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Mutation in *WWOX* gene cause severe neurological disorder with epileptic encephalopathy, west syndrome and psychomotor retardation

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Genetic testing including next generation sequencing have been increasingly used in the diagnosis of children with neurological disorders, including severe epilepsies and epileptic encephalopathy, intellectual disability and unexplained motor disorders. Here we report two pediatric cases from different consanguineous Emirati families. They both had early onset global developmental delay, quadriplegia and growth restriction. In addition, they both had epileptic encephalopathy and West syndrome. Case one had microcephaly and optic atrophy but normal MRI, while case two had cortical malformation in the form of polymicrogyria. Case two also had ventilator dependency and respiratory failure. Homozygous mutations involving the *WWOX* gene were found on whole exome sequencing in both cases (deletion affecting exons 3 to 4 in case one, and splice-site mutation c.606-1G>A in case two). Parents were heterozygous for the involved mutations, which confirm an autosomal recessive pattern of inheritance. WWOX is a cytoplasmic protein involved in many cellular processes including growth, differentiation and tumor suppression. WWOX mutations are reported in different human cancers. More recently WWOX mutations were described in a few cases with spino-cerebellar ataxia associated with epilepsy and mental retardation, a case of severe syndrome of growth retardation, microcephaly, epileptic seizures, optic atrophy, retinopathy and early death, a case of spasticity microcephaly seizures optic atrophy and psychomotor retardation, which is very similar to our 2 cases. The findings in our patients expand the phenotype associated with WWOX genetic abnormalities and confirm previous associations with microcephaly spasticity psychomotor retardation seizures and more importantly as a gene associated with epileptic encephalopathy and West Syndrome.

Biography

Jehan Suleiman is a Consultant Pediatric Neurologist at Tawam Hospital in UAE, and Assistant Professor at the UAE University. She is the Founder of Emirati Pediatric Neurology Network. She completed her training at the Children Hospital at Westmead in Sydney, Australia. She holds the Fellowship of Royal Australasian College of Physicians in Pediatric and Pediatric Neurology. She has completed her PhD in Medicine from the University of Sydney in the area of Autoimmune Epilepsy. She has many leading publications in this field in high impact journals. She is invited as a speaker to many national and international conferences. Her clinical and research areas of interest include neuroimmunology, complex epilepsy and neurogenetics.

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