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PAX3 and PAX7 mutations in children with rhabdomyosarcoma: A 10-year histopathological and genetic study

Kourosh Goudarzi Pour, Farzaneh Jadali, Roxana Aghakhani, Mehran Arab Ahmadi, Behdad Behnam and Maliheh Khoddami
Shahid Beheshti University of Medical Sciences, Iran

Background: Rhabdomyosarcoma is the most common soft tissue sarcoma among children which has two major subtypes: Embryonal rhabdomyosarcoma (ERMS) with more frequency and better prognosis and alveolar rhabdomyosarcoma (ARMS) with aggressive behavior and less survival rate. Distinction between these subtypes is mandatory to choose proper treatment. Histopathologic study is the main method, but nowadays cytogenetic studies like PCR are also used.

Objective: The aim of this study was to evaluate the frequency of PAX3 and PAX7 mutations in children with rhabdomyosarcoma.

Method: Paraffin blocks of 34 Rhabdomyosarcoma cases below 11 years, were gathered from our pathology department during a 10-year period. Tumoral areas dissected and embedded in paraffin blocks for PCR study (Tissue dissection method). Pure tissue and pure RNA extraction, cDNA synthesis and PCR process were performed according to iNtRON biotechnology company kits' protocols. All of these cases were analyzed for PAX3 and PAX7 mutations.

Results: Among total, 32 cases were ERMS and two of them were ARMS. None of the ERMS samples was t (2; 13) or t (1; 13) positive. Also, two ARMS cases were negative for PAX3 and PAX7 mutations.

Conclusions: This study revealed lack of PAX3 and/or PAX7 mutations in ERMS. However, careful morphological evaluation cannot replace by the PCR-based t (2; 13) and t (1; 13) assay of childhood sarcomas, but can be used to make certain current histopathological diagnosis.

Biography

Kourosh Goudarzi Pour is from Shahid Beheshti University of Medical Sciences, His research interests reflect in his wide range of publications in various national and international journals.

drkgoudarzipour@yahoo.com

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