ABSTRACT

Gaucher’s disease is a rare inherited lysosomal storage disease due to a genetic deficiency of an enzyme acid-B-glucosidase. Onset and clinical course is very variable but main features are hepatosplenomegaly, anaemia, thrombocytopenia and many bone features including osteopenia, lytic lesions, pathological fractures, chronic bone pain, acute episodes of excruciating bone crisis, bone infarcts, osteonecrosis, septic arthritis and skeletal deformities. It should be considered in the differential diagnosis of patients who present with unexplained organomegaly, easy bruisability and/or bone pain.

CASE REPORT

In February 2008 a 14 year old boy presented to our unit with complaints of a protruding distal left thigh bone, a left thigh wound and inability to walk. These were of a duration of two months.

Bone involvement is perhaps the source of the most variable symptoms attributed to Gaucher’s disease (14). The bone lesions include profound osteopenia, osteosclerosis, lytic lesions, pathological fractures, osteonecrosis, osteomyelitis, acute episodes of excruciating pain (bone crises), chronic bone pain and skeletal deformities which result from bone marrow replacement, compression of the intraosseous vasculature and erosion of osseous tissue (3,10). The skeletal manifestations are probably the most disabling aspect of the disease, with patients experiencing bone pain, some suffering bone crises and upto 20% getting impaired mobility while osteomyelitis and septic arthritis have also been reported (4,5,15). Vertebral compression has been noted to occur later in life while osteomyelitis has been observed to occur in the advanced stages of bone marrow infiltration, culminating in fracture. The impact on the patients quality of life is negative. The orthopaedic surgeon is concerned mainly with the skeletal manifestations of the disease, which usually begin in the femur, but have a greater impact on the patients quality of life than the haematological and visceral aspects (2,17). The purpose of this study is to present one patient that we consider to be a case of Gaucher’s disease with distal femoral metaphyseal infarction.
to contain a tender fluctuant swelling and incision and drainage was undertaken. The pus drained grew *staphylococcus aureus* sensitive to almost all antibiotics and patient was treated with clindamycin, augmentin and metronidazole, in addition to 2 units of compatible fresh frozen plasma, tranexamic acid, novoseven and potent oral and injectable analgesics. Haemoglobin electrophoresis returned a result of AA, while the HIV test was negative.

His family history revealed that he was the 5th born in a family of 5 siblings. All other siblings were alive and well. Specifically, no family member had bleeding tendencies.

Physical examination revealed a young boy, looking small for age and very pale. The left knee was swollen compared to the right, with the left lower limb held in external rotation. There was a wound on the medial aspect of the distal left thigh with the distal femoral bone protruding through the wound. Abdominal examination revealed a hepatosplenomegaly. X-rays done this time showed destroyed distal femoral metaphysis, osteosclerosis, irregular bone cortex and features of osteopenia in the left tibia and fibula.

Patient was worked up and prepared for surgery. Preoperative platelet count was 48.4x10⁹. Under vitamin K, cryoprecipitate, haematinics, antibiotics and whole blood, patient underwent two surgical procedures. First the patient was subjected to bone nibbling and debridement of all necrotic metaphyseal femoral bone. Culture grew *pseudomonas aeruginosa* and *proteus* which were sensitive to most antibiotics. Histopathological examination reported this as showing acute on chronic osteomyelitis. Later on he underwent a transarticular transfixing Kuntscher Nailing and bone grafting. Post operative haemorrhage was encountered and patient received 3 units of whole blood transfusion.

**Figure 1:** Destruction of distal femoral metaphysis. The distal epiphysis is spared.

**Figure 2:** Further femoral destruction with marked tibial and fibula osteopenia

**Figure 3:** Splenic ultrasound scan showing splenomegaly

**Figure 4:** Liver ultrasound showing hepatomegaly and peri-portal fibrosis
disease are universally acknowledged but with wide phenotypic variation. Gaucher’s disease is characterised by wide considerable variability in its clinical signs and symptoms, as well as its severity and course (13). This phenotypic variability is observed, even among siblings with the same genotype (3,10).

The patient presented above definitely fits into one of the clinical variants of Gaucher’s disease. The attendant osteomyelitis and subsequent bone infarction of the distal femoral metaphysis following a superficial abscess fits into the category of little attention given to osteomyelitis as a complication of Gaucher’s disease (12). Increased susceptibility to infection, by Gram positive cocci, *staphylococcus aureus*, was definitely demonstrated as was delayed surgical wound healing after draining of the superficial abscess.

The hepatosplenomegaly, recurrent bleeding tendencies, progressive osteomyelitis and ultimately distal metaphyseal femoral infarction as sequelae to a superficial thigh abscess add weight to a diagnosis of Gaucher’s disease in a patient who also has growth retardation. This is in the clinical picture of Type 1 Gaucher’s disease. Radiologically, the metaphyseal destruction with the attendant epiphyseal sparing in the femur are in keeping with the known pattern in Gaucher’s disease (6). Further weight is lent by the remarkable osteopenia witnessed in the tibia and fibula. Further skeletal changes could be demonstrated but the imaging done for this patient was guided by the symptomatic areas.

The orthopaedic manifestations of this disease are doubt important for the orthopaedic surgeon to recognize and understand (4). They are well exemplified in this case where the bone loss and protrusion through the thigh wound rendered the patient bedridden. With the currently available modalities of treatment available for Gaucher’s disease, Enzyme Replacement Therapy (ERT), which has dramatically reduced the incidence of bone crises (14), it is important that an orthopaedic surgeon be able to make this diagnosis and get the patient to commence specific treatment and thus prevent the patient from developing skeletal complications. These skeletal complications, are considered to be the most disabling and irreversible and once they have set in osteonecrosis, osteosclerosis and vertebral compression cannot be reversed by ERT (10). Initiation of genetic counselling is also in the realm of the orthopaedic surgeon. Later orthopaedic intervention may be called upon to improve the quality of life of the patients by such procedures like Total Hip Replacement in the face of severe skeletal damage. For our patient the expected challenge will be to correct the large limb length discrepancy.

Instances of the diagnosis of Gaucher’s disease not being made, despite later-recognized physical findings of a generalized sytemic disease or the syndrome of the “bone crises” not being appreciated in a known Gaucher’s patient have been documented (12).”
REFERENCES


