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Case report of malignant infantile osteopetrosis presenting with persistant infantile hypocalcemia

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Malignant infantile osteopetrosis is a rare autosomal recessive disorder with a incidence of 1 in 2,50,000 children, presenting in infancy. It is a bone dysplasia characterised by reduced resorption of bone and diffuse symmetrical sclerosis due to impaired function of osteoclasts. This leads to obliteration of marrow cavity by bony overgrowth resulting in inability of bone marrow to participate in hematopoiesis. It presents with pancytopenia resulting in abnormal bleeding, easy bruising, progressive anemia and failure to thrive. Hepato splenomegaly due to extra medullary erythropoiesis, delayed dentition, cranial nerve palsies (deafness, blindness), hydrocephalus and seizures due to hypocalcemia. We describe the case of a 4 day old male neonate a third born to a 2nd degree consanguinously married couple, who presented with fever and two episodes of convulsions from day 4 of life vitals were stable with mild hypotonia on examination. Suspecting neuro infection due to late onset sepsis, a lumbar puncture was done which showed features of pyogenic meningitis. Routine investigations revealed hypocalcemia and calcium correction was given. Antibiotics, anticonvulsants were started and calcium maintenance dose was given. Child had two further convulsions in NICU. Repeat Lumbar puncture was normal and MRI brain was normal. Investigations continued to show persistent hypocalcemia despite adequate maintenance dose of calcium. Other workup for persistent hypocalcemia were normal except for chest X-ray which showed sclerotic changes. X-ray of other bones also showed diffuse sclerosis, bone in bone appearance. Later DNA analysis was sent which showed mutation of TCIRG1(+), homozygous with autosomal recessive inheritance specific for malignant infantile osteopetrosis. Osteopetrosis remains as a rare cause by clinicians unrecognized for neonatal hypocalcemia, which often results in diagnostic confusion and delay. This is important as early intervention with curative hemopoietic stem cell transplantation before optic nerve compression can help preserve the eyesight and improve the survival of the infant.

Biography

Vivetha Elango has completed MBBS in the year 2016. She is pursuing her postgraduate in MD pediatrics at Kempegowda Institute of Medical Sciences Bangalore. This is currently the first paper that has been done.

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