ABCC8 Mutation as a Cause of Congenital Hyperinsulinism: A Case Report

Liliana Mejía de Beldjenna[1], *Cuellar Natalia*[1], *Margarita Jaramillo*[2], *Harry Pachahoa*[3]

[1]Universidad Libre GRINPED, Cali, Colombia; [2]Centro Médico Imbanaco, Universidad ICESI, Colombia

Introduction: Congenital Hyperinsulinism (CH) is the main cause of serious and persistence hypoglycemia in the neonatal period and it is secondary To inadequate insulin secretion despite hypoglycemia as the result of a genetic alteration of the sulfonylurea receptor of the beta cell. More than 150 mutations of the ABCC8 and 25 in KCNJ11 gene have been reported incidence is 1 in 5000 a live newborn babies, Higth metabolic fluxes, diazoxide, octreotide and even subtotal pancreatectomy are required.

Objective: To describe a case of CH to make people aware of its diagnosis and treatment.

Materials and Methods: We reviewed the clinical chart of a neonate with difficult to control hyperinsulinism. The propositus was a male burn at 36 week of gestation in no consanguineons parents. Weight 3.920 grams, height 52 cm. Patient presented at 13 hours of life, with a blood glucose of 3 mg/ dl, insulin of 99.1 Uu/ml and a serum cortisol of 3.65 ug/dl. A subtotal pancreatectomy was performed because of no response to metabolic flow including diozoxide, octeotride and hydrocortisone. Pathology described as generalized nesidioblastosis. Hypoglycemia and hyperinsulinemia persisted following surgery pancreatectomy subtotal. Genetic studies revealed a heterozygous ABCC8 gene, missense, variant p (Gly 228 Asp) wite paternal inheritance suggesting uniparental disomy. This mutation has not be.

Conclusion: Knowledge of the genetic alteration allows better management and parental counselling of this condition.

Pediatric Graves Disease Experience with Institutional Definitive Treatment

Olivia Ruiz, Patricia Papendieck, Eugenia Elias, Ignacio Bergadá, Ana Chies

Children's Hospital "Ricardo Gutiérrez"

Background: In pediatric Graves' disease (PGD), long term antithyroid treatment with drugs (DTAs) is the first line choice but a second alternative Often as radioiodine or thyroidectomy Such has to be considered.

Objective: To describe the outcome of PGD related to the therapeutic choice.

Methods: Medical records of the Patients diagnosed with PGD (2007–2015) in our center Were retrospectively reviewed. Demographics, treatment indication and evolution Were retrieved. In Those In Whom a second alternative treatment was indicated, the cause of esta switch, median time (MT) to indication and adverse effects (AE) Were registered.

Results: 66 records were reviewed (50 girls, median age 11.9 years (range (r): 1.5–17.1) received INITIALLY ATDs 65 and one was Treated with 131I. 24% (n: 16) Were lost to follow up after 2.6 years (r: 0.1–6.9), 28% (n: 18) continued on clinical follow up and in 32 (48.5%) was Indicated another option treatment: 18 received 131I and 14 underwent surgery. 9/18 Patients under DTAs remitted (hypothyroidism or euthyroidism) at a MT of 3 years (r: 1–6.3) and 9 are still Treated (median follow up: 3.7 years (r: 0.1–6.7)). 18 patients received 131I (16 girls, MT to indication 1.1 years (r: 0–4.9). 1 for autoimmune hepatitis, 4 for ATDs AE and 13 for poor compliance 14 (78%) after one dose remitted (median 10 mCi (r: 0.5–16)) and four required 2 doses rendered hypothyroid All Patients in a MT of 0.2 years (r: 0.1–0.8). No complications or AE Were registered. 14 Patients (10 girls) at a MT underwent thyroidectomy since diagnosis of 1.8 years (r: 0.1–6.8): 3 for ATDs AE, 6 for poor compliance (1 with severe ophthalmopathy, one with big goiter) and 5 for thyroid nodules. 10 (71.4%) presented postsurgical complications: such as 1 hungry bone, 6 transient hypoparathyroidism and 1 sepsis. Two Patients (14.3%) ADH definitive hypoparathyroidism. Transient and postsurgical complications definitive were significantly higher than those for 131I ($p = 0.01$).

Conclusion: Long term follow up in PGD is difficult and poorly Achieved. 131I treatment was safe without adverse outcomes correctly, Although surgery while the risk of Indicated entails transient and definitive complications.

Consumptive Hypothyroidism: Case Report

Maria Kuspiel, Marina, Maria Aguilar, Yamila Fittipaldi, Titania Pasqualini, William Alonso

Hospital Italiano de Buenos Aires

Introduction: Hemangioendothelioma (HHE) is a vascular lesion more frequent in the first year of life. The term includes a spectrum of vascular lesions ranging from benign and self-limiting to aggressive and life-threatening. The hepatic form of HHE is a rare tumor in infancy Typically presenting. The Natural course of HHE is Characterized by a proliferation phase and plateau phases followed by involution. Hypothyroidism in Patients of this group is Attributed to expression of type 3 iodothyronine deiodin-ase (D3) by the tumor tissue. The Increased activity leads to rapid enzyme degradation of thyroid hormones, RESULTING in severe hypothyroidism, thyroid hormone When inactivation by D3 Exceeds the synthetic capacity of the thyroid gland. Less than 100 Patients with This entity Have Been published in the literature.

Objective: To report The Importance of checking thyroid function in Children with neonatal massive hemangiomatosis.

Case Report: A 5 month-old male was derived by progressive hepatomegaly of 2 months of evolution. No Relevant perinatal and neonatal history Normal screening. Physical examination Showed rough facies, broad nasal bridge, puffy eyes, Hypotonia, tachycardia, hepatosplenomegaly with regular stools, and delay in neurocognitive milestone. Abdominal ultrasound Showed enlarged liver due to multiple solid hypoechoic lesions. Abdominal angio computed tomography confirmed multiple hypervascularized solid liver lesions Compatible with HHE. Propranolol treatment was indicated. With clinical suspicion of hypothyroidism, thyroid function test (TFT) Were Requested. Laboratory values Were: hemoglobin 9 g/dl, 500,800 platelets/mm^3, hepatogram elevated with gammaglutamyltranspeptidase 80 IU/L, 186.4 TSH mIU/m, T4 8.2 ug/dl, fT4 0.8 ng/dl, T3 0.3 ng/ml. Levothyroxine (LT4) (50 ug/d) treatment was started. At 17 months, I have presents adecuate neu-