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SINE QUA NON RADIOLOGY-PATHOLOGY

Disseminated Malignant Rhabdoid Tumor of the Head and Neck

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Abstract Disseminated extrarenal malignant rhabdoid tumors of the head and neck are very rare, but aggressive tumors. Although the features on radiological imaging may be nonspecific, the imaging is useful for assessing the extent of tumor involvement. Key pathologic features are those of a cellular "blue cell tumor" with variable rhabdoid appearance. These cells express a combination of markers usually viewed as characteristic of diverse lines of differentiation, including EMA, cytokeratins, smooth muscle markers, and GFAP, and occasionally synaptophysin. At a molecular level, the entity is defined by mutations or alterations in the SMARB1/INI1 gene resulting in loss of INI1 expression. Diagnostic features include rhabdoid cells, expression of keratin with absence of desmin, S100 protein and CD34, and loss of INI1 expression. These features are exemplified in this sine qua non radiology-pathology correlation article.

Keywords Malignant rhabdoid tumor · Head · Neck · CT · Pathology

Case Presentation

An infant born at 36-1/7 weeks to a 24-year-old G5P4 mother via vaginal delivery following decelerations at another institution. Apgar scores were 4, 5, and 7. At delivery, the amniotic fluid was found to be bloody with clots. Furthermore, the infant had a bulging parietal-occipital mass and was transferred to our institution with suspicion of an encephalocele. Upon transfer to our institution, physical exam showed a bleeding mass in the right scalp, swelling in the right parotid, and dysmorphic facies and a hemoglobin level of 1.5 g/dl. Due to profuse bleeding, decision for emergent resection of the scalp mass was made. Nevertheless, the infant developed multiorgan failure and disseminated intravascular coagulopathy and passed away. A complete autopsy was performed.

Radiographic Features

A non-contrast CT was performed (Fig. 1), which demonstrated a bulky heterogeneous mass within the right scalp soft tissues, measuring up to 6 cm, with erosions of the underlying calvarium, but no gross intracranial extension, and an additional mass within the right preauricular subcutaneous tissues, measuring up to 4 cm. The scalp mass also contained small areas of hyperattenuation, suggestive of hemorrhage. The imaging differential diagnosis included teratoma, infantile fibrosarcoma, and other neoplasms, perhaps with associated metastases.

Diagnosis

Pathologic examination of the surgical specimen obtained from the scalp lesion and the autopsy confirmed that the neck mass and the scalp tumor both exhibited identical histomorphologic features. Gross examination revealed skin sloughing overlying the tumors, which were comprised of poorly-circumscribed polypoid tan tissue, with

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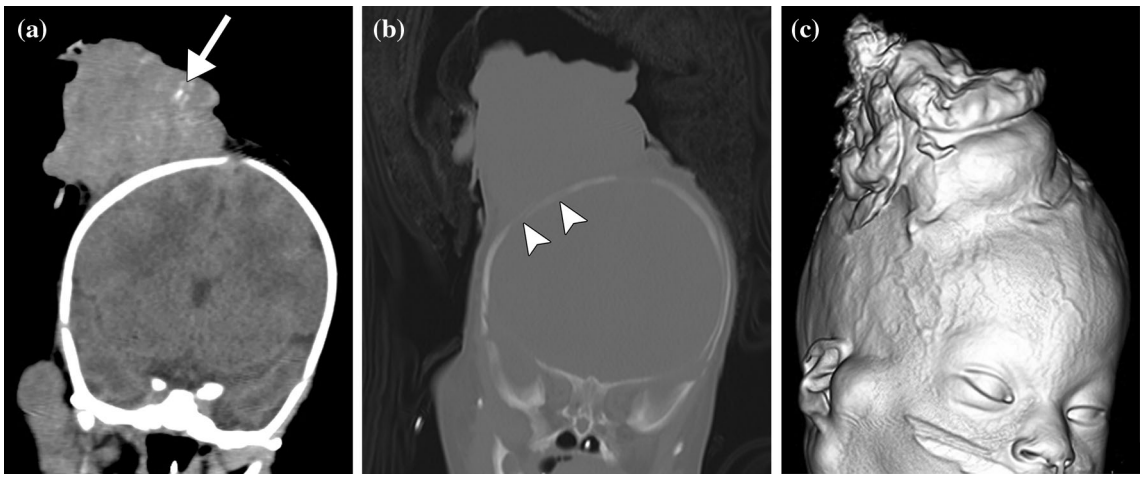

Fig. 1 Coronal soft tissue window (**a**), coronal bone window (**b**), and 3D (**c**) CT images show bulky right scalp and pre-auricular tumors with intratumoral hemorrhage (*arrow*) and osseous erosions (*arrowheads*) associated with right scalp mass

areas of hemorrhage (Fig. 2), as suggested by the CT. At both sites, the tumor was comprised of sheets of undifferentiated small cells that could be summarized under the descriptive term of a "small blue cell tumor". The lesional cells were arranged in vague nests and sheets. Nests of tumor cells were found to infiltrate between pre-existing tissue elements like adnexal structures. Rhabdoid features with perinuclear round eosinophilic staining attributed to whorled arrangement of cytoplasmic intermediate filaments was seen in part of the tumor (Fig. 3), but was not a universal feature of the tumor cells. The presence of whorled perinuclear aggregates of intermediate filaments was confirmed by ultrastructural studies. The tumor showed focal necrosis and high mitotic activity with over 25 mitoses per 10 hpf. There was no evidence of lymphovascular invasion in the scalp tumor, but in the neck mass, immunohistochemical studies identified focal areas of staining of residual dendritic cells suggesting that the neck lesion may represent spread of disease to lymph node(s). On immunohistochemical studies, the tumor cells exhibited patchy labeling for EMA and cytokeratin Cam5.2, as well as focal staining for desmin and synaptophysin. Patchy staining for CD99/MIC2 was seen but FLI-1 was negative. Also absent was expression of CD45/LCA and myogenin. Molecular studies by SNP array lastly showed homozygous loss of 22q of a region that includes the SMARB1/INI1 gene. Of note, no 22q loss was seen in the peripheral blood. Associated with this molecular alteration is the expected loss of INI-1 expression in the tumor cells (Fig. 4). The rhabdoid morphology, immunoprofile histochemical staining, and molecular features were consistent with diagnosis of a malignant rhabdoid tumor. The only other site of disease that was discovered at autopsy besides the scalp and the neck was a small deposit of tumor in the meninges overlying the right occipital lobe.

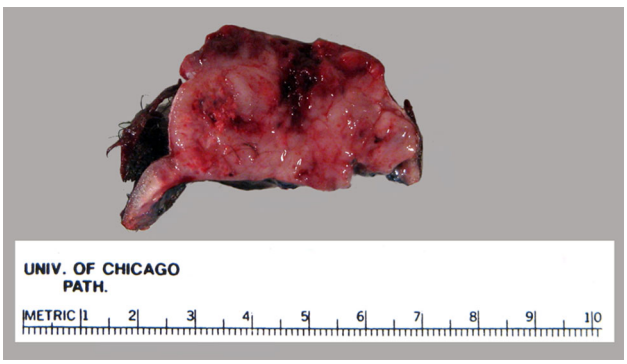

Fig. 2 Gross specimen photograph of the scalp tumor shows poorly-circumscribed polypoid tan tissue, with areas of hemorrhage

Discussion

Extrarenal malignant rhabdoid tumors are rare and aggressive neoplasms that are histologically distinct from Wilms tumor, but are analogous to atypical teratoid rhabdoid tumors of the central nervous system [1–5]. These tumors typically occur in infancy, but can also be congenital, particularly with the disseminated form, such as in this case. Although the tumors have been reported in soft tissues throughout the body, including the heart, musculoskeletal system, genitourinary system, liver, lungs, thymus, and thyroid, in most cases, the dominant

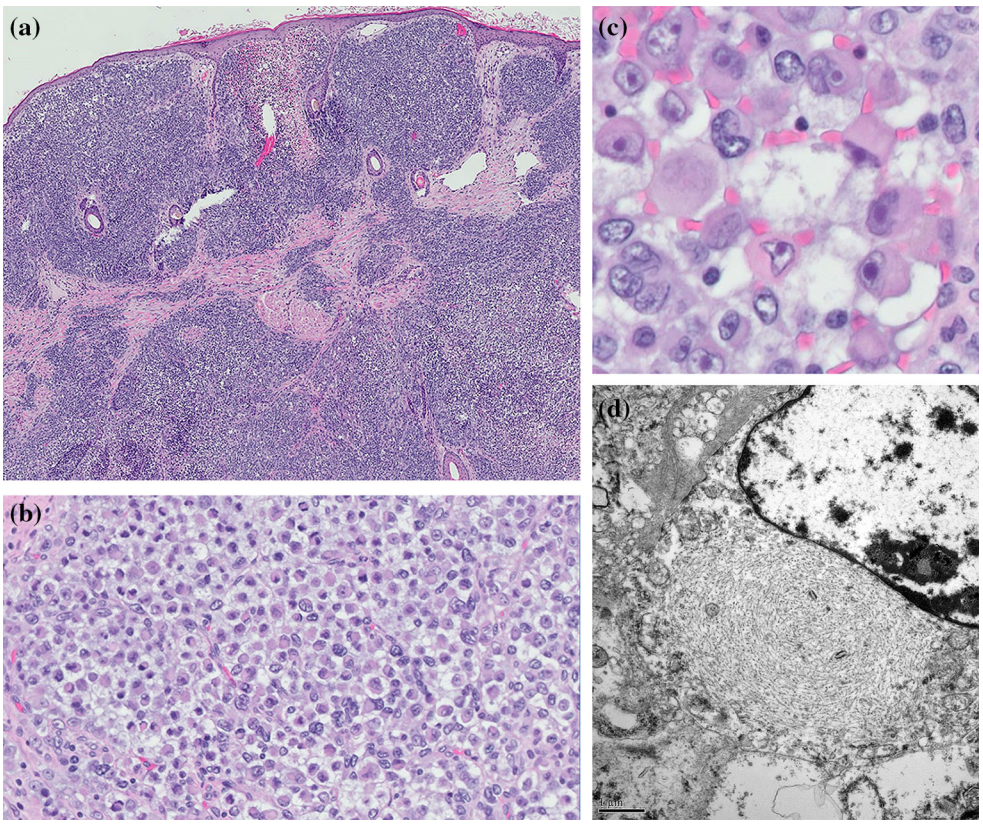

Fig. 3 Low power (**a**), intermediate power (**b**), and high power (**c**) hematoxylin and eosin stained photomicrographs show small round blue cells, sheets of small predominantly epithelioid cells. Cells with distinct eosinophilic cytoplasm imparting a rhabdoid appearance are present focally. Electron microscopy (**d**) shows the cytoplasmic inclusions are formed by the whorled filaments

mass involves the head and neck region. In particular, subcutaneous lesions are common with the disseminated form [1].

Extrarenal rhabdoid tumors tend to be bulky, lobulated, and hypoattenuating on CT, hypointense on T1-weighted MRI, heterogeneously hyperintense on T2-weighted MRI, and heterogeneously enhance on CT and MRI, often with areas of necrosis [2, 3]. On ultrasound, the tumors tend to appear as heterogeneously solid, moderately vascular masses [3]. Ultimately, the imaging features are not particularly specific [3], although the presence of aggressive features, such as bone erosions, may suggest a malignant neoplasm. Imaging is particularly useful for assessing the extent of disease with the disseminated form.

Ultimately, the diagnosis of extrarenal malignant rhabdoid tumor is mainly based on histopathology, immunohistochemistry and molecular studies. In general, soft tissue rhabdoid tumors are characterized by dyscohesive cells arranged in sheets, clusters, or single file pattern. Tumor cells are large with abundant glassy eosinophilic cytoplasm, eccentric vesicular nuclei, and large nucleoli, the so-called rhabdoid cells. Cells can also show spindled or primitive small cell morphology. A high mitotic rate and vascular permeation are present in the majority of tumors [6]. The characteristic immunoprofile is positivity for keratin and loss of INI1 expression due to biallelic loss of the SMARCB1 gene on chromosome 22q11.2 [2, 4, 5].

The histologic differential diagnosis for malignant mesenchymal lesions with rhabdoid phenotype includes the following: (1) rhabdomyosarcoma, which has positive muscle markers and retained INI-1 expression; (2) desmoplastic small round cell tumor, which demonstrates desmoplastic stroma and positive desmin staining, also with retained INI-1 expression; (3) epithelioid malignant peripheral nerve sheath tumor, which is characterized by relatively uniform atypical epithelioid cells with diffuse staining for S100 and loss of INI-1 expression in 50 % of cases [7, 8]; (4) epithelioid sarcoma, which also expresses keratin and loss of INI-1, but often shows intensely eosinophilic collagen in the center of the tumor nests and is CD34 is positive in 50 %. Furthermore, epithelioid sarcoma affects adults and rarely occurs in the head and neck region [8]. Finally, the possibility of metastases from a malignant rhabdoid tumor should be excluded by clinical correlation and imaging.

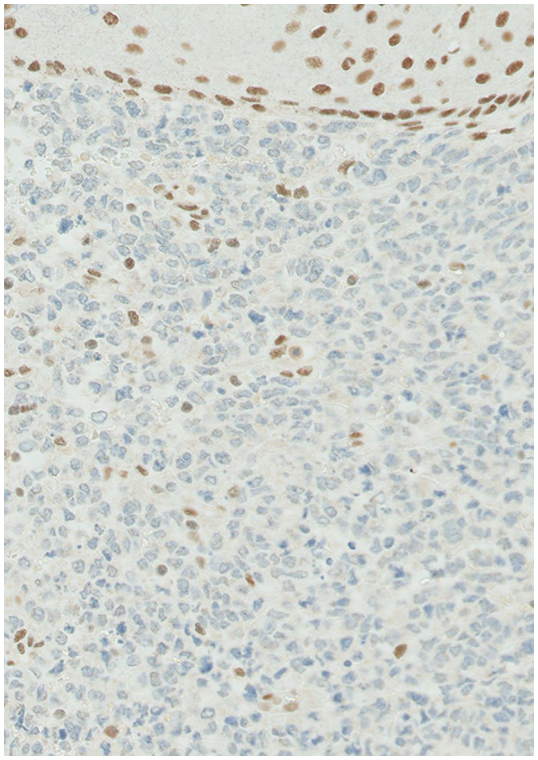

Fig. 4 Photomicrograph shows loss of INI1 nuclear staining in the tumor. INI1 is retained in epidermis, as an internal control

Treatment for extrarenal rhabdoid tumors is not well established and there is a high mortality rate for infants born with disseminated disease [1].

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