

Abstract

OBJECTIVES:

To determine the prevalence and to characterise Gaucher's disease in terms of socio-demographic data, clinical presentation, and management as seen at Kenyatta National Hospital.

DESIGN:

A retrospective record based study.

SETTING:

Kenyatta National Hospital, a referral and teaching hospital.

MAIN OUTCOME MEASURES:

Prevalence (number of cases seen a year), of Gaucher's disease, sociodemographic data, clinical presentation, mode of diagnosis and treatment modalities of Gauchers disease.

RESULTS:

Nine patients were studied, four males and five females giving a prevalence of 0.9 cases seen a year and a M:F ratio of about 1:1. The most common presentation was splenomegaly in nine (100%) cases and hepatomegaly in seven (78%) patients, neurological and bone symptoms were rare, in one (11%) cases and in two (22%) cases respectively. Diagnosis was mainly on basis of presence of Gaucher cells in bone marrow and splenic aspirate as enzyme assay was unavailable. Management was mainly supportive and enzyme therapy was only available for two (22%) patients. Anaemia was the most common complication with seven (78%) patients and one death occurred due to osteomyelitis. Only four (44%) patients were followed up for a period of four years.

CONCLUSIONS:

Gaucher's disease is a rare condition at the Kenyatta National Hospital (KNH). The presentation of most patients is organomegaly, (hepatosplenomegaly) and best fits the type 1 or non-neuronopathic Gaucher's disease. Neurological manifestations are rare. Management of this condition at the KNH is mainly supportive and enzyme therapy still remains out of reach for most patients.