PROPIONIC ACIDEMIA – A CASE REPORT

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Propionic acidemia is a rare disease which has not been reported in Chinese literatures yet. This article presents a two-year-and-eight-month old girl who was admitted to our hospital 5 times in the past year because of the same problems of recurrent vomiting, difficult feeding, lethargy, and hypotonia. The biochemical data revealed metabolic ketoacidosis, hyperglycinemia, and hyperammonemia, during the acute episodes. Urine gas chromatography disclosed propionate and its metabolites in a significant amount.

Then, this patient was treated intensively by eliminating dietary protein, administering sodium bicarbonate parenterally, and correcting serum electrolytes. After acute episodes, restriction of protein intake (1.0 gm/kg/day) and supplement with vitamins was advised.

Because propionic acidemia is an autosomal recessive inheritance, prenatal genetic counseling is required for her mother. Now her mother gets pregnant again. Aspiration of amniotic fluid was done during the 16th week of gestation. Methylcitrate in the amniotic fluid and propionyl CoA carboxylase activity in cultured amniotic fluid cells were reported to be normal. Her mother still under the care of our perinatal clinics. (Full text in Chinese)

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