Congenital CMV Assay on Dried Blood from Perinatal Cards

The UNC Hospitals Molecular Genetics Laboratory performs polymerase chain reaction (PCR) to detect cytomegalovirus (CMV) DNA in whole blood specimens. The test is used to assist in retrospective diagnosis of congenital CMV infection by testing blood that had been collected and dried on perinatal cards within three weeks of birth.

Utility and Clinical Significance of CMV PCR on Perinatal Cards

CMV infection is common in the general population and, when it occurs early in pregnancy, it can affect the developing fetal neurological system leading to sensorineural hearing loss and other neurological sequelae. Congenital CMV infection is defined as CMV infection acquired before three weeks of age, either in utero or perinatally via fluids such as cervical mucous, breast milk, urine or saliva. Congenital CMV infection is a significant cause of fetal and neonatal mortality and morbidity. It occurs in approximately 1% of births and is symptomatic in 10-20% of those infants. Disease in multiple organs is often manifest as jaundice, hepatosplenomegaly, petechial rash, microcephaly, motor disability, chorioretinitis, cerebral calcifications, lethargy, respiratory distress, and seizures. Almost all symptomatic neonates have neurological sequelae, most commonly sensorineural hearing loss, and often developmental delay ranging from subtle learning disabilities to severe mental retardation. The majority of congenitally infected infants (80-90%) have no signs or symptoms at birth, but 5-10% of these "asymptomatic" infected infants will later be found to have neurological sequelae, most commonly sensorineural hearing loss.

When a newborn is suspected of being infected, culture of the infant's urine or serologic testing of the mother can help determine if CMV infection is the cause. Due to the difficulty of diagnosing hearing loss and other neurological sequelae in infants, affected individuals may not be recognized until later in childhood when the active infection has resolved and culture is no longer informative. To overcome this problem, congenital CMV infection can be diagnosed retrospectively by testing residual blood samples collected from newborns and dried on filter paper (e.g. perinatal or Guthrie cards). Candidates for testing should be seen in Pediatric Genetics Clinic where consent for retrieval of the perinatal card from the NC State Health Laboratory may be arranged.

Laboratory Testing for CMV Viral Load:

DNA is extracted from dried whole blood and subjected to real-time PCR on an Applied Biosystems 7900 instrument using a TaqMan probe targeting a conserved sequence in the CMV polymerase gene. The assay is sensitive to as few as 1600 copies of CMV DNA per ml of blood. The result is reported qualitatively as positive or negative for CMV DNA. CMV is usually undetectable in dried blood from neonates, and negative results reduce the likelihood of congenital CMV infection.

References:

1. Sanchez, JL and Storch, GA. Multiplex, quantitative, real-time PCR assay for cytomegalovirus and human DNA. Journal of Clinical Microbiology, 40: 2381-2386, 2002

2. Barbi M. et al. A wider role for congenital CMV infection in sensorineural hearing loss. The Pediatric Infectious Disease Journal, 22:39-42, 2003.

Questions? Call the UNC Molecular Genetics Laboratory at (919) 966-4408
Website: http://labs.unchealthcare.org/directory/molecular_pathology/index_html