Systemic Mastocytosis: A Rare but Important Cause of Osteoporosis

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INTRODUCTION

Mastocytosis is a rare condition caused by excessive accumulation of mast cells in one or more multiple tissues. Mastocytosis can be divided into cutaneous and systemic mastocytosis. Cutaneous mastocytosis is limited to the skin. Systemic mastocytosis describes forms of mastocytosis in which pathologic mast cells infiltrate multiple extracutaneous organs with or without skin involvement. The exact incidence is unknown. Mastocytosis affects both male and female equally1,2.

CASE REPORT

We present a case of a 56 year-old man diagnosed with systemic mastocytosis by the dermatologists on presentation with classical skin lesions, confirmatory bone marrow biopsy and a tryptase level 105 ug/L (NR 2-14). Further genetic testing confirmed KIT D816V mutation.

DEXA bone densitometry at diagnosis revealed marked osteoporosis (Lumbar T score -3.7 (Z -3.5), Femoral Neck T score of -2.0 (Z -1.1)). He had no history of low trauma fracture and no relevant family history. Other osteoporosis risk factors included alcohol use, smoking history, minimal weight-bearing exercise, previous vitamin D deficiency and SSRI use.

He was treated with calcium and vitamin D supplements and weekly oral bisphosphonate. Due to gastrointestinal side-effects, he was switched to intravenous zoledronate. This has resulted in stabilisation of his osteoporosis on repeat DEXA bone densitometry and no fragility fractures.

DISCUSSION

The finding of osteoporosis in young men or pre-menopausal women must direct the clinician to carefully exclude any secondary causes of osteoporosis.

Systemic mastocytosis is a rare but important cause and requires a partnership with dermatologists. It comprises a heterogenous group of mast cell proliferation disorders, with infiltration of multiple organs including skin and bone. The precise pathophysiology of osteoporosis in systemic mastocytosis is poorly understood but bone involvement is common with osteoporosis being the most common manifestation. The risk of osteoporotic fractures is high especially in men. Furthermore, back pain secondary to osteoporotic fracture may be the only presenting symptom in systemic mastocytosis3,4.

This case highlights the need to consider systemic mastocytosis as a cause of osteoporosis especially in younger men and pre-menopausal women with or without associated skin lesions. Treatment with bisphosphonates remain the first-line treatment for mastocytosis-related osteoporosis and close liaison with dermatologists is advised5,6.

References: