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The significance of the minimal residual disease detection in Ewing sarcoma and primitive neuroectodermal tumor

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Background:The Ewing sarcoma and primitive neuroectodermal tumor are bone and soft tissue malignant tumors which occur mainly in children and young adults. A reciprocal translocation resulting in fusion of the EWS gene with a member of the ETS family of transcription factors is highly specific marker for this group of tumors.
Method: During the 5 year period, from 2004 to 2008, we have analysed 24 patients, tumor tissue, peripheral blood and bone marrow for the EWS FLI 1 type 1 and type 2 . The samples were analized with reverse transcriptase polymerase chain reaction method (RT-PCR)
Results: 24 tumor samples were analysed at the time of diagnosis. 19 (79,2%) were positive for type 1, 2 (8,3%) were positive for type 2 and 3 (12,5%) were negative. At the same time, 4 (16,6%) blood samples were positive for type 1, 1 (4,2%) was positive for type 2, and 16 (66,7%) were negative for EWS-FLI1 translocation. During the intensive chemotherapy treatment (Vincristine, Doxorubicin, iphosphamide, Etoposide), all the blood and bone marrow specimens were negative. During the follow-up 2(8,3%) patients hadpositive type 1, 1(4,2%) had positive type 2, 16 (66,7%) had negative blood samples, and due the technical error 5(20,8%) specimens were excluded. In the bone marrow, type 1 were positive in 2 samples, and type 2 in 2 samples. Overall distribution of the bone marrow samples was: 5(20,8%) type 1, 2 (8,3%) type 2.
Conclusions: EWS-FLI1 type 1, isolated from tumor tissue, showed to be prognostic factor for better outcome. The bone marrow positive translocation was more valuable prognostic marker then one in the peripheral blood. During our follow –up, all patients who had positive EWS-FLI1 fusion in the bone marrow had been detected with clinical progression or recurrence in the period of 2 to 11 months. Some patients with positive EWS-FLI1in the blood, after follow-up of even several years hadn't got the recurrence of the disease. EWS-FLI1 positive fusion in the peripheral blood therefore couldn't be reliable prognostic factor for the clinical outcome, and positive bone marrow would definetely be the strong predictive factor for the recurrence or disease progression.

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