Thoughts on the use of molecular genetic methods in newborn screening

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The established newborn screening in Germany can currently be considered as one of the most effective measures of secondary prevention. Against the background of improved diagnostic-analytical possibilities and new therapeutic options for severe congenital diseases, new genetic screening of newborns for an increasing number of target diseases as population-based screening is rapidly demanded by physicians, patient groups or politicians.

The aim of this population-based screening is the early diagnosis and initiation of therapy in as many newborns as possible in Germany affected by one of the target diseases. The practical implementation of newborn screening is subject to the regulations of the Children's Guideline of the G-BA.

Since most of the target diseases of newborn screening, like CF, are genetically determined, this genetic screening is subject to the Genetic Diagnosis Act (GenDG; § 3 No. 9 and § 16 GenDG). According to the GenDG, these are genetic examinations for medical purposes that are systematically offered to all newborns without there necessarily being any indication of the genetic characteristics whose presence is to be clarified by the examination.

The use of molecular genetic analysis procedures in the context of newborn screening raises a number of questions with regard to the provisions of the Genetic Diagnostics Act on diagnostic genetic examinations. Exemplary are the requirements for information, consent and communication of results in the context of genetic examinations according to the GenDG. In the course of the lecture, possible differences and similarities between genetic newborn screening and individually indicated diagnostic genetic examinations will be discussed in more detail.