

Thoracic Primitive Neuro-Ectodermal Tumor With Superior Vena Cava Syndrome: A Case Report

Dr. Sameer Peer¹, Dr. Iqbal Bashir², Dr. Junaid Kazimi³, Dr. Fahad Shafi⁴, Dr. Imran Nazir⁵, Dr. Sayar Ahmad⁶, Dr. Mir Wamiq Hussain⁷

(Department Of Radiodiagnosis & Imaging, Sher-I-Kashmir Institute Of Medical Sciences, Srinagar, Jammu & Kashmir, India)

Abstract : *Peripheral Primitive Neuro-ectodermal Tumors (pPNETs) are a group of rare but highly aggressive tumors which usually occur in children and adolescents with thoraco-pulmonary region being the most common site of occurrence. Patients usually present with cough, breathlessness, fever or hemoptysis. Superior Vena Cava Syndrome caused by a pPNET is a very rare clinical entity with literature limited to just a few case reports. We report a case of Thoracic PNET in a 15-year-old boy who presented with Superior Vena Cava syndrome. To the best of our knowledge, this is one of the few such cases reported in the literature.*

Keywords: *Askin tumor, Primitive neuro-ectodermal tumors, Superior vena cava syndrome*

I. Introduction

Primitive Neuroectodermal Tumors belong to the Ewing's sarcoma family of tumors which includes Ewing's Sarcoma (osseous origin), Extra-osseous Ewing's Sarcoma and Primitive Neuroectodermal Tumors (PNETs) [1]. These tumors have histopathological, immunohistochemical and cytogenetic similarities. PNETs occurring outside CNS are called peripheral PNETs (pPNETs) and are exceedingly rare [2]. They most commonly occur in thoraco-pulmonary region and are known as Askin Tumor [2,3]. They usually occur in children and adolescents [1,2,3]. Patients usually present with complaints of cough, breathlessness, fever, hemoptysis and chest pain [1]. Patients with paraspinal pPNETs can present with paraplegia or bladder/bowel dysfunction [4]. Peripheral PNET presenting with Superior Vena Cava Syndrome is a very rare occurrence and very few cases have been reported in the literature.

II. Case Report

A 15-year-old-boy presented to our hospital with complaints of progressively worsening breathlessness with orthopnea and cough for past 1 week. He also had intermittent fever for last 1 month which was documented as between 100°F to 102°F. There was also a history of anorexia and weight loss. No history of hemoptysis, night sweats or contact with tuberculosis patient was given

On Examination, the patient had tachycardia. His face was swollen with plethoric appearance. His neck veins were dilated. There were dilated veins all over the chest wall with flow directed downwards.

His laboratory investigations revealed haemoglobin of 7.2 g/dl, a Total Leucocyte count of 15,000 (N₅₇L₄₂M₁), ESR of 29 mm 1st hr. Kidney and Liver function tests were normal.

The patient was taken for an urgent Contrast Enhanced CT scan of the chest. CECT images revealed a large lobulated mildly enhancing mass involving the superior, anterior, middle and posterior mediastinum with obliteration of right main bronchus with extension of the mass to the right para-spinal area (figure 1). The mass was seen to infiltrate into and obliterate the Superior Vena Cava above the level of azygos vein (figure 2). Azygos vein was dilated and multiple dilated collateral veins were seen along the chest wall and multiple large soft tissue nodules were seen in both lobes of the lung which were suggestive of metastatic lesions (figure 3). CT guided biopsy was performed at a later date. Histopathological examination showed sheets of round cells with pseudorosette formation (figure 4). Immunohistochemistry revealed CD 99, vimentin, CD 56 and CD 117 positivity. Cytogenetic analysis was not available. Hence, a diagnosis of Thoracic PNET with Superior Vena Cava syndrome and lung metastasis was made and the patient was started on polychemotherapy and radiotherapy regimen. The patient was symptomatically better till the last follow-up.



Figure 1. CECT image showing a lobulated mass involving the mediastinum and right paraspinal region and causing obliteration of Superior vena cava and right main bronchus. Dilated Azygos vein and metastatic nodules in left lung can also be seen.



Figure 2: CECT image showing infiltration and complete obliteration of Superior Vena Cava by the mediastinal mass.

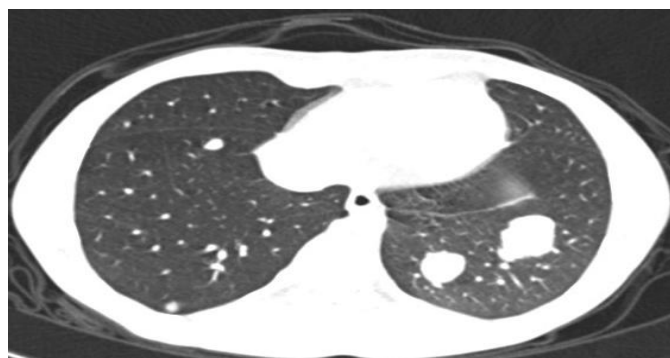


Figure 3. CT image in lung window setting showing multiple large soft tissue attenuation nodules in both lungs, suggestive of lung metastasis.

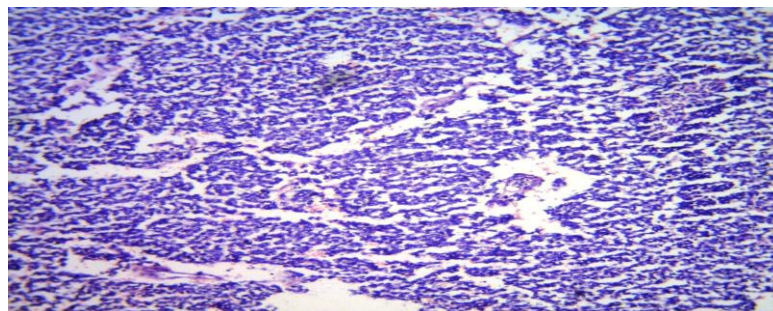


Figure 4. Histopathology examination of the mass (H&E stain) reveals sheets and cords of small round blue cells with scant cytoplasm with pseudorosette formations, features in keeping with Ewing's sarcoma family of tumors or PNET.

III. Discussion

PNET is a group of poorly differentiated small round blue cell tumors from neural crest origin [2-5]. PNET was first described by Hart and Earle in 1973 [2]. Batsakis et al divided PNET into three groups, (1) Central PNET (cPNET), (2) PNET arising from autonomic nervous system such as neuroblastoma; and (3) peripheral PNET (pPNET) [2]. Peripheral PNET was first introduced by Stout in 1918 [2,3].

Histopathologically, PNETs are characterized by sheets of small, round blue cells with scant eosinophilic cytoplasm and formation of Homer-wright pseudorosettes [1-5]. On immunohistochemical examination, these cells are found to express high levels of surface glycoprotein CD99 (or MIC2 or p30/32 MIC2), encoded by MIC2X gene. NKX2.2 protein encoded by NKX2-2 gene is a potential novel marker for PNETs and other Ewing sarcoma family of tumors [3]. On Cytogenetic analysis PNETs are characterised by t(11;22)(q24;q12) translocation [1-5] resulting in EWS gene mutation and EWS-FLI1 fusion protein formation [2].

pPNET represents approximately 1% of sarcomas. The most common site of occurrence is the thoraco-pulmonary region (Askin tumor) [2,3]. It is predominantly a tumor of childhood and adolescence with mean age of occurrence of Askin tumor being 14 years [3]. It is more common in Caucasian population and rarely seen in black population [3,4].

Superior Vena cava syndrome is a rare manifestation in peripheral PNETs. Gupta et al have reported lymphoma to be the most common cause for SVC syndrome [6]. Arya et al found T-cell acute lymphoblastic leukemia to be the most common cause of SVCS followed by lymphoma [7]. A review of literature shows only few case reports of PNETs presenting with SVCS [1,5,8,9]

pPNETs have an overall poor prognosis with disease free survival of <50% at 3yrs and 30-45% at 5 years. Local recurrence is common and 10% cases already have metastasis at the time of presentation [3]. Management is multidisciplinary with polychemotherapy and radiotherapy.

Our case was a 15-year-old boy who presented with typical clinical features of SVCS and was subsequently found to have a thoracic pPNET. Prompt diagnosis and treatment in this case led to symptomatic improvement.

IV. Conclusion

pPNET presenting as SVC syndrome is a rare clinical condition, however in a child or an adolescent presenting with clinical and/or radiological features of SVCS, pPNET should always be kept in differential diagnosis as prompt management with polychemotherapy and/or radiotherapy could be life saving for the patient.

References

- [1]. Nag VK, Nandan D, Chatterjee A, Bhardwaj M. Primitive neuroectodermal tumor presenting with superior vena cava syndrome. *J Assoc Chest Physicians* 2014;2:81-3.
- [2]. Kalantari M, Deyhimi P, Kalantari P. Peripheral Primitive Neuroectodermal Tumor (pPNET) of the Parotid: Report of a Rare Case. *Arch Iran Med.* 2015; 18(12): 858 – 860.
- [3]. Benbrahim Z, Arifi S, Daoudi K, Serraj M, Amara B, Benjelloun MC et al. Askin's tumor: a case report and literature review. *World J Surg Oncol* 2013, 11:10.
- [4]. Minne` C, Khan M. Paraspinal Primitive Neuroectodermal tumor (PNET) of the chest. *South African Journal Of Radiology* 2006; 10(1): 18.
- [5]. Nazari S, Malek F, Madani F, Hamoud M. Askin Tumor Presenting as Superior Vena Cava Syndrome. *IJBC* 2015; 7(3): 197-169
- [6]. Gupta V, Ambati SR, Pant P, Bhatia B. Superior vena cava syndrome in children. *Indian J Hematol Blood Transfus* 2008;24:28- 30.
- [7]. Arya LS, Narain S, Tomar S, Thavaraj V, Dawar R, Bhargawa M. Superior vena cava syndrome. *Indian J Pediatr* 2002;69:293- 7.
- [8]. Mehra S, Atwal SS, Garga UC. Primary Pulmonary Ewing's Sarcoma: Rare Cause of Superior Vena Cava Syndrome in Children. *J Clin Diagn Res* 2014;8(8): RD05-RD06.
- [9]. D Anirban, Roy PP, Sarkar A, Datta S, Dey S, Basu N. Mediastinal Primitive Neuroectodermal Tumour (PNET) – A Rare Case of Horner's Syndrome. *Journal of Medicine* 2012; 13 : 85-87