A Two-Year-Old Infant with a Myopathic Form of Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency

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A two-year-three-month old girl was hospitalized for detailed examination following repeated hyper-creatine kinasesmia and cervical muscle cramps induced by pyrexia and persistent hypertonicity of the cervical muscles. Physical examination showed mild hypotonia but no muscle weakness. Induction of symptoms by continuous cervical muscular exercise and the appearance of dicarboxylic aciduria during the fasting test indicated a disorder of fatty acid oxidation. Free fatty acid and acyl carnitine analyses using dried blood spots, and acyl-CoA dehydrogenase activity assays using cultured skin fibroblasts established a diagnosis of very-long-chain acyl-CoA dehydrogenase (VLCAD) deficiency. Currently VLCAD deficiency has been divided into three phenotypes; a severe childhood form, a milder childhood form, and an adult form. However, we suggest that the severe and milder childhood forms would be better described as a systemic form, and the adult form and our infant case as a myopathic form. An early onset of the myopathic form within the first year of life, as well as its diagnosis in early infancy, has never been described in the literature.

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＝ 訂正 ＝

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図1 頭部MRI（T1WI TR/TE=600/12）
(誤) 橫延軸断面(a)では、前頭葉と側頭葉がわずかに残存する。
横延軸移行部(b)では、脳幹部は保たれている。
(正) 橫延軸移行部(a)では、脳幹部は保たれている。
側脳室体部断面(b)では、前頭葉と側頭葉がわずかに残存する。

岩崎俊之

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