CASE REPORT

Small is challenging; distal femur fracture management in an elderly lady with achondroplastic dwarfism

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SUMMARY

Achondroplasia is an autosomal dominant dwarfing condition that represents the most common form of skeletal dysplasia. The disease is caused by a mutation in the gene encoding fibroblast growth factor receptor 3 (FGFR3) found at the p16.3 locus on chromosome 4 which results in severe inhibition of subchondral bone growth. Anatomic variations, including articular and periarticular deformities classically seen with this condition, amplify the complexity of fracture fixation. Blood volume loss, age-related bone fragility, component selection and positioning pose serious orthopaedic challenges. Concomitant cardiovascular, respiratory and neurological comorbidities pose additional high-risk perioperative considerations. Despite an estimated prevalence of 1:25 000 in the general population, there is little literature concerning the operative and postoperative treatment challenges faced by orthopaedic surgeons dealing with fracture management on a patient with this condition. We present a case of an intercondylar femoral fracture in an elderly achondroplastic lady successfully treated with percutaneous screw fixation.

BACKGROUND

This elderly achondroplastic patient admitted following trauma represented a highly challenging case. Preoperative workup uncovered significant cardiac comorbidities and required early involvement of anaesthetic colleagues. Meticulous templating of images with available fixation options was required due to the anatomical challenges of her condition. Tailoring surgery to technical and anaesthetic demands was successfully executed. With better medical care individuals with this condition are, like the general public, living longer and accruing more medical comorbidities. The presentation of similar osteoporotic fractures is likely to increase within this cohort. Owing to the numerous challenges—both medical and technical, and the paucity of literature regarding surgery for achondroplasia patients, we would advise a high degree of caution when faced with similar cases.

CASE PRESENTATION

An independent community-ambulating 73-year-old lady with achondroplastic dwarfism presented to accident and emergency department after she tripped at home and injured her right leg. She was unable to bear weight on this leg. Radiographs of the femur showed an intercondylar fracture of the right femur (figure 1).

The patient was a known smoker and hypertensive with dyslipidaemia although with no history of previous fractures, or any other surgery. Her regular medications included atorvastatin 10 mg once daily, doxazosin 2 mg once daily, captopril/hydrochlorothiazide 25 mg/12.5 mg and amlodipine 5 mg once daily. Physical examination revealed a height of 3 feet 4 inches (102 cm) and weight of 50 kg. She had a protuberant head with frontal bossing, megaloccephaly, mandibular enlargement and rhizomelic limb shortening. Neurological and vascular examinations were normal. Cardiovascular examination demonstrated a grade 4 ejection systolic murmur, which was previously undiagnosed. She had short conical-shaped limbs, with pain on movement of her right leg.

INVESTIGATIONS

Subsequent transthoracic echocardiography showed severe aortic stenosis. ECG also revealed a first-degree heart block with a PR interval of 0.21 s. Anaesthetic evaluation was performed; neck extension was deemed normal with a Mallampati grade of 2. Chest radiographs and c-spine images were obtained, as well as CT imaging of her knee to assist preoperative planning (figure 2).

TREATMENT

The surgery was performed under general anaesthesia, with the patient supine on a radiolucent table, using image intensification but no tourniquet. Percutaneous stabilisation was achieved using threaded K-wires, before three 4.5 mm AO cannulated screws were passed medial to lateral reducing the intercondylar fracture gap. Her recovery was uneventful following surgery. The patient was immobilised with an above knee cast, remaining non-weightbearing for the first 3 weeks, 25% weight bearing for the second 3 weeks and fully weightbearing free from cast after 6 weeks.

OUTCOME AND FOLLOW-UP

Interval radiographs were satisfactory (figure 3) and she progressed well with her rehabilitation, until discharge from the service 1 year after presentation.

DISCUSSION

The present case of an intrercondylar fracture of the distal femur (AO/OTA 33B2) in an elderly achondroplastic woman highlights a complex set of challenges, both technical and anaesthetic. Achondroplasia is characterised by defects in

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Figure 1 Anteroposterior radiograph of right knee on admission.

endochondral bone formation leading to retarded cartilage growth, short-limbed stature, flared metaphyses and metaphyseal angulation at the knee joint resulting in a common bowleg deformity.

In adults, percutaneous screw fixation, open reduction internal fixation (with or without locking plates) and retrograde intermedullary nailing have been well described for this fracture pattern. In the case of this achondroplastic patient, we were unable to find a plate (locking or non-locking) to control alignment of the distal articular segment due to the bone characteristics of this patient. Paediatric plates were templated to films, but same difficulties applied, and none were suitable. Percutaneous

Figure 2 Axial cut of CT right knee and three-dimensional reconstruction highlighting displaced intercondylar fracture.

Figure 3 Postoperative radiographs: anteroposterior and lateral right femur.

fixation has previously been described, however the stability of the construct is significantly weaker than plate fixation, especially for patients with poor bone quality. In this case absolute stability of the construct was sacrificed to perioperative considerations and percutaneous cannulated screws inserted under fluoroscopic guidance were selected to stabilise the fracture.

From an anaesthesia perspective, the patient had undiagnosed severe aortic stenosis—a narrowing of the aortic valve. This causes pressures to increase in the left ventricle, affecting blood flow leaving the heart, and results in an inability to obtain adequate systemic perfusion. This posed a significant challenge as it compromised the amount of anaesthetic agent in the systemic circulation. This was overcome through preoperative assessment and optimisation, administration of anaesthesia agents at small incremental doses, meticulous fluid management and monitoring of vitals via central venous and arterial methods to ensure adequate preload and contractility were maintained. In addition, due to the patient's small stature and the fact that a tourniquet could not be used with her leg length, there was significant concern that her mean allowable blood loss ($ABL = EBV \times (H_i - H_f) / HAV$) could not sustain a long procedure. As such, a percutaneous approach was favoured over a longer and more challenging locking plate technique, despite the compromise in stability.

Learning points

- ▶ Achondroplastic patients are living longer and accruing the normal spectrum of age-related medical morbidities.
- ▶ Involve anaesthetists early to optimise patient preoperatively.
- ▶ Plan early and carefully for technical considerations and implants.
- ▶ Perioperative management is challenging given the specific body habitus of achondroplasia patients.

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