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Unveiling the Intricacies of Neonatal Brain Injuries: A Case Study in Early Detection and Management

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CASE PRESENTATION

A premature newborn aged 33 weeks and 5 days, from an unsupervised pregnancy, experienced spontaneous labor. Despite initial signs of respiratory difficulty within the first hours of life, the infant adapted well to extrauterine life and received non-invasive ventilation for 12 hours, showing positive progress. The initial clinical examination was normal for gestational age. On day 1 of life, a cranial ultrasound (CUS) revealed evidence of agenesis of the septum pellucidum. Subsequent brain MRI unveiled an extensive anomaly of cortical development, characterized as polymicrogyric. An EEG performed on day 16 was indicative of several electroclinical episodes, characterized by extension of the left upper limb and generalized attenuation of cerebral electrical activity. Treatment with phenobarbital successfully resolved electroclinical crises. The infant underwent a normal ophthalmological evaluation, exhibited normal patterns of auditory evoked potentials in the brain stem, tested negative for urinary cytomegalovirus, and had a normal endocrine-metabolic screening. Discharged from the hospital on day 23, the infant underwent multidisciplinary follow-up (neonatology, neuropediatrics, hereditary metabolic disorders, physical medicine, and rehabilitation). Changes in the initial metabolic study revealed elevated ammonia levels (124 µmol/L, normal range 19-54 µmol/L) and a fatty acid profile showing slight amounts of succinic acid and alpha-ketoglutaric acid, which normalized in a subsequent study. The patient is currently under investigation, undergoing multidisciplinary monitoring and ongoing Whole Exome Sequencing, with evaluation of mitochondrial DNA, due to the suspicion of a neuronal migration disorders. At 31 months old, the infant exhibits weight status evolution below the 3rd percentile, along with adequate neurological examination and psychomotor development, and is receiving treatment with levetiracetam.

BACKGROUND

CUS plays a crucial role in the daily routine of a neonatal care unit, allowing for the early identification of brain injuries and facilitating appropriate clinical planning[1,2]. Malformations of cortical development can manifest as expressions of hereditary metabolic diseases, which can be challenging and time-consuming to diagnose due to their pathophysiological complexity[3]. This case underscores the significance of CUS in the early detection of brain injuries, particularly in newborns without prenatal surveillance[2]. It also emphasizes the role of EEG in evaluating these lesions in newborns, given the limited symptoms observed in this age group. This case serves as a testament to the significant impact of early detection and treatment on promoting positive developmental trajectories and enhancing the quality of life for vulnerable newborns, even if the underlying etiology has not yet been identified.

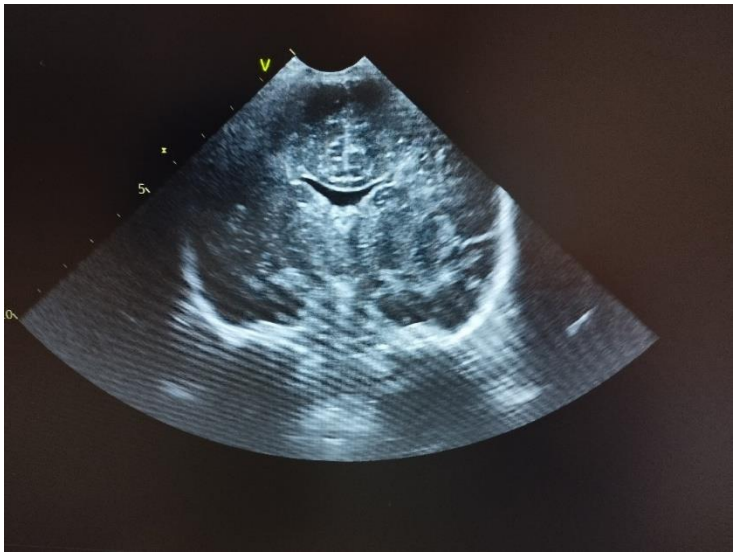

Figure 1: Cranial ultrasound revealing agenesis of the septum pellucidum

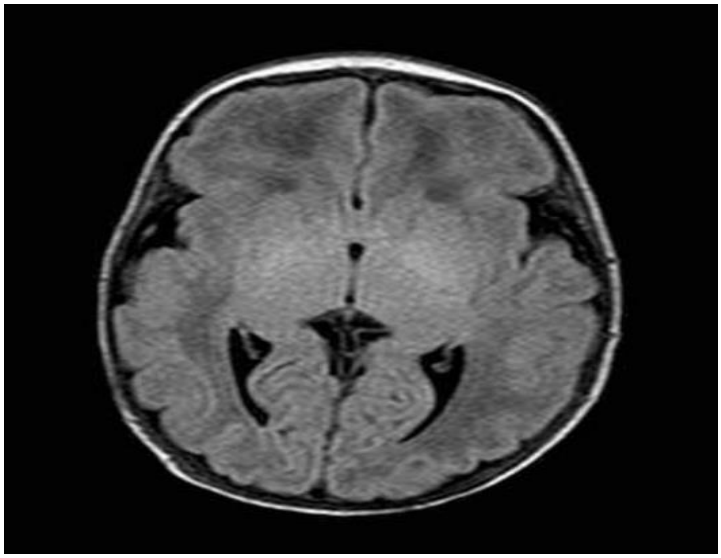

Figure 2: Brain MRI showing extensive anomaly of cortical development

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