A Case Report: Gross Anatomic Dissection and CT Scan of a 94-year old Achondroplastic Dwarf

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INTRODUCTION

- Achondroplasia is the most prevalent form of short-limbed dwarfism with over 200 possible types [1]
- Achondroplasia is an autosomal-dominant disorder found on chromosome 4 [1, 2]
  - The mutation is found in the gene for fibroblastic growth factor receptor-3 (FGFR3)
  - The mutation results in a significant decrease in endochondral ossification, resulting in shorter than normal long bones
- Specific anatomic characteristics are well documented and include:[2,3, 4, 5, 6, 7]
  - Short proximal limbs and trident hand
  - Genu varum: fibular overgrowth vs. LCL laxity
  - Hydrocephalus caused from either aqueduct stenosis or from raised intracranial venous pressure due to stenosis of the jugular foramen
  - Cardiovascular and respiratory complications
  - Neurologic complications caused from small sciatic notches, short inter-pedicle distances, narrow foramen magnum, spinal stenosis
  - Hip and elbow flexion contractions
- Life expectancy is estimated to be 15 years shorter then the average population[1]

METHODS

- A 94 year old female achondroplastic dwarf was evaluated through systematic gross dissection as part of a graduate level gross anatomy course
- Further evaluation included computed tomography (CT) scans
- Medical records were obtained on the subject covering the last 10 years of life
- A review of the literature was conducted to compare the subject to current findings in the literature

CASE HISTORY

- The subject died at the age of 94 years old, weighing 36 kg, measuring 124 cm
- History of coccygeal decubitus ulcer, stage III, measuring 6” x 4”
- Medical history from the past 10 years revealed a history of depression, dementia, congestive heart failure, bilateral cataracts leading to blindness in the right eye, hearing loss, osteoporosis, fracture of the right humerus (Fig. 11), and hydrocephalus
- Subject was bedridden for at least the last 6 years of life

Figure 4: Right radial artery

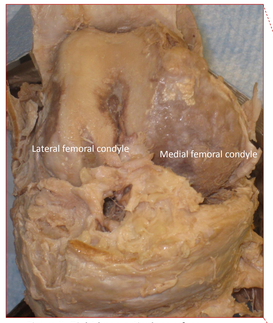

Figure 3: Right knee articular surfaces

Figure 2: Right foot: note the foot size relative to the size of the tibia

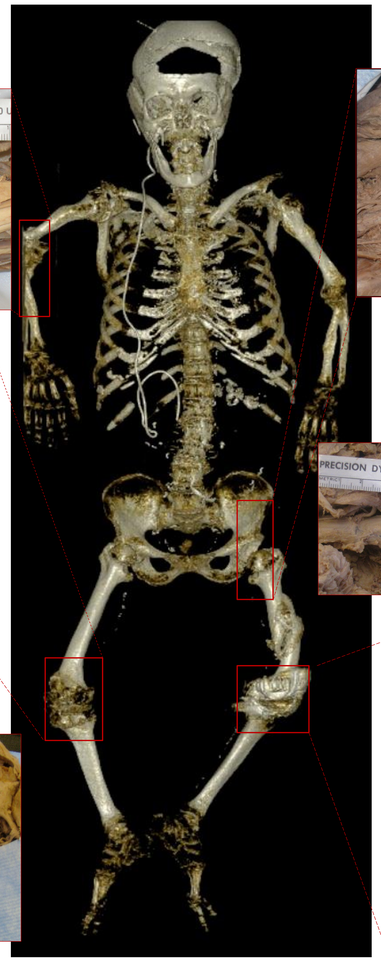

Figure 1: CT scan of subject

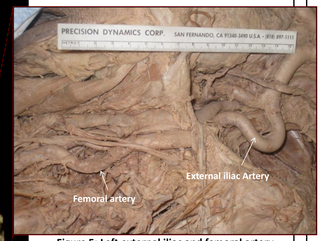

Figure 5: Left external iliac and femoral artery

Figure 6: Right sciatic nerve splitting into the common fibular and tibial nerve

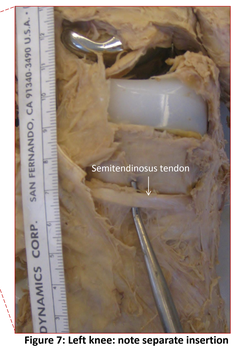

Figure 7: Left knee: note separate insertion of semitendinosus tendon

DISCUSSION

- The age of the subject is rare, as this population typically has a shorter lifespan of the average adult by approximately 15 years (the subject lived 30 years longer than average)[1]
- Genu varum is one of the most common bony malformations in this population and its cause is debated:
  - Many believe fibular overgrowth is caused by a differential growth rate between the tibia and the fibula [5,6,7]
  - An alternative theory is the belief that lateral collateral ligament laxity causes a varus deformity to develop[8]
- Osteoarthritis is relatively common in this population, especially for those who live longer[9]
  - Joint replacement surgery may provide a challenge and often requires a customized implant[10]
  - Surgical results are typically favorable and improve pain and function[10]
- There was no documentation of increased neurovascular size or of atrial malformations in this population

RESULTS

- **General Findings (Fig. 1):**
  - Visual observation revealed a subject with a relatively normal sized torso, short limbs (especially the proximal limb), and bilateral genu varum
  - Significant muscle atrophy noted, especially in the lower extremity
- **Orthopedic Findings:**
  - The CT scan and dissection showed evidence of severe degenerative joint disease, including the non-weight bearing joints (Fig. 3, Fig. 8)
  - A total knee replacement was found in the left knee (Fig. 7)
    - Bone quality around the implant was poor (Fig. 7, Fig. 9)
  - An ACL tear, partial PCL tear, and meniscal tear were also noted in the right knee (Fig. 3)
  - A mal-union, mid-shaft femur fracture with significant superficial deformity was initially discovered by dissection, and further evaluated by CT scan (Fig. 10)

Figure 8: CT right knee joint degeneration

Figure 9: CT left knee tissue overgrowth over knee replacement

Figure 10: CT of left mal-union femur fracture

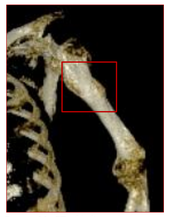

Figure 11: CT of left healed humerus fracture

- **Muscular Findings:**
  - Bilaterally the sartorius muscles were aligned along a vertical axis across the anterior thigh
  - The left semitendinosus tendon inserted separately from the pes anserinus muscles, virtually at the tibial tuberosity (Fig. 7)
- **Neurovascular Findings:**
  - Structures appeared normal in their branching, however, the size of the vessels were visibly large compared to overall size of the subject (Fig. 3, Fig. 5, Fig. 6)
  - Both the upper and lower extremities had tortuous arterial malformations, primarily in the femoral, brachial, and radial arteries (Fig. 3, Fig. 5)
  - Neurologic structures were excessively large, considering the size and degree of atrophy present (Fig. 6)
    - The sciatic n. and common fibular n. were most notable in size
    - The tibial n. split above the medial malleolus bilaterally

CONCLUSION

- There is very little anatomic research on achondroplastic dwarfism in the elderly
- The profound degenerative joint disease, large malformed vessels, and advanced age of the subject make this an interesting case study
- It is important for anatomists and/or future medical professionals to embrace anatomic variation in all populations

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