Case Report

Embryonal Rhabdomyosarcoma: A Case Report

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Abstract

Rhabdomyosarcoma is the most common childhood malignancy and can be occasionally misdiagnosed due to vague signs and symptoms. Current report of 11 year old girl stresses on early diagnosis and multi-disciplinary approach towards management.

Keywords:
Rhabdomyosarcoma, head and neck, maxillary antrum & embryonal variant

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**Introduction**

Rhabdomyosarcoma (RMS) is a malignant soft tissue neoplasm of skeletal muscle origin and accounts for around 6% of all childhood malignancies. It was first described by Weber in 1854\(^1\). Most common site of involvement is head and neck, genitourinary tract and retroperitoneum, while extremities are involved less frequently. The clinical features are variable, ranging from a small nodule to a fast growing swelling which may or may not be associated with pain, paresthesia, trismus or nasal discharge\(^1,2\).

Horn and Enterline in 1958 described four histopathological variants of RMS: embryonal, botryoid, alveolar and pleomorphic. Among them, embryonal variant being most common in children (<8 years of age) and affecting the head and neck region mainly, while the alveolar variant is commonly seen in adolescents involving the extremities\(^2,3,4\).

The present report is of an 11 year old girl who was diagnosed with embryonal RMS after histopathological examination and immunohistochemical confirmation.

**Case Report**

An 11 year old girl was referred to our institution with a painless, rapidly growing swelling in the middle third of the face which was noticed 5 months back. Previously she was administered parental antibiotics, but it was ineffective. Gross facial asymmetry was noticed with swelling in the right mid-face region and a yellowish nasal discharge (figure 1). The overlying skin was stretched and erythematous in appearance. On intra-oral examination, an ulcerated, mucosal coloured swelling in the right buccal vestibule was extending from canine to molar region and surface necrosis was observed (figure 2). On palpation the swelling was non tender, non fluctuant and firm in consistency. CT scan revealed an infiltrative, large space occupying lesion in the right maxillary antrum extending into the right nasal cavity and destroying the medial wall of nasal cavity as well (Figure 3).

After routine hemogram and patient consent, incisional biopsy was performed under local anaesthesia. Histopathological sections under scanner view revealed the presence of hypocellular, loose connective tissue with areas of necrosis and few blood vessels with surrounding inflammatory cells (Figure 4). Under higher magnification, tumor cells exhibited loss of cohesion, nuclear hyperchromatism, cellular pleomorphism, prominent nucleoli, abundant atypical mitotic figures and occasional giant cells. Occasional rhabdomyoblasts and strap cells were also observed in the connective stroma (Figure 5). On immunohistochemistry, tumors cells were immunoreactive for MyoD1 and...
desmin. Finally a diagnosis of embryonal rhabdomyosarcoma was established.

Patient was then referred to Oncology department, where a combination of chemotherapy (Vincristin, Actinomycin D & Cyclophosphamide) and radiation therapy was proposed. Tumor size regression was noticed after 10 weeks and the patient was regularly followed.

Figure 1: Extraoral examination showing midfacial swelling

Figure 2: Intraoral examination showing extent of the lesion

Figure 3: CT scan shows space occupying lesion in maxillary antrum and destruction of medial and lateral nasal wall

Figure 4: H&E stained section showing loose hypocellular connective tissue with blood vessels and inflammatory cells surrounding them (10X)

Figure 5: Connective tissue stroma showing strap or tadpole cells and tumor giant cells (inset) under 40X view
Discussion

RMS is the third most common neoplasm of childhood after neuroblastoma and Wilm’s tumor, comprising about 15% of all the solid tumors\(^1\). It is thought to arise from immature mesenchymal cells that are committed to skeletal muscle lineage, but it has been found in sites devoid of skeletal muscles e.g. urinary bladder\(^1, 2\). Its incidence is highest in children among the age group of 1-4 years, falling to a lower rate in 10-14 years age group and remaining steady between 15-19 years\(^3\). So, the present case of 11 year old girl does not fall in the category of high risk group. As per the pervious data, male predominance (1.3:1) has been observed, especially in the first decade of life\(^3\). However the present case has been reported in an 11 year old girl. Head and neck is the principal location for RMS (44% of the cases), but sinus is involved in only 8.4% of cases\(^3\). Maxillary sinus was involved in the present case with destruction of both medial and lateral nasal wall. The most common intra-oral site of involvement is tongue followed by soft palate, hard palate, and buccal mucosa\(^3, 4\).

The initial classification by Horn & Enterline has been modified by various researchers and societies. In 1992, collaborators of Pediatric branch at National Cancer Institute (NCI) modified the conventional classification based on review of 159 cases. They categorised RMS into favourable and unfavourable prognosis categories. Tumors with favourable prognosis involved embryonal rhabdomyosarcoma (conventional, pleomorphic, leiomyomatous & aggressive histologic features), while those with unfavourable prognosis include alveolar RMS (conventional & solid), pleomorphic and others\(^3\). More recently, international classification of rhabdomyosarcoma includes: superior prognosis (botryoid & spindle cell RMS), intermediate prognosis (embryonal RMS), poor prognosis (alveolar and undifferentiated RMS) and unknown prognosis (RMS with rhabdoid features). A new sclerosing variant of RMS has also been reported by some pathologists which demands more elaborate workup\(^3, 4\).

Histologic diagnosis of RMS is based on the identification of characteristic of skeletal muscle (cross striations), or rhabdomyoblasts. Cross-striations are seen in 50-60% of the cases. Histologically embryonal RMS is composed of rhabdomyoblasts and small round cells. Rhabdomyoblast, is characterized by bright eosinophilic cytoplasm. Sarcoma botryoides and spindle cell variant are two subtypes of embryonal RMS. Alveolar RMS consists of large round cells with prominent eosinophilic cytoplasm and rhabdomyoblasts arranged in cleft like...
spaces known as alveoli. The pleomorphic RMS is extremely rare in children and is characterised by anaplastic cells present in large aggregates or diffuse sheets. It commonly affects extremities and the trunk \(6, 7, 8\). Whenever, light microscopic diagnosis is inconclusive, electron microscopy and immunohistochemical analysis plays important role. On electronmicroscopy, visible z-bands are the diagnostic feature \(6, 9\). Further, skeletal muscle differentiation can be identified by immunohistochemical staining namely, antidesmin, muscle-specific actin and Myo D. Monoclonal antibodies, like those to desmin, muscle-specific actin, sarcomeric actin and myoglobin have also been used to confirm the myogenic lineage with very good specificity and sensitivity \(6, 10, 11, 12\).

The embroyonal RMS and alveolar RMS have been associated with distinct clinical characteristics and genetic alterations. Alveolar RMS is associated with 2; 13 or 1; 13 chromosomal translocations, resulting in formation of PAX3-FKHR and PAX7-FKHR fusion proteins respectively. On the other hand, embryonal RMS has been reported with 11p15.5 chromosomal deletion \(6, 7, 13, 14, 15\). Histologically, distinction of particular subtype is of prognostic significance. Newton et al had reported that spindle and botryoid variants of embryonal RMS have a better prognosis (95% & 88% respectively 5 year survival); followed by classic embryonal variant (66%); and alveolar variant (54%) \(^3\). Prognosis is not solely dependent on histologic variant, but on certain other factors also. Infants and young children have a reportedly better prognosis. Orbital and genitourinary location with < 5cm size of the lesion also has a favourable influence on the prognosis. Degree of cellular differentiation has also been found to be of major prognostic importance. Wijnaendts et al. had reported that a greater degree of cellular differentiation is associated with better prognosis, independent of the histologic variant\(^1\). Moreover, localized tumors without regional lymph node or distant metastasis which can be completely resected on initial surgery are also associated with better prognosis\(^3, 4, 5\).

Though, prognosis of RMS has drastically improved in the past decades due to advent of chemotherapy and radiotherapy along with the surgery, still the overall 5 year survival is around 70\%\(^2\).

Finally, whenever a non-resolving swelling of head and neck region is observed in young children, it should be critically evaluated to rule out RMS. Antibiotic therapy should be started only when an underlying cause can be visualised. In doubtful cases, biopsy is mandatory without delay as misdiagnosis and delayed diagnosis can adversely affect the prognosis.
References
