Von: Michaela Aubele [mailto:aubele@gsf.de]
Gesendet: Donnerstag, 2. November 2006 09:18
An: Hansen, Martina, Dr.
Betreff: Wiss.Highlight-Pathol

Liebe Frau Hansen,

anbei das Highlight 2006 der Pathologie; sollten wir etwas ändern müssen (z.B. verdeutschen) geben Sie mir bitte Bescheid.
Grüße
M. Aubele

Identification of a gene causing inherited endocrine cancer syndrome

Syndromic (familial) cancer syndromes result from the germ-line transmission of a mutant gene, predisposing recipients to the development of specific forms of cancer. Cancers of the endocrine tissues are frequently syndromic, but clinical management and genetic counselling of affected families is complicated because not all of the causal genes are known.

We have used a rat model that develops multiple endocrine cancers to identify a germ line mutation in the gene encoding the p27 cell cycle regulatory protein. A similar mutation was subsequently found in patients with a unique form of endocrine neoplasia affecting the pituitary and parathyroid glands.

The identification of p27 as the gene responsible for the disease provides the first evidence that confirms the long-suspected role of p27 as a classical tumor suppressor gene. Moreover, our studies establish that p27 is closely integrated into a functional pathway served by other genes responsible for related forms of endocrine cancer syndromes.

The rat model now will offer us a unique opportunity for translational studies to identify new therapeutic targets, develop imaging modalities for endocrine tumors, and a platform to study the molecular basis of cancer development and progression in different endocrine tissues.

Proc Natl Acad Sci U S A. 2006 Oct 17;103(42):15558-63.

'Germ-line mutations in p27Kip1 cause a multiple endocrine neoplasia syndrome in rats and humans'. Pellegata et al.