

Case of a Growing Congenital Mass

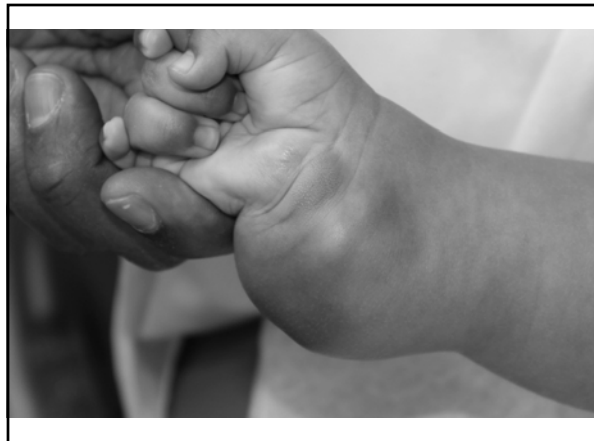
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Emory University Hospital
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Financial Disclosures

- I have no financial disclosures or conflict of interest

Patient History

- Healthy 2 month old Ethiopian Female
- At birth: slightly tender bluish nodule on right wrist
- X-ray from OSH: No evidence of bone involvement/ growth
- Ultrasound from OSH: "hemangioma"
- At time of presentation
 - Doubled in size
 - No bleeding or ulceration



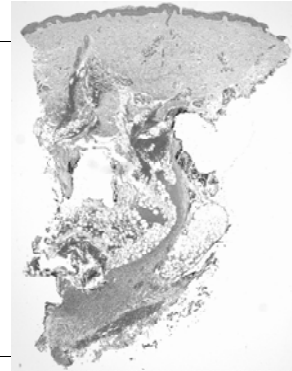
Differential Diagnosis

- Kaposiform Hemangioendothelioma
- Tufted angioma
- Congenital Hemangioma
- Congenital fibrosarcoma
- Infantile Myofibroma
- Congenital Dermatofibrosarcoma protuberans

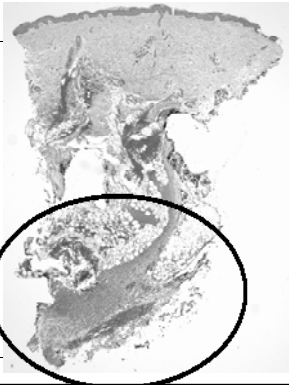
What Next?

- CBC including Platelet count- wnl
 - Kasabach-Merritt Syndrome
- Punch Biopsy

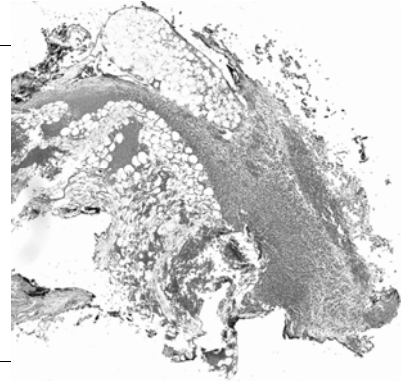
2x H&E



2x H&E

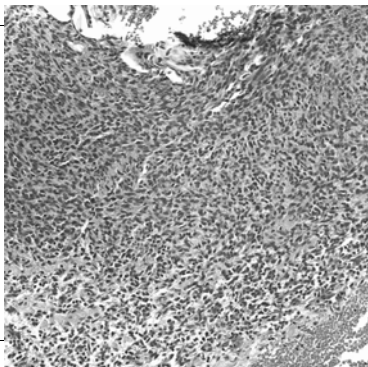


4x H&E



20x H&E

- Hyperchromatic ovoid & spindle shaped cells
- Focal moderate pleomorphism
- Broad cellular fascicles

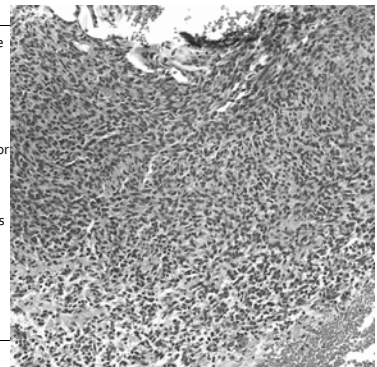



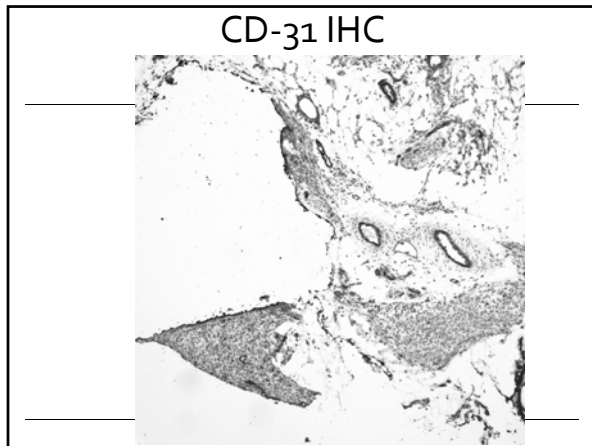
20x H&E

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Preliminary Biopsy concerning for fibrosarcoma:

- Tissue sent to Texas Children's Hospital for translocation studies

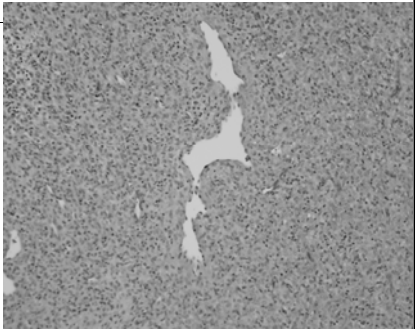




- Complications with hemostasis s/p biopsy
- Site continued to bleed despite multiple attempts at closure
- Admitted for dehiscence of biopsy site



H&E




- Dilated congested vessels
- Hemangiopericytoma-like pattern

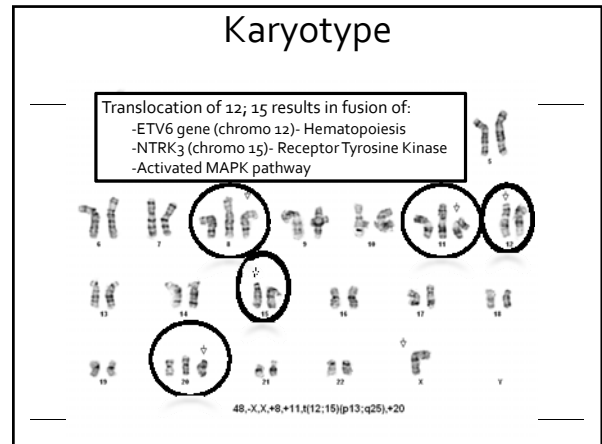
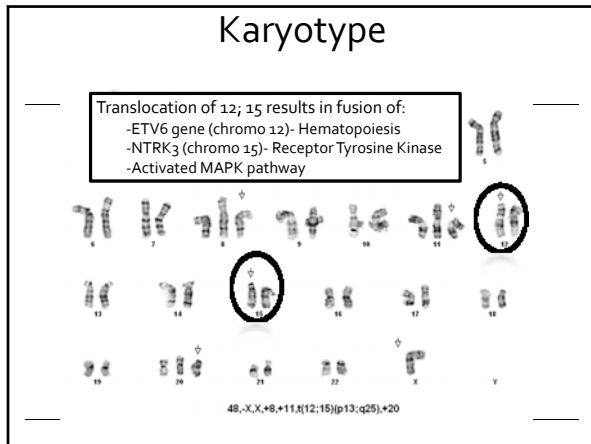
Molecular Studies

- Translocation studies (RT-PCR) from Texas Children's Hospital
- Chromosome analysis done at Emory
 - Karyotype of tumor

Karyotype



48,-X,X,+8,+11,t(11;15)(p13;q25),+20




Final Diagnosis

- Congenital Infantile Fibrosarcoma


Infantile Congenital Fibrosarcoma

- Relatively Rare
- Present at birth (1/3) or during 1st year of life
- Firm and fixed nodule or mass
 - Concern for malignancy
- Usually slow growing, non-tender mass
- Extremities: hand, wrist, forearm, ankle, foot
- Poorly circumscribed spindle cell tumor
- Highly vascular tumors
 - Ultrasound and MRI can be difficult to interpret
 - Often misread as hemangioma or lymphatic malformation (cystic spaces from necrosis)



Infantile Congenital Fibrosarcoma

- Morphologically resembles adult fibrosarcoma
- BUT separate entity:
 - Locally infiltrative, rare metastasis
 - Translocation t(12;15) results in fusion gene



Treatment

- Chemo-sensitive tumor
- Attempt to perform non-mutilating surgery for total excision
 - Positive surgical margins after resection may not indicate poor prognosis
- Amputation

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Our patient:

- Resection
- Port-A-Cath placed for OP low dose chemotherapy
- 12 wks of Vincristine, cyclophosphamide, actinomycin-D

References

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Thank you

- Dr. Mary Spraker
- Dr. Leslie Lawley
- Dr. Scott Lauer

Differential Diagnosis

- **Congenital Hemangioma**
 - MC benign soft tissue tumor of childhood
 - Fully formed at birth - 1-1% of neonates
 - Deep hemangioma: Subcutaneous nodules/tumor w/ overlying blue hue w/ telangiectases or prominent venous network
- **Kaposiform Hemangioendothelioma**
 - Rapidly expanding firm SQ nodules/plaques w/ violaceous discoloration
 - Associated coagulopathy, Kasabach-Merritt phenomenon (coagulopathy, thrombocytopenia, microangiopathic hemolytic anemia)
- **Tufted angioma**
 - Erythematous indurated annular nodules/plaques
 - Associated coagulopathy, Kasabach-Merritt phenomenon (coagulopathy, thrombocytopenia, microangiopathic hemolytic anemia)
- **Congenital fibrosarcoma**
 - Common on distal limbs
 - Red-red/blue firm tumors
 - Usually slow growing
- **Infantile Myofibroma**
 - MC benign mesenchymal tumor of infancy
 - Firm (rubbery colored) trantax nodules
 - Usually spontaneously disappear
- **Congenital Dermatofibrosarcoma protuberans**
 - Slow growing firm flesh-red nodules
 - Slow growing, high rate recurrence w/ excision, metastases uncommon