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Biotin-responsive basal ganglia disease: Catastrophic consequences of delay in diagnosis

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Background: Biotin-Responsive Basal Ganglia Disease (BBGD) is an autosomal recessive neurometabolic disorder caused by mutations in the SLC19A3 gene. The disease is characterized by subacute encephalopathy with confusion, dysphagia, dysarthria and seizures.

Methods: We diagnosed a family affected by BBGD and studied them including prognosis of cases when diagnosed and treated early in the disease process. We also review the literature comprehensively and summarize all published data about this disorder.

Results: Since its first description, a total of 89 cases (46 females and 43 males) have been published in the literature. We studied six patients in this article in which three died before a diagnosis was established, one was diagnosed lately and is currently severely affected, and two were diagnosed early and are currently stable on treatment. The clinical phenotype of each family member was studied in details and a genetic testing using whole exome sequencing and Sanger sequencing of the family members was done to confirm the diagnosis. The whole exome sequencing revealed a homozygous mutation in the exon 5 of the SLC19A3 gene c.1264A G (p.Thr422Ala) which is diagnostic of biotin-responsive basal ganglia disease.

Conclusion: BBGD is a treatable condition if recognized early and managed appropriately. Children presenting with unexplained encephalopathy and MRI abnormalities including bilateral signal alteration of caudate nucleus and putamen should raise the suspicion for BBGD and be started immediately on biotin and thiamine regimen since the prognosis of the disease is affected by the timing of treatment initiation.

Biography

Abdulrahman Bazaid is a medical intern at King Abdulaziz Medical City in Jeddah, Saudi Arabia. He obtained his MBBS degree from Battarjee Medical College in Jeddah, Saudi Arabia. He attended several seminars, courses and workshops in research methodology. He is determined to pursue his career as a physician, academician and researcher.

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