EMBRYONAL Rhabdomyosarcoma Presenting in a 1 Year Old Boy As a Diagnostic Dilemma and the Role of IHC-A Case Report

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Introduction

Rhabdomyosarcoma is the most common soft tissue sarcoma of childhood, representing 5% of all childhood cancers.

It is thought to arise from primitive mesenchymal cells committed to skeletal muscle differentiation and can occur in a variety of organs and tissues, including those that lack striated muscle.

The main histological subtypes of rhabdomyosarcoma are embryonal (with botryoid and spindle cell variants) alveolar and pleomorphic.

Approximately 60% of all newly diagnosed rhabdomyosarcomas are of embryonal histology.

Etiology:

ERMS occurs with many syndromes caused by germline mutations in RAS signalling pathway. E.g: Costello synd, Neurofibromatosis, Noonan synd, beckwith –weidmann syn etc.

The most common sites include head and neck and genitourinary regions.

At times, the myoid stroma and polypoid configuration of some RMS might be mistaken for nasal polyps or myxoma.

The diagnosis is most rapidly and reliably confirmed by immunohistochemical(IHC) by demonstration of one or more muscle antigens.

Materials and methods

- Small bits of tissue was sent for histopathological examination
- The paraffin blocks were used for IHC testing.

Case history

In this study, a 1 year old male patient presented to a local ophthalmologist with complain of painless swelling of left orbit (fig 1).

CT scan revealed picture suggestive of venolymphatic malformation (March 17,2016). Thereby, he was referred to a well known health care centre where MRI was advised which revealed a picture suggestive of rhabdomyosarcoma with differential diagnosis of pleiform neurofibroma.

Biopsy was done at the same centre and a diagnosis of orbital myxoma/angiomyxoma was given (April 29, 2016)

After about 2 months following the biopsy, patient’s parents noticed similar swelling of the same orbit. He was brought to the pediatric surgery department of AMCH and re-biopsied.

One specimen was sent to our department and the other to a private centre. The report from the private laboratory suggested myxoma. The report from our college was given as Embryonal rhabdomyosarcoma and further subjected to immunohistochemistry(IHC).

IHC was positive for desmin, myogenin and vimentin. Complete hemogram, ESR were within normal limits.

Results

Histopathological examination

- The HPE examination of the small bits of tissue sent for examination revealed tumor cells arranged in trabeculae and sheets. Individual cells were stellate shaped with sparse, amphophilic cytoplasm and central nuclei – known as Rhabdoblust. Rhabdomyoblasts progressively acquires more cytoplasmic eosinophilia and elongation, manifested by ” tadpole”, ”strap” and ”spider” cells. Some contain abundant ,myoid stroma resembling myxomas (fig 2,3).

IHC findings:

- In our case IHC came as a rescue measure to both patient and us as it confirmed the embryonal RMS diagnosis by depicting the muscle antigens- myogenin, desmin and vimentin positivity in the slides; thereby confirming the muscle tissue origin of the tumor(fig 4,5,6).

Discussion

Most RMS tumors are classified as embryonal; these tumors consist of sheets of poorly to moderately differentiated cells.

The most common sites of metastatic involvement are the soft tissues, serosal surfaces, lung, bone marrow, and lymph nodes. Cases associated with diffuse bone marrow involvement may simulate acute leukemia.

RMS arising from genitourinary sites or extremities are particularly prone to metastasize to lymph nodes, whereas tumors originating in head and neck structures adjacent to meningeal surfaces have a high incidence of direct meningeal extension.

Prognosis:

The prognosis of embryonal RMS has markedly improved following multimodality treatment with excision, radiation therapy, and multidrug chemotherapy. Over 80% of children now survive when the disease is localized to the region of origin.

Age is an independent risk factor with patients aged 1-9 years having better outcomes than infants and adolescents.

1. Good prognosis: spindle cell; botryoid variant.
2. Intermediate prognosis: ERMS.
3. Poor prognosis: Alveolar and pleomorphic variants.

Management:

Surgical excision followed by chemotherapy.

Conclusion

Orbital RMS is one of the few life-threatening diseases that presents first to the ophthalmologist; therefore prompt diagnosis and treatment is a life-saving issue.

In our case, the patient deteriorated within a very short span of 8-9 months due to mis-diagnosis owing to its similarity to myxoma picture histologically. The facility of IHC was not utilized resulting in huge tumor load of the patient. Thus, IHC as the confirming test in confusing cases as ours, should be made readily available to patients, thereby, saving patients valuable time.

References

- ERMS- Embryonal Rhabdomyosarcoma
- IHC-immunohistochemistry