CASE REPORT

Primary intracranial alveolar soft part sarcoma in a 22 years old female: a rare case report with literature review

Muhammad Saad Babar,[1] Haseeb Mehmood Qadri,[2] Ammara Tabassam,[3] Muhammad Imran,[4] Muhammad Hassan Raza[5]

Abstract

Alveolar soft part sarcoma is a very rare malignant neoplasm of peripheral muscular, adipose or neural tissue. Its occurrence, as a primary intracranial tumour, is even rarer. To the best of our knowledge, only nine cases of primary intracranial alveolar soft part sarcoma exist in English scientific literature. We attempt to write a comprehensive review on this poorly understood intracranial malignancy, with no evident systemic lesions, such as in the case of our 22 years old patient. In addition to absent definitive proof of benefit of radiologic or chemotherapeutic management, we highlight the role of surgery as the primary treatment. Younger patients with this tumour may have a worse prognosis than elderly patients.

Keywords: alveolar soft part sarcoma, intracranial, headache, radiosurgery.

DOI: 10.47391/JPMA.6925

Submission completion date: 10-05-2022
Acceptance date: 20-10-2022

Introduction

In general, the prevalence of soft tissue sarcomas in Pakistan has been calculated to be 3.5% of all malignancies, which is greater than those of western countries, i.e. 1%.[1] Alveolar soft part sarcoma (ASPS) is a rare soft-tissue sarcoma affecting adolescents and young adults between 15 and 35 years of age.[2] It accounts for 0.5%–1% of all soft tissue tumours and predominantly occurs in females at a frequency of 60%.[3] The tumour is usually located in the deep soft tissue, mostly in the thighs or gluteal region; however, it is also found in other areas including the arms, genital area and chest region.[4] Alveolar soft-part sarcoma usually presents as a slow-growing, painless mass, but different symptoms can occur depending on the location of the tumour and its metastasis. It metastasises early in its course especially to the lungs and brain. However, metastasis to the lymph nodes is uncommon.[3] Genetically, Alveolar Soft Part Sarcoma (ASPS) is caused by an unbalanced chromosomal translocation, resulting in the formation of ASPL-TFE3 fusion gene.[2] Histopathology reveals a distinctive pseudo-alveolar structure of tumour cells divided by a thin fibrovascular septum.[5] CT and MRI studies are suggestive of ASPS, with conspicuous venous vascularity, but the final diagnosis is based on tissue biopsy.[6] Treatment of ASPS includes complete resection of the primary, as well as the recurrent tumour manifestation.[4] Vascular endothelial growth factor receptor–targeted tyrosine kinase inhibitors might be useful with metastatic disease.[7] The case of a young female diagnosed with primary intracranial alveolar soft part sarcoma. is presented.

Case Report

A 22-years-old female was referred to our tertiary care hospital in September 2021 from the district Headquarter Hospital, Sheikhupura with the history of severe generalized headache with associated episodes of nausea and vomiting, occurring on and off for the past one month. She also complained of vertigo. Her parents described three to four episodes of "drowsiness and confusion" experienced by their daughter on various days. She was also believed to have lost weight in the previous two months, and also complained of decreased appetite. Her CT brain report without contrast from the referred hospital stated "an ill-defined mass occupying left frontoparietal lobe, with oedema and slight midline shift". We ordered an MRI scan of her brain, which showed similar findings (Figure 1). Her physical examination at our outpatient department was normal, except that she was pale and lean. Her funduscopic examination revealed mild bilateral papilloedema. A left frontal craniotomy and excision of the lesion was planned for this case. A 4.5x2.7x2.0 centimeters mass was excised and the resected specimen was sent for frozen section analysis, which ensured that adequate excision was done and safe margins were achieved intra-operatively. The patient recovered from general anaesthesia smoothly and was shifted to ICU for close monitoring. There were no post-

[1,5]Department of Neurosurgery, Punjab Institute of Neuro-Sciences, Lahore General Hospital, [2]Department of Surgery, Lahore General Hospital, [3]Department of General Surgery, Gurkhi Trust Teaching Hospital, [4]Department of Pathology, Allama Iqbal Medical College, Lahore, Pakistan.
Correspondence: Haseeb Mehmood Qadri. Email: haseebmehmood18@yahoo.com
ORCID ID. 0000-0003-0675-9934

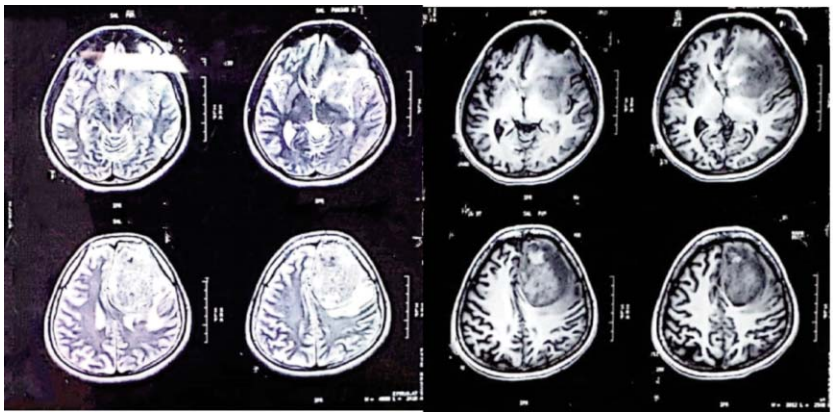

Figure-1: Pre-operative M.R.I. Brain Plain & Contrast (T1, T2): Sections showing the left sided frontoparietal space occupying lesion, creating a midline shift of the brain stroma towards the right side.

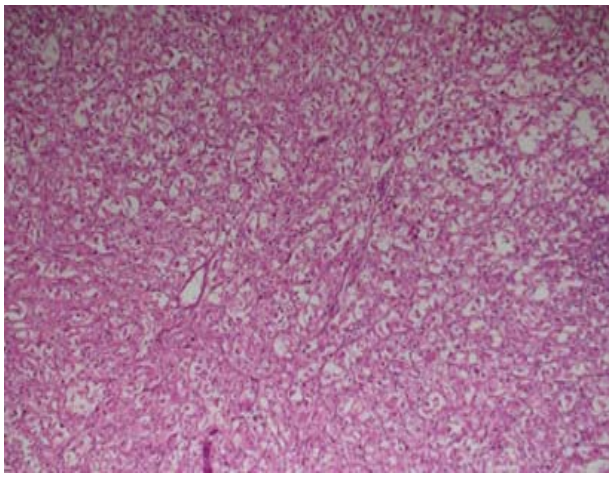

Figure-2: Histopathology of Alveolar Soft Part Sarcoma. Large, round to polygonal cells with well-defined cell borders; Abundant eosinophilic granular cytoplasm; Vesicular nucleus with a prominent nucleolus; Organoid & nest-like growth pattern; Central discohesion results in characteristic pseudoalveolar-like structures

operative complications of any kind. Her nutrition, mobility and wound care was encouraged as per protocol and she was discharged at her sixth post-operative day. The histopathology report was issued a week later, stating the suspicion of metastases of alveolar soft part sarcoma Figure-2. The immunohistochemistry was positive for cathepsin K and PAS. The patient was immediately called for follow up checkup and another detailed physical examination was performed, to especially assess the musculoskeletal system. However, the clinical examination was wholly normal. She was sent for a complete body computed tomography (CT). The CT showed no primary pathology anywhere in the body. Later on, she underwent a whole-body positron emission tomography (PET) scan, which was also unremarkable. Hence, it was concluded that this was a case of primary intracranial alveolar soft part sarcoma. Meanwhile, the patient remained symptom-free and she opted for a second opinion from field experts at a tertiary care hospital in Karachi. Two weeks after the primary surgery, an MRI scan was performed and radiosurgery was advised to cure the residual, fragmented intracranial lesion. She underwent one session of gamma knife stereotactic radiosurgery there and returned to our hospital in late October with the complaints of altered state of consciousness, vomiting and gastrointestinal upset. The patient was readmitted. The detailed clinical assessment revealed positive Brudzinski's and Kernig's signs along with neck stiffness. A clinical diagnosis of meningitis was made. Laboratory investigations revealed a clinical picture of bacterial meningitis, however no micro-organism was found on cultures, which had been sent to two different laboratories. A CT brain without contrast was ordered revealing changes suggestive of hydrocephalus. An intra-cranial pressure (ICP) catheter was placed and it pointed a severely raised intra-cranial pressure. External ventricular drainage was done to relieve the ICP. A post-drainage CT scan showed intraventricular haemorrhage and large areas of aerocoeles (Figure 3). The patient was managed conservatively. Her condition deteriorated and she expired after a week.

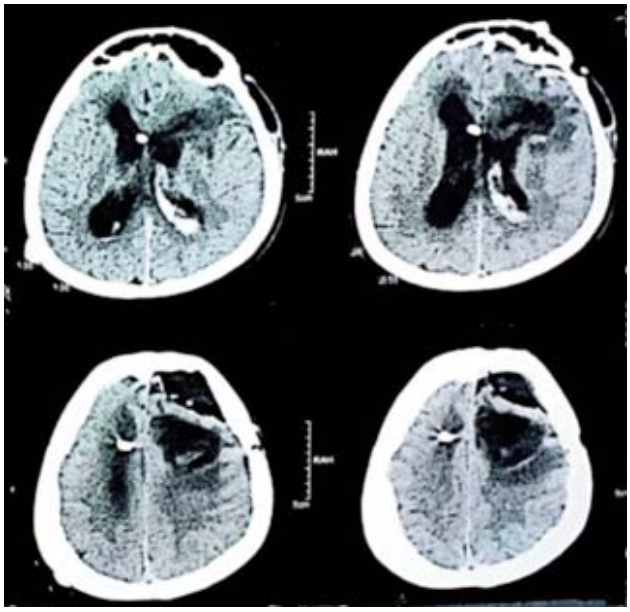

Figure-3: Post-drainage CT scan showing external ventricular drain in-situ, left lateral ventricular haemorrhage, dilated lateral ventricles and aerocoeles.

Discussion

Alveolar soft part sarcoma rarely develops as a primary intracranial malignancy.[8] To our knowledge, only nine such cases have been reported till now .[5]

The clinical presentation of ASPS is usually a painless mass and symptoms tend to be related to the mass effect of the tumour and metastasis.[2] In our study, the patient presented with a protracted course of vomiting with significant generalised weight loss.

Histologically, ASPS is characterised by large round to polygonal cells arranged in the form of nests containing ample vacuolated, eosinophilic cytoplasm. The 'pseudo-alveolar' pattern is attributed to the central necrosis and/or cell decohesion in the centre of the nests.[2] The histopathology report of our patient was consistent with the diagnosis of alveolar soft part sarcoma (ASPS), showing cells with rhabdoid morphology arranged in sheets as well as alveolar pattern with areas of necrosis. In our case, the immunohistochemistry showed cathepsin K and PAS positivity comparable to other studies in which tissue sections were well stained by the periodic acid–Schiff method after diastase digestion (PAS-D), with calretinin positivity in 46% and TFE positivity in 100%of the cases.[9]

Radiological investigations are contributory for the diagnosis and staging of ASPS. It usually shows hypervascular lesions with tortuous, dilated draining veins on contrast-enhanced CT or angiography. Magnetic resonance imaging (MRI) shows hyperintense images on T1 and T2 while contrast enhanced MRI shows intense post-contrast enhancement with peritumoural and intratumoural flow voids.[6]

The differential diagnosis may include arteriovenous malformations, haemangioendothelioma, haemangiopericytoma, rhabdosarcoma and metastasis from renal cell carcinoma, adrenocortical carcinoma, hepatocellular carcinoma, malignant melanoma and granular cell tumours. The median survival is 9 years for patients without metastasis and 3 years for those with metastasis.[6]

An adequate complete excision of primary tumour is the treatment of choice and is associated with a favourable outcome in localised disease.[3] Radical resection of primary cerebral ASPS is associated with a recurrence rate of 12% according to literature.[10] There are increased chances of local recurrence of ASPS. Hence a long-term follow-up along with clinical imaging is necessary for detecting any recurrence.[10]

Conclusion

Space occupying lesions of intracranial cavity present with non-specific symptoms as in our patient. Definitive management in the form of open surgical treatment was carried out timely, although no upfront chemotherapy was given. Detailed literature study reveals that no current guidelines exist on the radio-chemotherapy of primary intracranial alveolar soft part sarcoma. Despite elaborated efforts, the cause of intra-ventricular haemorrhage many days after the primary procedure could not be ascertained, which served as the terminal event for our patient.

Recommendations

For patients, with headaches of sudden onset and generalized distribution, it is advisable to consult physicians immediately. For physicians, it is highly inevitable to rule out all the possible causes of such headaches and refer the patient to subspecialty timely. Provision of a sterile microenvironment, a well-managed peri-operative and post-operative care is the responsibility of operating neurosurgeons. Research shall be sought in the areas of etiologic development of the primary intracranial entity of alveolar soft part sarcoma by oncologists.

Ethical Considerations: The written informed consent was taken from the parents of the patient for publishing her case. Detailed history was taken by the house officer and resident in charge of the patient after consent.

Disclaimer: None.

Conflict of Interest: None.

Source of Funding: None.

References

1. Hameed A, Sarwar G. Clinico-Pathological Features of Soft Tissue Sarcoma (STS). Ann King Edward Med Uni. 1999; 5:80. doi:https://doi.org/10.21649/akemu.v5i1.3423
2. Tanaka M, Homme M, Yamazaki Y, Shimizu R, Takazawa Y, Nakamura T. Modeling Alveolar Soft Part Sarcoma Unveils Novel Mechanisms of Metastasis. Cancer Res. 2017; 77:897-907. doi: 10.1158/0008-5472.CAN-16-2486.
3. Lin YK, Wu PK, Chen CF, Chen CM, Tsai SW, Chen PCH, et al. Alveolar soft part sarcoma: Clinical presentation, treatment, and outcome in a series of 13 patients. J Chin Med Assoc 2018; 81:735-41. doi: 10.1016/j.jcma.2018.01.006.
4. Pennacchioli E, Fiore M, Collini P, Radaelli S, Dileo P, Stacchiotti S, et al. Alveolar soft part sarcoma: clinical presentation, treatment, and outcome in a series of 33 patients at a single institution. Ann Surg Oncol. 2010; 17:3229-33. doi: 10.1245/s10434-010-1186-x.
5. Caporalini C, Giordano F, Moscardi S, Di Stefano G, Lenge M, Di Giacomo G, et al. Primary Intracerebral Alveolar Soft Part Sarcoma: Report of a Case and Review of the Literature. Int J Surg Pathol. 2022; 30:195-9. doi: 10.1177/10668969211027293.
6. Sood S, Baheti AD, Shinagare AB, Jagannathan JP, Hornick JL,

Ramaiya NH. Imaging features of primary and metastatic alveolar soft part sarcoma: single institute experience in 25 patients. Br J Radiol. 2014; 87:e20130719. doi: 10.1259/bjr.20130719.

7. Paoluzzi L, Maki RG. Diagnosis, Prognosis, and Treatment of Alveolar Soft-Part Sarcoma: A Review. JAMA Oncol. 2019; 5:254-60. doi: 10.1001/jamaoncol.2018.4490.
8. Tao X, Tian R, Hao S, Li H, Gao Z, Liu B. Primary Intracranial Alveolar Soft-Part Sarcoma: Report of Two Cases and a Review of the Literature. World Neurosurg. 2016; 90:699.e1-699.e6. doi: 10.1016/j.wneu.2016.02.005.
9. Chamberlain BK, McClain CM, Gonzalez RS, Coffin CM, Cates JM. Alveolar soft part sarcoma and granular cell tumor: an immunohistochemical comparison study. Hum Pathol. 2014; 45:1039-44. doi: 10.1016/j.humpath.2013.12.021.
10. Jo VY, Hornick JL. Pleomorphic Sarcomas. In: Jo VY, Hornick JL, eds. Practical Soft Tissue Pathology: A Diagnostic Approach 2nd ed. Philadelpha: Elsevier, 2019;186-8.