



Stone Man Syndrome a Rarest Disorder: Case Report

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Abstract

Stone Man Syndrome most of time referred to as Fibrodysplasia ossificans progressiva. Which is one of the rarest connective-tissue disorder. We report such a case of fibrodysplasia ossificans progressiva diagnosed by clinical and radiologically.

Keywords: Autosomal dominant, Fibrodysplasia ossificans progressiva, great toe, malformation, radiological examination, Stone Man syndrome.

Introduction

Stone Man Syndrome is one of the rarest connective-tissue disease. characterized by congenital malformation of the big toes and progressive heterotopic ossification of the fasciae, ligaments, tendons, and skeletal muscles¹.

Case Report

A 34 years female presented with classical postural deformity. Clinical and radiological examination revealed sheets of bone along spine, shoulder, hip and elbow. There was characteristic malformation of great toe which was present since birth. The age of presentation was 5 years. Heterotopic ossification typically found on cranial, axial, ventral, dorsal, aspects and distal regions and proximal regions of the body also. The diaphragm, as well as the extra ocular, cardiac, and smooth muscles, were characteristically spared.

Discussion

Fibrodysplasia Ossificans Progressiva is an autosomal dominant disorder, epidemiological occur sporadically (mainly). There is no ethnic, racial, gender, or geographic predisposition¹. Two clinical features define classic Stone Man Syndrome²: i. Congenital malformations of the great toes ii. Progressive heterotopic ossification of the ligaments, tendons, and skeletal muscles, before attending adolescent age group. Most patient with Stone Man Syndrome develop painful, episodic, soft tissue swellings and progressive, disabling heterotopic ossification of the fasciae, ligaments, tendons, and muscles (skeletal). These nodules may regress, but mostly they mature rapidly to form lamellar bone which immobilises the joints. The flare-up of disease occur by some triggers like-minor trauma, some viral infections, Excision of the heterotopic new bone. Plain x-rays can use to detect bony (big toes) abnormalities and presence of characteristic progressive myositis calcifications. So that Stone Man Syndrome should always come in mind as a differential

diagnosis of progressive myositis calcifications. Stone Man Syndrome is differ from Progressive Osseous Hetero-plasia by presence of congenital malformation of the great toes, lack of cutaneous ossification and “flare-ups” of disease by some triggers³. Acquired Progressive hetero tropic calcification also differ from Fibrodysplasia Ossificans Progressiva by occurrence of disorder at any age (rare in young age), and commonly follows severe trauma (despite of minor trauma. Stone Man Syndrome is commonly misdiagnosed as progressive juvenile fibromatosis, desmoids tumors, soft tissue sarcoma or calcinosis cutis etc.⁴

Conclusion

Stone Man Syndrome is one of the rarest connective-tissue disease. And most of time this is misdiagnosed due to common presentation of some congenital or acquired disorder. But by the use of some cheap and easily available radiological investigation (e.g.-X rays) and appropriate clinical examination we can easily diagnosed this rarest connective-tissue disease.

References

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