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.
- Achondroplasia is an autosomal-dominant disorder found on chromosome 4.
- 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:
  - 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 than the average population.

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.

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).
- 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.
  - An alternative theory is the belief that lateral collateral ligament laxity causes a varus deformity to develop.
- Osteoarthritis is relatively common in this population, especially for those who live longer.
- Joint replacement surgery may provide a challenge and often requires a customized implant.
- Surgical results are typically favorable and improve pain and function.
- 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).
- 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 malleous 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.

REFERENCES
- Heath Monat, John Bolte IV.