# Abstract – DGNS Annual Meeting

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## Title

Newborn Screening for Sickle Cell Disease in Germany: A Five-Year Review

## Introduction and Objective

Sickle cell disease (SCD) is the most common severe haemoglobinopathy in children with a migration background in Germany. Following four pilot studies and health insurance data analyses demonstrating a birth prevalence of approximately 1:5,000–10,000, the Federal Joint Committee (G-BA) resolved on 20 November 2020 to introduce universal newborn screening (NBS) for SCD. Screening commenced nationwide in October 2021.

## Methods

Data from the national screening reports (DGNS) for 2021–2023 were analysed. For 2024, the number of live births (677,117) served as a surrogate for the number of screened newborns, as coverage consistently exceeds 99%. The denominator for 2021 was restricted to approximately 25% of annual screened newborns (ca. 197,000) to account for the October start of SCD screening. Mean birth prevalence was calculated across all four cohorts.

## Results

A total of more than 400 newborns with SCD were identified across approximately 2.3 million screened newborns: 28 in Q4/2021, 137 in 2022 (prevalence 1:5,349), 129 in 2023 (prevalence 1:5,329), and a preliminary 109 in 2024. The mean birth prevalence across all four cohorts is approximately 1:5,700, consistent with pilot study projections. The predominant genotype was SCD-SS (2022: 68.6%; 2023: 55.8%), followed by SCD-SC (2022: 20%; 2023: 16.3%). Notably, a relevant

proportion of confirmed cases lacked complete follow-up data in the national reports (2022: 18/137; 2023: 9/129 cases without full confirmation records).

## Conclusion

Five years after the G-BA decision, NBS for SCD in Germany is functioning effectively in quantitative and technical terms, with approximately 110–140 affected children identified annually. The critical unresolved challenge is the insufficient enrolment of NBS-identified patients into the GPOH Sickle Cell Disease Registry. Without prospective long-term documentation of clinical outcomes, the ultimate benefit of NBS for SCD cannot be demonstrated. Ensuring complete registry enrolment must be the primary objective for the years ahead.

## References


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