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Visual Impairment and Deafness in Young Children: Consider the Diagnosis of Congenital Infection With Cytomegalovirus, Even Years After Birth

Infection with cytomegalovirus (CMV) is the most common congenital infection in humans and affects 1% to 2% of all neonates. However, only 10% show symptoms at birth, varying from slight developmental complaints to serious neurologic, auditory, or ophthalmologic abnormalities. Although the other 90% of infected neonates are asymptomatic at birth, symptoms of congenital CMV may not be discovered until many years later. Later in childhood, 5% to 17% of these children will develop ocular, audiologic, neurologic, or developmental sequelae. Symptoms of congenital CMV can be indistinct and the diagnosis may be overlooked for years, leading to developmental disorders without adequate treatment. This report demonstrates how visual impairment detected many years after birth may lead to the diagnosis of congenital infection with CMV.

Report of a Case. A 3-year-old boy was suspected of decreased vision and sent to our clinic. At the age of 7 months the patient was diagnosed with deafness of unknown origin for which he received a right-sided cochlear implant. Ocular examination using the Kay picture test revealed decreased visual acuity OD (2/5) and normal vision in the left eye. Funduscopv of the right eye revealed an atrophic chorioretinal scar of the macula (Figure), whereas the left eye was unremarkable.

Serologic tests were performed, the results of which showed both the mother and son to be positive for anti-CMV IgG antibodies. To differentiate between a congenitally and postnatally acquired CMV infection, the Guthrie card containing neonatal dried blood drawn within 7 days after birth and used for the screening of inborn errors of metabolism was retrieved (with the parents’ permission) from the regional screening center. The dried blood was eluted in isotonic sodium chloride solution and tested for CMV DNA (real-time polymerase chain reaction [PCR], Taqman ABI7700; sensitivity, 5 genome copies/100 µL; specificity >99.9%) and anti-CMV IgM antibodies (Enzygnost; Dade Behring; sensitivity on serum, 95%; specificity, >99%). Both tests’ results were positive, establishing the diagnosis of congenital infection with CMV.

Comment. Infants with symptomatic congenital CMV infection are at high risk of developing visual impairment. In one study up to 22% of patients with symptomatic congenital CMV infection developed chorioretinitis or optic atrophy and in most cases bilaterally. Conversely, during a 5-year follow-up in a longitudinal study that included 445 children with congenital CMV, ocular defects were found in only 7% of children who were asymptomatic at birth. In all of the cases visual impairment was associated with optic atrophy, macular scars (chorioretinitis), or cortical damage. Although hearing impairment is the highest risk associated with asymptomatic congenital CMV infection, isolated neurodevelopmental delay and ophthalmologic lesions can also be signs of congenital CMV. One way to diagnose congenital CMV years after birth is by performing a CMV-PCR and/or anti-CMV IgM enzyme-linked immunosorbent assay on the neonatal blood stored on the Guthrie card. Note that negative results do not exclude a congenital infection; both tests have limited sensitivity when used in this setting.

In conclusion, patients with asymptomatic congenital CMV are at risk for serious sequelae that need proper care. Among children with visual or hearing impairment of unknown cause, CMV-PCR and serologic testing of neonatal blood stored on the Guthrie card enables the diagnosis of a congenital infection with CMV, even years after birth.

Gunnar I. Andriesse, MD, PhD
Annemarie J. L. Weersink, MD, PhD
Joke de Boer, MD, PhD

Correspondence: Dr Andriesse, Department of Medical Microbiology, Amphia Hospital, Molengracht 21, 4818 CK Breda, the Netherlands (g.andriesse@planet.nl). Financial Disclosure: None.