De novo ADCY5 mutation in patient who showed early onset dystonia and paroxysmal choreoathetosis in Korea

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Background & Significance: Adenylyl cyclase 5(ADCY5) mutations is associated with heterogenous hyperkinetic movement disorders: familial dyskinesia, paroxysmal chorea and dystonia; autosomal-dominant chorea and dystonia; and benign hereditary chorea. We observed patient who identified ADCY5 mutation for the first time in Korea. Case: This 20-year-old man presented motor mile stone delaying since 9 month of age. He had difficulty in sitting, grab and standing, and walking difficulty in his 17 month of age. Psychomotor development was normal. The plasma amino acid, urine organic acid, X-ray and brain magnetic resonance image (MRI) was checked, but specific findings did not observed. At 2-year-old age, he had difficulty in supporting his neck, and he maintained head dropped posture. He had no pre and postpartum injury but diagnosed athetoid cerebral palsy at that time. He hospitalized for comprehensive rehabilitation at 20-year-old and cooperated with neurologist. Neurological examination confirmed axial hypotonia (neck) and mild dystonic posture of the four limbs. Gait is unsteady or nearly impossible because of the head drop. He showed hyperreflexia and episodic, paroxysmal choreoathetosis during sleep and when he fall asleep. The laboratory tests, EEG, EP, cognition test and follow up brain MRI were normal. De novo ADCY5 gene mutation (c.2088+1GT) was found by trio exome sequencing, and he was treated with clonazepam and clobazam. Conclusions or Comments: ADCY5 genetic analyses may be relevant in the diagnostic workup of early-onset dystonia and hyperkinetic movement disorders.