reported very often [4]. In this case, the patient was at a higher risk due to the possibility of permanent vision loss associated with neurological complications arising from the proximity of the tumour to the ethmoid. This potential unfortunate complication of the pathology is of greater importance considering the age of the patient and the long-term physical challenges she

would have need to overcome, had the optic nerve been damaged.

Conclusions

To conclude, the most important aspect of solitary fibromatosis is the recurrence. This is why, resection with clear margins must always be attempted, as well as thorough follow-up examination and CT investigation. Other important features of this pathology are its capacity to invade into surrounding structures as well as its inability to methastasise. The goldstandard treatment for this type of tumour is surgical excision with clear margins, particularly challenging in head locations due to the limited space allowed for surgical instruments.

Complications of Juvenile hyaline fibromatosis can be successfully overcome with extensive clinical and imagistic examination, accurate microsurgery neuronavigation systems that allow adequate margins and thorough follow-up examinations that prevent potential aggravating recurrences.

References

[1]Plaat BE, Balm AJ, Loftus BM, Gregor RT, Hilgers FJ, Keus RB. Fibromatosis of the head and neck. Clin Otolaryngol. 1995;20:103–108.

[2]Abdelkader M, Riad M, Williams A. Aggressive fibromatosis of the head and neck (desmoid tumours) J Laryngol Otol. 2001;115:772–776.

[3]Hoos A, Lewis JJ, Urist MJ, Shaha AR, Hawkins WG, Shah JP, Brennan MF. Desmoid tumors of the head and neck—a clinical study of a rare entity. Head Neck. 2000;22:814–821.

[4]Atul Gupta, Satish Nair, Ajith Nilakantan, and Amit Sood. Indian J Otolaryngol Head Neck Surg. 2015 Mar; 67(Suppl 1): 154–157.

[5]Ahn JM, Yoon HK, Suh YL, Kim EY, Han BK, Yoon JH, et al. Infantile fibromatosis in childhood: findings on MR imaging and pathologic correlation. Clin Radiol. 2000;55:19–24.

[6]Wang CP, Chang YL, Ko JY, Cheng CH, Yeh CF, Lou PJ. Desmoid tumor of the head and neck. Head Neck. 2006;28:1008–1013

[7]Nuyttens JJ, Rust PF, Thomas CR, Turrisi AT., III Surgery versus radiation therapy for patients with aggressive fibromatosis or desmoid tumors. A comparative review of 22 articles. Cancer. 2000;88:1517–1523

[8]Hidayat AA,Font RL. Juvenile fibromatosis of the periorbital region and eyelid: a clinicopatholgic study of six cases. Arch Ophthalmol 1980; 98: 280-5.

[9]Stout AP. Juvenile fibromatoses. Cancer 1954; 7: 953-78.

[10]Baer JW, Radkowski MA. Congenital multiple fibromatosis: a case report with review of the world literature. AJR 1973; 118: 200-5.

[11]Baird PA, Worth AJ. Congenital generalized fibromatosis: an autosomal recessive condition? Clin Genet 1976;9: 488-94.

[12]Benjamin SP, Mercer RD, Hawk WA. Myofibroblastic con- traction in spontaneous regression of multiple congenital mesenchymal hamartomas. Cancer 1977; 40: 2343-52.

[13]Briselli MF, Soule EH, Gilchrist GS. Congenital fibromatosis: report of 18 cases of solitary and 4 cases of multiple tumors. Mayo Clin Proc 1980; 55: 554-62.

[14]Schaffzin EA, Chung SMK, Kaye R. Congenital generalized fibromatosis with complete spontaneous regression: a case report. J Bone Joint Surg (A) 1972; 54: 657-62.

[15]Gupta LK, Singhi MK, Bansal M, Khullar R, Jain V, Kachhawa D. Juvenile hyaline fibromatosis in siblings. Indian J Dermatol Venereol Leprol. 2005;71:115–8

[16]Weiss SW, Goldblum JR. Fibrous tumors of infancy and childhood. In: Strauss M, editor. Enzinger and Weiss's Soft Tisssue Tumors. 4th ed. St. Louis: Missouri Mosby; 2001. pp. 363–7

[17]Katagiri K, Takasaki S, Fujiwara S, Kayashima K, Ono T, Shinkai H. Purifaction and structural analysis of extracellular matrix of a skin tumor from a patient with juvenile hyaline fibromatosis. J Dermatol Sci. 1996;13:37–48