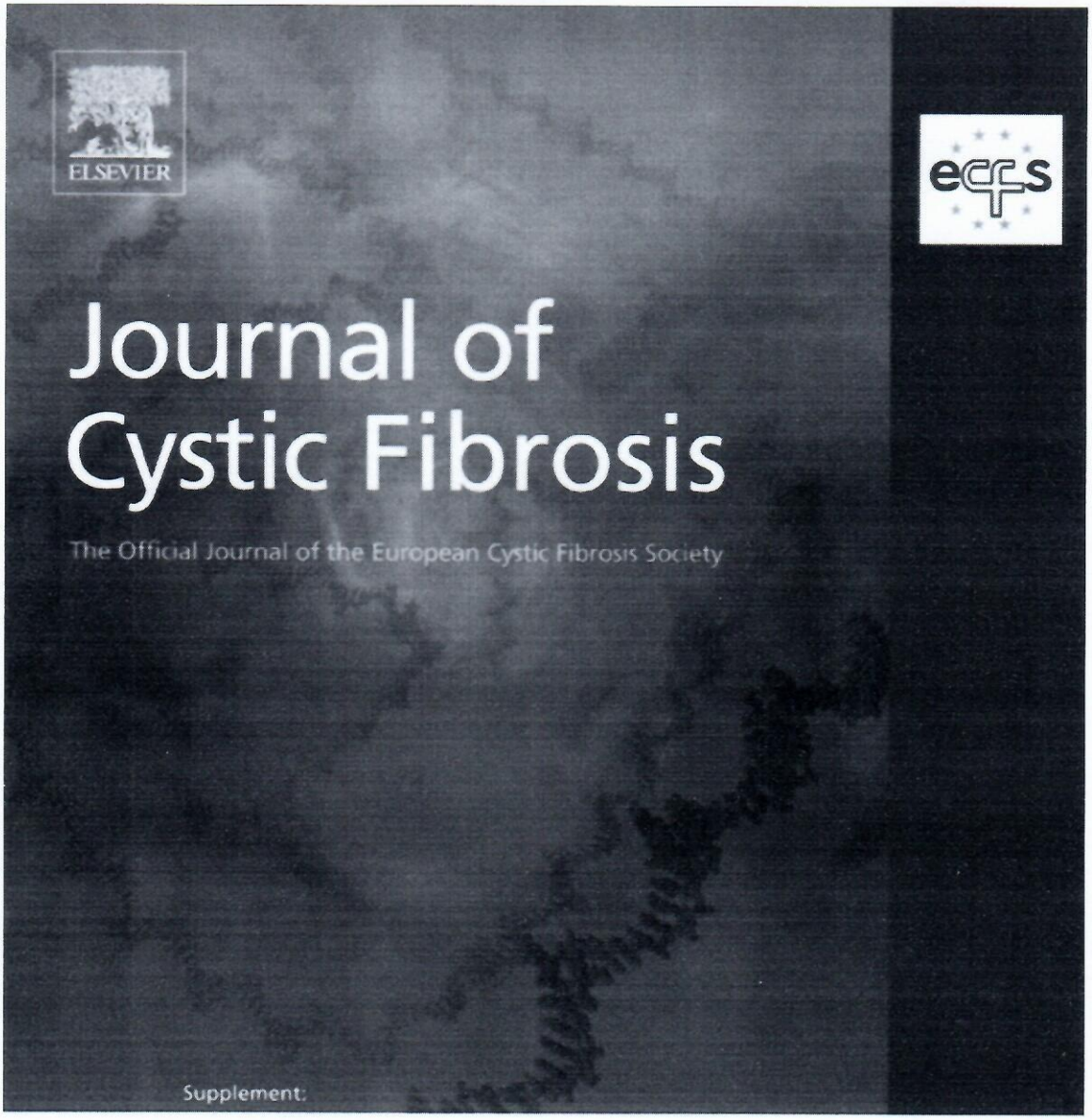

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bronchiolitis. Moreover, history of smoking by a household member and family history of allergy were recorded.
Results: Among the 160 newborns who presented to our clinic after newborn screening for CF, 117 (58.1%) could be contacted by phone. Cystic fibrosis was diagnosed in 6 (3.8%) of these subjects and 4 (2.5%) reported having had lower respiratory problems such as ever-wheezing, recurrent wheezing and pneumonia. Two children had died due to non-specified reasons. Smoking by a household member was reported in 33.8% of the parents and the prevalence of family history of allergy was 6.9%.
Conclusion: Frequency of every-wheezing in the first three years of life is about 20% in our community; thus the results of our study do not show an increase in respiratory problems with high IRT levels. However, follow up of these children for longer period of time for respiratory diseases is needed for more confident interpretations.

P015
Evaluation of the specificity and sensitivity of the cut-off values of immunoreactive trypsinogen in the cystic fibrosis newborn screening program

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Objectives: Cystic fibrosis (CF) newborn screening (NBS) programme with the two-stage immunoreactive trypsinogen (IRT/IRT) protocol was included in the screening programme in 2015 in Turkey. If the first IRT is 90 μg/L and above and the second IRT is 70 μg/L and above, the screening test is considered positive and the patient is referred to the CF centres for a sweat chloride test. Each country determines their own cut-off values for IRT. We aimed to evaluate cut-off values and to determine the specificity and sensitivity of them in current NBS.
Methods: The data of children with positive NBS between 2015 and 2020 were evaluated retrospectively in three paediatric pulmonology centres. Patients' age at diagnosis, genders, IRT values and sweat chloride test results were noted. The sensitivity and specificity of the IRT cut-off values were evaluated.
Results: In total, 1,122 children admitted to paediatric pulmonology centers with positive NBS. 58 (5.1%) patients were diagnosed with CF. 29 (50%) patients with CF were female. The median age at diagnosis of the patients was 44 (IQR:30–72) days. The median of the first IRT values was 105.0 (IQR:94.0–195.2) and of the second IRT values was 80.0 (IQR: 61.8–95.0) in the CF patients. The median of the first sweat chloride test results was 75.0 (IQR: 72.0, 95.0), of the second sweat chloride test results was 67.6 (IQR: 47.8, 90.6). When the cut-off value of the first IRT was 101.87 μg/L, the sensitivity was 81% and the specificity was 46%. When the cut-off value of the second IRT was 76.9 μg/L, the sensitivity was 82.8% and the specificity was 42.3%.
Conclusions: The detection power of screening tests is related to the cut-off value determined for the test. We found that the cut-off values we use in our country are effective in detecting the disease. The diagnostic power of IRT cut-off values may be evaluated periodically with multicentre studies.

P016
The role of clinical and laboratory findings in the diagnosis of cystic fibrosis in children with positive newborn screening

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Objectives: In newborn screening (NBS) of cystic fibrosis (CF), the diagnosis may be delayed due to difficulty in accessing the sweat test. The importance of laboratory and clinical findings in suggesting the diagnosis of CF was evaluated in children presenting with positive NBS.
Methods: The data of all children with positive NBS in three paediatric pulmonology departments between 2015 and 2020 were evaluated retrospectively. Children's age at admission and diagnosis, gender, parental consanguinity, daily weight gain, steatorrhea, clinical and laboratory findings were noted, the specificity and sensitivity of the clinical and laboratory findings of children were evaluated.
Results: A total of 1,141 children admitted to paediatric pulmonology departments with positive NBS were included, and 55.6% of them were female. The median age of children at admission was 27 (IQR: 21–35) days, and 59 (5.2%) patients were diagnosed with CF. The median age of patients at diagnosis was 43 (IQR: 30–71.5) days. Parental consanguinity was found in 193 (16.9%) children, 14 (1.2%) of whom were CF (specificity: 95.3%, sensitivity: 7.3%). Meconium ileus was present in 6 (0.5%) children, 2 (0.2%) of whom were CF (specificity: 99.4%, sensitivity: 9.1%). Doll-like face was found in 22 (1.9%) children, 18 (1.6%) of whom were CF (specificity: 99.6%, sensitivity: 33.3%). Metabolic alkalosis was found in 63 (5.5%) children, 16 (1.4%) of whom were CF (specificity: 91.8%, sensitivity: 30.8%). Steatorrhea was present in 181 (15.9%) children, 27 (2.3%) of whom were CF (specificity: 79.5%, sensitivity: 58.7%). There was a statistically significant difference in daily weight gain, pH, HCO_3, sodium, potassium, chloride, and albumin levels between children with and without CF ($p < 0.05$).
Conclusions: Doll-like face, meconium ileus, parental consanguinity, and metabolic alkalosis are powerful guides for the diagnosis of CF, since children with positive NBS may have difficulty in accessing the sweat tests.

P017
Different ethnical distribution of the incidence of cystic fibrosis in Republic of North Macedonia

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Objectives: Newborn screening (NBS) allows early diagnosis of cystic fibrosis (CF) and helps parents to keep their child as healthy as possible and delay or prevent serious, lifelong health problems related to this disease. In Macedonia, it was introduced as a national population program in 2019 after a pilot study in 2018.
Methods: Immunoreactive trypsinogen (IRT) was measured in dried blood spots sampled 48 hours after birth, using fluoroimmunometric assay (DELFIA) during the pilot study in 2018 as well as the period April 2019 - December 2020. Two-step IRT-IRT algorithm is performed, followed by the sweat test for confirmation/exclusion of the CF diagnosis when the IRT values were higher than the cut off values (70 ng/ml and 45 ng/ml, respectively).
Results: A total of 43,139 newborns have been screened for CF during the study period, and overall incidence of 1/2,538 was detected. Recall rate was 0.44% (n = 190). Out of 41 (0.095%) screening positive cases, the diagnosis of CF was confirmed in 17 (0.039%) newborns after the positive sweat test and molecular *CFTR* analysis. Twelve of the detected CF cases were ethnic Albanians (70.6%) and 5 were ethnic Macedonians (29.4%). Moreover, the CF incidence observed among the Albanian neonatal population (1/1284) was 4-fold higher than the incidence detected in the Macedonian newborns (1/4530). The more frequent consanguineous marriages in the Albanian population over the centuries, have probably led to a higher percentage of carriers of the CF mutations among them.
Conclusions: The detected overall CF incidence showed that CF is common in our country. We observed different ethnic distribution of the CF incidence in Macedonia. The estimation of the carriers for the *CFTR* mutations may explain the high CF incidence among the Albanian population.

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We thank you for your participation,

Isabelle Fajac
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