P-107 - SLEEP DISORDER IN AN INDIVIDUAL WITH DEFICIENCY OF CYTOCHROME C OXIDASE 15: A CASE REPORT

Salles C\textsuperscript{1,3}, Gomes IL\textsuperscript{2}G, Embiruçu EKE\textsuperscript{1,2}

(1) Hospital University Professor Edgard Santos, Salvador, Brazil. (2) Life sciences Department, Universidade do Estado da Bahia, Salvador, Brazil. ilsgomes@yahoo.com.br (3) Bahiana College of Medicine and Health, Salvador, Brazil.

BACKGROUND: Deficiency of cytochrome C oxidase 15 (COX15) is a rare cause of autosomal recessive mitochondrial disease in infants. They are related to the identification of pathogenic variant in MTFMT gene. Its prevalence is estimated at 1/1,000,000 live births and the patients exhibit multi-systemic impairment, most commonly with a Leigh Syndrome phenotype. Sleep disorders are associated with increased morbidity, but only a few cases of primary mitochondrial syndromes associated to sleep disturbances had been published in the literature.

METHODS: A case report of a patient with COX15 deficiency followed up at the Medical Genetics Service of the University Hospital Complex Professor Edgard Santos, Salvador, Bahia, Brazil. It was applied three different questionnaires to identify sleep disorders in children. They were: Sleep Disturbance Scale for Children (SDSC), ranging from 26 to 130 with a cut-off point of 39, which characterises a sleep disturbance; the Epworth Sleepiness Scale (ESS) to measure the daytime sleepiness: score >10 points indicates sleepiness; and the short form of Conners' Scale, used to suspect of hyperactivity disorders with a score ≤ 12 to girls and score ≤ 17 to boys as a positive finding.

RESULTS: A 3 years old male patient, non consanguineous parents, presenting at the service with previous recurrent infections and motor delay since 1 year of age. The parent refers no pregnancy or birth intercurrence. At neurological evaluation: bilateral palpebral ptosis, global muscle weakness, Gauss sign, global hipotonia and ataxia. Currently he does not climb stairs without support or ride the bike. He is also unable to raise the arms over the head. He underwent molecular analysis, which exhibited pathogenic variant in MTFMT gene (p.Arg332*/p.Ser209Leu), leading to Combined Oxidative Phosphorylation Deficiency 15 (COXPD15) diagnosis. The patient had positive finding at SDSC scale, with a 58 score. At Epworth and Conners' scales, 6 and 2 scores, respectively.

DISCUSSION: The result of this patient is compatible with sleep disorder. This condition can lead to increased morbidity and caregivers low quality of life. It is essential that geneticists identify the signs of sleep disturbances and try to manage critical findings regarding patients and caregivers quality of life.