Newborn screening in Germany - quality assurance in present and future

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Background: The German Society for Newborn Screening (DGNS) was founded 25 years ago in July 1996. The aim and purpose of the DGNS activities are the improvement and quality assurance of newborn screening. Therfore the screening data of the laboratories have been collected and merged since 1996. After the inclusion of newborn screening in Germany as ENS in the “Kinder-Richtlinie”, the quality parameters mentioned there were reported from the laboratories, analyzed and the results published annually in the DGNS Report. Here, the data from 2006 to 2019 are presented and interpreted in summary form.

Methods: The ENS should be continuously reviewed and evaluated according to a Public Health Action Cycle. The data required for this quality report are collected annually in a standardized way in an Access database, checked for plausibility, validated by experts, analyzed with SPSS 25. The report is published in German and English on the DGNS website.

Results: Among ca.10 million births, 7,528 neonates were identified with one of the 15 target diseases. The overall prevalence was 75 per 100 000 neonates; the most frequent disorders were congenital hypothyroidism (30 per 100 000) followed by phenylketonuria (PKU), medium-chain acyl-CoA dehydrogenase deficiency (MCAD) (10 per 100 000) and Cystic fibrosis (19 per 100 000). Of 272,205 requested follow-up screenings 80% were received. The rate of positive screening results (recall rate) declined over the observation period, from 0.90% in 2006 to 0.52% in 2019. However, this rate differs between diseases and laboratories. Influencing factors are the analytical methods (e.g. use of a 2tier procedure), but also the definition of conspicuous findings. In one out of five positive screening results, a target disorder was confirmed. A necessary treatment was started within two weeks in 79% of the children. Confirmatory diagnostic investigation was not performed or was inconclusive in 11.40% of the 51,531 neonates with positive screening results. The lost to follow-up rate after a positive screening result varied widely among the screening laboratories (1.14–37.28%).
Conclusion: The low recall rate and the early initiation of treatment in 79% of the affected children indicate that neonatal screening for metabolic and endocrine disorders in Germany is effective. The incorporation of tracking structures and a registry could further improve the quality of the program.