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Congenital CMV Infection Masking the Diagnosis of Syndromic Neonatal Diabetes

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Introduction

Cytomegalovirus (CMV) is one of the viruses that are thought to cause type 1 diabetes mellitus due to its ability to induce immunological B cell destruction. Wolcott-Rallison syndrome (WRS) is the most common cause of permanent neonatal diabetes mellitus (PND) in consanguineous families. In addition to PND patients have multisystemic clinical manifestations including hepatic and renal dysfunction, developmental delay, seizures, exocrine pancreatic deficiency (EPD) and neutropenia. So, it shares many features with congenital CMV infection like developmental delay, hepatic dysfunction and epilepsy.

Objective

To present a child with WRS in whom the diagnosis was initially masked by associated congenital CMV infection

Case Report

At 10 weeks old female, product of consanguineous marriage, presented with diabetic ketoacidosis. Since then, she was maintained on multiple daily doses insulin injection regimen. The patient has microcephaly, neurodevelopment delay and recurrent seizures. Her laboratory investigations revealed elevated liver enzymes and neutropenia. Skeletal survey was normal and brain computerized tomography showed brain atrophy without calcifications. Diagnosis of congenital CMV infection was confirmed by positive polymerase chain reaction (PCR) for both the infant and the mother. She received ganciclovir without improvement of liver function or neutropenia. Genetic testing for neonatal diabetes showed that she is homozygous for the previously reported EIF2AK3 missense mutation, p.Arg632Trp confirming the diagnosis of WRS.

Conclusion

This patient demonstrates that diagnosis of WRS might be obscured by coincident CMV infection and illustrates the importance of conducting genetic testing in all cases of neonatal diabetes

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