Division Molecular Genetic Epidemiology (C0400 / C050)

Head: Professor Dr. Kari Hemminki (from 10/2002)

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The new Division started to work in the Center in October 2002. Thus the listed references have been collaborations between DKFZ and Karolinska Institute, Sweden.

Work has proceeded in 2 overlapping areas, Genetic epidemiology and Molecular biology-mutations:

1) Genetic epidemiology [publications 1-22]: the goal has been to provide datasets for a reliable estimation in specific cancers of familial cancer risks, genetic and environmental components and modes of inheritance, and, additionally, to identify individuals in families for molecular studies of cancer. A family cancer dataset has been used to characterize familial effects at all main and individual cancer sites, such breast, prostate, ovary and skin, and to assess environmental effects by comparing cancer risks among spouses. Several modelling studies have been carried out to test recessive and polygenic effects in cancer. Cancer in immigrants has also been assessed.

2) Molecular biology-mutations [publications 23-27]: major effort has been directed to analysis of polymorphisms (SNPs) in cancer-related genes. Some 1000 samples have been collected, pathology assessed and prepared for PCR from familial cancers. Microsatellite mapping has been carried out on selected chromosomes, also analysed for mutations in candidate genes (p53, p16). In melanoma, the G1/S cell cycle checkpoint regulators have frequent abnormalities. Two linked polymorphisms in the 3′-untranslated region of the p16 gene were found to be prognostic factors. A tissue bank of 600 consecutive bladder cancers has been used in the analysis of homozygous deletions and mutations at the p16 gene locus and correlated with clinical markers. The role of BRAF and RAS has been elucidated in melanoma and other neoplasms.

References (published in 2002/2003 or in press)