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Section for Metabolic Diseases

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Program & Abstracts

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BRAINS FOR BRAIN

LATE DIAGNOSIS OF PHENYLKETONURIA - TIME FOR CHANGES

Natalija Angelkova[1], Vesna Sabolić[1], Elena Sukarova Angelovska[2], Tanja Zorčec[3], Elena Kochova[1], Filip Duma[1]

[1]Neurology department; [2]Department of Endocrinology and Genetics; [3]Psihophysiology; University Children's Hospital Skopje, Skopje, Macedonia

Hyperphenylalaninemia has been detected in four children in first 20 months of age during evaluation of psychomotor delay. The standard amino acid analysis done by liquid chromatography detected higher levels of PHA from just above the upper limit 460 μmol/L till 1200 μmol/L considering 120-360 μmol/L as normal ranges.

Three boys and a girl had been followed because of delayed fine motor skills, lack of attention, poor visual contact. There were episodes of anger and excitation including selfmutlation .There was no initiation of speech at all four of them. The severe deficits were in cognitive function and communication skills.

EEG recorded epileptiform activity in one case where tonic seizures occurred. The other 3 EEG findings were in normal ranges.

Areas of delayed myelination were detected on MRI as well as cortical thickness and atrophy in frontal and temporal regions

Phenylalanine free diet started, with difficulties to maintain PHA close to normal levels. The early outcome considered improvement in growth and nutrition. In 2 cases with good compliance there is improvement of communication but with poor speech which started at age 4 and 5. Hyperactivity was reduced, they started special education. Two severe cases, with poor diet are highly disabled, hyperactive and aggressive, they need permanent nursing and care.

Neonatal screening should obtain early diagnosis and treatment which is prerequisite of better developmental outcome and quality of life.

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