Clinico-Radiological profile of children with phenylketonuria - A tertiary care experience

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Phenylketonuria (PKU)
- Autosomal recessive IEM
- Deficiency of enzyme phenylalanine hydroxylase
- Deposition of amino acid phenylalanine in the body
- Irreversible brain damage and marked intellectual disability

Results
- Examination: microcephaly (n=2/5), pallor (n=4/5), hypopigmented hair (n=5/5), central hypotonia (n=5/5), Babinski’s sign (n=5/5)
- Biochemical: Elevated phenylalanine levels and phenylalanine-tyrosine ratios
- Phenylketonuria: 60% (n=3)
- Tetrahydrobiopterin deficiency: 40% (n=2)
- MRI: Demyelination, and non-specific white matter changes
- Clinical exome sequencing : Genetically confirmed cases (n=2)

Methods
- Objective: To describe clinical and radiological profile of children with PKU
- Study type: Retrospective review and prospective follow-up
- Study population: Consecutive children with elevated phenylalanine levels on mass spectroscopy
- Study setting: Pediatric Neurology Clinic of a tertiary care hospital
- Detailed evaluation, neurological examination and magnetic resonance imaging (MRI)

Results
- N=5 cases enrolled
- Males 60% (n=3/5)
- Mean age at presentation: 29 months (range 9-48 months)
- Clinical features
  - Excessive irritability 20% (n=1/5)
  - Feed intolerance 20% (n=1/5)
  - Abnormal body movements 60% (n=3/5)
  - Global developmental delay 100% (n=5/5)

Axial T2w image (a) shows presence of hyperintense signal (black arrows) in periventricular area. Axial T1w image (b) shows evidence of diffuse cerebral atrophy, hypomyelination and atrophic corpus callosum (thick white arrow). Axial DW images show restricted diffusion (thin white arrows) in bilateral parahippocampal (c) and periventricular regions (d)

Conclusion
- Absence of newborn screening in developing countries
- Imperative to diagnose PKU and hyperphenylalaninemia using biochemical screening followed by genetic confirmation
- Early diagnosis can improve the neurological outcomes

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