P-206 - EXPERIENCE IN THE NEONATAL SCREENING FOR CYSTIC FIBROSIS IN THE PUBLIC HOSPITAL DR GUILLERMO RAWSON, SAN JUAN- ARGENTINA.

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INTRODUCTION: Given the possibility of having numerous tools to diagnose Cystic Fibrosis (CF), a diagnostic algorithm, using different strategies of combined methodologies depending on the age of the patient, has been implemented. OBJECTIVE: Analyze the usefulness of the diagnostic algorithm for CF according to the age of the patient, using techniques available. MATERIALS AND METHODS: 68291 dried blood specimens were evaluated in newborns (NB), between 2 and 15 days of life, from March 2010 to December 2018. ImmunoChem Trypsin-MW ELISA, MP-Biomedicals was used for the IRT measurement. Positive cases with IRT >130 ng/ml were recited before 30 days and a 2nd IRT was determined.In those NB with IRT > 90 ng/ml in the 2nd sample, Molecular Biology (MB) or Sweat test (ST) was done, depending on their age. 10 mutations were investigated through 2014: DF 508, G542X, G551D, N1303K, 1717-1G - A, W1282X, S1255X, R334W, 3849 + 10 Kb and G85E. Then, 36 mutations were incorporated (CFTR test). The ST was made by iontophoresis (Gibson and Cooke). RESULTS: Out of 68291 samples, 255 were elevated for 1st IRT, 212 of them were negative in the 2nd IRT. 20 patients were positive for the 2nd IRT, MB was performed and mutations were found in 8 patients (incidence 1 in 8536). There were 12 patients not confirmed, 11 presented IRT near the cut off, concluding that they were false positives for the technique. One showed a value of IRT > 750 ng/ml and did not show any mutation. 23 patients (9%) did not attend to the recitation. One of them came back from clinical service within 18 months. A ST was performed and then an MB test, confirming CF diagnostic. CONCLUSION: The direct referral of these patients to MB would reduce the age range at the moment of the diagnosis as well as the false negatives in the ST. The different strategies proposed as diagnostic CF algorithm, will facilitate early diagnosis and adequate procedures depending on the age of the patient, helping to reach an accurate diagnosis, thus collaborating with the implementation of early therapeutic strategies to our hospital population with CF.