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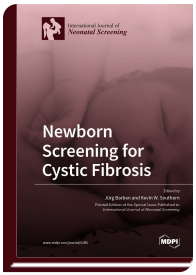
Newborn Screening for Cystic Fibrosis



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The introduction and widespread implementation of newborn bloodspot screening (NBS) for cystic fibrosis (CF) has offered earlier diagnosis and better outcomes for children with CF in many countries of the world. It represents a paradigm shift in the diagnostic pathway for these families. In contrast to a clinical diagnosis, infants are now referred for diagnostic testing after a positive NBS result. The introduction of NBS has enabled the provision of early appropriate treatment to prevent the manifestations of the disease. In the near future, early diagnosis will facilitate the prompt use of new CFTR modulator therapies that correct the basic underlying molecular defect. NBS for CF has been a global success but continues to raise questions with many varied approaches and the development of new technologies, in particular the ability to undertake extensive gene examination. Which is the best protocol to achieve high sensitivity and specificity, and how to evaluate and manage infants with inconclusive diagnosis are all subjects of ongoing discussion. It is also open to question: what is the best approach to informing and counselling the parents about a positive or inconclusive NBS result? These questions are not easy to answer and require a balanced solution that reflects the local health care system and may appropriately result in different answers around the globe. The articles in this book try to answer these questions and give an overview of the current state of knowledge in NBS for CF.

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