
Mastocytosis is an emerging differential diagnosis in patients with more or less specific mediator-related symptoms. In some of these patients, typical skin lesions are found and the diagnosis of mastocytosis can be established. In other cases, however, skin lesions are absent, which represents a diagnostic challenge. In the light of this unmet need, we developed a diagnostic algorithm for patients with suspected mastocytosis. In adult patients with typical lesions of mastocytosis in the skin, a bone marrow (BM) biopsy should be considered, regardless of the basal serum tryptase concentration. In adults without skin lesions who suffer from mediator-related or other typical symptoms, the basal tryptase level is an important parameter. In those with a slightly increased tryptase level, additional investigations, including a sensitive KIT mutation analysis of blood leucocytes or measurement of urinary histamine metabolites, may be helpful. In adult patients in whom (i) KIT D816V is detected and/or (ii) the basal serum tryptase level is clearly
increased (>25-30 ng/ml) and/or (iii) other clinical or laboratory features suggest the presence of occult mastocytosis or another haematologic neoplasm, a BM investigation is recommended. In the absence of KIT D816V and other signs or symptoms of mastocytosis or another haematopoietic disease, no BM investigation is required, but the clinical course and tryptase levels are monitored in the follow-up. In paediatric patients, a BM investigation is usually not required, even if the tryptase level is increased. Although validation is required, it can be expected that the algorithm proposed herein will facilitate the management of patients with suspected mastocytosis and help avoid unnecessary referrals and investigations.