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Clinical characteristics of mastocytosis in Poland based on Polish Registry of Mastocytosis

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Introduction

Mastocytosis (MS) is a clinically heterogenous, usually acquired disease of the mast cells with a survival time that depends on the time of onset. It ranges from a skin-limited to systemic disease, including indolent and more aggressive variants. The presence of the oncogenic KIT p. D816V gene somatic mutation is a crucial element in the pathogenesis. It is essential to assess the stratification risk of disease progression, and the specific clinical picture to establish an appropriate management and therapeutic strategy.

Aim

The aim of this study was to determine the clinical, biological, and molecular features of a cohort of 631 patients with mastocytosis.

Material and methods

The study comprised 631 patients included in the local registry and treated at the Allergology Department, Medical University of Gdansk, between 2012 and 2023. Mastocytosis was

diagnosed according to the WHO guidelines, which included a pathological examination of bone marrow aspirate (cytological evaluation, mast cell immunopheno-typing with assessment of CD2 and CD25 expression), identification of the presence of the activating point mutation in KIT, and the serum tryptase level. Molecular analysis of the c.2447A> T variant (KIT p. Asp816Val) in the KIT gene was performed using qPCR in bone marrow aspirate.

Results

Our cohort of patients showed female predominance (female/male ratio, 3:2), childhood onset of the disease in 32,5%. 426 adult patient with female predominance (n=281, 66%)m median age=42,2 years. Skin invovement recognized in 386 cases (91%). Diagnosis of subvariaons of MS: MPCM in 107 cases (25%), ISM in 257 (60%), BMM in 32 (7.5%), MIS in 14 (3%), ASM in 5 (1.2%) and SM-AHN in 2 (0.5%). Median tryptase level = 45.26 ug/l. Skin symptoms occured in 78% cases includin flush (80%), pruritus in 71%, blistering 12% cases.Type B symtops: hepatomegaly in 5%, spleenomegaly in 3%, lymphadenopathy in 2%,as well as malabsorption diagnosed in 1% cases and weight loss in 2%. Symptoms of allergy was reported by 203 cases (47,6%). The most common trigger of anaphylaxis was insect venom in 44,3% cases; food allergy was diagnosed in 19,2 % and drugs hipersensitivity in 23,6%.

Conclusions

This study confirms that patients with mastocytosis significantly differ in clinical presentation of the disease. The most common subvariant is indolent systemic mastocytosis and the skin is involved in more then 90% of patients. Almost 50% of patients reported anaphylactic reaction at the time of diagnosis. The prognosis of mastocytosis depends on the subvariant of the disease and the incidence of anaphylaxis with a low progression rate. The management and therapeutic strategy must Focus on the assessement possible triggers and the treatment of anaphylaxis.