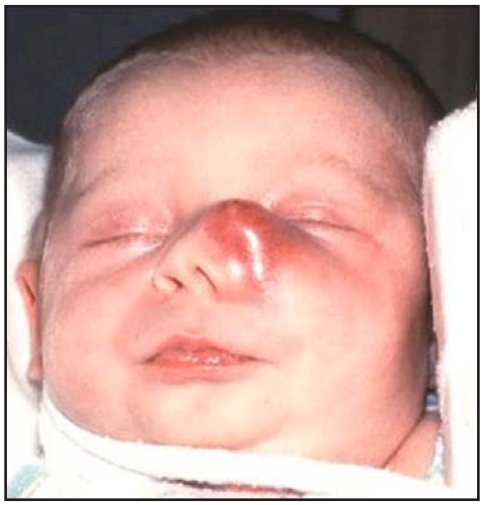
Pathological Condition of a Newborn Child

Dolphine Oda, BDS, MSc



The following Case Challenge is provided in conjunction with the American Academy of Oral and Maxillofacial Pathology.

Case Summary

This case challenge deals with a pathological condition of a newborn child.

A new born female presented at birth with a paranasal mass which appeared to be hemangiomatous. An ultrasound and aspiration biopsy of the lesion performed at the time of initial exam were not diagnostic. Due to the rapidly increasing size of the mass, the patient was referred for further evaluation to the Department of Oral and Maxillofacial Surgery at a major northwestern children's hospital. The clinical work up included a bone scan, chest and abdominal CT, and soft tissue biopsy.

After you have finished reviewing the available diagnostic information, make the diagnosis.

Diagnostic Information

Parental History
There were no known factors in the parent's history which contributed to this child's condition.

Chemical Factors
There were no known chemical risk factors in either the patient or parent's history which contributed to the condition.

Physical Factors
There were no known physical risk factors in either the patient or parent's history which contributed to this child's condition.

Histological Slides

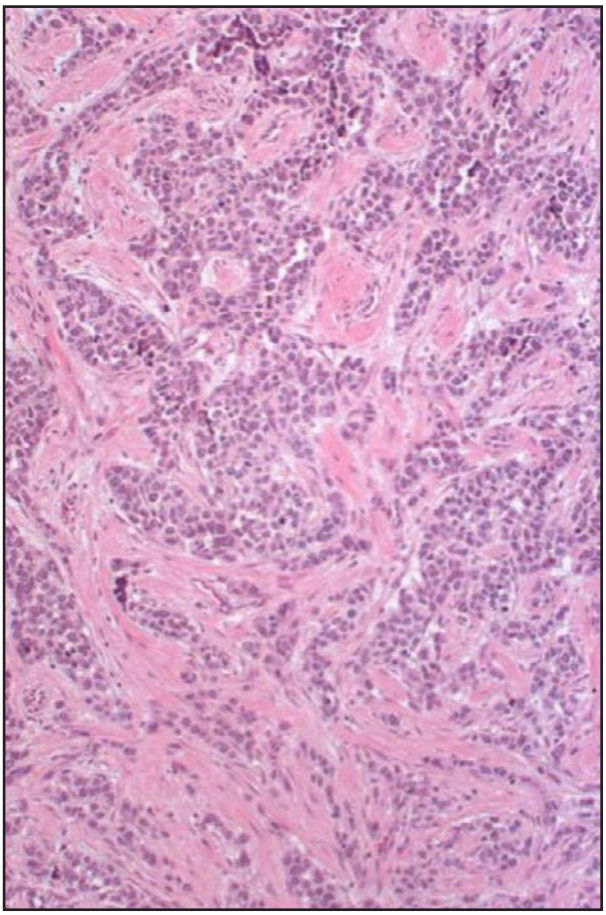


Figure 1. Low power x200

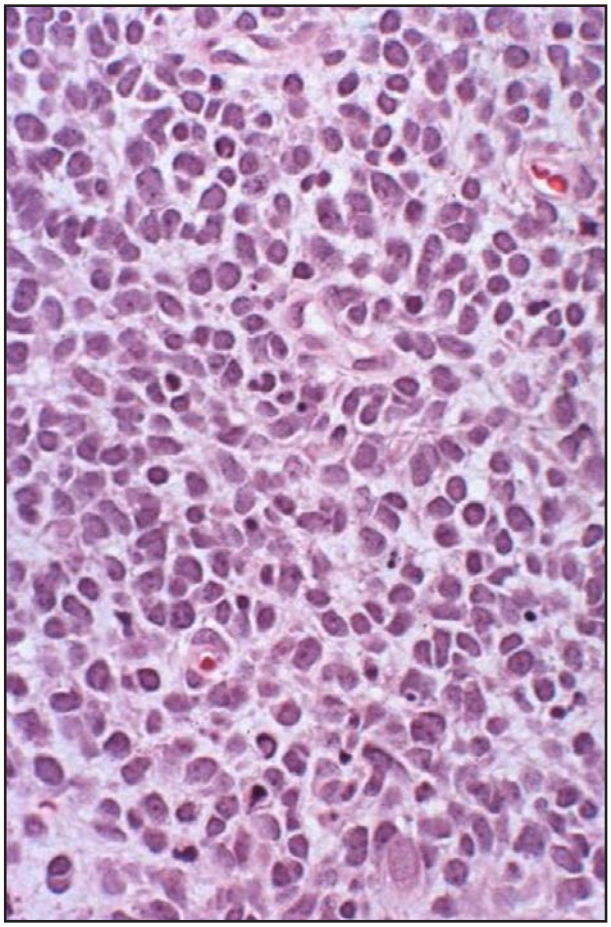


Figure 2. High power x400

MRI and CT Scan
A CT scan and an MRI of the head and maxillofacial region revealed a large endophytic and exophytically growth arising from the left paranasal region. An incisional biopsy was performed three weeks after birth and the lesion exhibited translocation T 2;13 which is typical for this lesion.

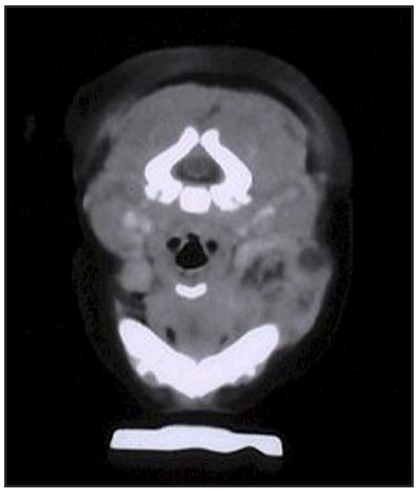
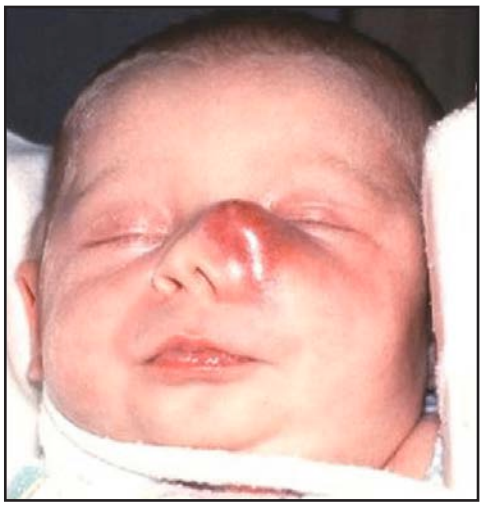


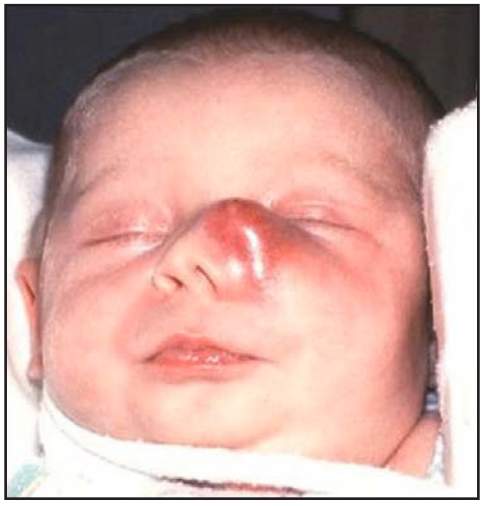
Figure 3. CT scan of area of the abnormality

Clinical Photograph



Can you make the diagnosis?

This case challenge deals with a pathological condition of a newborn child.



Select the Correct Diagnosis

A. Infection
B. Hemangioma/Hemangiopericytoma
C. Congenital Rhabdomyoarcoma
D. Histiocytosis X

Infection

Choice A. Sorry, this is not the correct diagnosis.

Infection-related erythematous swellings are the most common clinical presentation in newborn babies. Patients present with fever, general malaise, loss of appetite and urinary tract problems. This patient did not present with any of these symptons. The lesion was aspirated but did not yield any fluid, pus, or otherwise. The deep seated swelling may also represent sinus fungal infections such as mucormycosis and aspergellosis. Both of which are found in immunocompromised patients. The medical history was negative for AIDS.

Please re-evaluate the information about this case.

Hemangioma/Hemangiopericytoma

Choice B. Sorry, this is not the correct diagnosis.

Hemangioma is the second most common congenital lesion. It is the most common benign neoplasm in children and many occur at birth. The color is red but did not blanch with pressure. The fast growth is not consistent with hemangioma. It can not be definitively ruled out based on the clinical presentation. This is also true with hemangiopericytoma which may occur at birth, but does not exhibit rapid growth. The histology was negative for hemangioma or hemangiopericytoma.

Please re-evaluate the information about this case.

Congenital Rhabdomyoarcoma

Choice C. Congratulations! You are correct.

Discussion

Synopsis of the Disease

Rhabdomyosarcoma (RMS) is a malignant soft tissue tumor that accounts for 4-8% of all malignant disease in children under 15 years.[1] RMS was first described by Weber in 1854.[2] Stout (1946) was the first to give a detailed description of RMS in his report of 121 cases.[3] Stobbe and Dargeon (1950) identified the embryonal type of RMS in pediatric cases as a frequently misdiagnosed tumor in the head and neck region.[4] RMS of the head and neck is primarily seen in the first decade of life with a peak incidence between 2-6 years.[5] Bras and Batsakis noted that in their patients 77% were under 12 years.[6] Most studies have demonstrated that this tumor is slightly more common in males. In children, the head and neck (35%) is the most common site, followed by the genitourinary tract (23%), and extremeties (17%).[7,8] Depending on the location, RMS in the head and neck may be classified as: (I) orbital, (II) parameningeal, and (III) non-orbital-non-parameningeal. The orbit and nasopharynx, paranasal sinuses, middle ear and mastoid, soft tissues of the face and neck, oral cavity, pharynx and larynx are the predominant sites in the order of frequency in the head and neck.[6,7,8] In the oral cavity, RMS has been reported to account for 10-12% of all head and neck RMS.[9,10,11] The tongue, palate and cheek are the most common sites in the oral cavity.[10,11,12]

Clinical appearance may vary from a small cutaneous nodule to an extensive mucosal outgrowth. It may present as a painless facial swelling or may be occasionally associated with pain, trismus, paresthesia, facial palsy, aural or nasal discharge. It is because of this wide variation that the presenting clinical features are often non-diagnostic. This presentation in the head and neck often mimics other conditions like infection or an inflammatory polyp leading to delay in the diagnosis or misdiagnosis. Histologically, RMS is a neoplastic analogue of skeletal muscle embryogenesis.[7] The majority of newly diagnosed RMS can be classified as embryonal 60%, or alveolar 20% and the remaining as undifferentiated.[13] The morphologic subtype of the embryonal variety sarcoma-botryoides accounting for 5% of the cases derives its name mainly due to its gross form resembling a cluster of grapes (botrys in greek - a cluster of grapes).

The clinical grouping system used by the IRS committee is the most widely used staging system, although contemporary studies of RMS have relied on both the IRS clinical grouping system and TNM staging to assign therapy.[14]

Metastases of RMS is primarily by hematogenous route to lungs, bone, brain and other viscera. This tumor is very aggressive and, if undiagnosed, manifests high mortality. With the use of risk-adapted multidrug chemotherapy combined with radiotherapy, surgical excision, when possible, and more refined tumor grouping through ongoing research efforts of the Intergroup Rhabdomyosarcoma study (IRS), the five year survival rate has improved from 20 to 70%.[13,14]

Treatment

The infant was then enrolled in the MAYO Clinic's STS protocol and 12 weeks of chemotherapy was initiated with VCR, doxorubicin, cyclophosphamide-IV, etopside-IV and ifosamide. It was followed by surgery and radiotherapy.

Follow-up

The patient developed metastasis and was placed on chemotherapy. Her condition improved. The patient is alive and living with the disease with the assistance of chemotherapy.

Histiocytosis X

Choice D. Sorry, this is not the correct diagnosis.

Histiocytosis X is classified into three different types. The first two types occur at birth and involve both the bone and skin. The third type is eosinophilic granuloma which affects the bone alone. This lesion did not involve the bone and was isolated.

The histology was negative for histiocytosis X.

Please re-evaluate the information about this case.

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About the Author

Note: Bio information was provided at the time the case challenge was developed.

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