12. Clinical Spectrum of Glycine Encephalopathy in Indian Children

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Objective: Our aim was to determine clinical spectrum and outcome of NKH in Indian Children.

Material and Method: We identified 8 children with NKH in last 3 years. In all these 8 patients the Organic acidemia was ruled out by GC-MS of urine or MS-MS of blood or both. EEG was available in 7 children. CT Scan or MRI was available in 6 patients. Diagnosis of NKH was based on the ratio of CSF / Plasma Glycine > 0.08 with absence of Organic acidemia.

Results: Out of 8 children with NKH, 5 had neonatal onset of which 2 expired, one developed severe mental retardation, and two recovered completely (probably transient variety). 3 Children had late onset disease (after 3 months), of these none expired and all 3 had mental retardation.

Conclusion: NKH is fairly common in India and outcome is unsatisfactory. Only 2 babies with NKH showed good clinical improvement (probably transient). We found that Lactate > 4.5 mMol / L and presence of hiccups in a critically ill child with NKH are bad prognostic signs.