



CONFERENCE ABSTRACT

Siblings with autism, mental retardation, and convulsions in tuberous sclerosis: A case report

Farihan Helmy*

Taif University, Kingdom of Saudi Arabia

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*Corresponding Author,
Email: Farihanhelmy@gmail.com

Abstract

A 3 years old female patient born of consanguineous parents presented to the (development and behavioral clinic) in Taif children hospital, Western Saudi Arabia, her mother complained that her daughter had speech delay, no eye to eye contact, and was performing stereotyped behaviors (hand flapping). The girl developed convulsions at the age of 3 months and was on anticonvulsant medication since that age, her convulsions were controlled on anti-epileptic treatment. Family history revealed that the girl had a male sibling 6 years old who developed convulsions at the age of 4 months and is on antiepileptic medications, the boy suffered also from speech delay, absent social interaction, and repetitive behaviors. On examination the girl had characteristic features of angio-fibromas, hypo-pigmented macules on the trunk and legs, and moreover the boy had similar skin features plus hypo-pigmented tufts of hair. Both cases were diagnosed as Autistic spectrum disorder, tuberous sclerosis, and mental retardation. The family needed genetic counseling, while both cases needed as soon as possible behavioral and educational strategies.
