Clinical and Genetic Characterization of Femoral Duplication in Patients with Tibial Hemimelia

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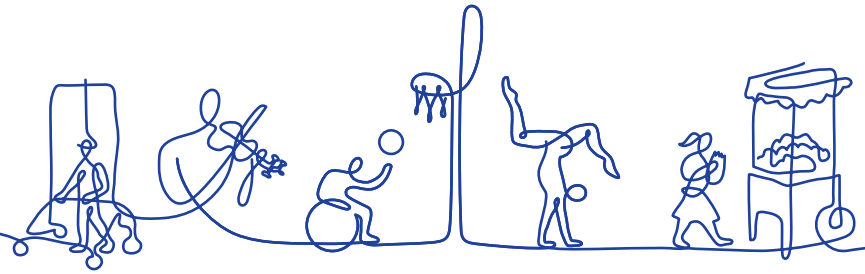
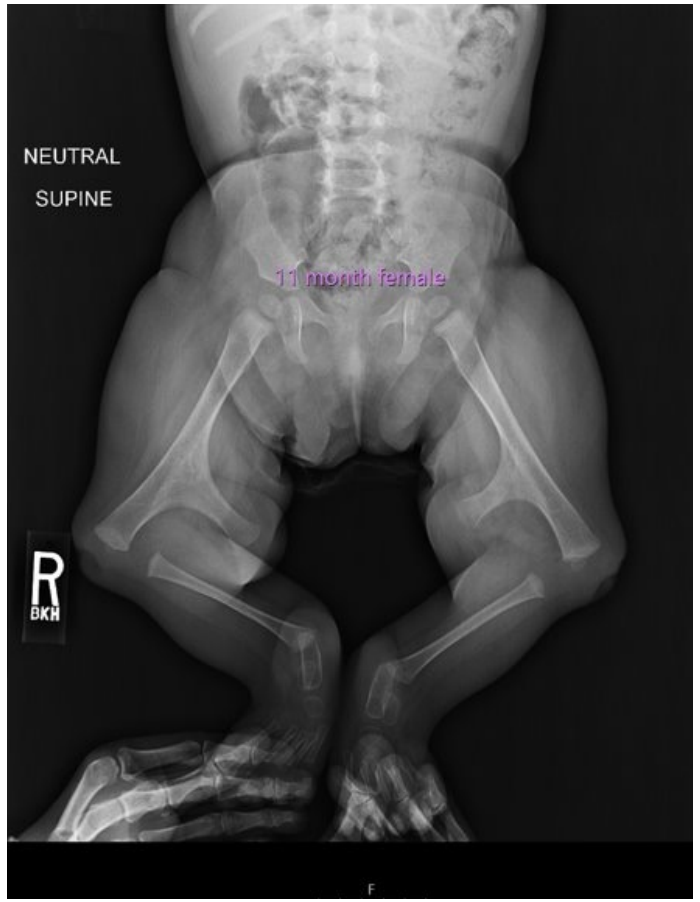


Femoral Duplication in Tibial Hemimelia

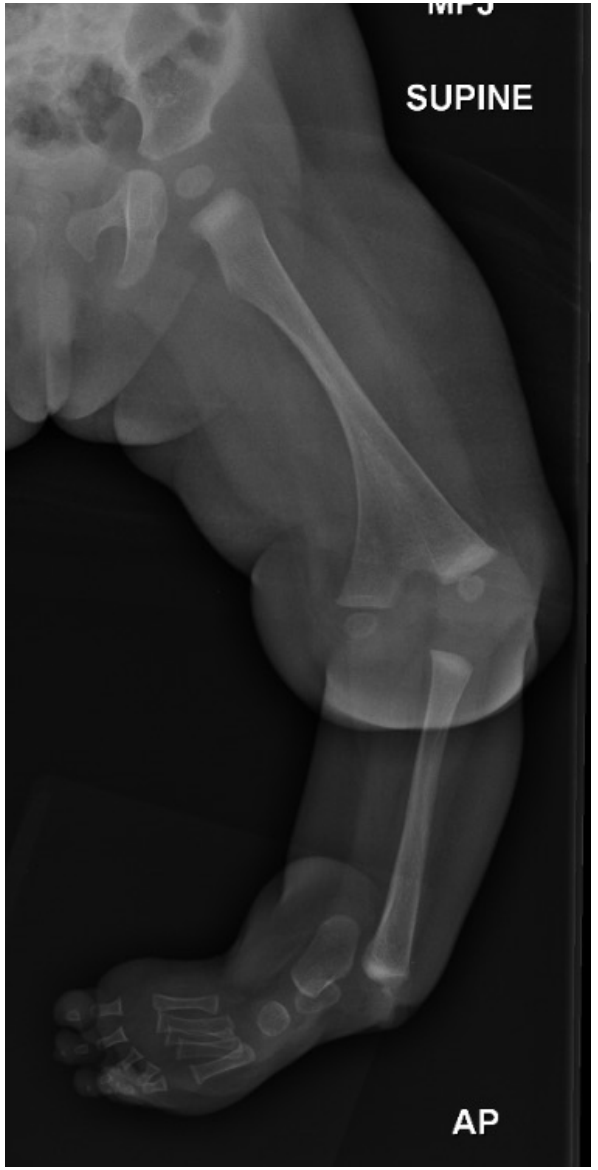
- First described by Erlich in 1885
- When associated with hand and/or foot ectrodactyly known as Gollop-Wolfgang complex
- Various inheritance patterns described (AD, AR, X-linked)
- Literature limited mostly to case reports
- Purpose
  - To define the clinical and genetic presentation of a cohort of femoral duplication patients treated at a single institution.



- No conflicts of interest to disclose

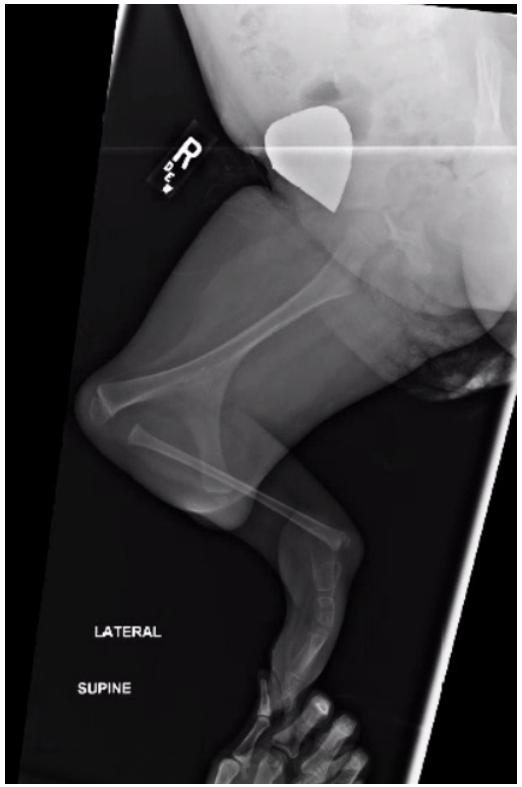
Methods

- Retrospective chart review
- Characterization of femoral duplication anatomy, associated conditions, upper extremity differences, surgical management, complications, and outcomes
- Prospective genetic evaluation of current patients



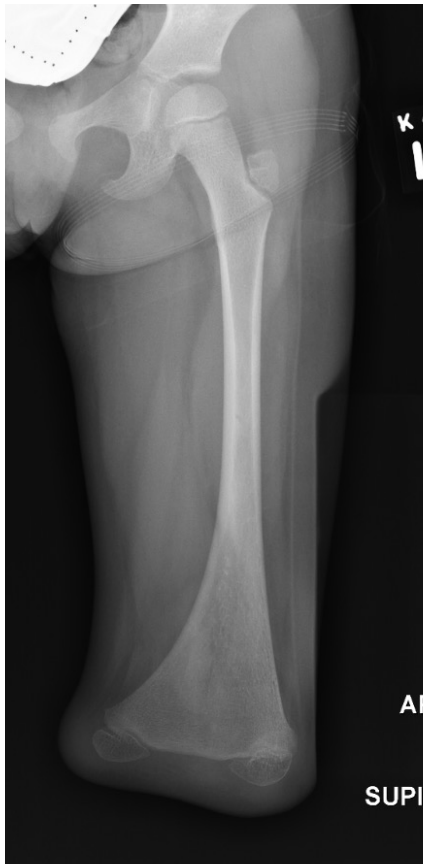
Results – Clinical Presentation

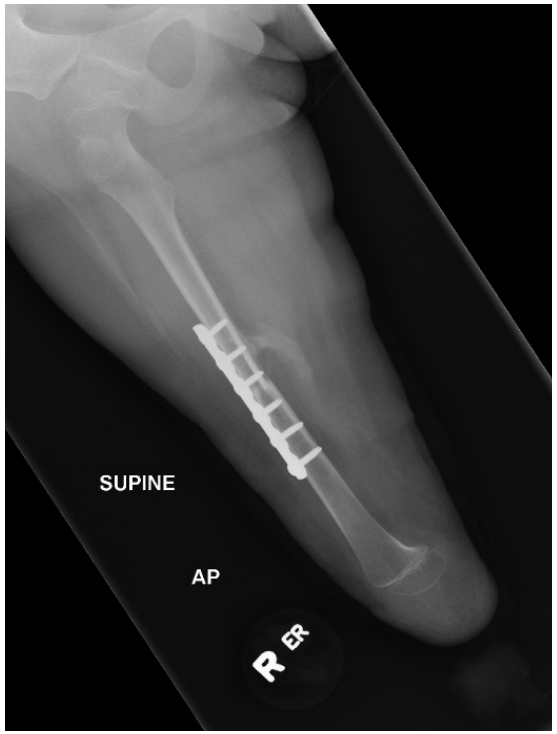
- 11 patients identified with femoral duplication
- Patients were most commonly female (64%) and Black (55%)
- All had Jones type 1 tibial hemimelia
- 4/11 (36%) of patients had documented congenital upper extremity anomalies
- 3/11 (27%) of patients had other nonorthopedic comorbidities
- Femur anatomy
  - In all patients, the accessory femur is medial, while the fibula and foot are attached to the more lateral primary femur
  - Accessory femur can branch off the primary femur at the level of the diaphysis, metaphysis, or epiphysis



Results – Treatment

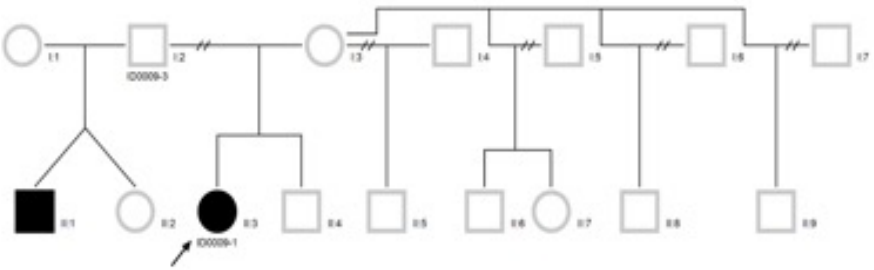
- All patients underwent knee disarticulation amputation
- 7/13 (54%) limbs underwent excision of the duplicated femur
- One patient had corrective osteotomy for angular deformity

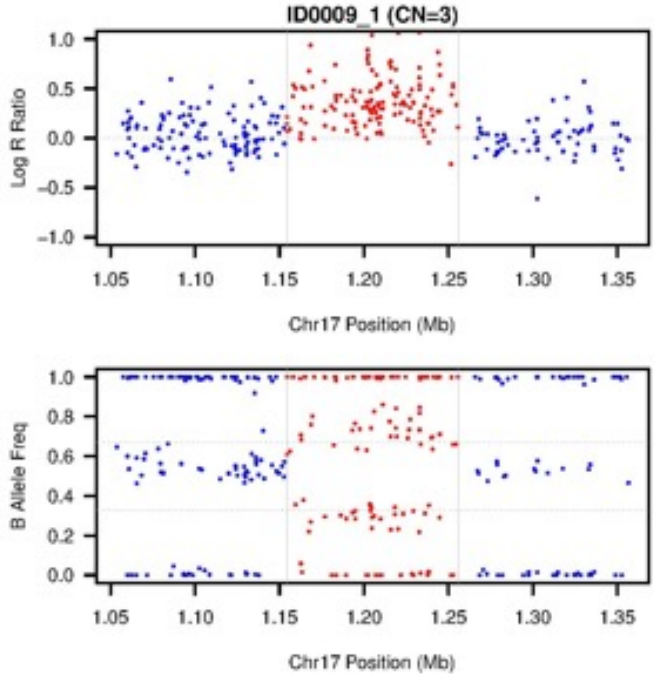




Results – Genetics

- 3/6 (50%) of patients with genetic testing had increased copy number, most likely duplication, of the chr17p13.3 locus encompassing the *BHLHA9* gene
- *BHLHA9* encodes a transcription factor with a well-described role in regulating apoptosis during limb development





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Conclusions

- Clinicians should screen all Jones type I tibial hemimelia patients for presence of duplicated femur
- Patients with duplication of the distal femur often require accessory femur excision
- Some, but not all patients with duplication of the femur have concurrent upper extremity limb deformities (Gollop-Wolfgang Complex)
- The BHLHA9 gene may contribute to development of femoral duplication in a subset of patients

Thank You!

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