Neurometabolic Disorders, Clinical and Neuroimaging Finding

How to Cite This Article: Jafari N. Neurometabolic Disorders, Clinical and Neuroimaging Finding. Iran J Child Neurol. Autumn 2014;8;4(Suppl.1):23.

Narjes JAFARI MD

Neuro metabolic disorders are the rare inherited disorders with most a defective gene that results enzyme deficiency. More common groups of neurometabolic disorders include organic aciduria, aminoacidopathies, storage disorders, Mitochondrial disorders, urea cycle defects, lysosomal storage disease and etc.

Inborn errors of metabolism are the difficult groups of disorders for the Physicians, including neurologists as there are few good clinical criteria for diagnosis.

In this study, Patients who were diagnosed withneurometabolic disorders in the Neurology Department of Mofid Children's Hospital in Tehran, Iran from October 2005 to February 2014were included in our Review. The disorder was confirmed by enzyme level detection, genetic mutation tests, assay for organic acid in urine, carnitine profile in serum, metabolic data from proton MR spectroscopy and etc.

We assessed and reported age, gender, family history, past medical history include pregnancy and neonatal period, developmental status (developmental delay and regression), clinical manifestations include : hair and skin involvement, dysmorphic facies, organomegaly, cranial nerveinvolvement, movement disorders, tonicity of body, type of seizures, behavioral problems and paraclinic and neuroimaging findings of 215patients with neurometabolic disorders, include 19 patients with biotinidase deficiency, 19 patients methylmalonicacidemia, 20 glutaricacidemia type 1, 20 GM2 gangliosidosis, 15 homocysteinemia, 13 canavan, 10 propionic acidemia, 9 glutaricacidemia type 2, and 8 patients with MCAD disorders, and etc. that results of which will be presented in this Congress.

Keywords: Neurometabolic disorders; Genetic disorders; Children

Pediatric Neurology Research Center, Shahid Beheshti University of Medical Sciences, Tehran, Iran
Corresponding Author:
Jafari N. MD
Shariati Ave, Pediatric Neurology Research Center, Mofid Children Hospital, Shahid Beheshti University of Medical Sciences, Tehran, Iran
Email: jafareadr@gmail.com