



Case Report

An Image for Differential Diagnosis

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Abstract

Male infant 16 month old of consanguineous marriage presented with macrocephaly (Head circumference > 97 % on growth charts) with delayed motor (stand with support) and mental development (No words, delayed recognition of his mother....). No family history of large head. No organomegaly or lymphadenopathy. Clinical examination and investigations excluded rickets as a cause for his macrocephaly. MRI brain revealed mega-encephalic leukoencephalopathy with subcortical cysts in which (diffuse hypo-density seen involving the white matter of both cerebral hemispheres including the deep, subcortical and periventricular matter with no significant mass effect or midline shift). What are your differential diagnoses? How can reach the final diagnosis?

Key words: Macrocephaly, Motor, Mental, Metabolic, Glycine

Van der Knaap disease

Megalencephalic leukoencephalopathy with subcortical cysts (MLC), or Van der Knaap disease, is a rare autosomal recessive disorder. It is characterised by macrocephaly that either presents at birth or develops during infancy. It occurs more commonly in some ethnicities where consanguinity is common