**O-006 - DESCRIPTION OF THE PRESENTATION, NEUROLOGICAL OUTCOME AND MORTALITY OF THE UREA CYCLE DISORDER IN 41 PATIENTS DIAGNOSED AT GARRAHAN HOSPITAL.**

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***INTRODUCTION:*** We describe a cohort of patients with urea cycle disorder (UCD) who have been diagnosed or are being followed up at the Pediatric Garrahan Hospital. OBJECTIVES: To correlate the biochemical data, at the time of diagnosis with the specific enzymatic deficiency and the type of presentation. To analyze the family history, the delay in diagnosis and the neurological outcome at the last control. ***METHODS:*** A retrospective study of the patients diagnosed with UCD between 2000 and 2017. The medical records of the hospital were reviewed.The biochemical data including the aminoacids profile were those obtained at the time of diagnosis. ***RESULTS:*** We assessed 41patients (33 families, 22 females, 19 males). A molecular study was performed on 20 patients. 17/41 patients had had a neonatal presentation, the remaining 24 a late-onset presentation; the majority of these patients had delay at diagnosis,(average 7,7months(1–57)), only 3/24 with late-onset UCD presentation were diagnosed during the first month as soon as the symptoms began. 20 patients had a family history. 19/41 showed some grade of developmental delay; in 10patients this delay was severe. 25/41 had ornithine transcarbamylase deficiency(OTCD); 7 had arginine succinic synthetase deficiency(ASSD); 5 had arginine succinate lyase deficiency(ASLD) and one had carbamoylphosphate synthetase deficiency. The remaining 3 patients were not categorized. 14/34 had hepatitis. 10patients died and 6 of them had had a neonatal presentation. ***CONCLUSIONS:*** UCD is still a severe disease with high mortality and morbidity and high developmental delay. The delay in diagnosis is significantly associated with intellectual disability(p: 0.016). Patients who had ammonia values higher than 250mmol/l have higher intellectual disability(p: 0.001). Glutamine values at debut were not associated with greater neurological compromise or mortality. Comparatively, the patients with ASLD were those that showed higher values of ammonia, all of them had a neonatal presentation. The patients with ASSD were those who showed higher values of transaminases. OTCD had shown the highest incidence and mortality, coinciding with the global incidence. To diagnose a severe disease early, in the immediate neonatal period due to the family history, improves the neurological result and death can be prevented.