

**Discussion**

The diagnostic features of Freeman Sheldon Syndrome include cranial, carpal and tarsal anomalies. The patient reported here had classical whistling face and hand abnormalities. However, several other malformations such as convergent strabismus, kyphoscoliosis(2), blepharophimosis, anti-mongoloid eye slant(3) limited mobility at shoulders and elbows, decreased pronation, supination(4), colobomata of nostril(5) and renal abnormality(6) were absent in this patient.

The condition is genetically heterogeneous. In most cases the mode of inheritance is autosomal dominant. However, an autosomal recessive mode of inheritance has also been suggested(4). In this case the father had restriction opening of mouth although no other stigmata were present. If it is taken as *formae frustee*, the possibility of dominant mode of inheritance in this case can be considered.

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**Askin Rosai Tumor**

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Askin Rosai tumor is one of the rarest of the rare group of chest wall tumors in the pediatric age group. It is a small round cell malignancy with a differential diagnosis of neuroblastoma, rhabdomyosarcoma, Ewing's sarcoma and malignant lymphoma. However, it can be differentiated from these by absence of certain light microscopic findings and by doing other special tests. Two cases of Askin Rosai tumor diagnosed by wedge biopsy are reported here. Both these children presented with a firm to hard mass on the posterior thoraco-abdominal wall.

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## Case Reports

**Case 1:** A 4-year-old girl was brought to us with a swelling in the paraspinal region below the scapula with a history of mild fever and anorexia for a period of 2 months. On examination, she was a well child, mildly pale and had a swelling in the region of right seventh rib, below the right scapular angle, measuring 5 cm × 3 cm, irregular, firm to hard, tender, nonpulsatile and was free from the skin but seemed to be attached to the rib beneath. There was no evidence of dissemination to other bones, joints and spine being normal. The examination of the respiratory system revealed diminished air entry in the right lower zone corresponding to the swelling.

Investigations revealed a hemoglobin of 8 g/dl with atypical lymphocytes (reactive) seen on the peripheral smear. Roentgenogram of chest showed 7th rib spreading on the right side (Fig. 1). Serum LDH and alkaline phosphatase were elevated. Urinary VMA was normal.

A fine needle aspiration done from the swelling on the chest showed a small round cell tumor. On wedge biopsy, it confirmed a small round cell tumor with pseudorosette formation, the cells had a faint blue cytoplasm with a nucleus with coarse chromatin. No cytoplasmic cross striations were seen. Mitotic figures were moderately present.

The diagnosis of Askin Rosai tumor was established by absence of investigative findings suggestive of other round cell tumors, i.e., Ewing's Sarcoma (PAS for glycogen-positive), rhabdomyosarcoma (acidophilic cytoplasm-spindle cells, cross striations), neuroblastoma (elevated urinary VMA levels) and lymphoma (other clinical features like lymphadenopathy, splenomegaly).

**Case 2:** A 6-year-old boy presented with a swelling on right lower chest wall and upper abdominal wall posteriorly for a

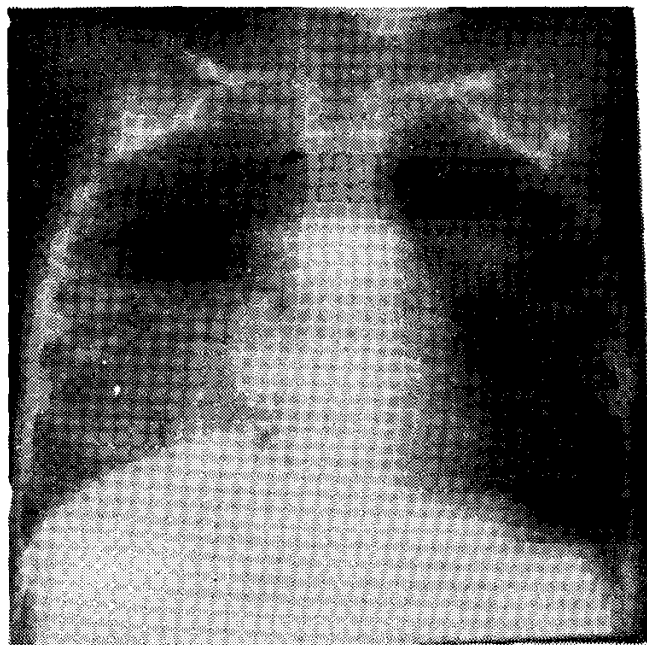


Fig. 1. The Seventh rib spreading on right side in the first patient.

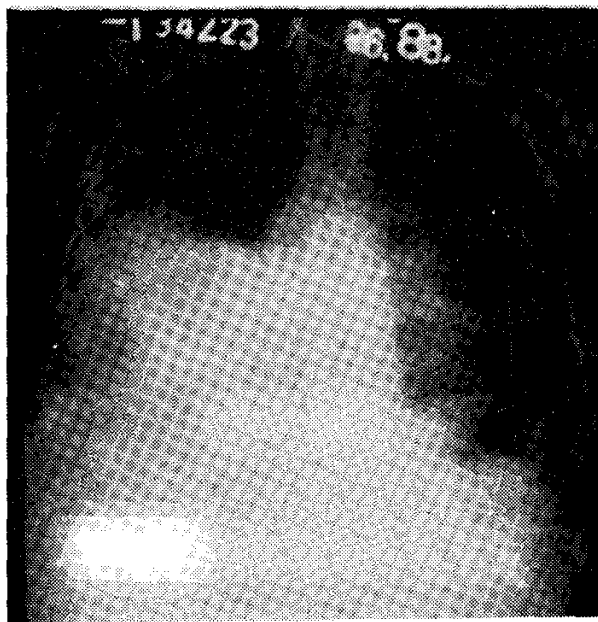


Fig. 2. Right sided pleural effusion in the second patient.

period of 15 days. On examination this child was also a well child, mildly pale and

had a swelling in the region of right 12th rib, on the posterior thoraco abdominal wall measuring 5 cm × 7 cm, irregular, firm to hard and non tender. On per abdominal examination the liver was 8 cm palpable, soft to firm, non tender, non pulsatile with a smooth surface and rounded edge. Rest of the systemic examination was normal.

On investigation, he had a hemoglobin of 9 g/dl with a typical lymphocytes on peripheral smear. His X-ray chest showed a right pleural effusion (*Fig. 2*). X-ray lumbo sacral spine showed a scoliosis and a soft tissue mass in the right hypochondrium with destruction of the 12th rib. Urinary VMA was normal. Ultrasonography of the abdomen showed a solid tumor arising from the posterior thoraco-abdominal wall with intra-abdominal extension causing displacement of upper abdominal organs. A diagnosis of Askin Rosai tumor was established as in *Case 1*, after FNAC and wedge biopsy; all other round cell tumors were similarly excluded.

Both these children were given weekly injections of a combination of vincristine and cyclophosphamide in the dose of 1.4 mg/m<sup>2</sup> and 600 mg/m<sup>2</sup>, respectively for seven doses followed by local radio-therapy up to a total of 4000 rads. The local swelling regressed with the first course of chemotherapy in both our patients but in the first child a maxillary metastasis was seen 8 weeks after diagnosis. She developed multiple metastasis within six months after diagnosis involving the right orbital region, vault of the skull, lumbo sacral spine and head of the femur and she expired within eight months of diagnosis. The second child was lost to follow up 6 months after diagnosis.

### Discussion

Primary malignant tumors of the chest

wall and/or lung excluding the mediastinum are infrequent in childhood and adolescence as is evident in various large series in literature. Askin Rosai tumor is even rarer. It was first described by Askin and Rosai in 1979, as a distinct clinicopathological entity of uncertain histogenesis. So far there have been only 3 major series reported in literature, first in 1979, by the original authors Askin and Rosai wherein they discussed 20 cases(1). All these consisted of a common histologic appearance of malignant round cells which had occurred at a particular anatomical site. The second series consisting of 15 cases was reported by Linnoila *et al.* in 1986(2). Recently in 1988, Parikh *et al.* from India have published their data on therapy in 15 cases(3). Askin and Rosai found a female preponderance with median age at presentation being 14.5 years and 25% of their patients were below the age of 6 years. Parikh *et al.* have described a male dominance, median age of presentation being 17.9 years. Our patients were 6 years and 4 years old respectively.

A thoracoabdominal mass with or without pain was the most consistent clinical presentation. The duration of symptoms prior to diagnosis ranged from a few days to six months. Certain atypical presentations described in literature are: (i) mass filling pericardial cavity, (ii) an abdominal mass which was found to be an extrapulmonary intrathoracic mass depressing the diaphragm and involving the chest wall. The latter presentation is similar to that found in the second case of ours.

The radiological findings, according to literature, could be variable and include pleural disease manifested by fluid and/or tumor, pulmonary parenchymal disease, rib destruction, rib spreading and diffuse osteoblastic metastasis. In the first of our

patients, the 7th rib was spreading (*Fig. 1*) and in the second patient, 12th rib destruction was seen along with right sided pleural effusion (*Fig. 2*).

**Pathological Findings:** The tumor may be round, ovoid, multinodular or lobulated measuring between 2 and 4 cm in the greatest dimensions. A greyish white appearance with a granular to glistening texture characterized the cut surface. Small, nonconfluent areas of hemorrhage and necrosis may be seen. Most of them are circumscribed but encapsulation is not a feature. Histopathologically, compact sheets of cells, a nesting arrangement of cells with an intervening fibrovascular stroma and serpiginous bands of cells with necrosis are the three basic microscopic pictures described. The individual cells measure 10-14 in diameter. The nucleus has a coarse but evenly dispersed chromatin.

The diagnosis of Askin Rosai tumor is made by light microscopic exclusion of the following: Ewing's sarcoma (light and dark cell pattern, glycogen on PAS staining), rhabdomyosarcoma (acidophilic cytoplasm; cross striations and spindle cells) and malignant lymphoma (other features of lymphoma). Neuroblastoma is excluded by urinary VMA estimation. This tumor does show neurosecretory like granules and rosette formation but without neurofibrillary material. Hence, it is thought to be neuroectodermal in origin, although this needs to be further proven. Neuron-specific enolase is the most diagnostic for this tumor(4). Our patients were, however, not subjected to this test.

Experience on treatment of this tumor is limited. Surgery, chemotherapy and radiotherapy were used in various combinations by Askin *et al.* However, the recent study by Parikh *et al.* has emphasized the need for aggressive chemotherapy and the

authors recommend complete excision of tumor whenever possible, followed by aggressive combination chemotherapy containing ifosfamide, etoposide and cisplatin with local radiotherapy. A higher rate of complete remission was experienced by them with this modality although long term survival, irrespective of initial treatment, was not encouraging as found by various authors. Thus, it does not differ from other round cell tumor in its management and prognosis, outcome being favourable if it is totally resectable (Advani SH, personal communication).

Death is usually due to local recurrence or complications consequent to contiguous spread rather than due to distant metastasis, as seen in other malignancies. More research needs to be done to improve the overall survival rate in patients with this tumor.

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