

Macrocephaly-Cutis Marmorata Telangiectatica Congenita Associated with Epilepsy: Case Report

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Abstract : Introduction: Cutis marmorata telangiectatica congenita (CMTC) is a rare cutaneous vascular malformation. It most often appears at birth or during the first days of life. Its origin is still unknown. It associates a livedo with telangiectasias of diffuse or segmental topography. In rare cases, it can be associated with neurological disorders such as macrocephaly and, less frequently, with epilepsy. Methodology: We report a case of an infant with Macrocephaly- Cutis marmorata telangiectatica congenita syndrome associated with epilepsy. Results: This is the case of a one month and 15 days old female infant from a non-consanguineous marriage, admitted for a status epilepticus in the context of apyrexia. Infectious and metabolic causes had been eliminated. Physical examination had shown non-infiltrated and reticular livedoid erythematous patches affecting the left upper limb and atrophic on the back of the left hand. Cerebral magnetic resonance imaging (MRI) showed thin layers of bifrontal, temporal, and left parietal hygromas associated with the widening of the bifrontal subarachnoid spaces. The electroencephalogram showed a well-organized sleep tracing with a single right occipital paroxysmal abnormality. Antiepileptic treatment has been administered with good clinical evolution and regression of the skin lesion and a control electroencephalogram without abnormality. Conclusion: This observation illustrates an association of CMTC with both macrocephaly and epilepsy. This pathology, which is relatively benign and has a good prognosis, generally does not require treatment. However, a detailed examination must be carried out, and a follow-up plan must be put in place for each patient presenting with CMTC, given the risk of association with other abnormalities, which can be potentially serious.

Keywords : cutis marmorata telangiectatica congenita, macrocephaly, epilepsy, children

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