DNA methylation profile in patients with indolent systemic mastocytosis.

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Introduction

The presence of oncogenic KIT somatic mutation D816V as the crucial element of pathogenesis in mastocytosis. Further epigenetic alterations are responsible for regulating the expression of genes. It is essential to identify indicators of disease progression or specific clinical picture to establish an appropriate therapeutic strategy.

Aim

To analyze the relation of mastocytosis symptoms and epigenetic changes and to identify epigenetic predictors of the disease.

Material and methods

Global DNA methylation profile analysis was performed in peripheral blood. Epigenetic interview questionnaires were completed by patients included to the study

Results

We examined the peripheral blood samples from 73 patients with ISM, and 43 healthy adult volunteers matched for gender and age. Significantly lower level of DNA hydroxymethylation (5-hmC) in blood DNA was found in patients with mastocytosis as compared to controls (0.022% vs. 0.042%, $p = 0.0001$). A significant effect of allergy on 5-mC and 5-hmC levels in patients with mastocytosis was observed: the trend of lower levels of 5-mC ($p = 0.057$) and simultaneously higher levels of 5-hmC ($p = 0.011$) were observed in patients with allergic symptoms compared to patients without allergies. 5-hmlevel was significantly associated with allergy ($p = 0.011$)

in patients with ISM, showing for higher level of 5-hmC in patients with allergy as compared to patients without allergy.

Conclusions

We observed the lower level of DNA hydroxymethylation in blood DNA in ISM. Moreover in patients with allergy we revealed the trend of lower levels of DNA methylation and simultaneously higher levels of DNA hydroxymethylation. Against patients with ISM without allergies have decreased levels of demethylation markers and therefore we can assume that the function of their mast cells is more impaired.