P-250 - NEWBORN SCREENING AND SELECTIVE SCREENING FOR CONGENITAL METABOLIC DISEASES IN A PEDIATRIC HOSPITAL, MENDOZA-ARGENTINA

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INTRODUCTION: Its important that the detection, diagnosis, treatment and follow-up of congenital endocrine-metabolic diseases are developed in Reference Centers. Therefore, our Program covered two areas of work: a) Newborn screening (NBS): primary congenital hypothyroidism (CH), congenital adrenal hyperplasia (CAH), phenylketonuria, galactosemia, biotinidase deficiency (BTDdef), cystic fibrosis (CF). b) Selective Screening (SS): for some non-mandatory congenital metabolic diseases (CMD). OBJECTIVES: Present results and indicators of NS and SS.

MATERIALS AND METHODS: a) From 1999 to 2009, NBS for CH and Phenylketonuria; in 2010 NBS for CAH, Galactosemia, BTDdef, CF was added. Analytical process in two stages, 1st stage: [Phenylalanine, Galactose, Biotinidase]: colorimetry; [TSH, 17OHP, IRT]: ELISA; 2nd stage: re-evaluation of samples with “border-line and positives” results: [Phenylalanine, Galactose, Biotinidase]: fluorometry; [TSH, 17OHP, IRT]: DELFIA. b) Since 2016, by medical order, SS CMD, prior to send samples to specialized centers in Argentina located 700-1100 km from our Hospital. Biochemical parameters: [Leucine-Isoleucine-Valine]: fluorometry, home-made methodologies; Amino Acids in plasma-urine and Monosaccharides in urine: TLC and Gal-1-phosphate-uridyltransferase: fluorometry. RESULTS: a) NBS, 1999-2018 = 410925 newborns (NB) (2010-2018 = 210921 NB). Indicators, mean, 2017-2018: Days of life (DOL) to the sample collection= 2. Time of transit of the sample from the Maternities to CE.P.E.I.I.= 3 days. DOL to get the result= 7. DOL to deliver the report= 9. Diagnosis and start of Treatment= 14 days. Rejected and Insufficient samples= 0.49%. Recall rate: Total/NB gestational age ≥ 37 weeks= 3.2%/2.1%; 99% of the NB were located. Coverage; Publics hospitals/Private= 99%/13%. Children in treatment-follow-up= 200 CH (Incidence= 1/2055); 9 Phenylketonuria and 18 persistent Hyperphenylalaninemas (Incidence= 1/15219); 15 CAH (Incidence= 1/14061) and 28 CF (Incidence= 1/7533). b) SS, 2016-2018: evaluation of 83 samples, patients admitted to our Hospital and outpatient. Results, average time= 3 days in CE.P.E.I.I., versus 30 days by referral of samples to specialized centers. SS= 2 children with MSUD. CONCLUSIONS: In the context of a Reference Center and cost-effectiveness: a.improved NS indicators, from sample collection to diagnosis and treatment b.the study of new CMD, SS, was expanded with acceptable costs for the validation of home-made methodologies and reduction of screening and diagnostic times.