PP-051 RAPID-ONSET OBESITY, HYPOTHALAMIC DYSFUNCTION, HYPOVENTILATION, AND AUTONOMIC DYSFUNCTION (ROHHAD)

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Aim To Report a rare and interesting case.
Material and Method Rapid-onset obesity, hypothalamic dysfunction, hypoventilation, and autonomic dysfunction (ROHHAD) is a rare syndrome that was initially described more than 50 years ago. However, the term ROHHAD and its criteria for diagnosis were not distinguished as a separate entity except in 2007. (Fishman LS) (Ize-Ludlow D). To date, literature shows fewer than 100 cases were described in the literatures. Despite that there is no diagnostic test that can detect ROHHAD; Keeping it in mind while excluding other etiologies is important; since the prognosis of ROHHAD mainly depends on how early, the case is diagnosed and managed in the aims to decrease severe complications and mortality. Symptoms of the disease, particularly rapid weight gain, can present as early as one year and a half of age, followed by gradually developing symptoms of hypothalamic dysfunction, autonomic dysregulation, and hypoventilation, leading the parents to seek medical attention. Currently, management of ROHHAD syndrome is largely symptomatic, focusing mainly on improving the quality of life and preventing potentially fatal sequelae in patients with this disease. As for medical management, clinical trials are still ongoing.
Results In this paper, we report a case of a 3-year-old girl who initially presented with rapid-onset obesity followed by multiple features of ROHHAD symptoms over time.
Conclusions In this paper, we aim to raise awareness of the presence of such cases in our community and to increase knowledge about the management of such cases.

PP-052 GROWING PAINS IN CHILDREN CAN MASK A RARE DISEASE

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Aim Growing pains are benign and recurrent limb pains that affect children between the ages of 3 and 12. This condition may mask various hereditary diseases, for which early diagnosis is key. Hypophosphatasia, an inherited disorder similar to rickets, has different clinical forms: perinatal, infantile, childhood, adult, and odontohypophosphatasia. The first symptoms in the childhood and adult forms of the disorder are often limb pain, poor exercise tolerance, lower limb deformity, poor posture, and fractures.
Material and Method We examined an 8-year-old patient who had been followed by a dentist since the age of 1.5 years due to early loss of deciduous teeth. Starting at age 3, the patient was found to have a delayed growth rate. Since the age of 4, he has been followed by a rheumatologist with a diagnosis of arthropathy and growing pains due to recurrent lower limb pain, arthralgias, and proximal muscle weakness when squatting and climbing stairs. At age 5, the patient's alkaline phosphatase level was 61U/L(156–369). Molecular genetic analysis revealed that the patient had a nucleotide sequence variant c.331_332insCCGGCA, p.Thr113Ala114insGlyThr in the ALPL gene in heterozygous state. This variant was also found in heterozygous form in
Results Clinical examination revealed short stature (z-score=1.93), vertical bone resorption and bone loss in the interdental septa, and pronounced lumbar levoscoliosis. The patient's alkaline phosphatase level was determined to be below the normal range at 81U/L(156–369).
Conclusions A child complaining of limb pain may be the first symptom of a rare genetic disease. In the presence of arthralgias, it is necessary to conduct a comprehensive examination, taking into account the family history and laboratory tests, including alkaline phosphatase, calcium, phosphorus, vitamin D, creatine kinase, rheumatoid factor, and a complete blood count.

PP-053 A CASE WITH BROWN VIALETTO VAN LAERE SYNDROME

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Aim Brown Vialetto Van Laere Syndrome (BVVL) is a progressive neurological disorder that is characterized by the mutations in the riboflavin transporter genes SLC5A2 and SLC52A3. Fifty-eight cases have been demonstrated in just over 100 years. Distribution in female and male cases have been reported as 3:1 ratio (female:male). Approximately 50% of BVVL cases have been reported as a familial with an autosomal recessive inheritance, however sporadic cases have been also demonstrated. BVVL affects pontobulbar area resulting as a sensorineural deafness, respiratory difficulties, limb weakness, slurred speech, muscle weaknesses. Moreover, optic atrophy, retinitis pigmentosa, macular hyperpigmentation, autonomic dysfunction and epilepsy may be observed as a symptom. Our patient is a 9 years old female who has a neuromotor retardation, scoliosis and difficulty in maintaining her posture. Furthermore, she is hypotonic and has both upper and lower limb muscle weakness, however lower limb muscle weakness is more prominent as she has little spontaneous movements. In addition to these symptoms, she has a hypoactive deep tendon reflexes. Aim of this case report is to identify the medical needs of the patient due to riboflavin transporter and advise a course of treatment.
Material and Method Whole Exome Sequencing genetic test was observed to identify the mutations. Electromyography was an another test to show any neuropathies. Moreover, laboratory analyses that detect the level of several acylcarnitine, hydroxy-acylcarnitine levels and riboflavin level.
Results Whole Exome Sequencing genetic test resulted in homozygote mutation at SLC5A2 gene. Electromyography shows peripheral neuropathy which supports the diagnosis. Laboratory analysis showed normal levels of several acylcarnitine and hydroxy-acylcarnitine levels. Moreover, riboflavin level was in normal range.
Conclusions The advised treatment is oral riboflavin supplementation and routine follow up will be preferred to observe improvements in fine motor skills.