P-160 - EVALUATION OF THERAPEUTIC RESPONSE TO SAPROPTERINE DICHLORIDRATE FROM A GROUP OF FENILCETONURIC PATIENTS CARRIED OUT OF THE EXTENDED NEONATAL DISTRICT SERVICE IN THE FEDERAL DISTRICT

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INTRODUCTION: Phenylketonuria (FNC) is an inherited disorder of phenylalanine metabolism (Phe), characterized by a loss of hepatic phenylalanine hydroxylase (PAH) function, limiting the hydroxylation of Phe to tyrosine (Tyr), using tetrahydrobiopterin as a cofactor. Sapropterine dihydrochloride, the pharmaceutical version of 6R-tetrahydrobiopterin (6R-BH4), increases its residual activity in sapropterin-responsive phenylketonurons and, together with dietary control, helps to reduce Phe concentrations. GOALS: To present the profile of the patients participating in the test and the percentage of responsiveness to sapropterin dihydrochloride.

MATERIALS AND METHODS: Ten samples were collected on filter paper from 12 patients on the drug for 28 days, two basal samples and two weekly samples. The Phe evaluation method was tandem mass spectrometry, using reference values between 2-6mg / dl and a cutoff value of 1.0 mg / dl to increase dietary Phe intake. Sapropterin was started after the second basal collection, at a dosage of 10mg / kg. After one week, the dose increased to 20mg / kg. Patients were evaluated weekly for drug tolerance, blood Phe and dietary Phe intake. RESULTS: The mean age was 13 ± 4 years, being 58% female and 42% male. All patients are classical phenylketonuric by oral tolerance of Phe. The weight variation was 0.9 ± 1.4 kg. Of the participants, 58% responded to sapropterin, with a mean baseline Phe of 6.2 ± 3.5 mg/dl and a final of 4.4 ± 2.1 mg/dl. The mean Phe reduction in responders was 38.1% and that of the increase in TYR was 7%. Of the respondents, 03 presented reduction of TYR by 20%. There was no discontinuity in the use of medication due to adverse effects, showing good tolerance of the studied group. DISCUSSION: The characteristics of the patients were considered in the interpretation of the results. There was good tolerance to the drug and an average response compatible with the international literature. Some genotypes are predictive of responsiveness and responsiveness is associated with specific alleles, so an unfolding of this study is needed to improve understanding of how FNC mutations influence PAH dynamics and increase understanding of the disease and its management.