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Title: Comparison of nanoduct versus macroduct sweat test for the diagnosis of cystic fibrosis in the newborn screening programme in Switzerland

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Body: Background: Newborn screening (NBS) for cystic fibrosis (CF), based on immuno-reactive trypsinogen (IRT) and 7 CFTR mutations, was introduced in Switzerland on January 1st 2011. In the pilot phase, we compared the performance of two sweat test methods for diagnosing CF in the NBS. Methods: All children with a positive screening result were referred to a CF center for confirmatory (diagnostic) testing with: a) the Nanoduct sweat test (conductivity); and b) the Macroduct test (chloride). If sweat test results were positive, borderline or inconclusive, an extensive DNA analysis was performed. Results: Within one year, 84 children were screened positive. In 30 children the diagnosis of CF could be confirmed, 53 had normal investigations, and 1 child was not yet fully investigated. All details of the investigations were available for 76 children. The children were seen in a CF center at a median age of 24 days. The Macroduct was attempted in 64 children, the Nanoduct in 71 children. A reliable test result was available in 66% (42/64) for the Macroduct and 79% (56/71) for the Nanoduct. In 37 children both sweat tests could be performed; in 19 only the Nanoduct and in 5 only the Macroduct was feasible. In 8 children none of the two sweat tests could be performed, and confirmation or exclusion of CF was based on extensive DNA analysis alone. Conclusions: In this pilot study, the Nanoduct sweat test showed a better feasibility for use in newborns compared to the Macroduct test, mainly because it needs a lower sweat volume. Analysis of a larger dataset will allow to compare sensitivity and specificity of the two tests for the final CF diagnosis.