Qualitative study on parental views on the most acceptable way to be told their child has a probable diagnosis for cystic fibrosis following neonatal screening

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Objectives: This study reviews parental views on how they were informed of the probable CF diagnosis, following neonatal screening, and explores which method parent’s found most acceptable.

Methods: Retrospective questionnaires were used to find parents’ views on how they were told that their child had a probable diagnosis of CF, who told them initially, were they told at home or by the GP followed by hospital, how acceptable was the mode of information sharing?

Results: 40 parents of children with CF were studied. Most parents (9 out of 12) who had been informed by their GP recall receiving inaccurate information about the condition. All parents (28 out of 28) who had received a home visit to be told about the CF diagnosis felt that the information received had been accurate and found it easier to accept.

Conclusion: Our qualitative study confirms that parents who were given accurate information by CF professionals during a home visit found this an acceptable means of being informed of the probable diagnosis of CF. Parents reported less negative recollections of that time than parents who were informed by their GP and met CF professionals in hospital. We consider that this is a superior method of sharing the information about the diagnosis of CF.

Cystic fibrosis (CF) prevalence derived from CF newborn screening (CFNBS) in the Czech Republic: comparison of previous epidemiological and current CFNBS-based disease prevalence data

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Objectives: More than 30 years old epidemiological estimates at birth CF prevalence are approximately 1 in 3,000 in the Czech (CZ) Republic. Since the introduction of the CZ CFNBS in 10/2009 we have documented substantially lower disease prevalence of 1 in 6,330. Based on the outcomes of the nationwide IRT/DNA/IRT protocol we assessed potential sources of the bias.

Methods: Data from the CZ Institute of Health Information and Statistics and CZ CF registry were utilized. Infants with equivocal diagnoses of CF were excluded.

Results: A total of 367,114 newborns underwent CFNBS (8/2009–12/2012). Within this period 58 patients were diagnosed (of which 52 via IRT/DNA/IRT) with classic CF leading to prevalence 1 in 6,330 in the CZ Republic.

Conclusion: The current prevalence based on CFNBS in the CZ Republic is markedly lower than the oral data. This could be due to a. possible false negativity of CFNBS; b. impact of prenatal diagnosis and pre-implantation genetic diagnosis (PGD) and increasing birth-rate of non-European immigrants.

Within the study period 15 pregnancies were terminated due to CF in the fetus resulting in modified prevalence of 1 in 5,029. Considering the existence of PGD for CF (statistics are unavailable) we can presume slightly higher population risk for CF. The historical higher prevalence could be due to regional bias (1 in 2,700 in Central Bohemia, 1 in 3,300 in the former Czechoslovakia) and/or smaller numbers. Finally, current CF prevalence data are in accordance with those from other European CFNBS programs.

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