Hereditary Osteochondromatosis of the Upper Extremity

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Objectives

Understand the Problem
• MHE biology & genetics
• MHE natural history
• Radiographic measurements

Intervention
• Osteochondroma excision
• Detethering
• Growth modulation
• Osteotomies / lengthening
Naming (Boyer 1814)

- Multiple Osteochondromas(-atosis) (OCE)
- Multiple Hereditary Exostosis (MHE)
- Hereditary Multiple Exostoses (HME)
- Multiple Cartilaginous Exostoses
- Diaphyseal Aclasia
- Etc....

- Maffucci’s syndrome (MHE + hemangiomas)
Biology and Genetics

- Autosomal dominant with very high penetrance and variable expressivity
- Most common genetic mutations in EXT-1 and EXT-2 genes
- EXT 1 more severe deformity and higher sarcoma risk
Abnormal proliferation of chondroblasts → subsequent defective remodeling of the metaphysis

Retardation of longitudinal growth

Exostoses migrate away from the physis with longitudinal growth

Forearm deformity in 30-60% of affected individuals

Can be very asymmetric
Natural History

- Decrease in objective measurements of hand and wrist function predicted
- Not necessarily limiting in work, recreational activities
- Pain variable reporting and significance of limitations
- Cosmetic issues

Arms et al JPO 1997
Noonan et al JBJS 2002
Dariek et al JPO 2005
Jager et al JOR 2007
Radiographic Measurements

- Masada Classification
- Carpal Slip
- Radial Articular Angle
- Proportional Ulnar Length
Effect of Osteochondroma Location on Forearm Deformity in Patients With Multiple Hereditary Osteochondromatosis

Hilton P. Gottschalk, MD, Yumiko Kanauchi, MD, Michael S. Bednar, MD, Terry R. Light, MD
RAA and Carpal Slip

RAA: angle between articular surface of distal radius and line perpendicular to a line joining the center of the radial head to the radial border of the distal radial epiphysis.

Carpal slip: percent of lunate radial to line from center of olecranon through the ulnar border of distal radius.


Normal 15-30 deg

Normal <50%
Proportional Ulnar Length

Forearm Deformity in Patients with Hereditary Multiple Exostoses
Factors Associated with Range of Motion and Radial Head Dislocation

N.D. Clement, MRCSEd, and D.E. Porter, FRCSEd(Tr&Orth)

Investigation performed at the Department of Orthopaedics and Trauma, The Royal Infirmary of Edinburgh, Edinburgh, Scotland

- N=106 skeletally mature
- EXT-1 44, EXT-2 46
- 91% of patients with forearm exostoses
- 619 forearm exostoses
Proportional Ulnar Length

Predicted Range of Motion According to Sex and Proportional Ulnar Length*

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<thead>
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<th>Proportional Ulnar Length (%)</th>
<th>Male</th>
<th>Female</th>
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Intervention?

- Natural History Treatment?
- Excision OCE?
- …and Detethering
- …and Osteotomy Ulna?
- …and Osteotomy Ulna, Lengthening Ulna?
- …and Osteotomy Ulna, Lengthening Ulna, Osteotomy radius?
- …Single bone forearm?
OCE excision

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Simple excision OCE

- Improves pain
- Improves motion
- Improves function
- May improve growth
- Ulna lesion isolated
- Growth deformity progression
- Multiple lesions, both bones

- Ishikawa J, Kato H, Fujioka F. Tumor location affects the results of simple excision for multiple osteochondromas in the forearm. JBJS 2007
Radiographic Outcomes of Hemiepiphyseal Stapling for Distal Radius Deformity Due to Multiple Hereditary Exostoses

Jason P. Kelly, MD and Michelle A. James, MD
Simple excision, osteotomies, lengthening

- Improves rotation
- Improves carpal, forearm alignment
- Improves appearance
- Lessens pain long term
- May reduce risk radial head dislocation
- Variable complications

- Ettl V et al Z Orthop Ihre Grenzgeb. 2005
- Etc
Ulnar Lengthening

RAA = 45°
RAA = 30°
Single bone forearm
Single Bone Forearm

**INDICATIONS**
- Marked radial and ulnar deformity with dislocated radial head
- Limitation forearm rotation with contracture
- Pain
- Limited function

**TECHNIQUE**
- Extra-periosteal excision distal ulna
- Extra-periosteal excision proximal radius
- Intercalary segment bone graft
- Subperiosteal exposure diaphyseal radius and ulna
- Internal fixation: 3 screws
Surgical technique
Results

• Elbow
  ‣ Full flexion/extension arc
  ‣ No instability
  ‣ No recurrent impingement

• Wrist
  ‣ Near full rotation through wrist
  ‣ No pain
  ‣ Full flexion/extension arc
  ‣ Normal strength

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