

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

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### Abstract:

A three year old female patient born of consanguineous parents presented to the (development and behavioural clinic) Taif Children Hospital, Western Saudi Arabia, her mother complained that her daughter had speech delay, no eye to eye contact, and was performing stereotyped behaviours (hand flapping). The girl developed convulsions at the age of three 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 six years old who developed convulsions at the age of four months and is on antiepileptic medications, the boy also suffered from speech delay, absence of social interaction, and repetitive behaviours. 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 counselling, while both cases needed possible behavioural and educational strategies.

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