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ATYPICAL PHENYLKETONURIA WITH MILD MENTAL RETARDATION CAUSED BY TETRAHYDROBIOPTERIN DEFICIENCY IN A CHINESE FAMILY.

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Most atypical phenylketonuria (PKU) due to defects in biopterin metabolism have been reported to be very severe and have been described as "Malignant hyperphenylalaninemia". Two boys (age: 4.7 and 6.7 years) from a Chinese family presenting with mild mental retardation (IQ: 53 & 65) were recently found to be hyperphenylalaninemia by blood spot phenylalanine (Phe) screening test (10.8 & 9.0 mg/dl). The parents have no consanguinity and their third boy is normal. PKU was confirmed in these two boys by serum amino acid analysis (Phe: 9.6 & 6.9 mg/dl, Tyrosine: 1.1 & 1.3 mg/dl). Although the results of urinary $FeCl_3$ tests were negative or weak positive, phenylpyruvate and other diagnostic metabolites were demonstrated in their urine by gas chromatography. The urinary pterins were determined by high performance liquid chromatography and the results showed that they had high neopterin (N) (5340 - 18902 umol/mol creatinine), low biopterin (B) (136 - 252 umol/mol creatinine) and low B/(B+N) ratio (1.3 - 2.5%). With oral loading of Phe (2200 mg/day, 140 mg/kg) for three days, their blood Phe was increased to 13 - 18 mg/dl and acute clinical symptoms (e.g. drowsiness) appeared simultaneously. But their blood Phe decreased drastically to normal (<2 mg/dl) and the clinical symptoms disappeared after oral intake of tetrahydrobiopterin (7.3 - 7.6 mg/kg). These data indicate that they are cases of atypical PKU due to defective synthesis of biopterin and the inheritance of the disease is autosomal recessive. They had a relatively high tolerance of serum Phe in response to Phe intake, but a low clinical tolerance of serum Phe level. This suggested that the boys may have a partial deficient form of defective synthesis of the cofactor.