

diagnostic features: C-Cardiac abnormality (tetralogy of Fallot); A-Abnormal facies (hypertelorism); T-Thymic aplasia; C-Cleft palate; H-Hypocalcemia /hypoparathyroidism; 22-Chromosome abnormality [2].

References.

1. Digilio MC, et al. Clin Genet. 2003 Apr;63(4):308-13.
2. Millichap JG. Neurological Syndromes : A Clinical Guide to Symptoms and Diagnosis. New York: Springer; 2013:279.

ENCEPHALOPATHIES

ANTECOLLIS AND PARKINSONISM IN ADULTS WITH DRAVET SYNDROME

Investigators at Toronto Western and The Hospital for Sick Children, Toronto, Canada, prospectively studied the motor abnormalities in a consecutive sample of adults with genetically proven Dravet syndrome (DS). Of 12 patients entering the study, 11 presented with severe cognitive delay, and 1 had mild spasticity. All had flexion of the head and 8 (66%) had dystonic antecollis, severity correlating with age. Mild parkinsonism in 11 patients (91%) was characterized by global bradykinesia and asymmetric cogwheel rigidity. Severity of parkinsonism was correlated with age but not with seizure frequency or use and dose of valproate. Crouch gait (in 5/12 cases), small steps, and/or wide base gait impairment occurred in 9 patients. Two of 4 most affected patients experienced sustained improvement of gait following levodopa treatment and were no longer wheelchair bound. (Fasano A, Borlot F, Lang AE, et al. Antecollis and levodopa-responsive parkinsonism are late features of Dravet syndrome. **Neurology** 2014 Jun 17;82(24):2250-1).

COMMENTARY. The cause of the postural and motor signs in adults with Dravet syndrome is unclear. The Toronto authors favor the progressive involvement of the nigrostriatal dopamine system but admit the possibility of adverse effects of antiepileptic drugs [1]. Crouch gait in 50% of adults with DS and SCN1A mutations was observed only in those patients with nonsense mutations or mutations in the pore-forming region of the Na 1.1 protein. Crouch gait, without spasticity, is identified as an element of the adult DS phenotype [2].

Investigators in Paris, France, discuss cognitive impairment in Dravet syndrome that it is not related to major epileptic factors (duration of seizures, fever-related status epilepticus) but more a consequence of genetic factors. SCN1A mutation directly contributes to cognitive impairment independent of seizure induction. Dravet syndrome should no longer be considered an epileptic encephalopathy [3].

References.

1. Fasano A, et al. Neurology. 2014 Jun 17;82(24):2250-1.
2. Rilstone JJ, et al. Epilepsia. 2012 Aug;53(8):1421-8.
3. Auvin S, et al. Editorial. Should we still consider Dravet syndrome an epileptic encephalopathy? Epilepsy Behav. 2014 May 31;36C:80-81.