51 Impact on physiotherapy workload at the Leeds Regional Paediatric CF Unit following the first full year of the UK National CF Screening Programme

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There is increasing consensus that early identification of CF by neonatal screening results in improved outcomes. Infants require early intensive specialist input if the benefits of early diagnosis are to be realised.

The Leeds Regional Paediatric CF Unit (LRPCFU) has screened babies born in Leeds for CF since 1975. From the beginning of 2007 all babies born in Yorkshire have been screened for CF using the National CF Screening Protocol (NCFSP). The NCFSP is prescriptive on how, and by whom, the diagnosis should be delivered, and where children should ideally receive care. Specialist Centres and experienced staff are central to this process. A typical UK District General Hospital expects no more than 1 diagnosis a year.

We present a retrospective audit of the impact of these changes on the workload of the LRPCFU. In 2007 16 newly diagnosed infants (8 Leeds & 8 non-Leeds) were referred to our unit compared to a median per year of 3 (range 0–5) from 1998 to 2006.

Changes in referral pathways have been associated with an increased number of babies with Meconium Ileus being cared for at the LRPCFU, 7 in 2007 vs a previous maximum of 2 per year.

For the 16 infants there were 24 inpatient admissions contrasting to a median of 2 per year from 1998–2006. Inpatient physiotherapy contacts during these admissions was 496 in 2007 vs a previous median per year of 44 (1998–2006). Outpatient physiotherapy contacts also increased, 76 vs 12.

The treatment of a newly diagnosed baby and education of their family requires considerable time from all members of the CF team. If the increase seen by our unit in the first year of NCFSP is predictive of the future then there must be appropriate targeted funding to ensure that these children receive optimal care and outcome.

52 Clinical outcome and CF care costs justify the need of newborn screening

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Within the Czech pilot CF newborn screening (NBS) project (2/2005–11/2006) 11 neonates fulfilling the criteria of CF were diagnosed. 9 of them are followed up at the Prague CF centre, 1 had meconium ileus (MI). The control group comprises of 18 non-screened patients born 2004–2007, diagnosed according to clinical symptoms. We compared anthropometric data and costs of care of 8 NBS patients and 16 patients from the control group (C) using F-test and t-test. MI patients were excluded.

The median age at diagnosis was 0.1 year in NBS patients and 1 year (0.2–2.8 years) in the control group. The period of follow up and care did not significantly differ in either group (median 1.5 years versus 1 year).

The total average cost of care in the NBS group was 37,659 CZK (30,754–57,251 CZK), which is an average of 29,691 CZK (19,217–40,410 CZK) per year. In the control group the total average cost of care was 125,258 CZK (18,124–622,020 CZK) which is an average of 84,760 CZK (21,480–806,482 CZK) per year. There was a much greater variation of cost of care in the control group and the cost of care per patient was much higher (total $p = 0.006$, per year $p = 0.011$). The patients did not significantly differ in body length: NBS -0.5 ± 0.6 SD versus C -0.9 ± 0.9 SD. However NBS patients had a better nutritional status at the latest examination: body weight NBS -0.2 ± 0.6 SD versus C -1.4 ± 1.4 SD ($p = 0.03$) and BMI 0.2 ± 0.6 vs -0.7 ± 1 SD ($p = 0.03$).

Patients diagnosed with NBS have a better nutritional status and lower costs of care than patients diagnosed according to clinical symptoms during the same period.

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53 Results of DNA-based cystic fibrosis newborn screening proficiency testing

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As newborn screening (NBS) for cystic fibrosis (CF) has expanded during recent years, many laboratories screening for CF include both biochemical and molecular testing with DNA analyses of CFTR mutations. With the growing need for a proficiency testing (PT) program for molecular testing, the Centers for Disease Control have created a repository of CFTR mutations using human blood specimens. PT panels consisting of 5 to 7 coded specimens from adult CF or CF carrier donors were sent quarterly to NBS laboratories worldwide with instructions to report the genotype, method used, and the presumptive clinical assessment of each specimen. As of 1/1/2008, there were XX participants using 16 different methods for DNA analysis. Due to the variety in mutation panels, laboratories were evaluated based on clinical assessments. Mutations that were not included in a panel were not evaluated. Overall, the laboratories performed well. Data compiled from 4 quarters showed that there were 336 (69%) correct assessments, 13 (3%) incorrect clinical assessments, and 8 (2%) amplification failures. There were 128 (26%) specimens that were not evaluated. Most recently, we completed PT assessment with blood spots from a homozygous F508del donor, and an F508del carrier spiked with IRT in an actual simulation of a NBS test. Developing a PT program for DNA-based testing is complicated by the number of methods and different alleles each laboratory chooses to test. Although molecular testing for CF may be complex, PT can monitor the laboratory's ability to test multiple alleles, including uncommon alleles, the limitations of various assays, and the different algorithms used for screening.

54 Hepatic enzyme changes in cystic fibrosis (CF) newborn screened babies: incidence, risk factors and evolution

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Objectives: Observe biochemical hepatic enzymes evolution of the CF newborn diagnosed by screening (starting 07/01/03), from diagnosis until 06/30/07, admitted to this Center; compare clinical manifestations, in the first year of age and follow the evolution of the above mentioned hepatic enzymes.

Methods: Analysis of medical records of 59 patients diagnosed as CF in 4 years. Outcomes recorded at the time of diagnosis: age, sex, gestational age, birth weight, nutritional status, mutations, meconium ileum, pancreatic insufficiency, neonatal jaundice, respiratory manifestations, edema, size of liver, level of serum albumin, hemoglobin, aspartate aminotransferase (AST), alanine aminotransferase (ALT), gama-glutamyl transferase (GGT) and alkaline phosphatase (ALP). For comparative analysis between patients, 5 were excluded (lack of information).

Results: 31/59 (52.5%) children were male. 6.5% were premies and 12% had low birth weight. Median age of diagnosis was 1.3 months. 64.4% presented Δ F508 mutation. 13.8% had meconium ileum. Respiratory manifestations were present in 12%. 23.5% were malnourished: moderate to severe ($\leqslant -2$ SD, Z score). 43/59 (73%) had pancreatic insufficiency. Almost half of them had hipoalbuminemia at the diagnosis. 39/54 (72%) presented at least one biochemical elevation. This was observed in ALP 46% (25/54) and in GGT 33% (18/54). By the end of one year of age, 27/39 (69%) had normal hepatic enzymes.

Conclusions: Hepatic enzymes were elevated in 72% of CF children diagnosed by neonatal screening, and these enzymes normalized in 69% children by one year of age.