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Case Report

Biphenotypic Sinonasal Sarcoma

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Summary

Biphenotypic nasal sarcoma has recently been described and included in the WHO 2017 classification. It is a low-grade sarcoma characterized by rearrangements of PAX3. Its rarity and its non-specific histological appearance make its diagnosis difficult.

Histological features, immunohistochemical profile and main diagnosis differentials of biphenotypic nasal sarcoma will be discussed with a case presentation.

Keywords: Sarcoma; Sinonasal biphenotypic sarcoma; Nasal cavity

Case Presentation

56-year-old patient with no particular pathological history whose history goes back to three years marked by the appearance of a bilateral nasal obstruction, anosmia and anterior rhinorrhea, ageusia with pain in the left hemiface and history of epistaxis.

The evolution was marked by a deformation of the nasal pyramid and an increased pain. The endoscopic examination reveals a tissue process that bleeds on contact and is lined with mucopurulent secretion.

On imaging, the CT scan reveals a lesional process in the left nasal cavity and the MRI reveals a left tumoral process to be compared with the histology.

The patient benefited from an endoscopic endonasal surgery with complete and wide ablation. The histological examination after immunohistochemistry came back in favor of a biphenotypic nasal carcinoma. the patient is referred for radiochemotherapy. Post-operative checks are unremarkable.

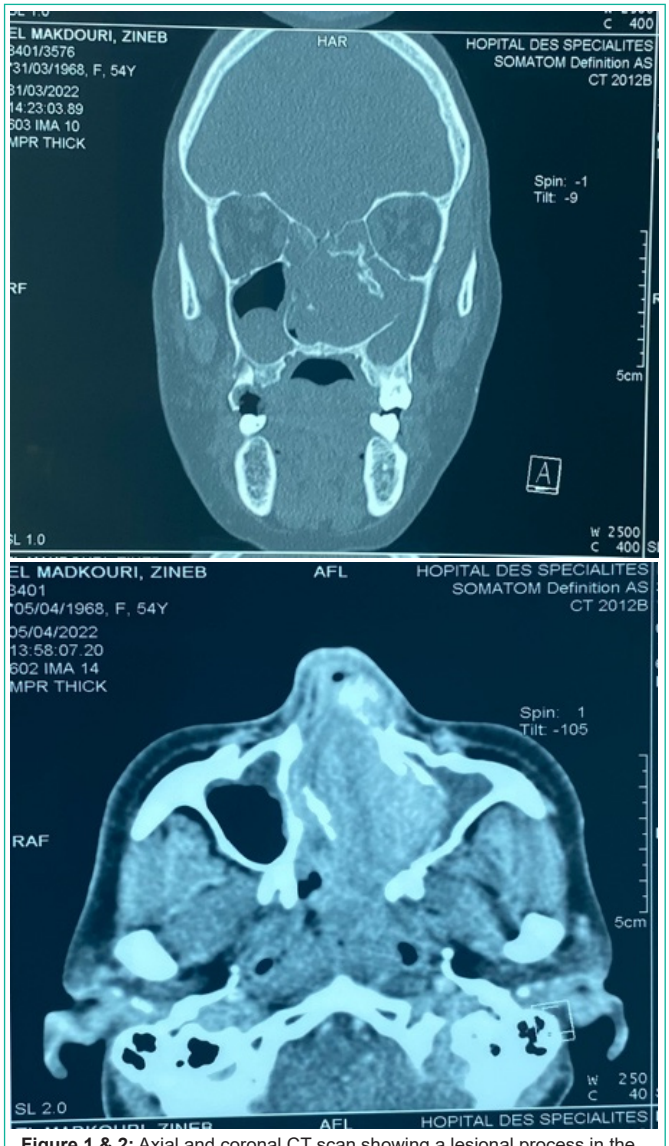

Figure 1 & 2: Axial and coronal CT scan showing a lesional process in the left nasal cavity.

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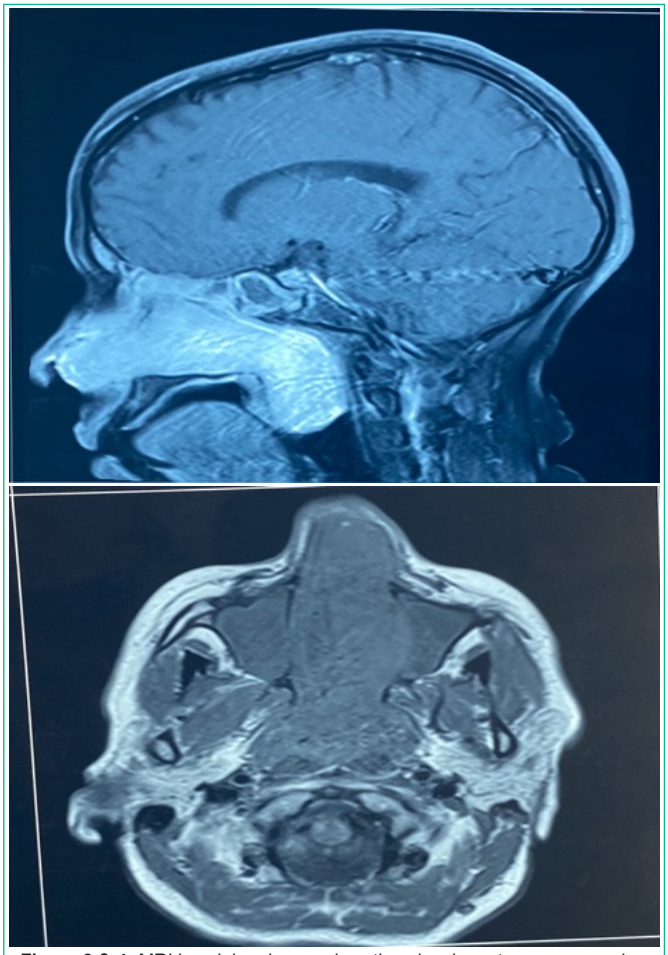

Figure 3 & 4: MRI in axial and coronal section showing a tumor process in the left nasal cavity.

Discussion

Lewis and al. [1] recently described a group of low-grade spindle cell sarcomas occurring exclusively in the nasal cavity and paranasal sinuses. They called this new entity "sarcoma". The same group then renamed the entity: Biphenotypic Nasal Sarcoma (NBS). The later nomenclature has been included in the latest version of WHO [2]. They usually occur in women (2 women for one man), in patients aged from 24 to 85 years old (52 years on average) and affects the nasal cavity and the paranasal sinuses.

Clinically, SNB is slow growing with a local aggressiveness, including bone invasion with sometimes intracranial and/or intraorbital extension. Close to a third of patients with SNB have recurred locally within 5 years [3], but none of the tumors gave rise to metastasis [1,3]. Recently, two cases of death linked to SNB have been reported. It was noted that these patients presented a SNB with intracranial extension [3]. Those data suggest that surgical treatment, even if it must be aggressive in case of orbital or intracranial invasion, must be as broad as possible in order to avoid any recurrence local.

Histologically, SNB presents as a uniform proliferation of spindle cells organized into fascicles within the chorion. This proliferation is poorly circumscribed, not encapsulated, with an infiltrative character. A "herringbone" arrangement of the beams is frequently observed and the vessels can take a "shaped" appearance of deer antlers. The nuclei of tumor cells are elongated and uniform. The presence of a respiratory epithelium invaginated, trapped by spindle cells is frequently observed, but not specific [3]. This histological characteristic has also been reported in fibrosarcomas and tumors of the peripheral nerve sheaths of the fossa nasal [3]. This surface epithelium can sometimes contain oncocytic or squamous metaplasia. The SNB is therefore histologically low grade, without pleomorphism nuclear, without hyperchromasia, without necrosis and weakly proliferating. It is possible to observe rhabdomyoblastic differentiation in some areas [4-6].

On immunohistochemical study, tumor cells express PS100, focally or diffusely, actin smooth muscle or calponin. Sometimes an expression of desmin, EMA, CD34, MYOD1, myogenin or pan-cytokeratin may be seen. The expression-catenin is generally nuclear, so focal or diffuse, sometimes cytoplasmic [3,6]. The SOX10 is always negative [6]. Recently, it was reported a nuclear and diffuse expression of PAX3 and PAX8 by immunohistochemistry with a specificity of 98% for PAX3 and 75% for PAX8 [7].

The SNB is characterized by the translocation t (2; 4) (q35; q31.1) which is responsible for protein fusion between the transcriptional factor PAX3 and most often with a co-activator of the NOTCH signaling pathway, MAML3 [8]. Huang and al. [9], Wong and al. [10] have described a subset of cases with another partner, NCOA1 or FOXO1, often seen during focal rhabdomyoblast differentiation with immunoreactivity to myogenin. The PAX3-NCOA1 fusion transcripts and PAX3 FOXO1 are also found in rhabdomyosarcoma alveolar.

Regarding differential diagnoses, positivity of PS100 may suggest a nerve tumor like a schwannoma or malignant tumor of the sheath of the peripheral nerves. Especially since schwannomas nasal sinuses are not encapsulated and may be hypercellular. The presence of muscular differentiation, an expression of the PS100 of lower intensity and more focal, and the absence of SOX10 expression are sufficient arguments to eliminate the diagnosis of schwannoma [6]. It must be emphasized that the presence of an inflection muscle or epithelial differentiation is compatible with the diagnosis of a malignant sheath tumor peripheral nerves. However, the SNB does not present any necrosis nor significant nuclear pleomorphism. Furthermore, due to the focal expression of the cytokeratin and the cytological appearance, the diagnosis of monophasic synovialosarcoma is also possible. However, the absence of the SS18 gene rearrangement allows to eliminate this diagnosis. Finally, the vascularization of appearance (antler) raises the possibility of glomangiopericytoma and a solitary fibrous tumor.

The appearance cytology and the expression of PS100 argue against the glomangiopericytoma which is more epithelioid. Unlike SNB, solitary fibrous tumor presents a variable cellularity, does not express PS100 and expresses STAT6. Finally, a rearrangement of the PAX3 gene by Fluorescence in Situ Hybridization (FISH) is not found in differential diagnoses [8-11]. More recently, an oropharyngeal tumor presenting the same morphological and immunohistochemical aspects as SNB was reported [12]. However, as this entity does not carry a rearrangement of the PAX3 gene, it has not been classified as an SNB. In this case it was found on RREB1-MKL2 fusion transcript, responsible for overe-expression of MKL2. Thus, it is necessary to confirm the diagnosis of SNB by the presence of a rearrangement of the PAX3 gene.

Conclusion

SNB is a low-grade sarcoma developing in nasal passages and paranasal sinuses in patients aged from 24 to 85 years. The lesion is very homogeneous, made of slightly atypical spindle-shaped cells, organized in bundles, oriented in a herringbone pattern. Mitoses are rare. It exists often a non-specific intussusception of the adjacent respiratory epithelium. Tumor cells express PS100 and smooth muscle actin. Immunohistochemical expression of PAX3 and PAX8 may help in diagnosis. A rearrangement of the PAX3 gene is necessary for diagnosis. SNB presents local aggressiveness and can recur within 5 years after initial treatment. No metastasis has been reported in any case. Only two cases of death linked to the disease have been reported.

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