عنوان مقاله:

Genetic diagnosis of a child with regression and accumulation of metal in the brain

محل انتشار:

دومین کنگره بین المللی و دهمین همایش ملی نوروژنتیک ایران (سال: 1396)

تعداد صفحات اصل مقاله: 1

نویسندگان:

Mehran Beiraghi Toosi - *Pediatric neurologist, Department of pediatrics, Faculty of Medicine; Mashhad University of Medical Sciences, Mashhad, Iran*

Farnoosh Ebrahimzadeh - *Department of Internal Medicine, Faculty of Medicine, Mashhad University of Medical Sciences, Mashhad, Iran*

Majid Mojarrad - *Department of genetics, Faculty of Medicine; Mashhad University of Medical Sciences, Mashhad, Iran*

خلاصه مقاله:

A 27 month-age girl came to us with regression form 5 months ago. She haddystonia and normal deep tendon reflexes. She had relative parents with nocomplication in delivery.Report of the brain CT scan was normal. We got Brain MRI and saw hyperintensity in glubuspallidus and dentate nucleus in T1 and hypo intensity of theregions in T2; so there was collection of some metals in the brain.In toxicology evaluation, we did not find any problem. After evaluation ofwhole exome sequencing, we found homozygote mutation in the SLC30A10gene; so the diagnosis hypermanganesemia with dystonia type 1 confirmed.Conclusion: Sometimes genetic testing is helpful for diagnosis of .raresyndromes that we can’t recognize with only clinical studies

کلمات کلیدی:

regression, hypermenganesemia with dystonia, genetic testing