Mastocytosis: recent advances in defining the disease

JournalArticle (Originalarbeit in einer wissenschaftlichen Zeitschrift)

ID 4507911
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Year 2001
Title Mastocytosis: recent advances in defining the disease
Journal British Journal of Dermatology
Volume 144
Number 4
Pages / **Article-Number** 682-95
Mesh terms Humans; Infant; Mastocytosis, diagnosis, etiology, therapy; Mutation; Prognosis; Proto-Oncogene Proteins c-kit, genetics; Urticaria Pigmentosa, diagnosis
Mastocytosis is a rare disease characterized by a primary pathological increase in mast cells in different tissues, which may present in a variety of clinical patterns. Major advances have been made in recent years in the understanding of the pathogenesis of mastocytosis. This review is aimed at familiarizing dermatologists with these recent findings, and at exploring their possible implications for the diagnosis and treatment of the condition. The heterogeneous clinical presentation of mastocytosis is detailed with respect to the type of skin lesions, age at onset, family history, organ systems involved, associated haematological disorders and prognosis. Recent genetic findings also indicate different pathogenetic forms of mastocytosis, as adult patients and those with associated haematological diseases usually express activating mutations of the stem cell factor receptor c-kit, whereas most cases of childhood-onset and familial mastocytosis seem to lack these mutations. Despite the presence of c-kit mutations, patients with cutaneous lesions generally have a good prognosis, even when there is involvement of other organs. Some patients, particularly those with childhood-onset disease, experience spontaneous remission, mostly by puberty. c-kit mutations do not explain the initial cause of mastocytosis, and their prognostic significance is as yet unclarified, as is the pathogenesis in patients without the mutations. Furthermore, these novel findings have as yet not resulted in a more effective treatment of the cause of the disease, so that counselling, prevention of exposure to mast cell secretory stimuli, and symptomatic treatment remain the mainstays of current patient management.
Publisher Wiley
ISSN/ISBN 0007-0963 ; 1365-2133
edoc-URL https://edoc.unibas.ch/70890/
Full Text on edoc No;
Digital Object Identifier DOI 10.1046/j.1365-2133.2001.04123.x
PubMed ID http://www.ncbi.nlm.nih.gov/pubmed/11298525
ISI-Number WOS:000168274900002
Document type (ISI) Journal Article, Review