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Original Article

Clinical and Laboratory Findings in Children with Positive Newborn Screening for Cystic Fibrosis: A Multicenter Retrospective Study

Pelin Asfuroğlu[1], Ayşe Tana Aslan[1], Tuğba Ramaslı Gürsoy[1], Gökçen Ünal[2], Aslı İmran Yılmaz[2], Büşra Sultan Kibar[3], Sevgi Pekcan[2], Melih Hangül[4], Mehmet Köse[4], Tuğba Şişmanlar Eyüboğlu[1]

[1]Gazi University Faculty of Medicine, Department of Pediatric Pulmonology, Ankara, Türkiye
[2]Necmettin Erbakan University Meram Faculty of Medicine, Department of Pediatric Pulmonology, Konya, Türkiye
[3]Necmettin Erbakan University Meram Faculty of Medicine, Department of Pediatrics, Konya, Türkiye
[4]Erciyes University Faculty of Medicine, Department of Pediatric Pulmonology, Kayseri, Türkiye

ABSTRACT

Aim: In some settings, the diagnostic evaluation of cystic fibrosis (CF) may be delayed due to limited access to sweat chloride tests. This study aimed to describe the clinical and laboratory findings observed during the evaluation of children with positive newborn screening (NBS) results for CF.

Materials and Methods: We retrospectively reviewed the data of children referred after positive NBS for CF who were evaluated at three pediatric pulmonology centers between 2015 and 2021. NBS was used as a referral tool, and the diagnosis of CF was established according to standard diagnostic criteria, including the sweat chloride test and/or genetic analysis. Demographic characteristics, clinical features, and laboratory findings were compared between those children diagnosed with CF after NBS and those not diagnosed with CF.

Results: A total of 1,469 children were included, of whom 76 (5.2%) were diagnosed with CF. CF was more frequently observed in those children with parental consanguinity, a history of meconium ileus, steatorrhea, doll-like facial appearance, metabolic alkalosis, hyponatremia, hypokalemia, hypochloremia, and having a sibling with CF (all $p<0.05$).

Conclusion: This large multicenter cohort study presents real-life data on the clinical and laboratory findings observed in those children with positive NBS for CF. This study does not propose an alternative diagnostic strategy to the sweat chloride test, but highlights supportive clinical features which may raise clinical suspicion and emphasizes the importance of timely referral and follow-up, particularly in settings where access to confirmatory testing may be delayed.

Keywords: Cystic fibrosis, immunoreactive trypsinogen, newborn screening, sweat chloride

Corresponding Author
Pelin Asfuroğlu, MD, Gazi University Faculty of Medicine, Department of Pediatric Pulmonology, Ankara, Türkiye
E-mail: pelinasfuroglu@hotmail.com **ORCID:** orcid.org/0000-0002-9575-3982
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Introduction

Cystic fibrosis (CF) is an autosomal recessive disorder caused by pathogenic variants in the gene encoding the CF transmembrane regulator (CFTR) protein, resulting in defective chloride transport across epithelial cells. This defect leads to the production of thick, viscous secretions, impaired mucociliary clearance, and progressive multisystem involvement, particularly affecting the respiratory and gastrointestinal systems (1,2).

Newborn screening (NBS) programs for CF aim to detect affected infants before the onset of clinical symptoms, thereby enabling the early initiation of treatment and improved long-term outcomes (3). Most NBS strategies are based on biochemical screening and begin with the measurement of immunoreactive trypsinogen (IRT) from dried blood spots (1). Elevated IRT levels indicate pancreatic duct obstruction and leakage of pancreatic enzymes into the circulation (4). Infants with persistently increased IRT levels are subsequently referred to specialized CF centers for confirmatory diagnostic evaluation, primarily using the sweat chloride test (5,6).

Early diagnosis through NBS enables the timely initiation of essential interventions, including pancreatic enzyme replacement therapy, salt and fat-soluble vitamin supplementation, and structured pulmonary care, which together reduce CF-related morbidity and mortality (7,8). The sweat chloride test remains the cornerstone and gold standard for the diagnosis of CF (9,10). However, access to sweat chloride tests may be limited due to the need for specialized equipment, standardized procedures, and experienced personnel (11,12).

The limited availability of sweat chloride tests has been recognized as a challenge in developing countries and increasing concerns have also been reported in parts of Europe (11,12). In our country, nationwide access to the sweat chloride test was interrupted between 2018 and 2019 due to a licensing issue related to pilocarpine. During such periods, diagnostic confirmation may be delayed, and reliance on genetic testing alone may not be feasible because of prolonged turnaround times and the high genetic heterogeneity observed in the population. Under these circumstances, clinicians may face uncertainty regarding early clinical decision-making for infants with suspected CF.

In situations where the sweat chloride test is delayed or unavailable, careful clinical evaluation and supportive laboratory findings may help raise clinical suspicion and guide interim management while awaiting confirmatory diagnosis. This study does not advocate for the replacement of the sweat chloride test as the diagnostic standard, but rather to describe the clinical and laboratory findings observed during the evaluation of children with positive NBS results for CF in real-life settings. By presenting data from a multicenter cohort, we aimed to highlight supportive features which may assist clinicians in identifying those infants who require close follow-up and timely referral during periods of limited access to confirmatory testing.

Materials and Methods

This multicenter retrospective study was conducted in three pediatric pulmonology centers between 2015 and 2021. Children who were referred to these centers following positive NBS results for CF were evaluated. Children without follow-up data or lacking sufficient core clinical information to ascertain their CF diagnostic status were excluded. Analyses for individual variables were performed using the available data.

Since January 1st, 2015, the Ministry of Health in Türkiye has implemented a nationwide NBS program for CF using the IRT/IRT protocol. Dried heel blood samples are analyzed for IRT levels. If the first IRT (at approximately 72 hours of life) is ≥90 μg/L and the second IRT (at 7-14 days of life) is ≥70 μg/L, the NBS result is considered positive. Infants with positive NBS results are referred by primary care physicians to specialized CF centers for further diagnostic evaluation (13).

NBS was used as a referral tool only and not as a diagnostic method. According to the European Cystic Fibrosis Society (ECFS), the diagnosis of CF is established by a sweat chloride concentration >59 mmol/L and/or the identification of two disease-causing CFTR variants in the presence of suggestive clinical findings, such as positive NBS, bronchiectasis, CF-related respiratory pathogens, salt loss syndrome, or exocrine pancreatic insufficiency (14). Only those children referred after positive NBS have been included in this study. Those children diagnosed with CF independently of NBS have not been included.

The following data were recorded for all of the children: age at first admission following NBS positivity, gender, gestational age (preterm or term), first and second IRT levels, duration of follow-up, age at diagnosis of CF (if applicable), parental consanguinity, a history of meconium ileus, the presence of steatorrhea (based on fecal fat excretion testing), daily weight gain, the presence of a doll-like facial appearance, having a sibling with CF, pancreatic elastase levels (if available), blood pH and bicarbonate (HCO_3) levels, and serum sodium, potassium, chloride,

and albumin levels measured at admission, regardless of presenting symptoms.

Although the sweat chloride test is routinely performed at participating centers, access was temporarily restricted for part of the study period due to a nationwide licensing problem with pilocarpine. When performed, the sweat chloride test results, obtained by either conductivity measurement or chloride titration, were recorded. All of the collected variables were analyzed in order to assess their associations with a confirmed diagnosis of CF.

The definitions used are as follows:

- Meconium ileus: intestinal obstruction caused by thickened meconium in the ileum (15).

- Doll-like facial appearance: a round facial appearance with puffy cheeks, small nose, and small chin; typically observed in infancy and associated with hypoalbuminemia and malnutrition; also seen in other malabsorptive conditions (16).

- Hyponatremia: serum sodium <135 mmol/L.

- Hypokalemia: serum potassium <3.5 mEq/L.

- Hypochloremia: serum chloride <96 mmol/L.

- Hypoalbuminemia: serum albumin <3.5 g/dL (17-22).

- Pseudo-Bartter syndrome: defined according to ECFS registry guidelines as metabolic alkalosis with blood pH >7.45, serum sodium <130 mmol/L, and serum chloride <90 mmol/L (23).

Ethical approval was obtained from the Clinical Research Ethics Committee of Gazi University (approval number: 11, date: 04.01.2021), and this study was conducted in accordance with the principles of the Declaration of Helsinki. Due to the retrospective design of this study, informed consent was not requested by the ethics committee.

Statistical Analysis

IBM SPSS Statistics version 22.0 (IBM, Armonk, NY, USA) and IBM SPSS Amos-24 were used for the statistical analyses. The conformity of the variables to the normal distribution was examined using visual (histogram and probability graphs) and analytical methods (the Kolmogorov-Smirnov test). Categorical variables are presented as numbers and percentages, while continuous variables are expressed as mean ± standard deviation or medians with interquartile range (IQR) as appropriate.

Binary logistic regression and multivariate logistic regression analyses were used in order to evaluate associations between clinical and laboratory variables and the diagnosis of CF. Variables with a p value<0.10 in univariate analysis and those considered clinically relevant were included in the multivariate model. Odds ratios (ORs) and 95% confidence intervals (CIs) were calculated. Sensitivity, specificity, positive predictive values, and negative predictive values were determined using 2 by 2 tables. A p value<0.05 was considered statistically significant.

Results

During the study period, 1,484 children had a positive NBS result for CF. Fifteen children were excluded due to a lack of follow-up or insufficient core clinical information, leaving 1,469 children for analysis. The demographic and clinical characteristics of the children with positive NBS for CF are presented in Table I.

The median first IRT level was 99 (IQR: 90-121.5) µg/L and the median second IRT level was 74 (IQR: 70-90) µg/L. The median daily weight gain was 30 (IQR: 23-38) g/day. Pancreatic elastase levels were available in 45 (3.1%) children; among these, 10 (22.2%) children had pancreatic insufficiency, and the median pancreatic elastase level was 369 (IQR: 204.5-476) µg/mL.

Following NBS positivity, 390 (26.5%) children were unable to undergo a sweat chloride test, while 1,079 (73.5%) children underwent at least one sweat chloride test. Among those tested, 204 (18.9%) had a sweat conductivity test, 948 (87.9%) had a sweat chloride titration test, and 73 (6.8%) children underwent both tests. The median of the sweat conductivity test results was 30.4 (10.8-120.0) mmol/L, and

Table I. Demographic and clinical characteristics of children with positive newborn screening for cystic fibrosis (n=1,469)

Gender Female (n/%) Male (n/%)	 818/55.7 651/44.3
Age at admission (days) [median (IQR)]	27 (21-37)
Gestational age at birth (n=1,330) Premature (n/%) Mature/Term (n/%)	 146/11.0 1,184/89.0
Parental consanguinity (n/%) (n=1,407)	249/17.0
History of meconium ileus (n/%) (n=921)	6/0.7
Doll-like facial appearance (n/%) (n=1,469)	29/2.0
Metabolic alkalosis (n/%) (n=858)	78/9.1
Hyponatremia (n/%) (n=902)	64/7.1
Hypokalemia (n/%) (n=839)	16/1.9
Hypochloremia (n/%) (n=880)	29/3.3
Hypoalbuminemia (n/%) (n=687)	95/13.8
Steatorrhea (n/%) (n=423)	94/22.2

IQR: Interquartile range

the median sweat chloride concentration was 26.2 (10.0-121.0) mmol/L.

Overall, 76 (5.2%) children were diagnosed with CF. The median age at diagnosis of CF was 40 (IQR: 30-73) days. Among the children diagnosed with CF, 60 (78.9%) underwent a sweat chloride test, while 16 (21.1%) children were diagnosed based on genetic testing in the absence of a sweat chloride test. Of all of those children with a sibling, seven (1.0%) had a sibling with CF.

The association between the clinical and laboratory variables and CF diagnosis in univariate analyses are presented in Table II. First and second IRT levels were higher in those children diagnosed with CF compared with those not diagnosed (Table II). Pseudo-Bartter syndrome was identified in 7 (9%) among the 78 children with metabolic alkalosis. When combinations of laboratory abnormalities were examined, CF was diagnosed in 12 (66.7%) of the 18 children with concurrent hyponatremia and hypochloremia [OR (95% CI): 26.9 (9.76-74.11), p=0.001]. Among the children with hyponatremia, hypochloremia, and hypoalbuminemia, 5 (83.3%) of the 6 children were diagnosed with CF [OR (95% CI): 60.8 (7.00-527.71), p=0.001]. CF was diagnosed in 6 (54.5%) of 11 children with hyponatremia, hypochloremia, and hypokalemia [OR (95% CI): 14.7 (4.38-49.52), p=0.001]. Additionally, 3 (75%) of 4 children with the combination of hyponatremia, hypochloremia, hypoalbuminemia, and hypokalemia were diagnosed with CF [OR (95% CI): 35.4 (3.64-345.24), p=0.002].

Albumin levels were measured in 23 (79.3%) of 29 children with doll-like facial appearance, with a median albumin level of 2.6 (IQR: 2.2-3.2) g/dL. Hypoalbuminemia was found in 18 (62.1%) of the children with a doll-like facial appearance.

The multivariate logistic regression analysis results are shown in Table III. The model explained 37.6% of the variance in CF diagnosis. When hypochloremia, doll-like facial appearance, and having a sibling with CF were included in the model, all three variables were significantly associated with a diagnosis of CF ($p<0.05$). The standardized beta coefficients were 0.117 for hypochloremia, 0.391 for doll-like facial appearance and 0.290 for having a sibling with CF.

The sensitivity and specificity of individual clinical and laboratory features are summarized in Table IV. The sensitivity and specificity values were as follows: parental consanguinity (27.4% and 82.8%), sibling with CF (20.0% and 99.8%), meconium ileus at birth (6.3% and 99.6%), doll-like facial appearance (32.4% and 99.6%), metabolic alkalosis (30.9% and 92.8%), hyponatremia (29.2% and 94.8%), hypochloremia (22.8% and 99.1%), hypokalemia (15.7% and 99.3%), hypoalbuminemia (40.8% and 89.3%), and steatorrhea (70.6% and 79.8%).

Table II. Univariate analysis of clinical and laboratory variables associated with cystic fibrosis

Variables	Diagnosed with CF (n=76)	Not diagnosed with CF (n=1,393)	Odds ratio (95% CI[a])	p*
First IRT level (µg/L) [median (min-max)]	136 (90-360)	98 (90-500)	1.01 (1.01–1.02)	**0.001**
Second IRT level (µg/L) [median (min-max)]	108 (70-275)	73 (70-285)	1.02 (1.02–1.03)	**0.001**
Prematurity [n/N (%)]	7/57 (12.3)	139/1,273 (10.9)	1.1 (0.50-2.56)	0.668
Parental consanguinity [n/N (%)]	20/73 (27.4)	229/1,334 (17.2)	1.8 (1.07-3.10)	**0.039**
Metabolic alkalosis [n/N (%)]	21/68 (30.9)	57/790 (7.2)	5.7 (3.21-10.27)	**0.001**
Hypoalbuminemia [n/N (%)]	29/71 (40.8)	66/616 (10.7)	5.8 (3.36-9.85)	**0.001**
Hyponatremia [n/N (%)]	21/72 (29.2)	43/830 (5.2)	7.5 (4.16-13.64)	**0.001**
Steatorrhea [n/N (%)]	12/17 (70.6)	82/406 (20.2)	9.4 (3.24-27.67)	**0.001**
Meconium ileus at birth [n/N (%)]	2/32 (6.3)	4/889 (0.4)	14.8 (2.59-83.69)	**0.016**
Hypokalemia [n/N (%)]	11/70 (15.7)	5/769 (0.7)	28.5 (9.58-84.71)	**0.001**
Hypochloremia [n/N (%)]	16/70 (22.9)	7/810 (0.9)	33.9 (13.41-86.14)	**0.001**
Doll-like facial appearance [n/N (%)]	23/76 (30.3)	6/1,393 (0.4)	110.3 (42.93-283.30)	**0.001**
Sibling with CF [n/N (%)]	6/30 (20.0)	1/645 (0.2)	161.0 (18.64-1,390.28)	**0.001**

*p<0.05 is significant
[a]CI: Confidence interval
[Odds ratios and 95% confidence intervals were calculated using binary logistic regression to evaluate associations with cystic fibrosis]
[Odds ratios for continuous variables represent the change in odds per unit increase]
CF: Cystic fibrosis, IRT: Immunoreactive trypsinogen

Table III. Multivariate logistic regression analysis of variables associated with cystic fibrosis

	Variables	B	S.E.	β (Beta)	p*
Cystic fibrosis	**Hypoalbuminemia**	0.025	0.022	0.038	0.256
	Hyponatremia	0.027	0.027	0.030	0.329
	Hypochloremia	0.166	0.047	0.117	0.001
	Parental consanguinity	0.009	0.013	0.016	0.475
	Doll-like facial appearance	0.614	0.040	0.391	0.001
	Sibling with CF	0.621	0.069	0.290	0.001

[Variance ratio: R^2=0.376]
*p<0.05 is significant
B: Unstandardized coefficient, S.E.: Standard error, β (Beta): Standardized coefficient, CF: Cystic fibrosis

Table IV. Sensitivity and specificity of clinical and laboratory features in children with cystic fibrosis

Variables	Sensitivity (%)	Specificity (%)	PPV[α] (%)	NPV[β] (%)
Sibling with CF (n=7)	20.0	99.8	85.7	96.4
Doll-like facial appearance (n=29)	32.4	99.6	79.3	96.6
Hypochloremia (n=23)	22.8	99.1	69.6	93.7
Hypokalemia (n=16)	15.7	99.3	68.7	92.8
Meconium ileus at birth (n=6)	6.3	99.6	33.3	96.7
Hyponatremia (n=64)	29.2	94.8	32.8	93.9
Hypoalbuminemia (n=95)	40.8	89.3	30.5	92.9
Metabolic alkalosis (n=78)	30.9	92.8	26.9	93.9
Steatorrhea (n=94)	70.6	79.8	12.8	98.5
Parental consanguinity (n=249)	27.4	82.8	8.0	95.4

[α]Positive predictive value, [β]Negative predictive value, CF: Cystic fibrosis

Discussion

This multicenter retrospective study aimed to evaluate the clinical and laboratory findings observed during the evaluation of children with positive NBS results for CF, particularly in situations where access to sweat chloride tests was limited or delayed. The findings highlight that having a sibling with CF and the presence of a doll-like facial appearance were the most prominent supportive clinical features associated with a diagnosis of CF among NBS-positive children.

In addition, a history of meconium ileus, steatorrhea, metabolic alkalosis, hyponatremia, hypokalemia, hypochloremia, and hypoalbuminemia were also associated with an increased likelihood of CF. Although CF is an autosomal recessive disorder and parental consanguinity is a well-known risk factor, it was less predictive than the other clinical and laboratory features in our cohort. This observation may be related to the high CF carrier frequency and marked genetic heterogeneity in our population. In populations where carrier rates are high, the presence of consanguinity alone may not be distinguishing; however, the coexistence of characteristic clinical findings may be more informative in raising clinical suspicion.

The doll-like facial appearance has historically been an underrecognized clinical feature in CF (16). Kose et al. (16) described this appearance in edematous, malnourished children with hypoalbuminemia. In the present study, doll-like facial appearance emerged as a strong clinical indicator of CF, even in some cases without documented hypoalbuminemia, supporting its value as a readily recognizable physical finding. Although this facial phenotype is not specific to CF and may also be observed in other malabsorptive or metabolic conditions, its presence in children with positive NBS should prompt careful evaluation and close follow-up, particularly when access to confirmatory diagnostic testing is limited (24).

Family history plays an important role in the early recognition of CF. In our study, having a sibling with CF

was the most strongly associated factor, emphasizing the importance of heightened clinical awareness and early evaluation in the siblings of affected individuals. While parental consanguinity was relatively common in our population, it did not emerge as a dominant distinguishing feature. This may reflect the widespread CF carrier status in Türkiye, where CF can occur even in the absence of consanguinity. In populations with lower carrier frequencies, however, consanguinity may retain greater clinical relevance as a supportive clue (25-28).

Meconium ileus is one of the earliest and most specific manifestations of CF and has been reported in 10-20% of affected newborns (25,29,30). Although the frequency of meconium ileus in our cohort was lower, its presence remained strongly associated with a CF diagnosis and should be considered an important early warning sign in those infants with positive NBS results.

Several laboratory abnormalities may support clinical suspicion of CF, particularly electrolyte disturbances such as hyponatremia, hypochloremia, and metabolic alkalosis, either alone or in combination as pseudo-Bartter syndrome, which have been reported as early manifestations of CF especially in younger infants (25,31). In our cohort, hypochloremia was the most frequently observed electrolyte abnormality among those children with CF. Additional findings, including hypokalemia and hypoalbuminemia, were also commonly encountered and may further strengthen clinical suspicion when present in combination (32).

Several studies have demonstrated regional variability in sodium and electrolyte imbalances among children with CF, influenced by climatic conditions, nutritional status and CFTR mutation distribution. Hyponatremia, hypochloremia, and metabolic alkalosis have been reported as early manifestations of CF, particularly in infants and in warmer climates or resource-limited settings. A narrative review synthesizing data from multiple geographic regions emphasized that salt loss and related electrolyte abnormalities remain clinically relevant across different age groups and health-care systems (33). In this context, supportive laboratory findings may provide valuable guidance for clinicians when confirmatory diagnostic testing is delayed or temporarily unavailable.

Previous studies evaluating laboratory findings in NBS-positive infants have emphasized that, while such markers do not replace the sweat chloride test, they may support clinical decision-making during the diagnostic process, particularly when confirmatory testing is delayed or unavailable (11). Our findings highlight the importance of a comprehensive clinical assessment and the integration of multiple supportive features, rather than relying on a single parameter.

Study Limitations

This study had several limitations. Its retrospective design and the absence of nationwide data may limit its generalizability. Some clinical findings, such as doll-like facial appearance, are subjective and may vary between observers. In addition, not all laboratory parameters were available for every child, and some abnormalities may have been transient and therefore not captured at the time of initial evaluation.

Conclusion

In children with positive NBS for CF, having a sibling with CF and the presence of a doll-like facial appearance were the most prominent supportive clinical features observed in those ultimately diagnosed with CF. While parental consanguinity remains clinically relevant, particularly in populations with lower CF carrier frequencies, it was not a distinguishing feature in our cohort. Careful anamnesis, physical examination, and basic laboratory evaluation remain essential components of the clinical assessment of NBS-positive children, especially in settings where access to sweat chloride tests is limited or delayed. In the presence of findings suggestive of CF, early referral and close follow-up may be considered while awaiting confirmatory diagnostic testing in order to minimize disease-related complications.

Ethics

Ethics Committee Approval: Ethical approval was obtained from the Clinical Research Ethics Committee of Gazi University (approval number: 11, date: 04.01.2021).

Informed Consent: Due to the retrospective design of this study, informed consent was not requested by the ethics committee.

Footnotes

Authorship Contributions

Surgical and Medical Practices: P.A., A.T.A., T.R.G., G.Ü., A.İ.Y., B.S.K., S.P., M.H., M.K., T.Ş.E., Concept: P.A., A.T.A., T.R.G., S.P., M.K., T.Ş.E., Design: P.A., A.T.A., T.Ş.E., Data Collection or Processing: P.A., T.R.G., G.Ü., A.İ.Y., B.S.K., M.H., Analysis or Interpretation: P.A., A.T.A., T.R.G., S.P., M.K., T.Ş.E., Literature Search: P.A., Writing: P.A., A.T.A., T.R.G., S.P., M.K., T.Ş.E.

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