P-112 - GLUT1 DEFICIENCY SYNDROME: DIAGNOSIS AND NUTRITIONAL TREATMENT WITH KETOGENIC DIET IN 8 CHILEAN CASES

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INTRODUCTION: Glucose Transporter type 1 (GLUT1) deficiency syndrome is an inborn error of glucose transport. Clinical manifestations are secondary to reduced glucose transport across the blood brain barrier, including: refractory seizures, developmental delay, microcephaly, persistent hypoglycorrhachia with normal glycemia. The diagnosis is suspected with glucose CSF/glycemia <40 (NV: > 60) and confirmed by molecular study in SLC2A1 gene (cr1p35-31.3). Treatment is based on a ketogenic diet (KD).

OBJECTIVE: To present the Chilean experience in diagnosis and follow-up of patients with GLUT1.

METHODOLOGY: 8 records of patients with GLUT1. Clinical picture, biochemical exams, molecular study, nutritional status and macro and micronutrient with KD were recorded.

RESULTS: 5/8 are men. Of the total 4/8 GLUT1 were diagnosed with mediana (Me) 9,0 years of age and 4/8 GLUT1 with (Me) 5,3 months. All had hypoglycorrhachia <40 mg/dl (range: 13-30) without hypoglycemia (80-90 mg/dl). The CSF/blood glucose ratio was (Me) 0,33. At the time of diagnosis, they presented myoclonic seizures, ataxia, and paroxysmal movements without response to drugs. They started KD and it has stayed between 18 years to 1 month. The seizures ceased after md 5 days of starting KD in children who were diagnosed at (Me) 5,3 months. The distribution of the caloric molecule was: Lipids: 87-85% (MCT oil, alpha linolenic acid and docosahexaenoic acid), Protein: 10-8% (0,8 to 2,0 g/wt/day), Carbohydrates: 3-6 %. Fasting levels of beta-hydroxybutyrate acid has been maintained over 2,0 mM/l and after meals lower 5 mM/l. The fasting blood sugar level was (Me) 80 mg/dL (NV: <200), LDL: (Me) 77 mg/dL (NV: <100), HDL: (Me) 62mg/dL (NV: >40), Triglycerides: (Me) 63.6 mg/dL (NV: <150), Vitamin D serum: (Me) 45,3 ng/ml (NV: >38), potassium: (Me) 4,4 mEq/L (NV: 3,4-4,7). All of them are supplemented with L-carnitine, vitamin C, sugar-free multivitamins, calcium. 7/8 GLUT1 have molecular study (C.177del.p; c.1088G>A; c.420delG; c57delA; 969del-C971T; c.143G>A; c.458G>C/p.R153P). According to nutritional status: 3/8 are eutrophic, 1/8 have malnutrition, 3/8 are overweight and 1/8 are at risk of malnutrition.

CONCLUSIONS: In patients with refractory epilepsy, GLUT1 deficiency screening and early diagnosis is important considering that KD is a safe and effective treatment for improving neurological manifestations.