GAUCHER'S DISEASE TYPE 1A: A CASE REPORT

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SUMMARY

A child with massive hepatosplenomegaly was diagnosed on bone marrow biopsy to have Gaucher's disease. The clinical progress in this form of Gaucher's disease is highlighted and the clinical diagnosis correlated with published criteria.

INTRODUCTION

Gaucher's disease is a group of recessively-inherited inborn errors of glycosphingolipid metabolism, characterised by the deficient activity of lysosymal acid β-glucosidase. Accumulation of glucosyl ceramide occurs, primarily in the reticuloendothelial system, presenting with gross hepatosplenomegaly and varying degrees of neurological and skeletal dysfunction.

CASE REPORT

A one-year-old Chinese male was first seen in April 1983. NKH is the second of two siblings, the elder sibling being a three-year-old girl, who is well.

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The main reason for admission was the presence of a large mass in the left hypochondrium, progressively enlarging over the previous six months. There were no other evidence of ill health, except that NKH was retarded in physical growth, weight and height being at the third percentile of the normal growth chart.

Enquiry revealed that a paternal aunt had expired at four-years of age, with some illness presenting with convulsions; also, the child's parents are first cousins.

On examination, NKH was found to be of small stature and looked emaciated but was active, alert and able to feed well. There was gross hepatosplenomegaly with the liver palpable 8 cm below the right coastal margin and the spleen palpable at the level of the umbilicus. There were no other significant abnormalities and no fundoscopic abnormalities.

On assessment, his motor development was noted to be appropriate for age but language development was thought to be slightly delayed.

Initial investigations revealed the following: Hb 11.2g%; wbc count 11,800/mm^3; platelets adequate in film, no abnormal cells seen. Haemoglobin analysis: HbA_2 2%; HbF 2.0%. TORCH screen for Toxoplasma, Rubella, Cytomegalovirus, Herpes Simplex and Syphilis: no significant antibody titres seen. Blood culture

was negative. Bone marrow aspiration showed presence of large, foamy cells characteristic of Gaucher's disease and this was subsequently confirmed (Fig. 1).

NKH was subsequently seen again in August 1983 at the age of 17 months, and December 1983 at the age of 21 months. He was noted to have regressed on his motor milestone and there had been no further development of language skills. His spleen had increased in size (Fig. 2) and he was anaemia (Hb 8.8g%) and thrombocytopenic. Hypochromia was present. No abnormal cells were seen.

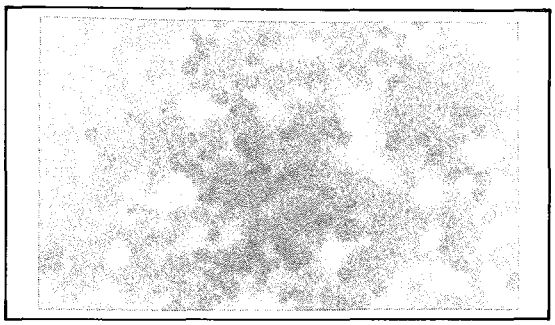

Fig. 1 Bone marrow aspirate of patient NKH. Note presence of large foamy Gaucher's cells at centre of field.

Fig. 2 Patient NKH at 17 months of age. Note massive hepatosplenomegaly.

DISCUSSION

This patient illustrates an interesting subtype of Gaucher's disease. Diagnosis was inferred from clinical presentation and histology of bone marrow. We were unfortunately unable to assay β-glucosidase or even acid phosphatase level.

Three main subtypes of Gaucher's disease have been described and cases of different subtypes within one family have also been reported.[1]

Type 1 disease is further delineated into: malignant childhood; benign adult; moderate severity – variable course variants.

Type II disease is an acutely neuronopathic form that presents as either early onset or late onset variants respectively presenting in infants and toddlers (two to three) years. In Type II disease, severe opisthotonus and bulbar signs like trismus, dysphagia and laryngeal spasm present with generalised spasticity and gross developmental retardation.[2]

Our patient hence illustrates the malignant form of Type 1 Gaucher's disease with massive splenomegaly, stunted growth, retarded social and intellectual development but no objective central nervous system involvement. His retarded motor development is probably a function of his severe abdominal distension.

In view of the gross splenomegaly with hypersplenism, a splenectomy was planned for this child. We have considered all the problems associated with splenectomy at this age, in particular fluminating septicaemia but we have no practical alternative. Partial splenectomy for Gaucher's spleen or partial splenic embolization[3] is still experimental and not feasible in our centre.

The patient however defaulted and has been lost to follow-up.

ACKNOWLEDGEMENTS

I am indebted to Dr K.L. Yong, formerly of the Pathology Department for confirmation of the histological diagnosis and Dr Abdul Halim of the Paediatric Department for help with the developmental assessment. Thanks also to Medical Illustrations UKM, for excellent photographic work.

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