LETTER TO EDITOR (VIEWERS' CHOICE)

SEGAWA SYNDROME - A FAMILY CASE REPORT

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Keywords: segawa syndrome, levodopa, spasticity

Segawa syndrome is a rare genetic disorder characterized by an uncoordinated or clumsy manner of walking. Symptoms of Segawa syndrome usually become apparent around six years of age. Intelligence is not affected. (1) Mutations in several genes have been shown to cause dopamine-responsive dystonia. (2) The symptoms worsen during the course of the day and with increasing age until the third decade of life.(3) Other names of Segawa syndrome are Tyrosine Hydroxylase Deficient-Dopa - Responsive Dystonia (T-H deficient DRD), Tyrosine Hydroxylase deficiency.(4) Family of four children suffering from this syndrome is being reported. The index case is a 10 years old male child who presented with complaints of inability to stand and walk along with spasticity increasing progressively. Higher functions were normal. On examination, weight was 12 kg (< 3rd percentile), height was 105 cm (< 3rd percentile). Intelligence was normal. On central nervous system examination, he had dysarthria, cogwheel rigidity, brisk deep tendon reflexes with clonus. Scissoring was present. Other systems were normal. Routine cerebrospinal fluid (CSF) analysis was normal. CSF neurotransmitter dopamine 16 pg/ml (normal 30- 160 pg/mL)and biopterin13.5uM (19.2 - 22.0uM) were low.A phenylalanine loading test was done and phe/tyr (phenylalanine/tyrosine) ratio was 5.25 after 4 hours(normal 2.4 to 14.74). MRI of brain was normal. Diagnosis of Segawa syndrome was made. Siblings were also noted to be suffering from same disorder. (Table 1) All children were put on levodopa. Three years follow up revealed full recovery on drug. Whenever L-dopa is withdrawn, condition worsens, so medication is regular.

Dopa-responsive dystonia (DRD) is a genetically heterogeneous syndrome that typically presents in children as leg dystonia and parkinsonism. Similar to juvenile-onset Parkinson disease, dopa-responsive dystonia is due to dopamine depletion, but unlike Parkinson disease, dopamine deficiency arises secondary to a defect in neurotransmitter synthesis rather than a loss of dopaminergic neurons. It responds dramatically to low-dose levodopa therapy, independent of patient age or disease duration.Clinical features, and response to levodopa have remained the diagnostic gold standard for DRD.(5) CSF biopterin (measured after chemical oxidation of BH4) and neopterin are also markers for autosomal dominant DRD, and both are decreased in manifesting and non-manifesting gene carriers.Our patient also had a lower CSF biopterin levels. We report a family of Segawa syndrome for its rarity.

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DOI: 10.7199/ped.oncall.2014.10

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Table 1: Details of each sibling

CASE	AGE/SEX	AGE OF ONSET	COMPLAINTS	DEVELOPMENT	CNS EXAMINATION
2nd	6 years/ male	4 years	Similar to index case	Normal for age	Similar to index case with normal reflexes
3rd	4 years/ female	2 years	Same as index case with deviation of neck towards right side with bruxism	Normal for age	Similar to index case
4th	2 years/ female	2 years	Abnormal movements of all the limbs	Normal for age	Normal