一個台灣法布瑞氏症家族的臨床表現與酵素療法一年經驗

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Clinical Features and One-year Experience with Enzyme Replacement Therapy in a Taiwanese Kindred with Fabry Disease

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Fabry disease is an X-linked recessive inheritance with defective activity of lysosomal enzyme, α-galactosidase A. Clinical diagnosis is sometimes difficult because of diverse manifestations. Confirmation of the disease is based on enzyme levels as well as on molecular biology. The prognosis is related to renal, cardiovascular and neurological complications. We report a family with Fabry disease, mother and her two sons (aged 25 and 22 years old, respectively). Both sons had numerous angiokeratomas on the lower trunk and thighs. In addition to cornea verticillata and acroparesthesia, the older one had left ventricular hypertrophy with severe mitral valve regurgitation, and the younger one had coarse liver surface on abdominal sonography, ST change on V2-4 leads and hypohidrosis. Furthermore, decreased α-galactosidase A activities (0.9 and 1.0 nmol/hr/ml, respectively, normal range: 7.6-16.5) were found. The mother, a Fabry carrier, had no abnormal findings except mild acroparesthesia, cornea verticillata and subnormal enzyme activity (2.6 nmol/hr/ml). A deletion mutation (c.1072_1074delGAG) in exon 7 of α-Gal A gene on Xq22 was detected. Both siblings are undergoing enzyme replacement therapy.

Compared with pretreatment, after one-year enzyme replacement therapy, the glomerular filtration rate (GFR) of the proband improved slightly (70.6 vs. 82.8 ml/min). There was also no tendency of progression in the younger brother after enzyme supplement. (Dermatol Sinica 22 : 159-165, 2004)

Key words: Fabry disease, Angiokeratoma, α-Galactosidase A, Mutation, Enzyme replacement therapy

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