LETTER TO EDITOR (VIEWERS CHOICE)

CAN RETINA BE NORMAL IN AICARDI SYNDROME?

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A three and half years old muslim girl, born of a non-consanguineous marriage was admitted with uncontrolled recurrent infantile spasms since 4th day of birth for which she was hospitalized 15 times and had received anticonvulsive treatment initially with monotherapy and then gradually polytherapy. She was delivered by caesarean section and cried immediately after birth. She had no other perinatal complications. There was gross developmental delay. She neither developed social smile nor could she sit or stand. She was not able to hear or see. Her mother, 29 years old had two successive abortions in early months, first one at 4 months and second one at 2 months of gestation. On examination the patient had spasticity. Examination of the fundus was normal. Other systems were normal. Investigations showed cortical dysrhythmia on EEG. MRI of brain showed partial agenesis of corpus callosum mainly in posterior portion of the body and splenum, thalamus and periventricular region. Cross sectional MRI study revealed mild hyper intensities in bilateral putamen. Thus she was diagnosed as Aicardi syndrome without retinal involvement.

Aicardi syndrome is a severe neurodevelopment disorder initially thought to be the triad of partial agenesis of corpus callosum, choroidoretinal lacunae and infantile spasm. It is an X- linked dominant disorder affecting only females, usually males are not compatible with extrauterine life except some 47XXY (1). Infantile spasm usually starts in earlier and is very difficult to control. There is global mental retardation, visual defects and hearing defects. Other features present in subsets of affected individuals are costovertebral anomalies, facial dysmorphism, hand abnormalities, hypopigmentary skin lesions, multiple nevi, and an increased occurrence of rare vascular malformations and tumors along with other brain malformations (2,3). Chorioretinal lacunae are considered one of the pathognomonic sign of Aicardi syndrome though in some cases it may not be present (4) as was seen in our patient.

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