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Abstracts

Invited Paper

What can molecular pathology do for you?

Author: David Hughes
Contact: hughesd13@sky.com

Department of Histopathology, Sheffield Teaching Hospitals NHS Trust

The term “molecular pathology” is generally applied to using the detection of abnormalities in DNA for diagnostic purposes. These abnormalities can be found in either chromosomes or genes. In the sarcoma field, use of these techniques is becoming increasingly important in clinical practice. Genetic abnormalities can cause tumours in several ways. Some individuals have “germ line” genetic abnormalities that occur in every nucleated cell in their body and predispose to the formation of tumours, for example in neurofibromatosis.

Most sarcomas are caused by “somatic” abnormalities that only effect the cells of the tumour. A number of techniques, including karyotyping, fluorescence in situ hybridisation (FISH) and the polymerase chain reaction (PCR) can be used to detect genetic abnormalities in a highly-specific way. Most sarcomas have multiple genetic abnormalities, but some have very specific and characteristic abnormalities such as the chromosomal translocations seen in Ewing’s sarcoma and synovial sarcoma and the gain of function mutations seen in gastro-intestinal stromal tumours. Detecting these abnormalities is useful in making specific diagnoses that are difficult to make by other means, such as morphology and immunohistochemistry, and in guiding therapeutic choices, for example mutational analysis in gastro-intestinal stromal tumours.

The use of these techniques is likely to increase in the future and it is possible that novel approaches such as microarrays, examining the analysis of thousands of genes in a single tumour may come into clinical practice.