Case Communications

Osteonecrosis in a Patient with Gaucher’s Disease Treated with Enzyme Replacement

1Ehud Lebel MD, Deborah Elstein PhD, Daniel Hain MD, Irith Hadas-Haltem MD, Ari Zimran MD and Menachem Itzchaki MD

1Department of Orthopedic Surgery, 2Gaucher Clinic, and Departments of 3Nuclear Medicine and 4Radiology, Shaare Zedek Medical Center, Jerusalem, Israel

Keywords: Gaucher disease, enzyme replacement therapy, osteonecrosis, magnetic resonance imaging, bone mineral densitometry

Gaucher disease, the most prevalent lysosomal storage disorder, is caused by an inherited enzymatic defect with consequent accumulation of undegraded glucocerebroside in monocyte-macrophage cells, known as the ‘Gaucher cells’ [1]. The most debilitating, albeit most variable symptom, is bone involvement: pathologic fractures after slight trauma, destruction of heads of the femur or humerus in the hip or shoulder joints (avascular necrosis), and compression fractures of the spine, as well as “bone crises” of pain, which although self-limiting and diminishing after puberty may require pain relief. It was hoped that the introduction of enzyme replacement therapy for Gaucher disease [2] would eliminate this devastating involvement of bones. We present the case of a patient with an osteolytic lesion of the proximal tibia that developed under enzyme therapy. The use of various imaging techniques for differential diagnosis is discussed.

Patient Description
A 69 year old patient with type I Gaucher disease was admitted to the emergency room with acute right knee pain. Pain was aggravated by weight-bearing and relieved by rest. Gaucher disease was diagnosed at age 47 years, secondary to severe knee pain. Skeletal evaluation revealed old infarcts of the left distal femur and right femoral head. Visceral involvement was minimal with very mild hepatosplenomegaly, low-normal hemoglobin, and normal platelet counts. Enzyme replacement therapy was begun in 1999 with the low dose regimen of 30 u/kg per month.

On physical examination of the right knee, local medial joint-line tenderness was found with vague tenderness of the proximal tibia. No joint effusion was noted and there was no limitation in movement. Varus stress was severely painful, but other stability tests were negative. Laboratory tests revealed: white blood cells 5,400/mm³ (86% polymorphonuclears), hemoglobin 14.0 g/dl, and platelets 145,000/mm³; sedimentation rate was 13 and C-reactive protein 0.2, all other tests were normal. Plain knee radiographs were normal; a computed tomography scan that was ordered to rule out an occult fracture showed a low density region of the medial tibial metaphysis without fracture or collapse of the articular surface. A positron emission tomography scan of bone showed “hot” regions in the medial tibial metaphysis and “cold” region under the tibial eminence. Bone mineral density evaluation revealed only very mild osteoporosis; z-score for the lumbar spine was -0.5 (in the low-normal range for age and gender).

All imaging results supported the diagnosis of osteonecrosis of the proximal tibia rather than infection or a neoplastic process. The patient was treated symptomatically with oral opioids and given

Intractable Hypotension in Septic Shock: Successful Treatment with Vasopressin in an Infant

Leah Leibovitch MD, Ori Efrati MD, Amir Vardi MD, Ilan Matok MSc Pharm, Zohar Barzilay MD and Gideon Paret MD

Department of Pediatric Intensive Care, Sheba Medical Center, Tel Hashomer, Israel
Affiliated to Sackler Faculty of Medicine, Tel Aviv University, Ramat Aviv, Israel

Key words: septic shock, hypotension, catecholamines, vasopressin

Sepsis and septic shock constitute an important cause of morbidity and mortality in critically ill children worldwide. The mortality rate of septic shock remains over 50%. Apart from antibiotic administration, most available therapies are limited to supportive strategies. Early mortality is usually due to refractory hypotension with progressive acidosis, which is unresponsive to fluid resuscitation and catecholamine infusions [1].

In case of no response to fluid resuscitation, inotropes and vasoactive agents are commonly used to increase cardiac output, maintain adequate blood pressure and enhance oxygen delivery to the tissues [1]. Recently, several case reports have described the potential benefits of vaso-