Tyrosine Hydroxylase (TH) Deficiency: Hong Kong Experience

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Tyrosine hydroxylase (TH) deficiency is a rare autosomal recessive monoamine neurotransmitter disorder. Tyrosine hydroxylase converts L-tyrosine to L-dihydroxyphenyalanine (L-dopa), which is essential for biosynthesis of catecholamines - dopamine, norepinephrine and epinephrine. The phenotype of TH deficiency ranges from 1) mild form with dopa-responsive dystonia, 2) moderate to severe form presenting as infantile parkinsonism with motor delay to 3) very severe form of progressive infantile encephalopathy. Only a few dozen cases have been reported in medical literature by 2003. Since our first case of TH deficiency in a Chinese family was diagnosed genetically in 2004, we have had more than 16 cases in Hong Kong over the past 13 years. Their clinical manifestations, biochemical findings, genetic mutations and treatment response will be discussed.