DIAGNOSTIC SLIDE SESSION 2016 CASE 2016-3

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DISCLOSURES

No relevant financial relationships to disclose.

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CLINICAL HISTORY

HPI:

- 23-year-old previously healthy female
- Presented to Ophthalmology for bilateral scleritis with new symptoms of right ear chondritis
- Referred to Rheumatology for further work-up

Neurologic exam:

- Alert and oriented x 3, CN 2-12 grossly intact
- 5/5 strength, normal sensation, normal range of motion
- Reflexes intact



RADIOLOGIC STUDIES

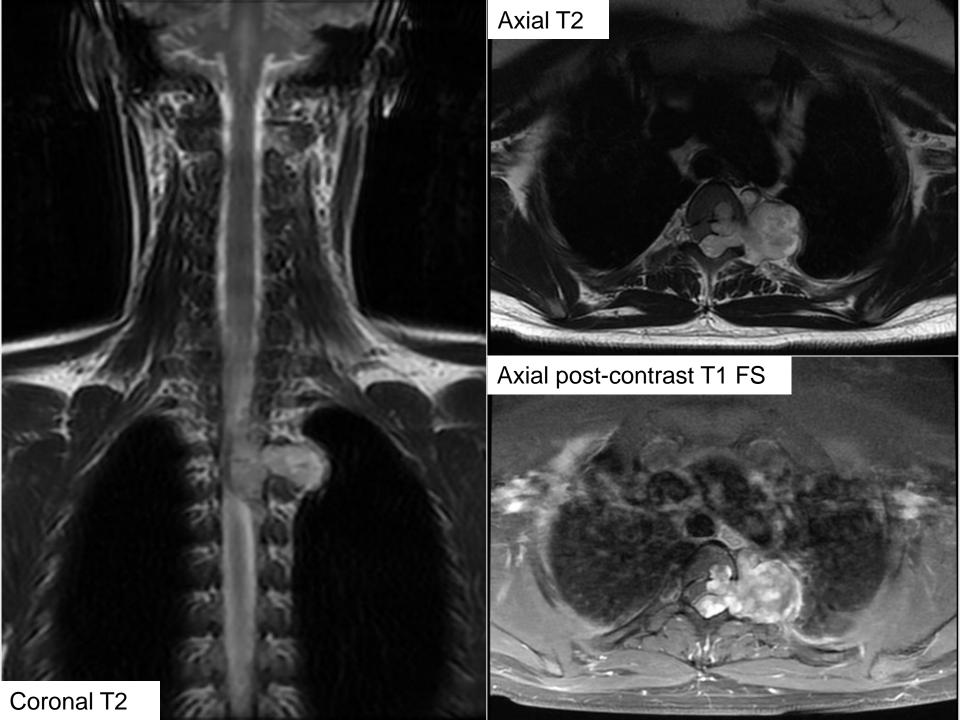
Chest radiograph:

2.9 cm round opacity projecting just above the aortic arch

CT:

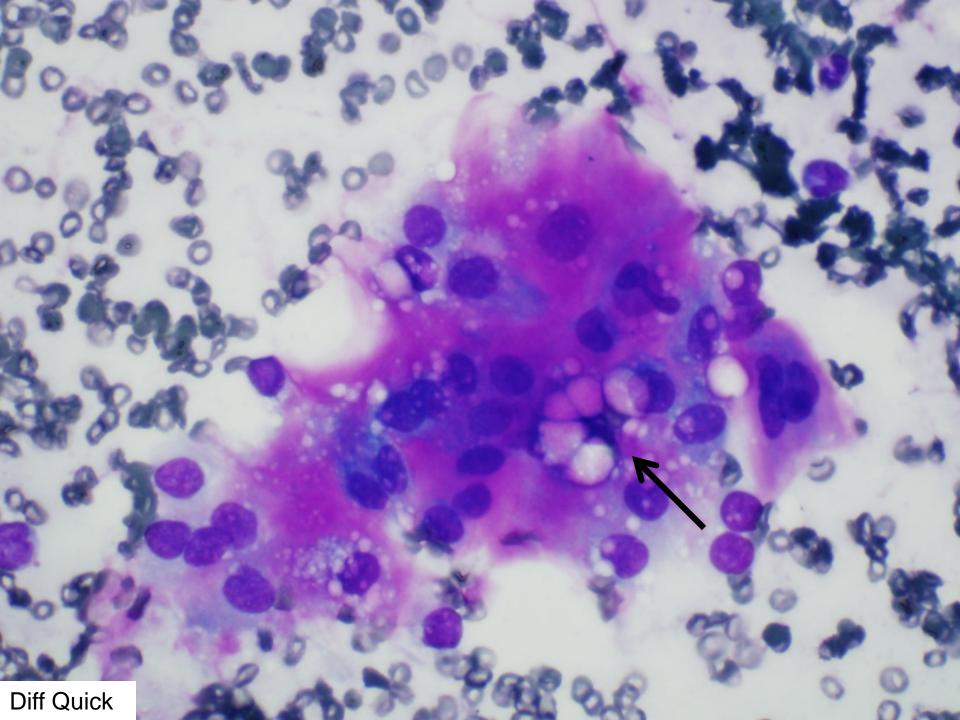
Posterior mediastinal mass extending from the left T3-T4 neural foramen

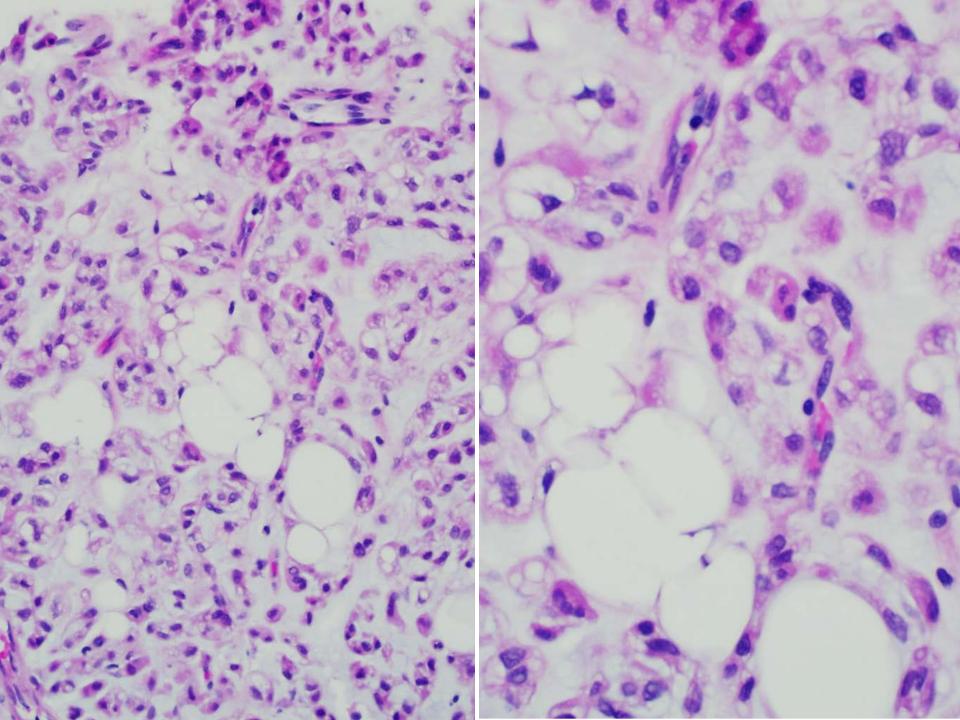




BIOPSY

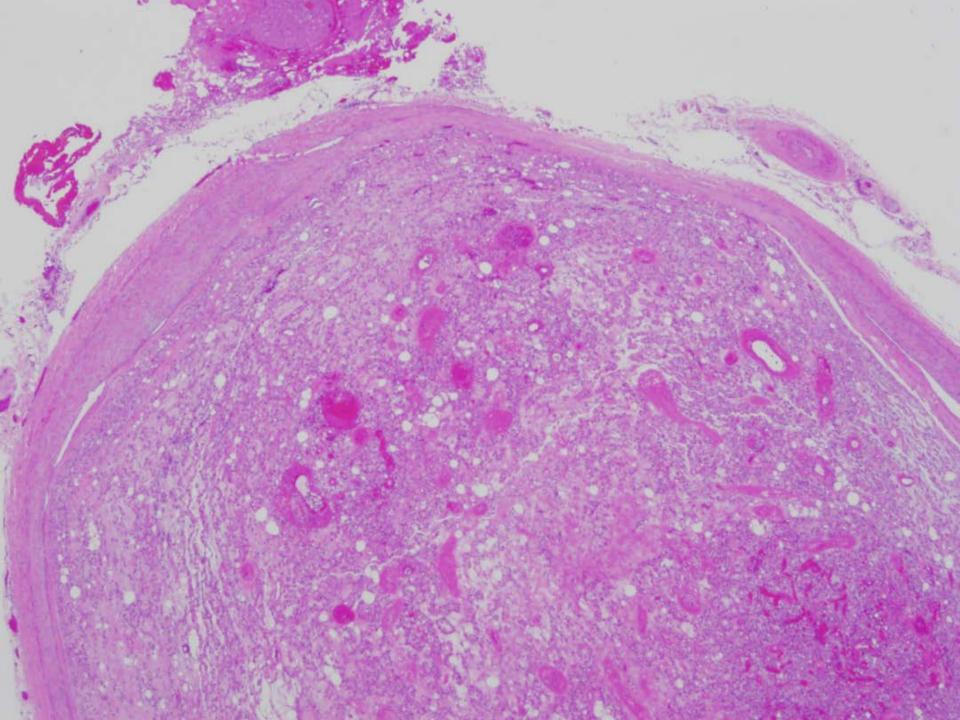


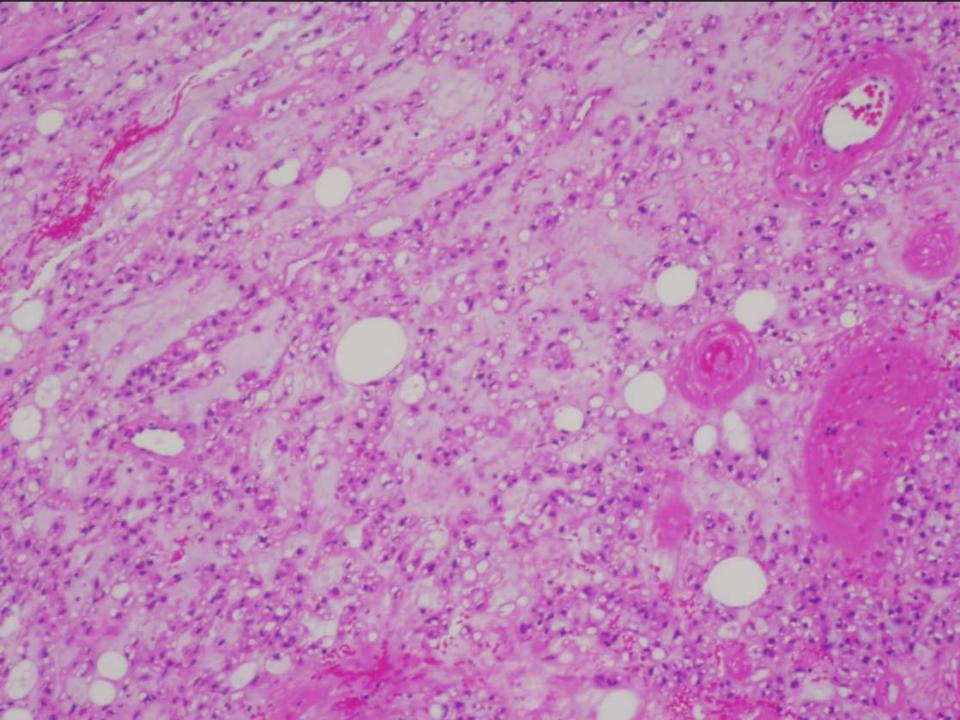


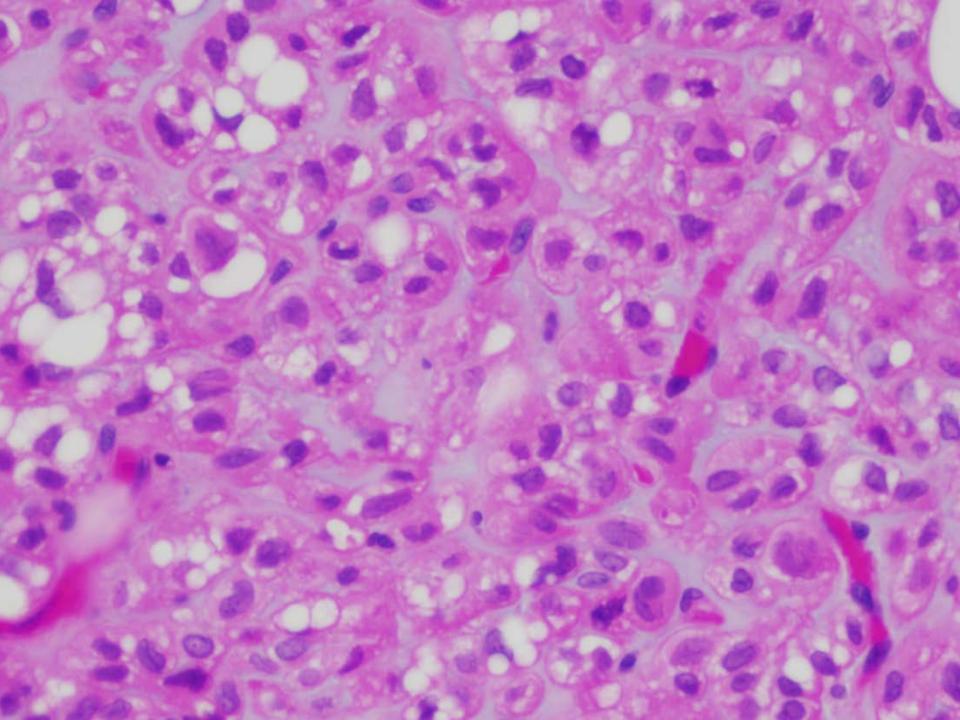


RESECTION







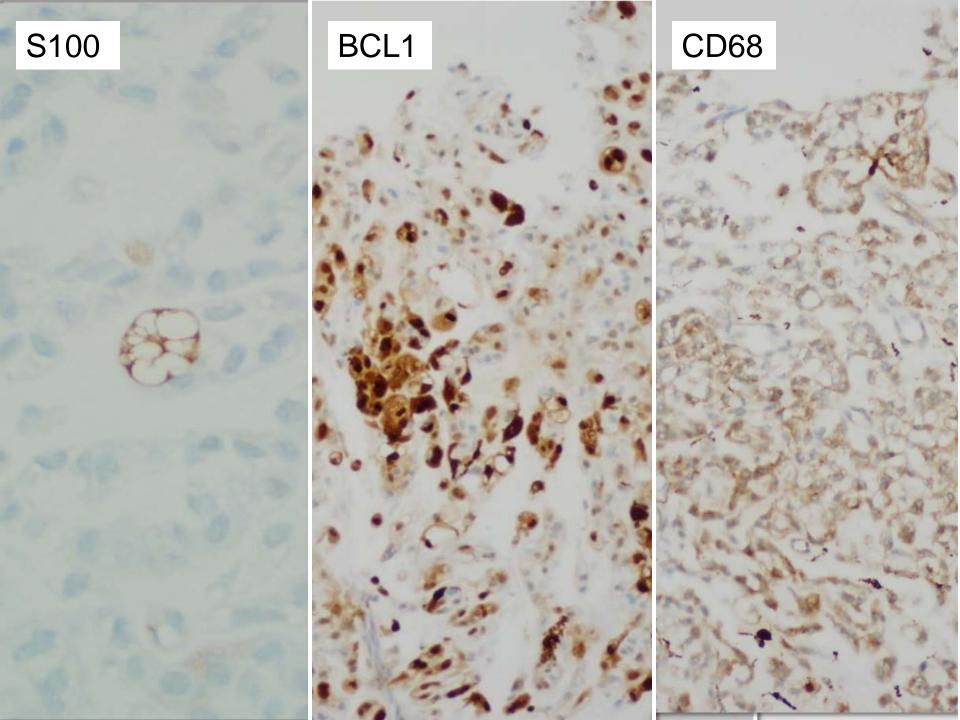


DIFFERENTIAL DIAGNOSIS?



IMMUNOSTAINS





IHC SUMMARY

- Immunopositive: \$100 (lipoblast-like cells), BCL1, and CD68
- Immunonegative: AEI/AE3, SOX10, and EMA
- Ki67 proliferative index: <1%



CYTOGENETICS AND MOLECULAR GENETICS

Fluorescence in situ hybridization (FISH)

- FUS gene rearrangement: Negative
- Cllorf95-MKL2 fusion gene: Positive



C11orf95 Spanning probe MKL2 Spanning probe

FINAL DIAGNOSIS:

CHONDROID LIPOMA



Chondroid Lipoma

A Unique Tumor Simulating Liposarcoma and Myxoid Chondrosarcoma

Jeanne M. Meis, M.D., and Franz M. Enzinger, M.D.

The American Journal of Surgical Pathology 17(11): 1103-1112, 1993

© 1993 Raven Press, Ltd., New York

Clinical

- Subcutaneous or deep soft tissues
- Extremities or trunk
- 2nd-3rd decade (females > males)
- No recurrence or metastasis
- Local excision is curative

Pathologic

- Mature adipose, myxochondroid matrix, vacuolated cells
- +\$100, CD68, keratin (1/3)
- -EMA, HMB45, Actin
- t(11;16)(q13;p12-13) c11orf95-MKL2



Genes Chromosomes Cancer. Author manuscript; available in PMC 2011 September 1.

Published in final edited form as:

Genes Chromosomes Cancer. 2010 September; 49(9): 810-818. doi:10.1002/gcc.20788.

C11orf95-MKL2 is the Resulting Fusion Oncogene of t(11;16) (q13;p13) in Chondroid Lipoma

Dali Huang¹, Janos Sumegi², Paola Dal Cin³, John D. Reith⁴, Taketoshi Yasuda¹, Marilu Nelson⁵, David Muirhead¹, and Julia A. Bridge^{1,5,6,*}

MKL2 gene

- Gene in the myocardin/megakaryoblastic leukemia gene family
- Codes for a 1049 amino acid myocardin-like protein that can act as a transcription factor
- Cllorf95 protein
 - Function yet to be determined

PATIENT UPDATE

- Patient doing very well: denies fevers, chills, sweats, wound difficulty, drainage, weakness of extremities, or incontinence
- Post-operative pain/spasm still present but improving daily
- MRI: No evidence of residual or recurrent disease



CONCLUSION

- Rare soft tissue neoplasm most often occurring in young women in the extremities
- May cause concern due to size and presence of lipoblasts
- Behave in a benign fashion
- Small biopsies and uncommon locations may cause diagnostic confusion



REFERENCES

- Huang D, Sumegi J, Dal Cin P, Reith JD, Yasuda T, Nelson M, Muirhead D, Bridge JA. C11orf95-MLK2 is the resulting fusion oncogene of t(11,16)(q13;p13) in chondroid lipoma. Genes Chrom Ca 2010;49(9):810-18.
- 2. De Vreeze RSA, van Coevorden F, Boerrigter L, Nederlof PM, Haas RL, Bras J, Rosenwald A, Mentzel T, de Jong D. Delineation of chondroid lipoma: an immunohistochemical and molecular biological analysis. Sarcoma 2011;2011:638403
- 3. Thway K, Rashpal S, Fisher C. Chondroid Lipoma: an update and review. Ann Diag Pathol 2012;16(3):230-4.



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