Picture Story

A case of chronic neuropathic Gaucher disease

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Sri Lanka Journal of Child Health, 2010; **39**:31

(Key words: myoclonic seizures, organomegaly, Gaucher disease)

A three year old only girl of non-consanguineous parents, presented with progressive myoclonic jerks confined to the neck and upper limbs for the past three months. Her birth was uneventful with a birth weight of 3.3kg and she was apparently well up to eighteen months of age.

On examination, her height and weight were below the 3rd centile and occipito-frontal circumference was at the 5th centile without any coarse facies. Cranial nerves, cerebellar function and fundi were normal with appropriate higher functions. All four limbs were spastic and power grade IV with exaggerated reflexes and positive Babinski. Her abdomen was grossly distended with a 3cm firm liver and a 20cm massive spleen. Respiratory and cardiovascular systems were normal.

Full blood count showed a pancytopenia. Blood picture did not reveal any abnormal cells. Liver function, renal function and serum calcium were normal. Serum ferritin was elevated (197ng/ml). EEG showed chronic encephalopathy and abdominal scan revealed massive splenomegaly and hepatomegaly. A provisional diagnosis of Gaucher disease was made by demonstrating typical Gaucher cells in the bone marrow (Figure 1). Gaucher cells are 20-100µm in diameter and have a characteristic wrinkled paper appearance resulting from the presence of intracytoplasmic substrate inclusions[1]. Gaucher cells may also be found in patients with granulocytic leukaemia and myeloma[1]. Definitive diagnosis requires the determination of the acid β-glucosidase activity in isolated leucocytes or cultured fibroblasts[1]. This was not done.

References:

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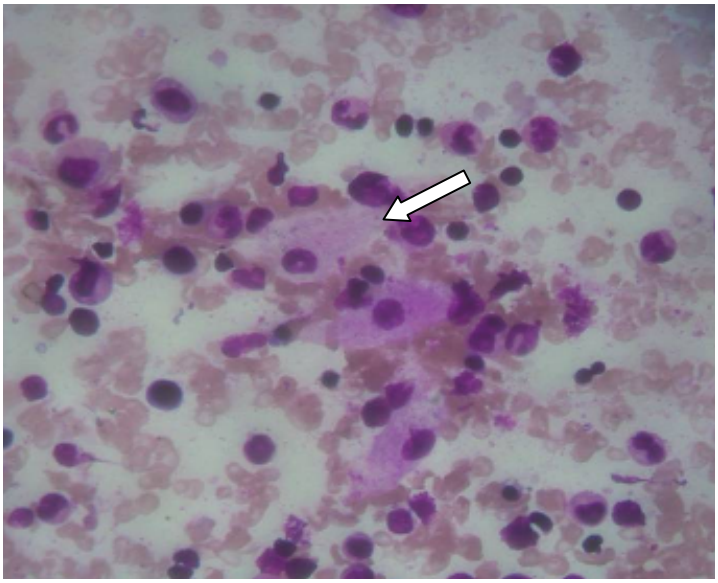

Figure 1 *Bone marrow showing typical Gaucher cells*

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(Received on 11 September 2009. Accepted after revision on 23 October 2009)