

CF-screening positiv – what´s next? Diagnostics und therapie for infants with cystic fibrosis

*Jutta Hammermann, UniversitätsMukoviszidoseCentrum „Christiane Herzog“,
Universitätsklinikum Carl Gustav Carus, Klinik und Poliklinik für Kinder- und Jugendmedizin,
TU Dresden*

With introducing newborn screening for cystic fibrosis (CF) in Germany the diagnose of cystic fibrosis can be made much earlier in life in most cases.

Positive effects of this early diagnosis for the patients and their families are early intervention and preventive care after detailed diagnostics and education. Specialized cystic fibrosis care in certificated CF-centers leads to routinely standardized care and possibility of early detection and therapy of changes in health status. Early approach to new therapies can clearly improve quality of life and life expectancy.

After positive CF-screening already sweat testing for confirmation or exclusion of the diagnose CF should be performed in a specialized CF-center, to be able to give clear and profound information about the results and their consequences for further life.

In the case of confirmed diagnose of CF especially in the first time next to diagnostics a close contact to the patients and their families, teaching, social and psychological support is extremely important. Therefor CF-care does not only mean consultation of physicians and medical diagnostics, but team work of specialized CF-caregivers like nurses, physiotherapists, social workers, psychologists and dieticians in addition.

National and international standards of care for CF have been established, which have to be followed, but also have to be individualized for the special needs of every patient.

In some cases results of CF-screening and confirmation diagnostics still leave the diagnose unclear. Those cases have been characterized as CFSPID: Cystic Fibrosis Screen Positive Inconclusive Diagnose. Those children and families need specialized care and follow up as well.

In summary CF-screening is the first step to improve quality of life and life expectancy for CF-patients. The success of newborn screening depends on valuated confirmation of diagnose, specialized and standardized care and after all ongoing evaluation of processes through patient registries with consequences according to the results.