Mastocytosis is a heterogeneous group of disorders with different severity and prognosis, which are characterized by excessively increased numbers of mast cells in the skin or in other tissues. The diagnosis of mastocytosis is made on the basis of typical clinical skin manifestations or symptoms of mast cell mediator release and is confirmed by biopsy. In cutaneous mastocytosis, which is generally found in children, mast cell proliferation is restricted to the skin. In the majority of adults, systemic mastocytosis is diagnosed with involvement of the bone marrow or other extracutaneous organs. Evidences for severe systemic involvement are high concentrations of tryptase, soluble CD25 and/or soluble CD117 (KIT) in serum or plasma as well as the extent and density of the skin involvement. Bone marrow examinations are crucial to confirm the diagnosis of systemic mastocytosis. Sometimes specific immunophenotyping and analysis of KIT-mutations in mast cells are necessary to establish the diagnosis. The severity and form of disease is assessed by screening procedures. Indolent systemic mastocytosis is the most common form of systemic disease. Whereas the prognosis of this disease variant is good, that for systemic mastocytosis with associated non-mast cell lineage hematologic disease, aggressive mastocytosis or mast cell leukemia is less favourable. At the moment research projects look for new therapeutic approaches to improve the prognosis of these aggressive forms of systemic
mastocytosis.