

Anna Greta Odendahl  
Dr. med.

## **Knowledge and Use of Apolipoprotein E Genetic Testing among Cardiologists and Neurologists**

Geboren am 18.7.1980 in Karlsruhe  
Staatsexamen am 30.6.2007 an der Universität Heidelberg

Promotionsfach: Neurologie  
Doktorvater: Herr Prof. Dr. med. Stefan Schwab

The APO E genetic test is a genetic susceptibility test that determines the risk of getting cardiovascular disease, Alzheimer's disease, or hyperlipoproteinemia type III. Several laboratories within the United States offer this test for the risk assessment of CVD and to define the subtype of hyperlipoproteinemia type III, only one laboratory is allowed to offer the test for the risk assessment of Alzheimer's disease. The laboratories only report on the genotype, they don't have special disclosing forms for the physicians or the patients on how to interpretate the test results. Cardiologists and neurologists may order the APO E genotype on their patients. Test results always have implications on the patient's health in both medical fields. No research has been done- to our knowledge- on how a genetic test with pleiotropic outcomes is applied in these medical fields and no comparison of two medical fields using the same genetic test could be found. The APO E genetic test serves as an excellent example for the present- day analysis of this matter. In our study, general awareness and order practices, physicians' disclosing and referral practices after receiving test results, and their general knowledge were assessed and compared. Almost all of the responding neurologists and half of the cardiologists know the test. It is ordered more often in the field of neurology. Physicians in both medical fields order it in order to do risk assessment and confirm a diagnosis, only neurologists order the test in response to patients' requests, even if the test is not recommended by several consensus statements in both medical fields to do risk assessment. Few physicians would inform their patients about the implications of their APO E genotype in the other medical field which often might result from a lack of knowledge. Nor were patients often referred to other specialists such as the physician from the other medical field or to a professional genetic counselling. The general knowledge among both specialists groups about alleles, genes and disease related to the APO E genetic test was insufficient. As the Human Genome Project advances, many more genetic markers will be invented and a lot of new genetic tests with pleiotropic implications will compete to be introduced in the offer catalogue of laboratories. Laboratories should adapt a new role in providing physician with the necessary information when to order these tests and how to interpretate test results since physicians might not be able to keep up with the high number of test offers in the future. Research based guidelines should further be developed to inform clinical use

of susceptibility testing. There is a strong need for a better genetic education, not only for medical students, but for physicians already in daily clinical practice in order for them to keep up with the quickly developing field of genetics and to be able to provide the patient with the best possible care. There is also a strong need for federal and health providing institutions to set control instances who apply their rules in a critical way to ensure that the clinical implications of pleiotropic genes as the apolipoprotein E gene are fully appreciated.