An unusual case of knee pain: A bony complication of Maffucci syndrome

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Outline

**Part 1:** Evaluating knee pain in a patient with Maffucci Syndrome

- Patient Presentation
- Review of Bone Anatomy
- Introduction to Maffucci Syndrome
- Menu of Tests for Knee Pain
- Differential Diagnosis
- Confirming the Diagnosis
Patient Presentation

- **Chief Complaint:**
  - 49 y/o female with a history of Maffucci syndrome presents with “burning pain in right knee”

- **History of Present Illness:**
  - She has had increasingly severe pain in her right knee over the past year with no history of trauma.
  - She has 5/10 pain at rest, 8/10 pain when she walks and it awakens her at night.
  - She takes ibuprofen, tylenol and lyrica, which provides moderate relief from the pain
Patient Presentation Cont’d

- **Focused Physical Exam**
  - Walks with an antalgic gait.
  - Extreme pain sensitivity over right distal femur, and around patella.
  - Possible mass around right distal lateral femoral condyle, exquisite pain sensitivity when palpated.
  - Limited range of motion in right knee.
  - Full range of motion and no pain on left knee.
Review of Long Bone Anatomy

Source: http://biologyonline.us
What is Maffucci Syndrome?

- Maffucci syndrome is a rare, non-hereditary condition characterized by multiple enchondromas and soft tissue hemangiomas.
- First described in 1881.
- In 78% of the cases, the symptoms start before puberty.
- Patients with the syndrome demonstrate a normal intellect and generally live a productive life.
What are Enchondromas?

- Common benign proliferation of cartilage within the medullary cavity
- In Maffucci syndrome, multiple enchondromas present in various parts of the skeleton, especially the long bones of the hands and feet.
- This causes bone changes including shortened length of long bones, pathologic fractures and malunion of fractures. However, malignant transformation of enchondromas to chondrosarcoma is the biggest risk.
What are Enchondromas?

- Oval, well circumscribed radiolucent lesion.
- Cortex can be scalloped and thinned if the lesion abuts the cortical border.
- Densities grow in lobules and some lesions may calcify:
  - Rings - Complete calcification around cartilage lobule
  - Arcs – partial calcification of cartilage lobule
  - Less than 1% may show eccentric protuberation.

Source: Skinner, 2006

Enchondromas in Maffucci Syndrome.

Source: PACS, BIDMC

Source: Abdelmak and Stanko, 2008
What are Hemangiomas?

- Hemangiomas are a benign proliferation of blood vessels.
- Maffucci syndrome is characterized by multiple hemangiomas, located in the subcutaneous tissue.
- They appear as blue or reddish blue, soft to firm subcutaneous nodules that blanch upon compression.
- Thrombi can form within the vessels leading to phleboliths (local calcification) that are visible on radiographs.
- Ollier’s syndrome is a similar condition to Maffucci syndrome, characterized by a proliferation of endochondromas without hemangiomas.
Hemangiomas in Maffucci Syndrome.

Source: PACS, BIDMC

Source: Abdelmak and Stanko, 2008
Back to Patient: Menu of Tests for Knee Pain

- Plain x-ray
- CT
- MRI
- Ultrasound
- Nuclear Medicine

- Convenient and inexpensive
- Can identify broad range of aggressive and non-aggressive lesions.
- Limited radiation
# American College of Radiology Appropriateness Criteria

**Variant 2:** Child or adult: patellofemoral (anterior) symptoms. Initial examination.

<table>
<thead>
<tr>
<th>Radiologic Procedure</th>
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<td>US knee</td>
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</table>

*Rating Scale: 1,2,3 Usually not appropriate; 4,5,6 May be appropriate; 7,8,9 Usually appropriate

*Relative Radiation Level*
X-Ray Results

- Intramedullary ring and arcs calcification in the distal femoral diaphysis.
- Exostosis of the anterior distal femur.
- Expansile lytic lesion within the anterior lateral femoral diaphysis with ring and arcs calcification.
- Multiple soft tissue calcifications consistent with hemangiomas.
- Focal lytic lesion within the right tibial metaphysis with endosteal scalloping.
Differential Diagnosis

- Enchondroma
- Chondrosarcoma
- Metastases

- What is the next step?
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MRI – T1 Sequence

- Multiple focal lesions that were low intensity on T1.
- A lesion with an exophytic component within the mid right femoral shaft with endosteal scalloping.
- An exophytic lesion projecting from the medial cortex of the proximal right tibia measuring approximately 1.9 cm in diameter with deep endosteal scalloping.
- Large lesion that extends through the cortex of the right lateral femoral condyle. (4.6 cm in anteroposterior dimension x 4.1 cm transversely x 6.8 cm in the craniocaudal dimension). Exophytic component is intra-articular abutting the lateral patellar facet.

Source: PACS, BIDMC
Focusing on the knee

- Multi-lobular lesion with complete erosion of the cortex.
- T1: Lesion has mostly low signal intensity, with areas of high intensity.
- T2/STIR: Lesion is mostly high signal intensity with low signal intensity septa.
- Consistent with a cartilaginous lesion, but Enchondroma or Chondrosarcoma?
Diagnosis

• Differentiating chondrosarcoma from an enchondroma (Murphy et al., 1998, Brian et al., 1997)
  ▫ Pain related to lesion
  ▫ Palpable mass.
  ▫ Deep endosteal scalloping (greater than two-thirds of cortical thickness)
  ▫ Lesions greater than 5-6 cm in diameter
  ▫ Cortical destruction and soft tissue mass visible on imaging.
  ▫ Focal areas of cortical thickening

• What next?
  ▫ MRI, X-ray and clinical history would have been enough to confirm the diagnosis of a chondrosarcoma.
  ▫ CT-Guided Biopsy was performed to establish histologic grade and demonstrate any cortical abnormalities.
CT demonstrations extensive cortical destruction
Biopsy Results: Chondrosarcoma – Grade 2

“Permeating growth pattern, myxoid matrix, increased chondrocyte cellularity and mild atypia is seen. There are areas that show cytologic features of an intermediate grade chondrosarcoma”

Source: www.webpathology.com
Outline

**Part 1:** Evaluating Knee Pain in a Patient with Maffucci Syndrome

**Part 2:** Overview of Chondrosarcoma

- Introduction to Chondrosarcoma
- Types of Chondrosarcoma
- Diagnosis
- Staging
- Treatment
Chondrosarcoma

- Malignant tumor of cartilage producing cells.
- Chondrosarcomas are the third most common primary malignancy of bone after multiple myeloma and osteosarcoma.
- They account for 20-27% of all primary bone neoplasms.
- Histologic grading is an important marker of prognosis.
  - Grade 1: Moderately cellular with abundant hyaline matrix. Mitoses are absent. 1% risk of metastases. Ten year survival is 83 to 95%.
  - Grade 2: More cellular with less chondroid matrix that Grade 1. Mitoses are present but scattered. 10 to 15% risk of metastases. Ten-year survival is 64 to 86%.
  - Grade 3: Highly cellular with nuclear pleomorphism and easily detected mitoses. Chondroid matrix is sparse. 32 to 70% risk of metastases. Ten year survival is about 29 to 55%.
- Patients typically present with progressively worsening pain over months to years that is worse at night, palpable mass or pathologic fractures.
Classification

- Conventional chondrosarcoma are the most common type, accounting for 90% of tumors. Long bones of the appendicular skeleton are the most common sites.
  - **Central chondrosarcoma**
    - Arise from the medullary cavity and constitute approximately 75% of all chondrosarcomas.
    - 40% are malignant transformations of enchondromas, the rest are primary.
  - **Peripheral chondrosarcoma.**
    - All are malignant transformation of pre-existing osteochondromas
  - **Periosteal chondrosarcoma**
    - Arise from the surface of the bone.
- Rarer types include: dedifferentiated chondrosarcoma, mesenchymal chondrosarcoma, clear cell chondrosarcoma and myxoid chondrosarcoma.
Radiologic Diagnosis

- **Radiographs:**
  - Low grade tumors can appear similar to enchondromas with lytic regions that may have ring and arcs calcification patterns. They may have geographic margins and some endosteal scalloping.
  - As lesions grow, borders are moth eaten. Cortical thickening, deep endosteal scalloping (>two thirds of the depth of the cortex), cortical penetration and a soft tissue mass which is sometimes seen on radiograph (best seen on MRI) are suggestive of a chondrosarcoma.

- **CT:**
  - Provides more sensitive detection of depth and degree of cortical destruction.

- **MRI**
  - **T1:** Cartilaginous lesion has low to intermediate signal intensity. Some areas are void of signal due to matrix mineralization and some areas have high signal intensity due to entrapped fat marrow.
  - **T2:** Cartilaginous lesion has a high intensity on T2 because of the higher water content of cartilage. There are lobules of chondral tissue separated by hypointense areas of matrix mineralization.
The Enneking Staging system is generally used for bone sarcoma

<table>
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<tr>
<td>III</td>
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- Full staging involves at thoracic CT to evaluate for the presence of pulmonary metastases.
Treatment

- Low-grade tumors are treated with intralesional curettage followed by local adjuvant chemical treatment of phenol or cryotherapy and cementing or bone grafting
- Intermediate and high grade tumors are treated with wide local excision and endoprosthetic replacement
- Chondrosarcomas are considered relatively refractory to chemotherapy and radiation therapy because they are slow growing.
- Nevertheless, radiation therapy may be of benefit after an incomplete resection of a high grade tumor to maximize local control or palliatively in situations were resection is not feasible.
Outline

**Part 1:** Evaluating Knee Pain in a Patient with Maffucci Syndrome

**Part 2:** Overview of Chondrosarcoma

**Part 3:** Clinical outcome of patient and follow up
Clinical Course and Follow up

- Chest CT was done which showed no metastases to her lung.
- Underwent wide local excision of the chondrosarcoma and reconstruction of distal femur with a distal femoral replacement and rotating hinge knee replacement.
- Unfortunately, she had another recurrence of chondrosarcoma one year later.
Summary

**Part 1:** Evaluating Knee Pain in a Patient with Maffucci Syndrome
- Review of Bone Anatomy
- Introduction to Maffucci Syndrome (Multiple Enchondromas and Hemangiomas)
- X-ray is the initial imaging of choice for non traumatic knee pain.
- How to differentiate endochondromas from chondrosarcoma through radiologic imaging.

**Part 2:** Overview of Chondrosarcoma
- Overview of chondrosarcoma including histologic classification, radiologic features, staging and treatment

**Part 3:** Clinical outcome of patient and Follow up
- Patients with Maffucci syndrome have a high risk of chondrosarcomas.
References

- Skinner H & McMahon P. Current Diagnosis & Treatment in Orthopedics. 4e. Lange Series, The McGraw-Hill Companies, 2006
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