Familial Infantile Globoid Cell Leukodystrophy (Krabbe’s Disease)

Krabbe’s disease is a neurodegenerative disease characterized by severe destruction of myelin and presence of globoid bodies in white matter. The biochemical defect is marked by deficiency of enzyme galacto-cerebroside beta-galactosidase resulting in accumulation of galactocerebrosides. The mode of inheritance is autosomal recessive. We report a case of Krabbe’s disease in an infant.

A two and half moths old female child born to consanguineous parents was referred to us with generalized convulsion with occasional myoclonic episodes and unexplained fever since sixty days of life. The antenatal and birth history was uneventful and birth weight was 2.5 kg. There was no abnormal odor in urine and breath. The child had no neurocutaneous marker nor there was any gross facial and skeletal dysmorphism. There was history of deaths of two siblings at the age of around 3 months. Previous two infants were apparently symptom free till sixty days of life then they developed seizures which were refractory to anticonvulsants. Both of them expired around 90 days of age due to seizure.

Her weight and head circumference was 4 kg and 39 cm respectively. No obvious cranial nerve palsy was detected clinically except for visual impairment. Skull and fontanels were normal. Moro’s reflex had disappeared and there was weak palmar and plantar grasp. Tone and tendon jerks was slightly increased and plantar response was bilaterally extensor but no restriction of limb movements was present. The child had not developed social smile and head control was poor. Fundoscopic examination revealed both discs were pale. No organomegaly was present on examination of abdomen. Other systemic examination was normal.

Investigations revealed that the complete blood count, blood culture, urine culture, CSF culture were within normal limit. Arterial pH and blood ammonia level was normal. There was no evidence of aminoaciduria. Blood sugar and calcium was normal. There was no abnormality in chest X-ray. CSF protein was 125 mg/dL. TORCH screening was non-contributory. EEG finding revealed bilateral sharp and slow wave discharge. CT scan examination of brain of the living 3rd issue done at 20 days of life revealed bilateral symmetrical hypodensities noted in both frontoparietal and occipital regions in the white matter zone. Both thalamus and basal ganglia appeared hyperdense on NECT scan. Cerebellum was unaffected. Ventricles were normal, septum was in midline. Repeat NECT Scan examination done at 85 days of life revealed more prominent features of demyelination in above mentioned areas of brain with atrophic changes in cerebrum. Interestingly similar findings were also present in CT scan of the previous female child. The above features are consistent with the diagnosis of Globoid cell leukodystrophy or Krabbes Diseases. Axial T2 weighted MR Scan revealed periventricular white matter demyelination with hyperdense basal ganglia and thalami. However, enzyme assays for confirmation of the diagnosis could not be done.

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