SFB 35 Colloquia in Membrane Transport

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"Pendrin, a transporter involved in hearing loss and Asthma bronchiale"

The sequencing of the human genome was a milestone in science and marked the beginning of the post-genomic era. Genetic make-up, proteomic interaction and epigenetic regulation of gene expression are important for normal cellular physiology and play key roles in both disease progression and responses to therapeutic intervention. The knowledge of the genomic make-up, however, does not by itself give insight into the function of single genes; nor does it give conclusive answers on the effect of single genes or gene products on regulatory circuits established in living cells. The challenge now and in the immediate future is to combine the genomic and epigenetic information with the functional information already known regarding gene products in their native environment. The research in our laboratory is focused on the genomics, epigenetics and functional-proteomics of pendrin. The SLC26A4 gene codes for the pendrin protein, which exchanges bicarbonate, iodide and other anions for chloride. Pendrin is mainly expressed in the thyroid gland, inner ear and kidney. The identification of the pendrin coding gene led to investigations regarding the origin of some types of hereditary deafness. 170 mutations located both in exons and introns of the pendrin gene have been linked to various human diseases. Individuals with disease-causing mutations can present distinct phenotypes, collectively named Pendred syndrome (PS, OMIM n° 274600), DFNB4 (OMIM n° 600792) or LVAS (OMIM n° 603545). PS is a rare disease causing 5-13% of childhood deafness. It is an autosomal recessive disorder characterized by sensorineural.