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CASE REPORT

A case report on Winchester syndrome

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ABSTRACT

Winchester syndrome is a rare inherited disease characterized by severe osteolysis particularly in the hands and feet, generalized osteoporosis and absence of subcutaneous nodules. It is a group of hereditary metabolic diseases in which certain enzymes are lacking or defective that would normally break down molecules into smaller units, which leads to the accumulation of molecules in cells and tissues. A middle-aged female who is a known case of Juvenile inflammatory arthritis presented with complaints of deformities in hands, feet, knees, and hip joints. She was short stature and the random of motion of her ankle, knee, and hip was restricted. Winchester syndrome is an extremely rare disorder inherited as an autosomal recessive trait. Winchester syndrome is caused by an alteration in a gene called MMP2. Symptomatic treatment can be given with anti-inflammatory drugs and skeletal muscle relaxants. Physical therapy may be advised as a supportive measure to improve the functioning of affected limbs. Genetic counseling on the basis of the inheritance of this disease may be beneficial. Our patient was symptomatically managed with anti-inflammatory drugs and discharged with calcium supplements. Furthermore, regular physiotherapy and replacement surgeries for joints were done. This syndrome was first diagnosed in 1969; only a few cases have been reported in the medical literature till now.

KEY WORDS: Winchester Syndrome; Osteoporosis; Osteolysis

INTRODUCTION

Winchester syndrome is a rare inherited disease characterized by severe osteolysis particularly in the hands and feet, generalized osteoporosis and absence of subcutaneous nodules.[1] The pathologic changes include of dwarfism, corneal opacities, coarsening of facial features, leathery skin, and hypertrichosis.[2] It is a group of hereditary metabolic diseases in which certain enzymes are lacking or defective that would normally break down molecules into smaller units, which leads to the accumulation of molecules in cells and tissues.[3] Here, we report a case of a middle-aged woman with Winchester syndrome. Rheumatoid factor was negative. Later, she was diagnosed to have Winchester syndrome. Usually, the onset of this syndrome varies from 3 months to 22 years.[3] In this case, the diagnosis was delayed because it was overlapped with that of Juvenile inflammatory arthritis (JIA) which was also present.

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CASE REPORT

A 36-year-old female who is a known case of JIA for 25 years has come to AIMS for further evaluation and management for complaints of deformities in hands, feet, knees, and hip joints for the past 15 years. She was short stature and the random of motion of her ankle, knee, and hip was restricted. She had flexion postured hip and knee, also extended postured ankles. No joint pain, swelling or morning stiffness was present. A positive familial history of arthritis was present. She had hypertension for 20 years but not taking any medications.

Anti-cyclic citrullinated peptide antibody was strong positive while rheumatoid factor was negative. Inflammatory markers and baseline liver and kidney functions were normal. X-ray pelvis showed fused/fibrosis hip joints and Sclerosis of sacroiliac joints. X-rays of other joints (knees, hands, and feet) also showed degenerative changes. Pediatric genetics consultation was taken, and possibility of Winchester syndrome was made. As she does not have any sign of active arthritis, she was given symptomatic medicines. DEXA Z score was −0.6 in spine, 0.2 in left femur, and −1.3 in right femur. She was symptomatically managed with anti-inflammatory drugs and discharged with calcium supplements. Furthermore, regular physiotherapy and replacement surgeries for joints were done.

DISCUSSION

Winchester syndrome is an extremely rare disorder inherited as an autosomal recessive trait.[3] Winchester Syndrome is caused by an alteration in a gene called MMP2. Alterations in the same gene can lead to conditions such as Torg syndrome and multicentric osteolysis with nodulosis and arthropathy which are very similar to Winchester syndrome.[4]

Winchester is more common in females than males with female to male ratio of 3:1.[4] This syndrome was first diagnosed in 1969; only a few cases have been reported in the medical literature till now. Some of the affected individuals may be undiagnosed or misdiagnosed.[3] Usually, the onset of this syndrome varies from 3 months to 22 years. In this case, the diagnosis was delayed because it was overlapped with that of JIA which was also present. The main characteristic feature of this disease is growth restriction and marked short stature due to the degenerative changes occurs in the vertebrae of the backbone and the long bones of the limbs.[5,6]

Osteolysis and osteoporosis are the other significant characteristics. The mostly affected joints include joints of the hands, feet, knees, shoulder, elbow, and hip.[7] An Z score of −1.3 of the right femur and −0.6 of the spine indicated the low bone mass density of the patient.

Symptomatic treatment can be given with anti-inflammatory drugs and skeletal muscle relaxants. Our patient was managed with tramadol and got better. Physical therapy may be advised as a supportive measure to improve the functioning of affected limbs. Genetic counseling on the basis of the inheritance of this disease may be beneficial.[8]

CONCLUSION

Here, we report a case of a middle-aged woman with Winchester syndrome. Rheumatoid factor was negative. Later, she was diagnosed to have Winchester syndrome. Usually, the onset of this syndrome varies from 3 months to 22 years. 3 In this case, the diagnosis was delayed because it was overlapped with that of JIA which was also present.

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