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# Long-term outcome of 30 patients with 6-pyruvoyl-tetrahydropterin synthase deficiency

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**Objective** To evaluate the long-term outcome of patients with 6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency. **Methods** Patients were identified as tetrahydrobiopterin (BH<sub>4</sub>) deficiency based on the results of urinary pterin analysis, detection of dihydropteridine reductase (DHPR) activity in blood from a total of 550 patients with hyperphenylalaninemia (HPA), and then BH<sub>4</sub> loading tests in suspected patients with abnormal pterin profiles at our outpatient clinic since 1992. BH<sub>4</sub> deficient Patients were treated with BH<sub>4</sub>, levodopa and 5-hydroxytryptophan (5-HTP). Development and intelligence quotient (DQ/IQ) and magnetic resonance imaging (MRI) of the brain were followed up. **Results** A total of 30 cases were diagnosed as BH<sub>4</sub> deficiency, all of them were revealed as PTPS deficiency. They were diagnosed at the age of 2.5~27 months and the follow-up duration was 36~118 months. The average full-scale DQ/IQ at diagnosis and after treatment of at least 3 years were 53 ± 16, 78 ± 15 respectively. The improvement of abnormalities in the white matter was also seen on the MRI of the brain after treatment. A significant negative correlation was observed between the level of the DQ/IQ and the age of treatment commenced ( $r = -0.751$ ,  $P < 0.001$ ). **Conclusions** Long-term follow up demonstrates that outcome of patients with PTPS deficiency benefits from treatment as early as possible with three drugs combined.

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