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Guidelines for cytogenetic investigations in tumours

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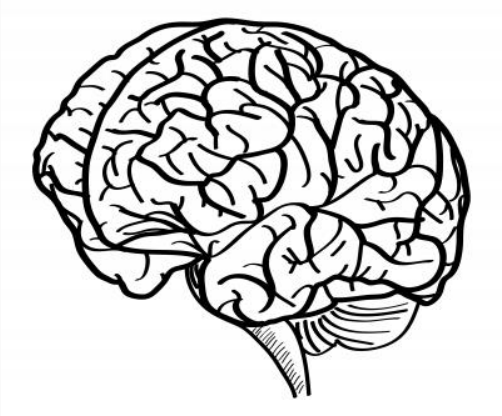

Cytogenetic and molecular genetic data are of paramount importance in the diagnosis, prognosis, and risk stratification of patients with malignant diseases. Sometimes they even directly guide the choice of therapy. Disease-specific abnormalities, particularly translocations, can provide essential information to assist the Pathologist and/or Oncologist in assigning a diagnosis. In several diseases, tumour genetics correlate strongly with clinical risk; thus, cytogenetic information may help the Oncologist counsel the patient, choose a specific treatment, and/or modulate treatment intensity. Clinical trials may involve cytogenetic classification of patients to the appropriate treatment regimens.

Currently, the provision of specific assays for acquired neoplasia-specific genomic changes varies among and within countries as a range of laboratories offer diagnostic solid tumour genetics; these may include Cytogenetic, Pathology, Haematology, and Molecular Genetics laboratories. Technical standards and general guidelines for the analysis and the report of results on most solid tumours are lacking.

To address these deficits, a tumour best practice meeting with invited tumour experts without conflict of interest was held on 23rd April 2013 in Oxford, United Kingdom. The aim was to produce professional guidelines for tumour genetic laboratories and to incorporate the standards imposed by generic European guidelines, regulatory bodies (ISO15189, 2012 Medical laboratories – requirements for quality and competence), reporting guidelines, ISCN, and acquired best practice guidelines, while taking into account the current practice in Europe.

http://www.nature.com/ejhg/journal/v24/n1/full/ejhg201535a.html