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Case Report

Rhabdomyosarcoma Mimicking Olfactory Neuroblastoma with Aberrant Expression of Immunohistochemical Markers -: A Diagnostic Pitfall

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Abstract:

Primitive round cell neoplasms are difficult to distinguish from one another without ancillary techniques. Many tumors such as lymphoma, melanoma, sinonasal undifferentiated carcinoma, neuroendocrine carcinoma mimic olfactory neuroblastoma leading to misdiagnosis [1]. This causes wrong treatment regimen and poor prognosis for certain treatable tumors.

We discuss in this report of case of orbit/ paranasal sinus tumor diagnosed as olfactory neuroblastoma, turned out to be Rhabdomyosarcoma with neuroendocrine differentiation. And also discuss the importance of immunohistochemistry in diagnosing such cases.

Keywords: Rhabdomyosarcoma, Immunohistochemistry, Neuroendocrine

Case Report:

17 year old boy presented to a tertiary center with complaints of diminution of vision since 6 months which progressively increased. On examination found to have proptosis due to frontoethmoid sinus mass .CT showed an hyperdense soft tissue mass in right orbit causing expansion of right ethmoidal sinus and extension into frontal sinus, sphenoidal sinus and nasal cavity. Patient underwent FESS assisted biopsy. Frozen and H and E Sections were reported as malignant neoplasm arranged in lobules with round to oval cells , scanty cytoplasm, nuclear pleomorphism and surrounding neurofibrillary matrix and was given possible diagnosis as Olfactory neuroblastoma advised IHC for confirmation .

We received block and slide of the same for immunohistochemistry which was carried out in 3 micron thick sections by Horse radish peroxidase technique. A panel of markers were employed which includes Myogenin, chromogranin, synaptophysin , CK 7,CD 99,EMA,S100 and ki67. Of which myogenin and chromogranin A showed bright positivity with ki67 -30%.

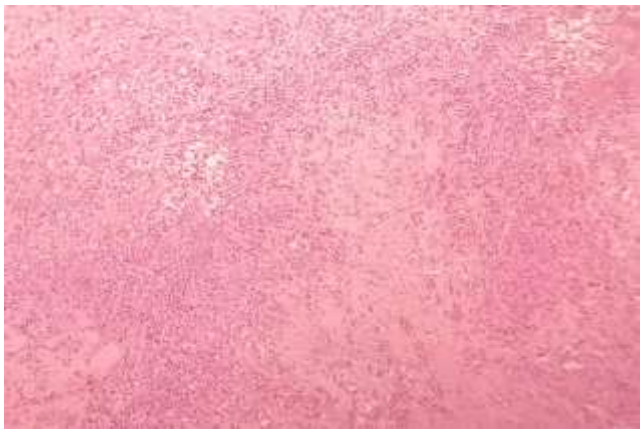

FIG-1 : H&E (LP) Sections show round to oval cells arranged in sheets and foci of necrosis noted

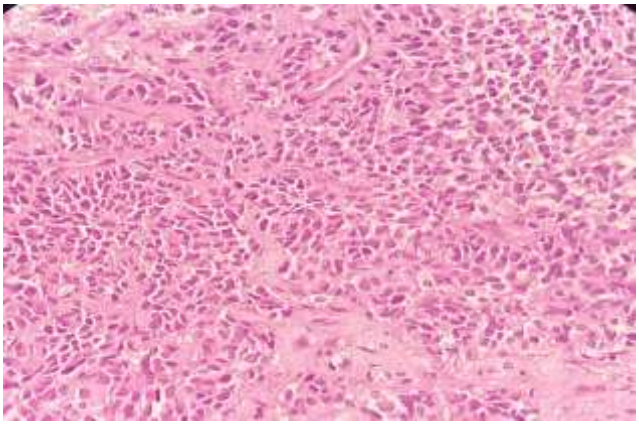

FIG 2: H&E (HP) Section shows round to oval cell with scanty cytoplasm and pleomorphic and hyperchromatic nuclei

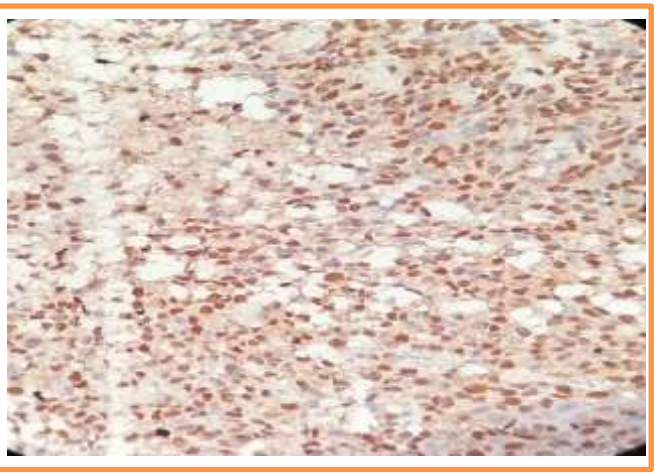

FIG-3: Section shows strong positivity for Myogenin

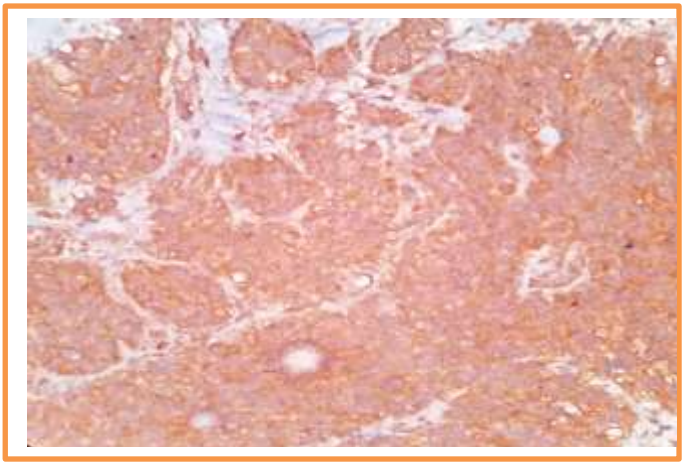

FIG -4: Section shows strong positivity for Chromogranin A

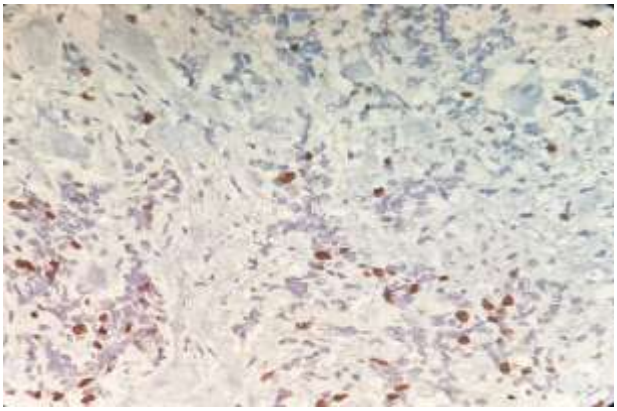

FIG 5- Section shows KI67 index of 30%

Discussion:

Rhabdomyosarcoma is a relatively uncommon aggressive tumor of sinonasal tract. Rhabdomyosarcoma has many variants of which embryonal RMS and solid type of alveolar variant mimic small round blue cell tumor. [2,3]

Alveolar RMS may involve the head and neck region (40%), the genitourinary system (20%), the extremities (20%), the trunk (10%) and other sites (10%) [4, 5]

Alveolar RMS histopathology shows distinctive nests of primitive-appearing round cells, which grows in a discohesive fashion, surrounded by hyalinized and highly vascular fibrous septa. Obvious rhabdomyoblastic differentiation, in the form of strap cells and cytoplasmic cross striations, is rarely identified.

Solid forms of alveolar RMS lack the prominent nested pattern and cellular discohesion seen in the classical type and may closely mimic a variety of other small round cell tumors, such as small cell carcinoma, lymphoma, neuroblastoma and malignant melanoma [6]

Diagnosis of small round cell tumor is very much challenging without immunohistochemistry particularly in head and neck region where close common differentials include small cell carcinoma and olfactory neuroblastoma. It usually present as midline mass of short term duration.

Certain tumors show aberrant immunoreactivity which pose further challenge for diagnosis. And it need to be differentiated from other tumors in head and neck that shows rhabdomyoblastic differntiation like undifferentiated carcinoma of thyroid , malignant triton tumor etc by judicious selection of markers .[7]

In a study by Armita bahrami et al showed of the 44 alveolar rhabdomyosarcoma which occurred with mean age of 18, of which 50% showed positivity for epithelial markers .32%expressed synaptophysin and 22% expressed chromogranin. 11%expressed both. [1]

In a study by Houreih MA et al presented 3 cases with alveolar rhabdomyosarcoma of which one involves orbit of 20 year old man of which all 3 showed desmin , myogenin and synaptophysin positive and 2/3 showed chromogranin positive .[8]

In a study by lester D.R Thompson showed sino nasal alveolar rhabdomyosarcoma of 52 cases of which myogenin, myoD1 was positive in all cases .MSA (96%) SMA (52%),CAM5.2 (50%) Synaptophysin 35% , Chromogranin (13%). Also said 10 of 48 cases (21%) tested for epithelial and neuroendocrine markers true neuroepithelial differentiation in a significant subset of sinonasal ARMS.[9]

In our case we found positivity for myogenin which points towards muscle lineage and chromogranin positivity points towards neuroendocrine differentiation. Above studies and our study states that rhabdomyosarcoma with neuroendocrine differentiation in more common in sinonasal tract, commonly occurs in adolescent and have aberrant expression of markers.

Conclusion:

In cases with primitive round cell neoplasm morphological evaluation alone is not sufficient. Immunohistochemical study

is mandatory and evaluation of right panel of markers are important for prognostic and therapeutic implications .In doubtful cases cytogenetic and molecular testing can be useful in establishing the correct diagnosis which plays a major part in treatment and prognosis.

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