A Rare Cause of Limbic Encephalitis

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A 16-year-old female first presented as generalised tonic-clonic convulsion in November 2011. She had two more attacks afterwards. She was admitted into the intensive care unit in December for status epilepticus. There was low-grade fever. Neurological examination revealed no focal neurological deficits. Computed tomography of brain was unremarkable. Lumbar puncture (LP) showed normal protein level and leukocyte count. Intravenous acyclovir and ceftriazone were started empirically. Electroencephalography (EEG) showed subclinical bitemporal independent seizure without generalisation. MRI brain showed symmetrical increased T2-signal intensity involving both mesial temporal lobes, suggestive of limbic encephalitis. CSF HSV PCR was negative. A course of intravenous immunoglobulin was given because autoimmune limbic encephalitis was suspected. Voltage-gated potassium channel antibody, anti-NMDA receptor antibody turned out to be negative. CSF later revealed positive VDRL. Serum VDRL was negative and FTA was reactive. HIV antibody was negative. A course of penicillin G was initiated. Serial EEG showed resolved epileptiform activities. However she had episodic fluctuation of conscious level. Five sessions of plasmapharesis were given. She gradually recovered and repeated LP showed negative VDRL and follow-up MRI brain showed resolution of both temporal lobe lesions. She later confessed that she had unprotected sex with two to three sexual partners. She returned to school and remained seizure-free till last follow-up in July 2012.

A high index of suspicion is necessary to make a correct diagnosis of neurosyphilis, especially when approaching a young patient presenting with limbic encephalitis. Negative serum RPR and HIV testing, negligible sexual history and absence of CSF leukocyte hampered a prompt recognition of neurosyphilis in our case. Early diagnosis and treatment are important because the infection can be treated with appropriate antibiotic.

Dravet Syndrome: Genetic Analysis of SCN1A and PCDH19 Mutations for 17 Chinese Children

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Background: For Dravet syndrome (DS), 80% had mutation in SCN1A gene, which encoded a voltage-gated sodium channel. Recent study demonstrated that 16% of SCN1A-negative patients had mutations in protocadherin-19 (PCDH19) genes. The present study examined the genetic mutations in Chinese DS children and assessed the relationship between mutation and phenotype.
Methods: DNA of 17 DS in the University of Hong Kong was screened for SCN1A mutation using polymerase chain reaction and direct sequencing. SCN1A-negative female patients were then screened for PCDH19 mutation.
Results: For DS, 82% (14/17) had SCN1A mutations—truncating mutations (6), splice site mutations (2) and missense mutations (6). These mutations affected Nav1.1 protein functions by pathogenicity assessments including conservative, SIFT and Align-GVGD analyses. We found a relationship between the type of mutation and the degree of intellectual disability ($P<0.05$), with truncating/splice site mutations associated with moderate/severe mental retardation. At the evolution of the disease, 79% (11/14) of DS patients with SCN1A mutations had features which fit into the diagnostic criteria of autism spectrum disorder (ASD). 57% (8/14) had a history of vaccination-induced seizures. One of the two female SCN1A-negative patients had PCDH19 mutation.
Conclusion: High percentage of genetic mutations was identified in our Chinese cohort of DS. Pathogenicity assessment demonstrated that the mutations were linked to the phenotypes of DS. Our detection of high frequency of ASD (79%) and vaccination-induced encephalopathy (57%) in those DS with SCN1A mutation suggested evaluating ASD with epilepsy or vaccination-induced encephalopathic children for any relationship between SCN1A mutations.