Case Report

Diagnostic Difficulties in Sanfilippo Disease

DKH Chan, CW Lam, STS Lam

Abstract

We reported on a 21 months old girl, diagnosed as Sanfilippo disease, MPSIIIB. Her mother was at 21 weeks of pregnancy when she first presented to us. The diagnostic pitfalls in the clinical, biochemical and genetic aspects were highlighted. The limited time frame for urgent molecular diagnosis, prenatal diagnostic tests and counselling issues were discussed.

Keyword: Alpha-N-acetylglucosaminidase; Diagnostic pitfalls; MPSIIIB; Sanfilippo disease type B