P-127 - CLINICAL AND BIOCHEMICAL EVOLUTION OF THE FIRST PATIENT WITH MAPLE SYRUP URINE DISEASE WHO RECEIVES A LIVER TRANSPLANT AT GARRAHAN HOSPITAL

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**INTRODUCTION:** Since 2005, elective liver transplantation (LT) has been implemented as a therapeutic alternative for maple syrup urine disease (MSUD). In 2017, the first transplant was carried out on a patient with MSUD. **MATERIALS AND METHODS:** Review of clinical history and collection of biochemical data. **OBJECTIVES:** To describe the clinical and biochemical outcome after LT of a patient with MSUD. **RESULTS:** We describe a patient with a neonatal diagnosis of MSUD with a severe form of the disease who has recurrent episodes of acute metabolic decompensations despite optimal medical management. For this reason, an elective LT was performed from an unrelated cadaveric donor at 3 years of age. The year prior to transplantation the values of leucine and isoleucine were of average 651.9μmol (range 166-990μmol) (normal values 70-250μmol) and valine average 209μmol (range 36-511μmol) (normal values 40-200μmol). The natural protein intake was of 0.21gr/kg/day. Prior to the transplant, she had been hospitalized 12 times, two of which required admission to the ICU due to sensory deterioration. At 24 hours after LT, the branched chain amino acid were within normal parameters. Natural protein, was increased to 2gr/kg/day. After 33 days, she presented a mild acute rejection of the graft and required pulses of methylprednisolone with reduction of the natural protein intake at 1g/kg/day for 6 days without alteration in the amino acids. After transplantation, the values were stable: MS/MS: leucine+isoleucine average 215μmol (101-348μmol) and valine average 235μmol (92-375μmol) plasma aminoacids were performed by Biochrom HPLC: Leucine average 300μmol/L (290-300μmol/d) (normal value 50-175μmol/L), isoleucine average 189μmol/L (180-190μmol/L) (normal values 6-110μmol/L) and valine average 380 μmol/L (330-420μmol/L) (normal values 64-294μmol/L). Currently she has a diet without protein restriction, the protein intake is estimated at 3gr/kg/day. **CONCLUSION:** According to what is described in the literature, the patient presents amino acids with values higher than normal. Currently, she is 4 years old, without decompensation and without the need for dietary treatment. The child attends kindergarten with improved integration and acquisition of new maturing milestones.