Thoughts on genomic newborn screening on the basis of the Genetic Diagnostics Act and the GEKO guidelines

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The established newborn screening (NGS) enables the early diagnosis and initiation of therapy for as many newborns affected by one of the target diseases as possible in Germany. Against the background of improved diagnostic-analytical possibilities and new therapeutic options for serious congenital diseases, the inclusion of new target diseases in the extended newborn screening is increasingly being discussed and demanded.

Newborn screening as genetic screening for medical purposes is legally subject to the regulations of the Genetic Diagnostics Act (GenDG). According to Section 16 (2) GenDG, genetic screening may only begin if the Genetic Diagnostics Commission has assessed the examination in a written statement. On the basis of the documents submitted to it, the GEKO examines and assesses whether the requirements of Section 16 (1) GenDG are met, whether the application concept for carrying out the test corresponds to the generally recognized state of the art in science and technology and whether the test is ethically justifiable in this sense. The examination is based on the “Guidelines of the Genetic Diagnostics Commission (GEKO) for the requirements for the performance of genetic screening tests in accordance with Section 23 (2) No. 6 GenDG”.

The concept of genomic newborn screening, which would have the technical potential to examine a large number of genetic characteristics with significance for a disease or health disorder in one screening run, raises new questions of a scientific, legal and ethical nature, considering the existing legal requirements for genetic screening.

Some thoughts on these questions, considering the Genetic Diagnostics Act and the guidelines of the GEKO, will be presented in this lecture.