Malignant Peripheral Nerve Sheath Tumour In Von Recklinghausens disease a rare case report.
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Abstract : Malignant peripheral nerve sheath tumors (MPNSTs) are rare soft tissue tumors that arise from a peripheral nerve or exhibit nerve sheath differentiation. Most of these tumors arise from trunk, extremities, or head and neck regions. They are very rarely located in the abdominal cavity. We report a rare case of MPNST which presented as mass abdomen.
Keyword : Malignant peripheral nerve sheath tumor, soft tissue tumor, peripheral nerve.

INTRODUCTION:
Neurofibromatosis (NF-1) is an autosomal dominant disorder characterized by developmental changes in the nervous system, bones, and skin. An abnormality in human chromosome 17 leads to the development of Von Recklinghausen’s disease. Gastrointestinal (GI) involvement of neurofibromatosis (NF-1) occurs in 10-25% of cases (1); they usually originate from the intramuscular plexus of Auerbach (4) and are generally submucosal but may extend to the serosa (1). Neurofibromas can occur as sporadic lesions in the stomach and jejunum, and rarely involves the mesentery (3). The abdominal tumors in NF-1 can be divided into five basic categories: neurogenic, neuroendocrine, embryonal, non neurogenic GI mesenchymal and miscellaneous (2). Common clinical manifestations of GI involvement include abdominal pain, constipation, anemia, melena and a palpable abdominal lump.

CASE SUMMARY:
A 29 year old male presented to our hospital with mass abdomen for 4 months duration associated with abdominal pain, progressive dyspepsia and weight loss. On examination patient had multiple small swellings and hypopigmentations (café au lait spots) all over the body. (Picture 1&2)
General examination were within normal limits. Per abdomen a firm mobile mass of 10*8cm was palpable occupying the umbilical and hypogastric region. The mass was dull on percussion. Patient was investigated- routine blood parameters were normal .Patient was further investigated. Computed tomography (CT) of abdomen revealed a well circumscribed solid heterogeneously enhancing soft tissue density noted in left mesenteric bed in lumbar region. The lesion was measuring about 9.0 x 8.2 x 8.1cm . Non enhancing, hypodense areas were found within the lesion - suggestive of necrosis. Lesion showed persistent enhancement in delayed phases without any calcification. Lesion appears to have lamellar /whorled appearance. Mesenteric vessels noted supplying the mass lesion. The mass was suspicious of neurogenic in origin. (Picture 3)
Imaging techniques used for the evaluation of patients with MPNSTs have been reported in the literature. To date, very few cases of MPNSTs arising from the mesentry have been reported.

On histopathological examination the mesentery showed an infiltrating neoplasm composed of spindle cells arranged in fascicles. Intermixed with the cellular areas, are seen hypocellular, inconspicuous nuclei, and moderate amount of cytoplasm with indistinct cytoplasmic borders. Bundles of spindle cells exhibiting neural differentiation - elongated wavy nuclei were observed. Areas of necrosis and mitotic figures (10 Mitotic Figure/10 High Power Field) are noted. Focal collections of cells with abundant dense eosinophilic cytoplasm and pleomorphic nuclei resembling rhabdomyoblasts were also seen. A few entrapped mature adipocytes are seen amidst the neoplastic spindle cells. Bowel wall is not infiltrated by tumor. On Immunohistochemical marker study, some of the tumor cells are Glial fibrillary acidic protein(GFAP) positive and occasional cells are S100 positive. The neoplastic cells are negative for muscle markers (Smooth Muscle Actin, Desmin, and Myogenin). Immunohistochemical features are consistent with MPNST. Post operative period was uneventful. Patient underwent adjuvant chemotherapy and he is under regular follow up.

**DISCUSSION:**

MPNST with Von Recklinghausen's disease is considered the result of the malignant transformation of a benign neurogenic mass such as neurofibroma or schwannoma (5). Growth tendency, subjective symptoms of pain or numbness, and local fever have been reported as good indicators of malignant transformation (5). These tumors are associated with lower 5-year survival rates, higher recurrence rates and higher frequency of metastasis compared with neoplasms at other body sites (14). The most effective treatment appears to be early diagnosis and wide surgical excision. Complete tumor removal is the mainstay of treatment and the most significant prognostic factor of MPNST. It remains uncertain whether chemotherapy and radiotherapy have a positive impact on the survival of patients with MPNSTs. The results of most case series indicate limited benefits and high morbidity on using adjuvant radiotherapy or chemotherapy. Despite aggressive combined radiation and systemic chemotherapy, the 5-year survival rates for MPNSTs range from 35% to 50% (24,25). The current recommendation is that this therapy be reserved for recurrent tumors, suspected residual microscopic disease, and high-grade tumors. We recommend wide excision of MPNSTs with postoperative follow-up.

**CONCLUSION**

MPNST associated with NF1 will have an aggressive behavior and poor prognosis. It is recommended that a MPNST associated with NF1 be treated aggressively with multi-modalities such as a radical tumor excision with a wide margin of normal tissue followed by adjuvant chemoradiotherapy. In addition, the patients should be closely monitored for the early detection of a recurrence.

**References**


They are useful in early detection of malignant changes in plexiform neurofibromas (7) contributing to staging, therapy planning and subsequent follow-up (8). Importantly, MPNST tends to be heterogeneous in CT attenuation and MRI signal intensity because necrosis is frequently present within the mass (9). Although asymmetry in size or in CT attenuation suggests malignant degeneration of the larger mass, it has been shown that these criteria are not reliable indicators of malignancy (10, 11). The pathologic diagnosis of MPNST is facilitated by features such as palisading arrangement, nuclear atypia, bizarre giant cells, mitotic figures, and necrosis. These tumors have morphological heterogeneity, and staining analysis of such tumors reveals spindle cells with a fascicular pattern (12). Histological and immunohistochemical markers specific for MPNSTs are not available. The S100 protein is the antigen most commonly used to identify nerve sheath tumors of various types. However, S100 protein immunoreactivity is detected in only 50-60% of MPNSTs, and this protein is also expressed in a range of other tissues and tumor types (13). MPNSTs in patients with NF-1 are diagnosed in younger age group and has poorer prognosis than sporadic MPNSTs.