O-014 - CYSTIC FIBROSIS: A 6-YEARS EXPERIENCE OF NEWBORN SCREENING IN THE PUBLIC HEALTH SYSTEM OF SOUTHERN BRAZIL

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INTRODUCTION: Cystic fibrosis (CF) is a genetic disease of a great clinical variability. The morbidity and mortality is mostly caused by progressive respiratory impairment and gastrointestinal disorders. In Southern Brazil, the Newborn Screening (NBS) is based on immunoreactive trypsinogen (IRT) and sweat tests, medical appointment and molecular screening for the most frequent mutations in our state. ***OBJECTIVE:*** to describe the most frequent clinical, laboratory characteristics and mutations/polymorphisms found in babies with a confirmed diagnosis of CF during the first medical evaluation. ***MATERIALS AND METHODS:*** it was a cross-sectional and descriptive study of the newborns diagnosed with CF, screened from 2012 to 2018 by the Reference Service in NBS from Rio Grande do Sul state/ Brazil. Laboratory, clinical and genetic data were collected. ***RESULTS:*** In total, 61 patients were included, of whom 20% had low birth weight and only 9.8% were asymptomatic at the time of the first clinical evaluation. Steatorrhea (31.1%), cough (29.5%) and meconium ileus (24.6%) were the most observed clinical manifestations and the median age at the first visit was 35 days. IRT had significantly lower values in neonates with a history of meconium ileus, and significantly higher values in patients who had the p.Phe508del mutation. The allelic frequency of the mutations found were: F508del (65.6%), G542X (4.1%), 3120+1G>A (3.3%), N1303K (2.5%), R1162X (1.6%) and others mutations (2789+5G>A, G85E, 1717-1G>A, R334W, 711+1G>A, 3191delC, R1066C, 3272-26G>A, 1812-1G>A and D1152H) with a frequency of 0.82% were also found. The genotypic frequency of the IVS8TGmTn polymorphism variants found were: TG10T7 (68.4%), TG11T7 (18.4%), TG10T9 (13.2%) and TG12T7 (1.3%). The T5 allele was not observed in these patients. ***CONCLUSIONS:*** this work shows the importance of a NBS for CF because provides the opportunity to undertake preventive and treatment before the development of irreversible changes in the respiratory tract and other complications, contributing to quality of life and patient survival.