CORRESPONDENCE

One-Year Evaluation of a Neonatal Screening Program for Cystic Fibrosis in Switzerland

by Dr. phil. Corina S. Rueegg, Prof. Dr. med. Claudia E. Kuehni, Prof. Dr. phil. nat. Sabina Gallati, Prof. Dr. med. Matthias Baumgartner, Dr. phil. nat. Toni Torresani, PD Dr. med. Juerg Barben in volume 20/2013

Problems

All the problems associated with screening that would need clarification before screening is introduced can be deduced from the article by Rueegg et al. (1).

The first screening step will result in psychological stress for the parents because of numerous false-positive findings that will require further investigation. Furthermore, there are those false-positive findings which cannot be excluded by further diagnostics, due to the nature of the genetic findings and the disease itself—with subsequent de facto unnecessary treatment and impaired quality of life (2, 3). For it is known that among those being positive in the genetic a nalysis (CFTR variants) as well as in the sweat test, some will be affected by the effects not at all or only later in life, and these effects then can also be mild in some cases (2, 3).

The biggest problem with introducing screening, however, is one that is regularly left out in German discussion—in spite of the German Medical Association's hesitant prioritization committee: There have to be economical limits concerning medical interventions, including screenings.

According to Rueegg et al. and other publications (2, 3), the incidence of cystic fibrosis is 1 in 3500 neonates; this means 200 children per year in Germany with its 700 000 births. If the 20-year mortality in cystic fibrosis is 25%—as, for example, found in Dijk et al. (4) and Scott et al. (3)—and owing to screening this can be reduced by 50%, this means that mortality and lung replacement (according to [2], but not achieved according to [1]) would be reduced in a maximum of 25 children in Germany per year. For their sake a very large program requiring the highest quality (and expenditure) would be conducted—one which also includes undesirable side effects.

To counter any accusation of heartlessness on my part I would like to mention: Priorities have to be set, if a healthcare system is to be maintained that remains open to everybody.

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Conflict of interest statement
The author declares that no conflict of interest exists.

In Reply:

In addition to the undisputable medical benefits (1) it was the economic considerations in particular that have persuaded many health authorities to introduce neonatal screening for cystic fibrosis (CF). In England, the cost savings for drug consumption alone were calculated to be considerably higher than the costs of neonatal screening (2). The authors of this study therefore request that screening should be introduced internationally, since the benefits are important not only for the children and their parents, but also economically.

In Switzerland, screening costs are reimbursed at 5 Swiss Francs (SFr.) per child. For 83 198 screened children and 31 diagnosed cases of CF in 2011, this equates to 13 419 SFr. per treatable CF patient. Economically, this is a small amount if one assumes that due to neonatal CF screening early therapy can be initiated, and prevent unnecessary visits to doctors, expensive antibiotic therapies, and cost intensive hospital admissions (reimbursed at a cost of 21 186 SFr. for 14 days) in case of delayed clinical diagnosis.

For each screening procedure, the aim is to achieve high sensitivity while keeping the number of false-positive cases as low as possible. In neonatal CF screening, the cut-off for the initial test (immunoreactive trypsinogen) and the selection of CF mutations that are being screened for are the deciding factors. The more CF mutations are included in the screening, the more often asymptomatic CF carriers will be detected, or children with mild variants of the disease, who may not need treatment for many years. Therefore, it has to be the aim of each neonatal CF screening program to avoid capturing children with mild forms and to keep the number of asymptomatic carriers low. In Switzerland this is done by screening only for the seven most common mutations, and only looking for further mutations in children with a positive sweat test. This approach was evaluated carefully before the start of the screening program (3). Furthermore, according to our prospective study (5) and a French study (4) the parents' fears are minimal and short-lived, if a sweat test has to be undertaken subsequent to a false-positive screening result.

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