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Title: Investigation of characteristic genetic marker abnormalities in soft tissue tumours

Subject: Medical Science, Genetics, Oncology, Molecular Genetics

Keywords: Sarcomas, Oncogenes, FISH

Proposed Language for the PhD Thesis: Greek

Short description of the topic:

Mesenchymal tumors are difficult to be histologically classified (eg distinction between lipoma and liposarcoma or various microglobular tumors) and exhibit a variety of biological behavior. The above features and the fact that sarcomas occur more often than other malignancies in younger ages, emerge the need for accurate diagnosis to treat them more effectively. Genetic analysis can be a valuable tool for investigation of diagnostic markers in order to classify this heterogeneous group of tumours.

Their classification requires the combination of morphological, immunohistochemical and genetic analysis (1). In the majority of these tumors specific genetic abnormalities (mainly chromosomal aberrations) can be detected, which in many cases are the basic diagnostic feature.(2).Most of these genetic abnormalities are investigated by the application of molecular and molecular-cytogenetic techniques such as PCR, FISH and recently NGS (3). Among these, the most widely used is FISH analysis. The large amount of data obtained using this technique in soft tissue tumours makes FISH a method particularly reliable for their study. However, despite the effectiveness of this methodology, questions often arise from the use of FISH, which require further investigation. Such issues are the presence of atypical signals, discordance of findings with other techniques or even with histological analysis (4,5). Of particular scientific interest can also be the finding of new genetic markers in certain soft tissue tumours that have not been characterized so far. This category of tumors, which remain essentially unexplored, needs further investigation with the appropriate methodological approach (6).

Suggested Thesis Contribution:

Mesenchymal tumours are a group of neoplasms with enormous heterogeneity and not sufficiently genetically characterized. Their genetic analysis will lead to the emergence of new reliable diagnostic markers that can contribute to their classification and further treatment.

Main Purpose of Doctoral Thesis:

(a) Interpretation of the range and significance (biological-clinical) of atypical FISH signals in a number of archival specimens relating to soft tissue tumours.

b) Comparison of the results of FISH analysis to the findings of other methods (PCR, NGS) and their correlation with the original histologic diagnosis, as well as investigating the cause of the discrepancies that may occur.

c) The possible detection of novel molecular genetic markers in soft tissue tumours, which are not included in the hitherto known markers.

Research Methodology and Research Center where the study will be devised or done:

1) Analysis by FISH technique of tumours, mainly sarcomas, that have been previously analyzed at the Department of Genetics of Anticancer Hospital of Athens "AGIOS SAVVAS" by PCR.

2) Analysis of specimens from the Pathology Department of the General Hospital for Children “AGIA SOFIA” which have not been investigated via molecular techniques.

3) Investigation and interpretation of atypical signals derived from FISH method using different types of techniques, namely: RT-PCR, Sanger sequencing and NGS analysis

The study will be carried out at the Department of Genetics of Anticancer Hospital of Athens "AGIOS SAVVAS".

ΒΙΒΛΙΟΓΡΑΦΙΑ

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