A NOVEL FOCAL EPILEPSY AND INTELLECTUAL DISABILITY SYNDROME

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PURPOSE: To describe the clinical features and genetics in a new focal epilepsy and intellectual disability syndrome, focal seizures with intellectual disability, characterized by mild to moderate mental retardation, with prominent eye-blinking, facial and limb jerking beginning at around two months of age and persisting throughout life.
PATIENTS AND METHODS: We describe an Arab Israeli family consisting of 4 patients over 4 generations, reporting their clinical examination, EEG and MRI.
RESULTS: All the patients presented with focal seizures starting at age 2 months. Convulsive seizures also occurred but were generally controlled by anti-epileptic medication, carbamazepine and valporic acid. Motor and speech development were mildly delayed in some children and in adult life they had borderline to moderate intellectual disability (ID), associated with mild dysarthria and ataxia. There were no pathognomonic abnormalities in the EEG, MRI in one patient showed subtle cortical thickening most obvious in the antero-mesial frontal areas. The patients described an aura with the sensation of the tongue being anaesthetized. A mutation in *TBC1D24* was found in all patients
CONCLUSIONS: Focal epilepsy with intellectual disability must be considered in the differential diagnosis of children who present with early focal epilepsy together with intellectual disability and important dysarthria. This diagnosis is backed by the typical findings in the MRI.
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