Report two cases of dopa-responsive dystonia

JIANG Zhuannan

Objective: Understand the research progress of dopa-responsive dystonia by reporting two cases of this rare disease.

Methods: Analysed the clinical manifestations, clinical laboratory examination and gene sequencing of two cases of dopa-responsive dystonia patient and reviewed the related literatures.

Results: Clinical and Laboratory diagnosis two cases of dopa-responsive dystonia.

Conclusion: Dopa-responsive dystonia is a rare disease, its clinical characteristics include;

1. childhood-onset dystonia with marked diurnal fluctuation and female predominance, for the not enough supplement of BH4 during the day;
2. A dramatic and sustained response to relatively low doses of levodopa, but little effectiveness of treatment of BH4;
3. Mutations of GCH-I gene and TH gene;
4. Increased prolactin by regulation of dopamine D2 receptor clinical diagnosis of DRD remains difficult.The children who suspected the disease should be taken genetic test oonce diagnosed, the Comprehensive Treatment, low doses of levodopa therapy, should be taken to improve the quality of life as soon as possible.

55ESPE 797--P2

Pituitary and Neuroendocrinology

Jiang Zhuannan

DOI: 10.3252/pso.eu.55ESPE.2016

Poster presented at: ESPE 2016 PARIS