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Congenital cytomegalovirus infection presenting as cerebral palsy

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Main Text

A 9 month old boy presented with history of delayed attainment of milestones. He was born to a primigravida mother, with an uneventful perinatal period, but had a low birth weight (2.1 kg) for gestational age. There was no history of seizures, abnormal movements, loss of previously gained milestones or prior sibling deaths.

On examination, he had severe microcephaly, failure to thrive and hepatosplenomegaly. Neurological examination revealed severe axial hypotonia and spastic quadriplegia with brisk deep tendon reflexes and intermittent scissoring of lower limbs. Fundus examination and hearing evaluation were normal. His current developmental age was 4 months and developmental quotient was 30.

Investigations revealed anemia, thrombocytopenia with elevated transaminase. Neuroimaging revealed hyperintensities in periventricular white matter on T2/FLAIR weighted images (Fig 1). Workup for toxoplasmosis and human immunodeficiency virus was negative. Real-time polymerase chain reaction using Cobas Taqman 48, showed a cytomegalovirus load of 35900 copies/ml. A diagnosis of congenital CMV infection was made and he was started on oral valganciclovir.

Congenital cytomegalovirus infection is the commonest intrauterine infection, with a global prevalence rate of 0.2-2.2%. Transmission is usually secondary to maternal viremia, which may be asymptomatic.

The diagnosis is often missed in clinical practice, due to lack of physician awareness and its myriad ways of presentation. Only 10% of congenital CMV infection is symptomatic, masking its true prevalence. Subtle clues like hepatosplenomegaly and cytopenia can help in suspecting diagnosis [3].

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