

Bone Impairment in Systemic Mastocytosis (SM)

Up to 70%

of patients with SM have
bone involvement¹

SM is a rare disease with frequent bone impairment, leading to bone pain, fractures and an urgent need for differential diagnosis^{1,2}

What is SM, and how does it affect bone health?

- SM is a rare clonal mast cell (MC) disease characterised by abnormal proliferation of atypical MCs in various tissues, including the bone marrow.²
- MCs regulate bone homeostasis and remodelling.¹



~70% of SM patients experience bone impairment, including osteopenia, osteoporosis, pain and fractures:¹



Localised bone pain

- Frequent pain in long bones, e.g. femur, pelvic bones²
- Sometimes pain experienced in joints and smaller bones, e.g. skull, spine, ribs and hands²



Fractures

- Fragility fractures are common (**33%–43%**)²
- Vertebral fractures are common (**30–40%**), with a higher prevalence reported in men.^{1,3} Many fractures occur in patients with T-scores ≥ 2.5 , showing that BMD is not an indicator of fracture risk¹

Bone manifestations vary between SM subtypes¹

Indolent SM (ISM) is the most common subtype of SM (~90% of patients).⁴



ISM

- Low BMD and osteopenia^{2*}
- Osteoporosis (20–38% of ISM patients)¹



Advanced SM

- Increased BMD or osteosclerosis (up to 75% of Advanced SM patients)¹

- In one survey, **32% of ISM patients** had experienced severe bone pain during the previous 60 days (n=53)⁵

Could your patients with unexplained bone-related symptoms be experiencing SM?

*Low BMD was defined as a Z-score ≤ -2.0 SD at the lumbar spine or hip.²

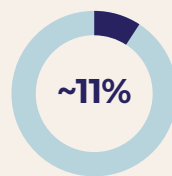
BMD=bone mineral density; ISM=indolent systemic mastocytosis; MC=mast cell; SM=systemic mastocytosis.

Bone involvement, unexplained osteoporosis in men and elevated serum tryptase could raise suspicion of SM^{2,6}

Bone involvement can be the only sign of SM^{2,6,7}



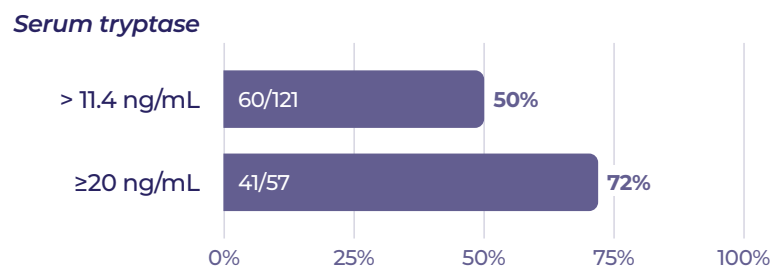
Unexplained osteoporosis was the **only** symptom in almost one-third of patients with SM⁶



Bone impairment (osteoporosis, osteosclerosis or osteoporotic fracture) was the **first** presenting symptom of ISM in ~11% of patients in a cohort study (n=157)^{2,7}

Among individuals (n=121) with unexplained osteoporosis, low BMD or fragility fractures – and **elevated serum tryptase** – the proportion diagnosed with SM increased with higher serum tryptase levels.⁶

SM diagnosis rates by serum tryptase in patients with isolated bone manifestations⁶



Cases of osteoporotic fractures in men under 50 years of age could raise suspicion of SM⁷



In 157 patients with ISM, **osteoporotic fracture** occurred in **35% of men aged under 50 years**, vs 6% of women aged under 50 years⁷



A history of mast cell mediator symptoms in cases of unexplained osteoporosis could raise suspicion of SM⁸

If you suspect that bone impairment in your patient is potentially indicative of SM, consider a serum tryptase test to investigate a diagnosis of SM^{2,6}

Patients with SM face a median of 7 years to diagnosis.^{9**} Despite bone-directed treatment, some patients with ISM may continue to experience fractures¹⁰

Osteoporosis and fragility fractures in low-risk demographics could be an indicator of ISM²

If your patient aged under 50 years presents with osteoporotic fractures, consider performing a serum tryptase test to help screen for ISM.⁷

If baseline serum tryptase levels are elevated, consider a high-sensitivity *KIT* D816V mutation assay if available or refer to known centre of excellence for SM in your region.¹¹

*Low BMD was defined as a Z-score ≤ -2.0 SD at the lumbar spine or hip.²

**Based on time of symptom onset to diagnosis of 149 patients with SM in the Mast Cell Connect registry.⁹

ISM=indolent systemic mastocytosis; SM=systemic mastocytosis.

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