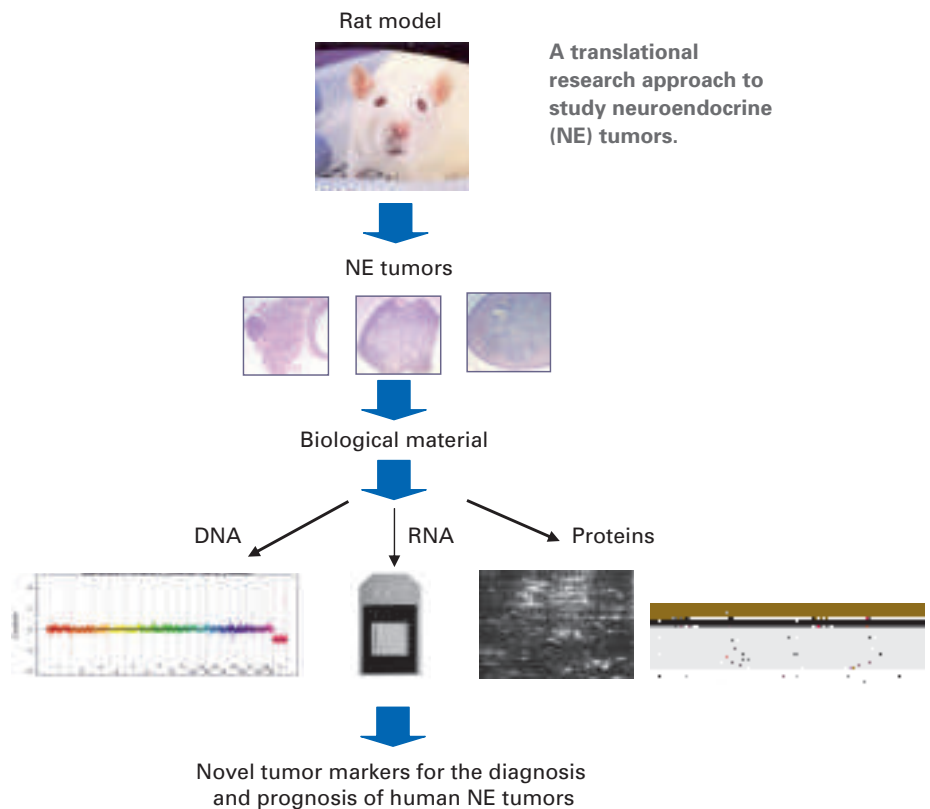


Identification of a New Mutation Enhances the Prevention of Inherited Tumours

Institute of Pathology

Multiple endocrine neoplasia (MEN) is a generic term for a rare, inherited disease, in which tumours can arise in internal hormone producing (endocrine) organs. Organs that can be affected by tumours include the thyroid gland, pituitary gland, pancreas, and adrenal gland. So far mutations have been identified in two different genes which cause predisposition to tumour development and that are inherited as autosomal dominant traits. This means that statistically the disease will be passed on to every second child if one parent is a carrier. Once it is known that the gene mutation is present in a family, tumours can be identified at an early stage through early and regular investigation and treatment is more effective. However, many MEN patients don't display mutations in either of the known genes. Scientists from the Institute of Pathology working together with colleagues from the University of Goettingen have identified a further gene – Cdkn1b – in a rat model of MEN that can lead to the development of multiple endocrine tumors also in humans. The gene codes for a protein (p27kip1) that has a regulatory function in the cell cycle and can thus affect cell division. Thus genetic counselling of affected families can now use a third gene to further support prevention of MEN. Furthermore, the rat model offers a



promising experimental system in which the development of endocrine tumours can be investigated further and potential targets for new therapy identified.

Literature:

■ Pellegata, N.S. et al.: Proc. Natl. Acad. Sci. USA 103, 15558-15563 (2006)



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