A female infant was born to a healthy 28-year-old primiparous O-positive kindergarten teacher, and weighed 2.5 kg, measured 47 cm in length and had a head circumference of 31 cm. The pregnancy was uneventful, except for flu-like symptoms occurring at the end of the second trimester.

At 30 h of age, the infant was slightly jaundiced but not toxic. She had no hepatomegaly and no signs of bruising. Laboratory investigations revealed a normal complete blood count, an AB-positive blood grouping, a negative Coombs test and a serum total bilirubin level of 150 µmol/L with a direct component of 7 µmol/L. The infant was breastfed, and she voided well. A repeat serum total bilirubin level of 104 µmol/L was obtained 6 h later. Before the infant was discharged home, a routine metabolic screening was performed. There was also a hearing screening performed because this was recently instituted in the nursery. The attending physician was informed that the infant did not pass her hearing assessment. A repeat hearing test was scheduled within two weeks and another laboratory investigation was performed. What is this investigation? Why is it important?

**LEARNING POINTS**

- Congenital deafness occurs in one of 1000 to 2000 births. While autosomal recessive inheritance is the most common form (more than 75% of cases), congenital cytomegalovirus (CMV) infection is the leading nongenetic cause of hearing impairment in children.

- During three years of active surveillance (March 2005 to February 2008), the congenital CMV study of the Canadian Paediatric Surveillance Program confirmed 48 cases. Of the 35 of 48 cases with test results in the newborn period, nine cases had an abnormal hearing assessment.

- In asymptomatic infants not passing their newborn hearing screening, physicians should complete their evaluation by performing a CMV investigation and ensuring that a second hearing screening will be performed in the near future.

- The gold standard investigation for the diagnosis of congenital CMV infection is isolation of the cytomegalovirus or detection of viral DNA using polymerase chain reaction, usually in the urine or in the saliva where massive amounts of CMV are excreted. For diagnostic purposes, both methods are equally useful and the testing method will be determined by the reference diagnostic virology laboratory at each centre.

- The diagnosis of congenital CMV infection can only be made with certainty during the first three weeks of life. Virus isolation beyond that age may indicate acquired infection from exposure to the virus in the birth canal or in breast milk. Consequently, there is some urgency for the primary attending physician to collect the right specimen and order the right test before this window of diagnostic opportunity is lost.

- Infants infected with CMV should have a hearing screening performed in the neonatal period and regular audiology follow-ups for several years for the detection of progressive sensorineural hearing loss.

- Infants infected with CMV should also have an ophthalmology assessment as well as neuroimaging for documentation of central nervous system disease in the neonatal period. If affected, consultation with an infectious diseases specialist and consideration of ganciclovir therapy is important.

- Early diagnosis and intervention are essential to improve outcomes in hearing-impaired children.

- Handwashing and avoiding direct contact with potentially contaminated body fluids, especially urine and saliva, remains the cornerstone of CMV prevention. These preventive measures should be reinforced for all pregnant women and especially those who work with children.

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**RECOMMENDED READING**