Images in Aicardi-Goutieres Syndrome

Kallol Set

Department of Pediatric Neurology, Children’s Hospital of Michigan, Detroit, Michigan 48201, USA

Corresponding author: Kallol Set, MD, Department of Pediatric Neurology, Children’s Hospital of Michigan, Wayne State University, Detroit, MI 48201, USA, Tel: (1) 313-745-5788; E-mail: dr.kallolset@gmail.com

Received: Sep 28, 2016; Accepted: Sep 29, 2016; Published: Sep 30, 2016

Citation: Set K. Images in Aicardi-Goutieres syndrome. Transl Biomed. 2016, 7:3

Abstract

Aicardi-Goutieres syndrome (AGS) is a very rare genetic condition. A 16-month old girl one of twin, was born from consanguineous marriage with microcephaly, global developmental delay, hypertonia, failure to thrive has been presented here. Extensive lab work and MRI images suggested differential diagnosis of Aicardi-Goutieres, megalencephalic leukoencephalopathy with subcortical cysts, CMV TORCH intrauterine infection. The molecular testing for Aicardi-Goutieres syndrome (AGS) was performed which revealed an apparent homozygous deletion of exons 14 and 15 in the SAMHD1 gene. These exons repeatedly failed to amplify for sequence analysis which is consistent with a diagnosis of Aicardi-Goutieres syndrome. Symptomatic management and Genetic counselling done.

Introduction

Aicardi-Goutieres syndrome (AGS) manifests as an early-onset encephalopathy with severe intellectual and physical handicap after the first few weeks of life, frequently after a period of apparently normal development [1-3]. Typically, they present with sub-acute onset of a severe encephalopathy characterized by extreme irritability, intermittent sterile pyrexias, loss of skills, and slowing of head growth. Over time, as many as 40% develop chilblain skin lesions on the fingers, toes, and ears [3]. I present a patient with all the above mentioned features.
Figure 1 MRI of Brain shows microcephaly, moderate ventriculomegaly, diffuse decreased caliber of the brainstem, thin corpus callosum. Increased amount CSF signal in the posterior fossa likely represents a prominent cisterna magna. Extensive abnormal hyper intense T2/FLAIR signal seen throughout the periventricular and subcortical white matter of the frontal, parietal, temporal and occipital lobes. There are subcortical cystic changes of bilateral anterior temporal lobes and anterior right frontal lobe. MR spectroscopy with voxel positioning in the left frontal lobe demonstrates decreased choline, creatinine and NAA on the TE 38 and 144 signal. Evaluation of the lactate peak is difficult due to artifact. Overall nonspecific.

References