

IMAGES IN CLINICAL RADIOLOGY

Macrocephaly in an 8-month-old infant

K. Ramakers¹, W. Siemons², L. Meylaerts²

An 8-month-old infant presents with macrocephaly and a mild motoric deficit. The pediatrician asks a complementary MR examination to exclude hydrocephalus.

MR-images show diffuse swelling of white matter with preservation of the corpus callosum and the internal capsule. Also the occipital periventricular and subcortical white matter is better preserved than the rest of the cerebral white matter (A, axial T2 weighted image). The cerebellum seems normal (B, coronal T2 weighted image). The anterior temporal white matter is rarified (C, sagittal T1 weighted image, arrow). Axial T2 FLAIR image (D, arrow) shows a lower signal intensity in the anterior temporal subcortical white matter than in the cortex.

Diagnosis of van der Knaap's disease or MLC (megalencephalic leukoencephalopathy with subcortical cysts) was made.

Comment

Megalencephalic leukoencephalopathy with subcortical cysts (MLC) is an infantile-onset inherited disease of the brain white matter, also called van der Knaap disease. It is very rare and consanguinity and inbreeding contribute to its occurrence. MLC is a genetic disease with a defect in brain water and ion homeostasis and volume regulation by astrocytes. This defect results in chronic white matter edema.

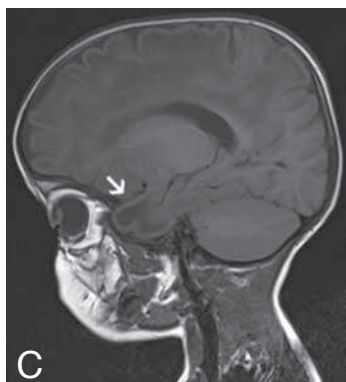
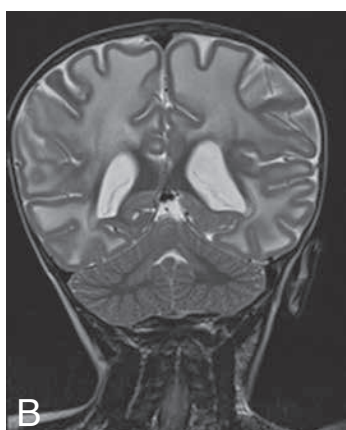
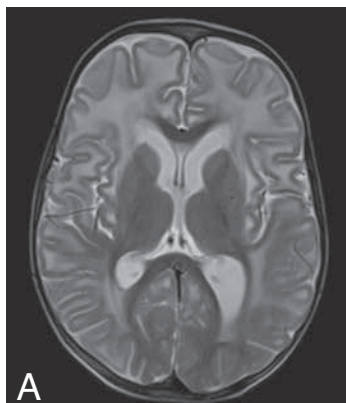
At birth, most infants are healthy and develop a macrocephaly in absence of other neurological signs. Most patients are mildly delayed in achieving unsupported walking and have an unstable gait. They become wheelchair-dependent as teenagers. Cognitive capabilities are normal or mildly decreased. Many patients living in their 40s and 50s are known.

MRI shows diffuse white matter abnormality with swelling of the abnormal white matter. Subcortical cysts are invariably present in the anterior temporal region but frequently also in the frontal and parietal regions. In children cysts may not yet be present, but anterior temporal white matter can be rarified, as in our case. Over time, the abnormal cerebral white matter becomes less severely swollen and atrophy follows, leading to widened lateral ventricles and subarachnoid spaces. Prominent atrophy is usually seen only in adults.

The combination of macrocephaly and diffuse cerebral white matter abnormalities can be seen in Alexander disease, Canavan disease, L-2-hydroxyglutaric aciduria, congenital muscular dystrophy with merosin deficiency, and sometimes in infantile-onset GM1 and GM2 gangliosidoses. In MLC The severity of the white matter disease contrasts with the presence of no or only mild clinical signs at this age.

Reference

1. Van der Knaap M.S., Boor I., Estévez R.: Megalencephalic leukoencephalopathy with subcortical cysts: chronic white matter oedema due to a defect in brain ion and water homeostasis. *Lancet Neurol*, 2012, 11: 973-985.



1. Department of Radiology, UZ-Gasthuisberg, Leuven, 2. Department of Radiology, Ziekenhuis Oost-Limburg, Genk, Belgium.