P-173 - SLEEP DISORDERS CHARACTERIZATION OF CLASSICAL HOMOCYSTINURIA IN PEDIATRIC PATIENTS FROM THE REFERENCE CENTER OF BAHIA, BRAZIL

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\textbf{INTRODUCTION:} Classical Homocystinuria (HCU), an autosomal recessive inherited disorder, worldwide prevalence estimated in 1: 900.000 to 1: 1.800. Sleep disorders are common and affect sleep quality and quantity, leading to increased morbidity. \textbf{OBJECTIVE:} To describe a serial of individuals diagnosed with HCU followed in a reference center in Salvador, Bahia, Brazil and their sleep disorders. \textbf{METHOD:} Observational, descriptive study, from data review of patients with HCU biochemical diagnosis followed in a reference center. Three types of questionnaires were applied to identify sleep disorders: the Sleep Disturbance Scale for Children (SDSC), range from 26 to 130 with a cut-off point of 39 that characterizes sleep disturbance; the Epworth Sleepiness Scale (ESS) to measure the daytime sleepiness, score >10 points indicates sleepiness; and the Conners’ Scale short Form to suspect of hyperactivity disorders with a score ≤ 12 to girls and score ≤ 17 to boys as a positive finding. \textbf{RESULTS:} Three pediatrics patients with HCU. Patient 1, male, onset age 3y, diagnostic age 8y, current age 12y, presents EKG alterations, crystalline dislocation, spinal deformities, marphanoide appearance, cave foot, behavioural problems, intellectual disability. Patient 2, male, onset age 4y, diagnostic age 4y, current age 8y, presents EKG alterations, severe myopia, spinal deformities, behavioural problems, intellectual disability. Patient 3, female, onset age 3y, diagnostic age 4y, current age 5y, presents severe myopia, crystalline dislocation. The molecular diagnosis indicated a mutation in homozygosis, the p.W323X/p.W323X, non-responsive to pyridoxine supplementation to all patients. The 3 pediatric patients have family positive cases of HCU, only the Patient 3 has parental consanguinity. The SDSC questionnaire observed that the score of Patient 2 and Patient 3 were the following values 34 and 33, respectively, while Patient 1 presented value = 54, above the cutoff point, characterizing the sleep disorder. Though Abridged Conners Scale, Patients 2 and 3 presented the values = 5 and 12, respectively. While Patient 1 presented value = 21, characterizing hyperactivity. In this study, no change was found on the Epworth scale. \textbf{CONCLUSION:} The present study draws attention to the the medical geneticist over suspicion about the presence of sleep disorders which are pervasive in HCU.