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A Case of Osteopetrosis Diagnosed with Nystagmus

Authors : Zerrin Orbak, Busra Demir

Abstract : Osteopetrosis is a rare genetic disease characterized by impaired bone resorption and increased bone sclerosis. Clinical presentation is very different in osteopetrosis. It can be asymptomatic or can be seen with typical symptoms. Here, a case of osteopetrosis was presented when evaluated for nystagmus. She was 10 months old. Parents were second-degree relatives. On physical examination, pigeon chest deformity and horizontal nystagmus were observed. There was a failure of thrive but no fracture. The cardiovascular examination was normal. Cranial, vertebral and long bone roentgenograms revealed characteristic deformities of osteopetrosis and diffuse sclerosis. The diagnosis was confirmed by genetic testing. A Homozygous mutation was detected in the TNFRSF11A gene (c.508A>G p.(Arg170Gly)). RANKL is encoded by the tumor necrosis factor ligand superfamily member 11 (TNFSF11) gene, and the binding to its receptor RANK, encoded by the TNFRSF11A gene, determines the activation of the downstream pathway that drives osteoclast differentiation and activation (51). The complete absence of osteoclasts is the key feature of the osteoclast-poor form of osteopetrosis (46). Patients are characterized by the absence of TRAP-positive osteoclasts in bone biopsies. The osteoclast-poor subtype of osteopetrosis caused by mutations in TNFSF11 gene is ultra-rare in humans. Clinical presentation is usually severe, with onset in early infancy or in fetal life. But here, a case was presented with horizontal nystagmus. A case presented with horizontal nystagmus, which was evaluated by neurology and diagnosed incidentally, was shared.

Keywords : osteopetrosis, nystagmus, bone, osteoclast-poor
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