

regular basis. This work reviews the outcomes of positive and borderline results for patients at the Royal Manchester Childrens Hospital (RMCH) who were not referred through the NBS pathway.

Methods: Sweat chloride data were gathered for all sweat collections from April 2015 to March 2018. Patient outcome data was then reviewed for sweat chloride results ≥ 40 mmol/L (≥ 30 mmol/L for patients <6 months) and for insufficient sweat collections.

Results: Excluding NBS referrals, 566 individual sweat tests were performed on 440 patients. 50 (8.8%) of the tests performed were insufficient (26 patients (5.9%). Sufficient sweat was subsequently achieved for 17 of these patients leaving 9 for which a result could not be obtained. 35 patients were found to have either borderline or elevated levels of sweat chloride. 21 of these were newly referred RMCH patients, all of whom underwent confirmatory genetic testing with the following outcomes: 7 CF positive, 9 CF negative/unlikely, 2 CF carriers, 1 CFTR related disorder and 2 with an inconclusive diagnosis (deceased/lost to follow up). The average sweat chloride concentration for CF positive patients was 74 mmol/L (43–116 mmol/L) compared to 42 mmol/L (23–74 mmol/L) for unaffected patients. The average age at referral for these patients was 6 years (3 months to 15 years).

Conclusion: This review demonstrates the effectiveness of sweat testing in the referral pathway for CF diagnosis at RMCH.

P022

Earlier contact with a cystic fibrosis centre is associated with better nutritional outcomes in infants with cystic fibrosis

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Objectives: Newborn screening (NBS) for CF was fully implemented in the US in 2010, but process variations may affect timeliness of evaluation for infants with positive NBS tests. We hypothesised that earlier age at first CF Centre contact is associated with better nutritional outcomes at age 1 year in infants with CF.

Methods: We studied infants born during 2010–2018 enrolled in the CF Foundation Patient Registry. We defined CF Center contact as age at first event (AFE: sweat test, encounter, or care episode). Exclusions were meconium ileus, prematurity, missing gestational age, sweat chloride <30 mEq/L, CFTR modulator use before age 1, missing zip code, diagnosis before birth, and age at first event >365 d. We compared outcomes of infants from the lowest quartile (early, E) to the highest quartile (late, L) AFE, matching for sex, median income by zip code, race, ethnicity, genotype (McKone class) and pancreatic status.

Results: Among 6879 infants, 3607 met inclusion criteria. After quartile assignment and matching, 579 infants were in each group. Median AFE was 11d in the E quartile and 47d in the L quartile ($p < 0.001$). At age 1y, E had higher median WFA Z-score (0.09 vs -0.07 , $p = 0.024$) and HFA Z-score (-0.45 vs -0.6 , $p = 0.004$) than L. At 3 years, WFA Z-score was similar, but E ($n = 358$) had higher HFA Z-score (median -0.104 vs 0.24 , $p = 0.007$) than L ($n = 420$). This persisted at 5 years (media -0.075 vs -0.26 , $p = 0.11$; E $n = 226$, L $n = 283$). There was no difference in *Pseudomonas aeruginosa* infection rates at 1y (22 vs. 20%, $p = 0.33$) or 3–6 y (29 vs. 30%, $p = 0.60$). L were older than E at the end of 2018 (mean 5.2 ± 2.5 vs. 4.3 ± 2.6 , $p < 0.001$).

Conclusion: Earlier evaluation at a CF Centre is associated with improved WFA and HFA at age 1 year, and improved HFA at age 3 and 5 years. The infants diagnosed later were older at the end of the study period, and a cohort effect cannot be excluded. Further evaluation is underway. (CFF MCCOLQ01).

P023

New delayed diagnosed cases of cystic fibrosis in Cystic Fibrosis Centre at the Institute for Respiratory Diseases in Children in Skopje, Republic of Macedonia

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Objectives: Our institute is the largest of its kind in our country and it specialises in paediatric pulmonology. During the last year at the institute, 33 763 children were examined at the institute, out of which 3344 were hospitalised. The majority of patients with CF are diagnosed in the first decade of life, but it is difficult when you do not have newborn screening for CF. In our country screening started at April 2019. We want to describe the clinical presentation and delays in diagnosis of patients with cystic fibrosis (CF) with the goal of raising physicians' awareness of CF.

Methods: We diagnosed 5 paediatric cases of CF, based first on the clinical picture, then on a sweat test, faecal elastase, sputum isolates and finally all confirmed by genetic analysis. Period of examination was one year.

Results: Mean age at diagnosis of children with CF was 6.5 years (range 30 days to 5 years). Three children with CF remained undiagnosed at 1 year old. All respiratory and gastrointestinal symptoms were present before diagnosis. All 5 children were with Weight ≤ 5 th percentile and height or length ≤ 5 th percentile. Also all of them have vitamin D levels <8 mmol/L. Sputum cultures were positive for *Pseudomonas aeruginosa* in two child. Two patients had positive family histories of CF. But only 1 case was diagnosed on the basis of family history; the patient had no symptoms at diagnosis. Average amount for Cl in sweat were 110 mmol/L. Faecal elastase was 42 $\mu\text{g/ml}$.

Two of this patients were homozygous for F508del/F508del, two were F508del/S466(TAG) and one was with F508del/G542X.

Conclusion: Our only one year research has documented considerable morbidity and delay in diagnosis of CF when identification of the disease was based on clinical suspicion. By time of diagnosis, some patients were already seriously ill. So we must always make sweat test and think of CF when the respiratory and malnutrition persist.

P024

Minority infants with cystic fibrosis are initially evaluated at a later age than non-Hispanic white infants

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Objectives: Most US states perform newborn screening (NBS) for cystic fibrosis (CF) using IRT and DNA panels. DNA panels may miss more CFTR mutations in ethnic and racial minorities (M). We hypothesized that M infants would be evaluated for CF at an older age and have a different distribution of CFTR mutations than non-Hispanic White (NHW) infants.

Methods: We compared median age at first event (AFE: sweat test, encounter or care episode) of M to NHW infants born in 2010–2018 in the CF Foundation patient registry. M infants had race entered as African-American or other, and/or ethnicity entered as Hispanic, consistent with US census categories. We compared rates of prematurity, clinical symptoms at presentation, CFTR mutation class and lowest quartile (LQ) of cohort median income by zip code (MIZ).

Results: We identified 1335 M and 5019 NHW infants. M had later AFE than NHW infants (median, 31d vs 22d (range, 0–365 d), $p < 0.001$). More M infants had CFTR mutations characterized as “other” (table, $p < 0.001$). Fewer M infants had a positive NBS or prenatal test. The prematurity rate was similar and M infants had lower mean birth weight Z-scores (-0.34 ± 1.17 vs -0.11 ± 1.12 $p < 0.001$). M infants were more likely to have