24 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.

25 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 original data. This could be due to
a. possible false negativity of CFNBS;
b. impact of prenatal diagnosis and pre-implantation genetic diagnosis (PGD) and
c. 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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26 Risk factors for poor outcomes in cystic fibrosis newborns diagnosed by neonatal screening in Italy: years 2009–2011

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Objectives: To identify risk factors associated with poor clinical outcomes in CF diagnosed by NBS in Italy.
Methods: Our survey include CF infants born and diagnosed in the years 2009–2010–2011 recruited in the 16 Italian CF care Centres of Regions where CFNBS is performed. They were evaluated according to: gender, ethnicity, genotype, pancreas status, meconium ileus (MI), age at diagnosis and longitudinal outcomes: weight, height, Ps infection, pulmonary exacerbations at age of 6 mo and 1–2–3 y.
This is a longitudinal cohort and nested case–control study: cases were infants presenting at least ONE of the following variables (primary outcome): stunting, wasting, Ps chronic infection at age of 1 y; controls were infants matched for age not presenting the primary outcome.
Results: We have collected data from 399 children. The total rate of cases was 20%: 19.8% among patients with classical CF diagnosed by screening, 1.8% among "equivocal" CF, 40% among MI, 33% among CF diagnosed by symptoms and 40% among CFNBS false negatives.
In order to identify risks factors associated to adverse outcomes we considered 34 cases and 162 controls. Not statistically significant associations with: CF Centre, gender, genotype, ethnicity, nationality, occupation of parents, age at diagnosis, age of onset of physiotherapy was found. Significant association was found with pancreatic insufficiency (OR = 3.065, 95% CI 1.02–9.18, $P < 0.05$).
Subjects presenting the primary outcome have length or weight at age of 2 and 3 y significantly lower than controls.
Conclusion: Clinicians should pay close attention to nutritional status of infants during the first year of life.

27 Infants with an equivocal diagnosis at the Oslo CF centre following newborn screening for CF in Norway

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Objectives: Norway started newborn screening (NBS) for CF on March 1st 2012 with an IRT/DNA protocol. Infants with a positive NBS for CF are defined as either having significantly increased immunoreactive trypsinogen (IRT) (cut off value 99.5 p) and two identified CFTR mutations or markedly elevated IRT levels. The aim of the study was to assess the frequency of an equivocal CF diagnosis after positive NBS with corresponding CFTR mutations and to assess whether sweat chloride (SC) levels and pancreatic status could clarify the diagnosis.
Methods: NBS positive infants with an equivocal diagnosis were registered from March 1st 2012 until December 31st 2013. SC was measured initially and after six months age. Pancreatic status was determined by quantifying fecal elastase.
Results: 22 infants were followed up at the Oslo CF centre in this period, and 10 infants (45%) were heterozygous to indeterminate CFTR mutations (R117 H with different poly T tract status). At follow up two infants (n1 and n7) with initial indeterminate SC levels had positive SC levels (⩾60 mmol/L) (Table 1).

Table 1.

Infant	Poly T tract status	Initial SC (mmol/L)	Follow up SC (mmol/L)	Fecal elastase (μg/g)
n1	5T/9T	53 and 54	62	442
n7	7T/9T	37 and 40	62	>500

Eight infants had negative (<30 mmol/L) or indeterminate (30–59 mmol/L) SC levels. Fecal elastase was normal in all infants.
Conclusion: At the Oslo CF centre the rate of NBS positive individuals with an initial equivocal diagnosis was 45%. All of these had at least one R117H CFTR mutation and were pancreatic sufficient. The SC levels could not clarify the diagnosis in all individuals. SC levels in two infants turned positive in the follow up.