Care structures for SMA Newborn Screening

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Introduction: 5q-associated spinal muscular atrophy is one of the most common causes of severe disability and early death in childhood. Molecularly, most cases are due to a homozygous deletion in exon 7 of the SMN1 gene. In the most severe form, SMA type I, the majority of untreated children die within the first two years of life. There are now 3 different causal therapies available, all of which are based on providing sufficient SMN protein. When therapy is initiated in asymptomatic patients, the course can be dramatically improved, and in some cases the onset of symptoms can be prevented completely(1).

Results: to demonstrate the feasibility and requirements for successful newborn screening pilot projects were launched in parts of Germany beginning in 2018 (2, 3). Between January 2018 and January 2020, a total of 297163 newborns were screened. Homozygous deletions in the SMN1 gene were found in 43 infants. All suspected findings were confirmed in a second genetic analysis(4).

Based on the experience in the pilot projects and the data in the literature, which showed clear indications for the necessity of an early therapy, the GbA decided at the end of 2020 to include SMA screening in the general newborn screening.

However, some preconditions are mandatory to ensure a successful nationwide implementation:

1) A close cooperation with the obstetric clinics to ensure that as many newborns as possible are covered by SMA screening. The pilot projects showed a high willingness of 87% for optional SMA-screening.
2) Availability of specialized neuromuscular centers to give advice to parents and to treat the childrens in case of suspicious findings. A list of neuromuscular centers which have been selected by the German neuropediatric Society and patient advocacy group (DGM) is available to the screening laboratories. The selected centers have neuropediatric and physiotherapeutic expertise in the field of SMA and are able to provide psychological care if necessary.
3) Expert centers have to take care that genetic confirmation of the diagnosis, including determination of the SMN2 copy is obtained within a few working days.
4) Payment for treatment must be guaranteed by the health insurance company within a few days
5) a tracking system must ensure that all children with suspicious findings can actually be guided to an adequate therapy.

Conclusion: SMA newborn screening is feasable and will dramatically change the prognosis of spinal muscular atrophy. However, close cooperation between obstetricians, screening laboratories, geneticists, pediatric neurologists and insurance companies is indispensable in the best interest of the children.

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