centiles, corresponding to 45 ng/mL and 76 ng/mL of blood, respectively. Hundred fifteen samples surpassed the cut-off and underwent PAP analysis; eight exceeded the ultra-high cut-off and were further assessed for *CFTR* gene variants. Among the 115 samples, 25 had an IRT×PAP product exceeding 135 $\mu g^2/L^2$ and proceeded to DNA analysis. Of the 33 samples analysed, three were found to carry one F508del variant and were classified as NBS positives. Further confirmatory evaluations, including sweat testing and full *CFTR* gene analysis are pending to finalise CF diagnoses. An additional 10.000 samples will be tested to complete protocol validation and to refine performance metrics, including the positive predictive value and the identification of false-negative results.
Conclusion: These interim data suggest that the used screening strategy effectively identifies neonates at increased risk of cystic fibrosis. Further pilot expansion will enable refinement of performance metrics and screening strategies.

P026

Five-year evaluation of the cystic fibrosis Newborn Screening Program in the city of Buenos Aires

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Introduction: The benefits of early cystic fibrosis (CF) detection using newborn screening (NBS) has led to widespread use in NBS programs. Since 2002, a two-stage immunoreactive trypsinogen (IRT/IRT) screening strategy has been used as a CFNBS method in all public maternity units in the City of Buenos Aires, Argentina. This approach has been changed to the IRT/PAP (pancreatitis-associated protein) strategy since 2018.
Objective: to evaluate the cystic fibrosis newborn screening (CFNBS) program in the city of Buenos Aires during the period 2018-2022.
Material & Methods: All newborns born between January 2018 and December 2022 were included. IRT/PAP were obtained after 36 hours of life in a single dry blood sample. IRT was measured by immunofluorometric method (PerkinElmer, cut-off value 60 ng/ml) and PAP by ELISA method (Dynabio, cut-off value 1.6 ng/ml for samples with IRT values between 60 and 99 ng/ml and 0.5 ng/ml for values ≥100 ng/ml). Sweat Chloride Tests (ST) were performed using the Gibson and Cook method to confirm the diagnosis. Genetic studies (50 mutations) were done for all the detected and died patients.
Results: A total of 89082 newborns were screened; 533 had high IRT values (0.60 %). A total of 319 patients with elevated IRT/PAP were recalled to perform ST (0.31%). CF was diagnosed in 15 patients, 33 patients died without being able to perform ST, all of them had normal genetic studies. 53% were homozygous, 20% heterozygous F508del, and 13.3% had other mutations. The prevalence of CF was 1/5939. The median IRT value (x ±SD) in those affected was 192 ±78.4 ng/ml. CF diagnosis was confirmed at a median age (X ±SD) of 78 ±115 days of life.
Conclusions: The CF prevalence in the city of Buenos Aires is 1: 5939 live births. The recall rate was acceptable, optimizing the CFNBS of our program in this five-year evaluation.

P027

A ten-year outcome of cystic fibrosis newborn screening programme at Hacettepe University, Türkiye

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In Türkiye, cystic fibrosis (CF) newborn screening (NBS) was started in 2015 and performed by measuring immunoreactive trypsinogen (IRT) in heel-prick blood samples. If IRT levels are above the threshold, a second IRT is measured. Infants with both IRT values above the threshold are referred to sweat testing. If sweat chloride (Cl) is ≥60 mEq/L patient is diagnosed as CF, if between 30-59 mEq/L genotyping is done, if <30 mEq/L, no genotyping is done and the baby is considered to have no CF.
Objective: This study evaluates the outcome of babies referred to Hacettepe University Children's Hospital, the biggest CF center in Türkiye for sweat testing in 10-years, since the beginning of national NBS.
Method: In this retrospective cohort study, infants who had positive NBS for CF between January 1st 2015 - December 31st 2024 and had sweat testing in our hospital were evaluated. Genotyping and sweat Cl results were obtained from hospital information system and patient files.
Results: A total of 1,370 infants were included in the study. 48 patients (3.5%) had sweat Cl ≥60 mEq/L and were diagnosed as CF and 13 patients (0.9%) had intermediate (30-59 mEq/L) sweat Cl. Of those 13 patients, 4 were lost to follow-up and 9 patients were diagnosed with CFSPID/CRMS with genotyping results. During follow-up, conversion to CF occured in 5 of these patients (55.5%); 4 due to elevated sweat test results and 1 due to the identification of pathogenic mutations. Our CF/CFSPID ratio was calculated as 5.3:1. On follow-up, 1 patient with CFSPID/CRMS was diagnosed with CFTR-Related Disorder, with a history of Pseudo-Bartter syndrome.
Conclusion: Our CF/CFSPID ratio is significantly higher compared to the literature. One reason for this is that genetic testing is not done as part of NBS for those with sweat Cl<30 mmol/L. If genetic analysis is included for this group as a part of NBS, CFSPID patients will not be missed, and the follow-up of these individuals can be appropriately managed.

P028

Cystic fibrosis newborn screening in Luxembourg: 7 years' experience

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Objectives: 1) To describe the Cystic Fibrosis (CF) Newborn Screening (NBS) protocol in Luxembourg and evaluate its performance compared to recommendations for standards of care, 2) To calculate the incidence of CF in Luxembourg.
Methods: Retrospective analysis of CF NBS performed in Luxembourg during a 7 years' period (January 2018- December 2024) and comparison with current recommendations for standards of care.
Results: The Luxembourgish protocol for CF NBS relies on Immunoreactive Trypsinogen (IRT) on day 3 of life (D3) as the primary test. DNA analysis for *CFTR* gene variants is performed in case of high IRT (>60ng/mL), if the specific informed consent is signed by legal authorised representatives. The genetic testing is performed with the CF-EU2v1 panel (Yourgene® Cystic Fibrosis Base) which includes 50 pathogenic variants and 4 polymorphisms of the *CFTR* gene. A safety net based on IRT on day 21 was introduced in June 2022. Patients screened positive are referred to the Paediatric National center where sweat test is performed. Genetic counselling is provided in case of detected *CFTR* variants. During the period 2018-2024, IRT was above the threshold level for 1.16% of live births. CF was diagnosed in 14 infants of a total of 52,777 births. All patients had at least one *CFTR* variant identified by the genetic panel. Identification of the second variant with extensive gene analysis (EGA) was required in 4 cases (4382delA, E664X, I502T, S489L). All CF patients were seen by a CF specialist within the first month of life. The overall positive predicted value (PPV) is 0.13. The calculated incidence of CF was 1/3770 live births in Luxembourg. No missed cases have been identified to date.
Conclusion: CF NBS was introduced in Luxembourg in 2018. The incidence of CF in infants born in Luxembourg is comparable to neighboring countries. Further assessment is required in order to validate the diagnostic protocol.

P029

Newborn screening for cystic fibrosis in North Macedonia

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Introduction/Objective: The main goal of the newborn screening (NBS) programme is to detect infants with treatable diseases early. NBS for cystic fibrosis (CF) was introduced in The Republic of North Macedonia in April