CORRESPONDENCE

Hereditary Nonpolyposis Colorectal Cancer (HNPCC)/Lynch Syndrome

by Dr. med. Verena Steinke, Dr. med. Christoph Engel, Prof. Dr. med. Reinhard Büttner, Prof. Dr. med. Konrad Schackert, Prof. Dr. med. Wolf H. Schmiegel, Prof. Dr. med. Peter Propping in volume 3/2013

Law on Genetic Diagnostic Testing Needs to Be Borne in Mind

The authors rightly highlight the subject of microsatellite instability analysis, which is the focus of attention especially for heads of certified colorectal centers. In this context, however, it should be pointed out for the purposes of routine clinical practice that the law on genetic diagnostic testing stipulates not only genetic counseling but also obtaining written consent from the patient before such an investigation can be initiated.

The primary referring physician is responsible for obtaining written consent. This doctor is also obliged to provide information about the nature, importance, and possible consequences of the investigation and to document this.

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Conflict of interest statement

The author declares that no conflict of interest exists.

In Reply:

Germany's Genetic Diagnostics Act (*Genetikdiagnostikgesetz*, GenDG) defines in paragraph 3 No 4 as follows: in the sense of this law (verbatim, in translation), "genetic attributes are inherited genetic information, or genetic information acquired during fertilization or in the time leading up to the birth, which originate in the human being." In the rationale for the law, the following explanation is given: "Somatic genetic changes—that is, changes that occur in only a proportion of the body's cells and as a rule not in the gametes, are not captured by Germany's Genetic Diagnostics Act because the regulatory requirement of the act assumes the particularity of genetic data. These have a predictive value beyond the tested individual, and their validity is not time limited (that is, they are valid for an entire lifespan). These characteristics do not apply to somatic genetic changes."

Microsatellite instability (MSI) or loss of mismatch repair protein (MMR) expression are found only in the tumor in HNPCC. Once this has been surgically removed, MSI and MMR loss are gone from the patient's body. The Genetic Diagnostics Act therefore does not apply.

Germany's HNPCC consortium found a pathogenic germline mutation in an MMR gene in 46% of all families that met the Amsterdam I criteria and in 54% of families that met the Amsterdam II criteria. When MSI or MMR loss were also included, 69% and 78% of families, respectively, had a pathogenic mutation in an MMR gene. The molecular pathological changes are a necessary condition, but not an adequate one for detecting a clinically relevant risk increase for a germline mutation in an MMR gene. Familial findings are of far greater relevance.

In our opinion, investigating tumors for MSI or MMR expression is not subject to the genetic diagnostics law. Patients are therefore not required to give written consent according to Germany's Genetic Diagnostics Act.

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