The Clinical Characteristics, Diagnosis and Management of patients with Mastocytosis in a Tertiary Centre

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BACKGROUND

Mastocytosis is a rare heterogenous group of haematological neoplasms characterised by an accumulation of neoplastic mast cells in the skin alone (cutaneous mastocytosis, CM) or involving other organs (systemic mastocytosis, SM). The majority of patients with SM have an indolent disease variant whilst others present with more aggressive forms, collectively called "advanced systemic mastocytosis". With a prevalence of approximately 1 in 10,000[1], it is a very rare disease with little systematic study. Both diagnosis and management require a multi-disciplinary approach.

AIMS & OBJECTIVES

-To find out how many patients within the South Yorkshire and North Derbyshire region have Mastocytosis and which variant.
- To evaluate the investigations and ongoing management of these patients.

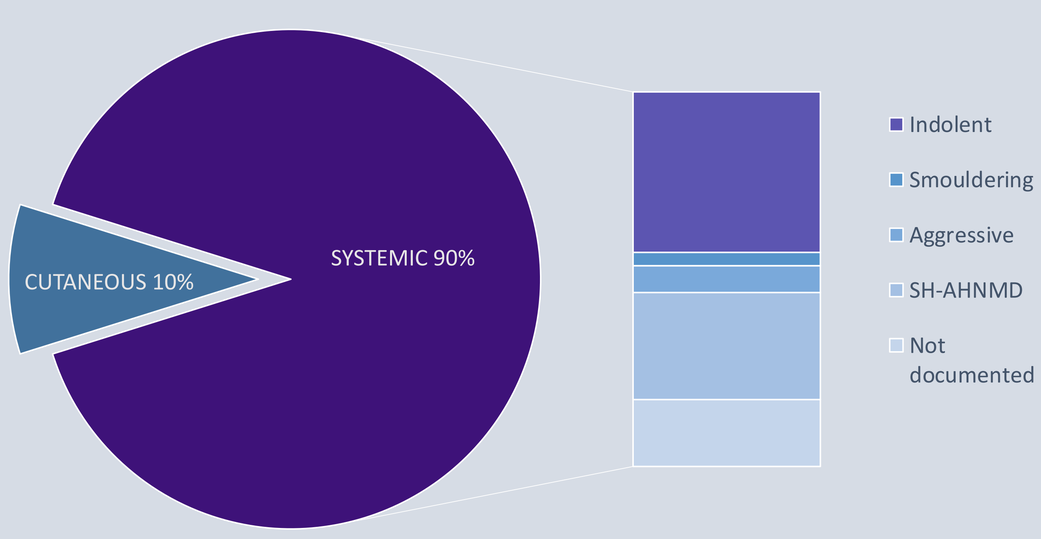
METHOD

Retrospective data for 31 patients across South Yorkshire and North Derbyshire was collected and analysed.

RESULTS

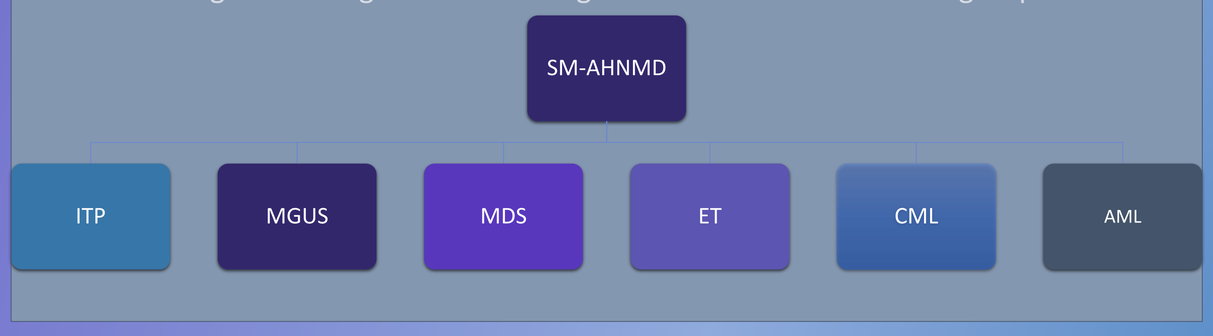
Of the 31 patients with mastocytosis, 21 were referred from district general hospitals in the region. 28 patients had SM (8 of whom are since deceased) and 3 had CM alone (see figure 1). Of the SM group, 13 cases had a prior diagnosis of CM. The average time from CM diagnosis to SM diagnosis was 5 years.

Figure 1: Variants of Mastocytosis in the Patient Population



The most common variant of SM was indolent, followed by SM with associated clonal haematological non-mast cell lineage diseases (SM-AHNMD), see figure 2. No patients had a diagnosis of mast cell leukaemia.

Figure 2: Range of Haematological diseases in SM-AHNMD group

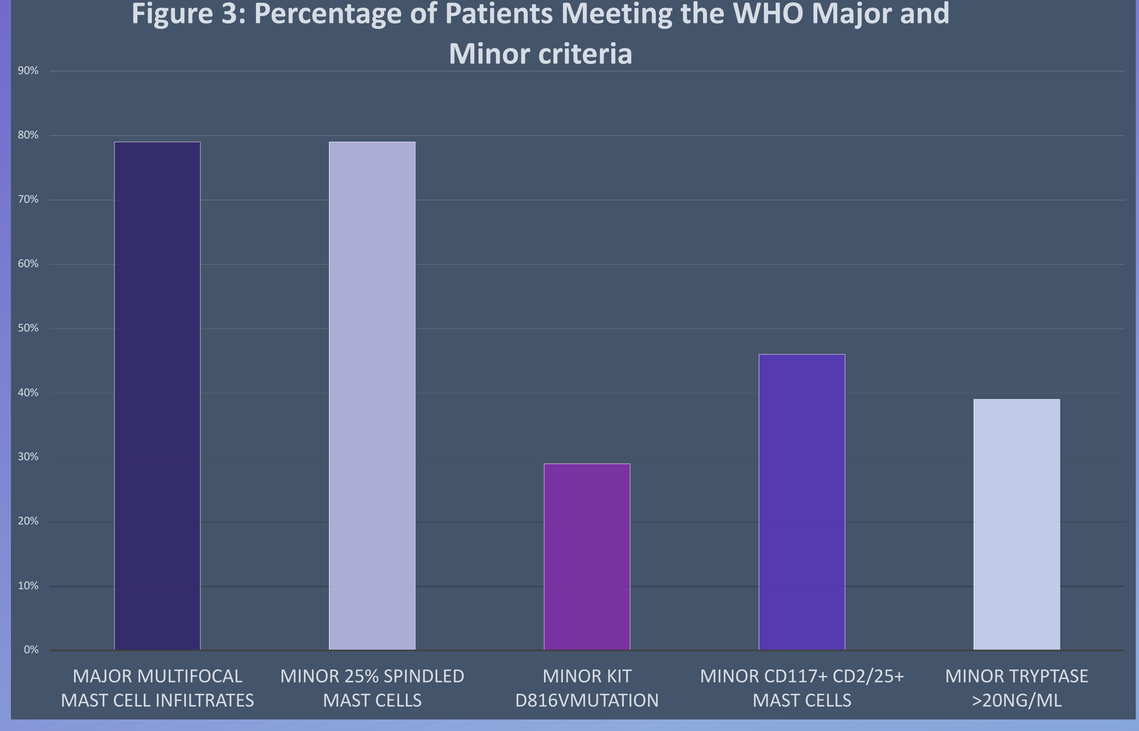


The average age at diagnosis for those with SM was 59 years compared with 17 years for CM. There was no gender bias in the SM group.

As per the WHO guidelines, major and minor criteria must be met to reach a diagnosis of SM. In our patient population, the diagnosis was reached based on one major and one minor criterion in 85% of cases and on three minor in 15%. The most common minor criterion to be fulfilled was the finding of more than 25% morphologically atypical or spindled mast cells (see figure 3).

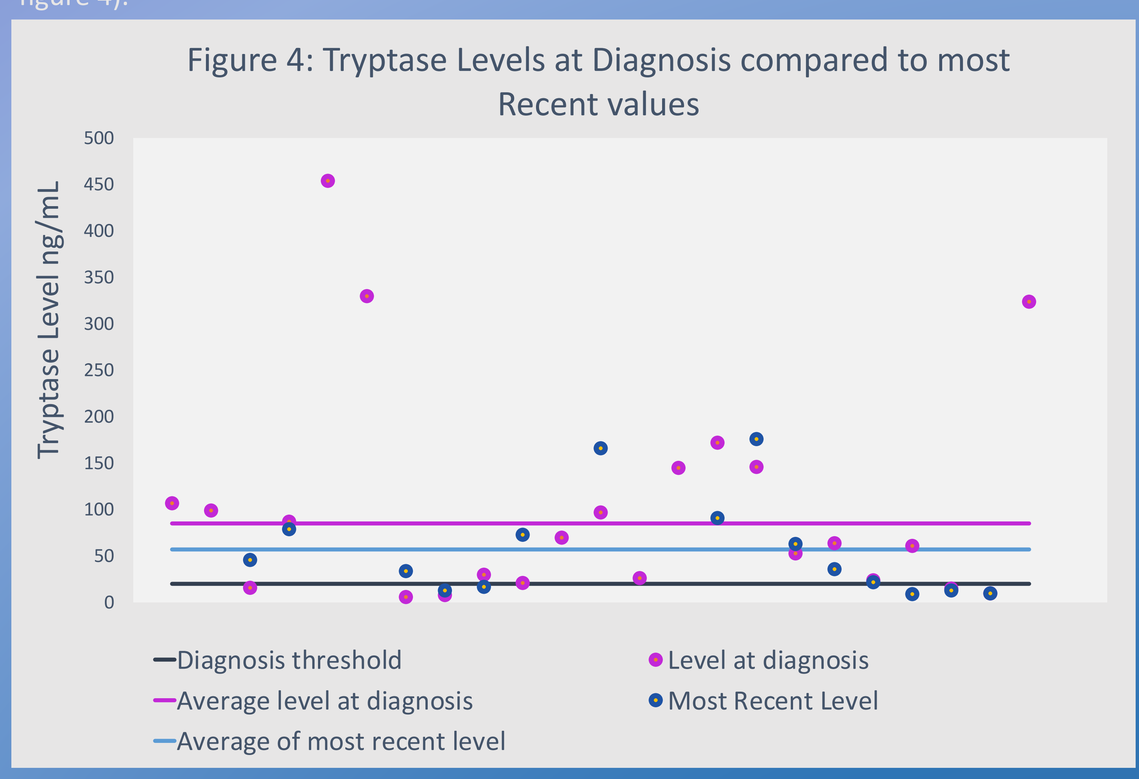
The following immunohistochemistry (IHC) was used in diagnosis; CD25 (6 cases), CD2 (13 cases) and CD117 (16 cases). There was no immunochemistry performed in 7 cases. Of the 17 cases known to have had KIT mutation analysis performed, 86% were tested on BM and 18% on a blood sample.

Figure 3: Percentage of Patients Meeting the WHO Major and Minor criteria



The tissue used to diagnose SM was bone marrow (BM) in 90% of cases. In one case the diagnosis was made on duodenal tissue as the patient was unable to tolerate a BM trephine. The average tryptase level at diagnosis in the SM patients was 85ng/dL (see figure 4).

Figure 4: Tryptase Levels at Diagnosis compared to most Recent values



Clinical findings in SM are categorised into 'B' and 'C'. Of the patient's with SM, 20 had B findings with BM hypercellularity being the most prevalent. Six patients had C findings of which weight loss was the most common. Two patients had suffered pathological fractures. On-going management of these patients requires a holistic approach and monitoring of bone density is important. Of the 28 patients with SM, 57% had a DEXA scan with 2 cases showing osteoporosis. 11 patients were receiving bisphosphonate treatment and/or vitamin D supplementation

Further investigation in SM includes identification of organomegaly in order to assess disease severity. Within the patient cohort there were 8 cases of splenomegaly and 2 cases of hepatosplenomegaly

CONCLUSION

SM has been shown to be a disease of middle age with CM manifesting in young adults. Almost half of the SM cases had a prior diagnosis of CM with a relatively short interval between each diagnosis. In-depth examination of BM was required to reach the diagnosis of SM using IHC and molecular testing in the majority of cases. Determining which mutation is present is important for risk stratification models[2]. The most common group of SM patients had indolent disease which requires monitoring of symptoms and regular assessment of bone density. Patients are best managed in centres with experience in Mastocytosis where possible[3], and a multidisciplinary approach is essential.

REFERENCES

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