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8. DETERMINATION OF PLASMA α-GALACTOSIDASE AND ITS APPLICATION TO DIAGNOSIS OF FABRY'S DISEASE IN CHINESE.

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Fabry's disease is an X linked inborn error of glycosphingolipid metabolism resulting from the deficient activity of the ceramide trihexosidase, a specific α-galactosidase. The plasma α-galactosidase activity was determined at 37°C and pH 4.6 by using 4-methylumbelliferyl-α-D-galactopyranoide as substrate. The reaction was stopped after 90 min incubation by the addition of 0.1M ethylenediamine (pH= 11.4). Fluorescence (ex.365nm; em.450) was read on an automatic fluorometer (Auto FP-1). The within-run C.V. of the test was 1.5-2.7%. The plasma α-galactosidase reference range was estimated to be 9.7-20.3nmol/hr.ml in 197 healthy Chinese adults. The pH optimal was 4.6 and Km value was 8.0mM for normal control. A case (female, 35 years old) of cornea vortex epitheliopathy with family history of cardiac disease was diagnosed as heterozygote of Fabry's disease by this enzymatic assay. Her plasma α-galactosidase activity was 5.14 nmol/hr.ml, which was about 40% of the mean of the normal control. The result indicated that this assay of plasma α-galactosidase could be used for diagnosis of Fabry's disease and its heterozygotes in Chinese. This assay system could be further extented to determinate α-galactosidase activity in fibroblasts and amniotic fluid cells for genetic consulting and prenatal diagnosis.

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