

Parent survey on the benefits and burdens of newborn screening

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Introduction: Newborn screening (NBS) is one of the most effective secondary prevention measures, but a continuous risk-benefit assessment is relevant to ensure that the benefits clearly outweigh the risks. While the beneficial outcome for true-positive (TP) individuals has been proven, NBS can cause stress and harm, especially in case of a false-positive (FP) or a negative (NEG) NBS results. Therefore, the aim of this survey was to investigate the acceptance and perception of NBS and possible experienced adverse effects of parents of children with NEG, FP or TP NBS results.

Methods: A prospective, monocentric questionnaire study was conducted at the NBS laboratory of Heidelberg University Hospital (UKHD). Parents whose child received NBS, including the NGS2025 pilot study, at the UKHD NBS laboratory in 2024 or 2025 were eligible for inclusion. They were assigned to three subgroups (NEG, FP, TP) and contacted by mail 3-6 months after the NBS was performed.

Results: For 2024, 739 questionnaires of 2477 contacted families were included in the study (NEG: 519/1800, FP: 170/578, TP: 50/99). Nearly all participants (96%) remembered NBS, but only 67% were aware of it before birth. The informed consent for NBS was perceived as not (75%) or mild (89%) burdensome. The benefit of NBS was rated very high (63%) or high (28%), with higher rating by TP parents. The majority of participants would consent again (94%) and recommend participation (91%), with a trend of lower consensus among FP parents. The follow-up examinations were perceived overall burdensome, with higher burden for parents than for children. Free-text comments frequently criticized insufficient information during the IC process and high uncertainty following an FP result.

Conclusion and Outlook: Parents have shown a positive attitude towards NBS. A main criticism was the lack of sufficient information prior NBS, which could be overcome by providing prenatal information. There was a high burden after an FP result, underlining the unmet need to improve

communication in this case. With this aim, the German legal regulations for communicating NBS results changed on 1 January 2025. Data collection and analysis for this survey for 2025 is ongoing.