

Session: Update - Newborn Screening for Cystic Fibrosis

Title: Parental experience with positive newborn screening for cystic fibrosis – Results from a survey in Bavaria and Southwest Germany

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Background: Cystic Fibrosis (CF) has been added to the national German newborn screening (NBS) panel in 2016. According to the national regulation of the Joint Federal Committee (G-BA) parents are informed about a positive NBS result by the sender of the NBS sample and consecutively have to arrange an appointment for confirmatory diagnostics (sweat test) in a hospital of their choice. In contrast, other NBS systems (e.g. Switzerland [1]) employ a centralized approach, where parents are informed about positive CF-NBS directly by the CF center offering confirmatory testing.

Methods: In a questionnaire-based survey parents of children with positive CF-NBS in Bavaria and Southwest Germany (Heidelberg NBS center) were interviewed. This allowed for comparison of parental perceptions in a system with decentralized organization of confirmatory testing in Southwest Germany according to the G-BA and a more centralized organization in Bavaria [2].

Results: Questionnaires were sent to 343 families whose children had positive CF-NBS results at the Heidelberg NBS center between October 2016 and January 2020 and 192 families with confirmatory testing until August 2018 in Bavaria. Of these in Heidelberg 178 (51.9%) and in Bavaria 105 (54.7%) were returned.

Information provided about NBS at birth was satisfying for 78% (Heidelberg, HD) and 71.4% (Bavaria, BY) of parents. Although required by the national regulation, only a minority of parents received this information by a physician. 52.9% (HD) and 65.7% (BY) of parents were satisfied with the information given about the positive CF-NBS result. Most parents expressed negative emotions at this point (HD 89.5%, BY 86.4%). Parents of children with false-positive CF-NBS remained anxious in 17% (HD) and 15.9% (BY) after confirmatory diagnostics. This percentage was significantly higher if CF was confirmed (HD 66.7%, BY 69.2%). 70.7% (HD) and

77.1% (BY) of parents were satisfied with the information given during confirmatory diagnostics. Waiting time for sweat testing was >3 days for 56.1% (HD) and 35% (BY) of families. Parents who waited for a maximum of 3 days were significantly more satisfied (HD 76.4%, BY 88.1%). Overall, 91.4% (HD) and 83.5% (BY) of parents were satisfied to have participated in CF-NBS.

Conclusion: CF-NBS stands in high regard with parents. Its smooth organization, timely initiation of confirmatory testing, and professional communication are of utmost importance to limit parental anxiety. A more centralized system of confirmatory diagnostics seems to be advantageous in several of these aspects.

[1] C.S. Rueegg, J. Barben, G.M. Hafen, A. Moeller, M. Jurca, R. Fingerhut, C.E. Kuehni, G. Swiss Cystic Fibrosis Screening, Newborn screening for cystic fibrosis - The parent perspective *Journal of cystic fibrosis : official journal of the European Cystic Fibrosis Society* (2016) 15: 443-451.

[2] I. Brockow, U. Nennstiel, Parents' experience with positive newborn screening results for cystic fibrosis *Eur J Pediatr* (2019) 178(6): 803-809.