GP287 FANCONI BICKEL SYNDROME AND RENAL TUBULAR DYSFUNCTION

Gözde Atasever Yıldırım*, Fatma Derya Bulut, Bahriye Atmış, Sebile Kilavuz, Deniz Kör, Aysun Karabay Bayazit, Halise Neslihan Önenli Mungan. *Cukurova University, Adana, Turkey*

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Introduction Fanconi Bickel Syndrome (FBS) is a rare autosomal recessively inherited inborn error of metabolism due to impaired utilization of glucose and galactose. The mutations of the responsible gene *SLC2A2* cause defects in glucose transport protein-2. Clinical findings are severe growth retardation, polyuria, polydipsia, and hypophosphatemic rickets. Laboratory findings are proximal renal tubular dysfunction, fasting hypoglycemia and postprandial hyperglycemia leading to diabetes, in some cases even in the neonatal period, due to hepatorenal glycogen accumulation.

Method Data of 10 patients from 8 families diagnosed as FBS with molecular analysis were reviewed retrospectively for renal involvement.

Results 3(30%) were female and 7(70%) were male. All had parental consanguinity. Mean age at diagnosis was $10,8 \pm 8,5$ months. Mean current age was $7,3 \pm 6$ years. One patient died at 1 year of age. Height measurements of 7 patients were below 3rd percentile and weight measurements of all patients were below 3rd percentile. 6 patients (60%) had hepatomegaly. Hypertransaminasemia, hypertriglyceridemia and metabolic acidosis with normal anion gap were observed in all patients. 9(90%) patients had hypophosphatemia, hypokalemia and 6(60%) patients had hyponatremia. All patients had glycosuria and proteinuria. Tubular phosphate reabsorption levels of 4 out of 8 patients were below 78%. Mean HbA1C levels were $5,6 \pm 0,6$, oral glucose tolerance test was performed on 6 patients over three years of age. Plasma glucose levels at 120 minutes were higher than 140 mg/dL in 4 (66,7%), higher than 200 mg/dL in 2 patients (33,3%) despite none of them had overt diabetes. 8 patients (80%) showed severe manifestations of rickets. All patients had homozygous *SLC2A2* gene mutations [6 had p.G162Rfs*17 (c.482_483insC), 1 had p.H192R(c.575A>G), 1 had p.R301*(c.901C>T), 1 had p.D344Y(c.1030G>T), 1 had p.V357Nfs*37(c.1069delGinsAATAA)]

Discussion FBS is a well-defined and a very rare inherited metabolic disease characterized with hepatic and renal involvement. Here, we report our single center experience of 10 patients with FBS, due to the high frequency of parental consanguinity in our region. Different from the other glycogen storage diseases and disorders of carbohydrate metabolism such as galactosemia and hereditary fructose intolerance, hyperglycemia is an important factor contributing to early proximal renal dysfunction in FBS. Six out of our 10 patients had impaired glucose tolerance. In conclusion the early occurrence of renal tubular acidosis, especially under the age of one years with elevated levels of HbA1C is a key point of thinking FBS in the differential diagnosis.

GP288 THE ROLE OF MYOFUNCTIONAL THERAPY IN TREATMENT OF ENURETIC CHILDREN WITH SLEEP-DISORDERED BREATHING

¹Pietro Ferrara*, ²Diletta Saitta, ²Michela Pulcino, ²Federica Di Ruscio, ²Giulia Franceschini, ²Roberta Autuori, ²Alessandro Di Lucia, ²Flavia Dosa, ²Margherita Zona, ²Federica Sandullo, ³Andrea Ianni, ⁴Massimo Pettoello-Mantovani. ¹*Institute of Pediatrics, Catholic University of Sacred Heart, Rome, Italy;* ²*Campus Bio-Medico University, Rome, Italy;* ³*Public Health and Statistics, Campus Bio-Medico University, Rome, Italy;* ⁴*Department of Pediatrics, Scientific Institute Casa Sollievo della Sofferenza, University of Foggia, Foggia, Italy*

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Objective Nocturnal enuresis (NE) is one of the most common disorder in children. Even if NE is considered a benign condition, a timely treatment is important because it can interfere with children's psychosocial development. The presence of a sleep-disordered breathing (SDB), from habitual snoring to obstructive sleep apnea, is a worsening factor in NE and an unfavorable prognostic factor for the response to drug therapy.

The purpose of this study was to investigate if the combination between pharmacological treatment of NE and a non-invasive treatment of SDB, such as myofunctional therapy, determines a statistically significant increase in the response rate.

Design This is a longitudinal cohort study.

Setting The study was carried out within our pediatric outpatient clinic in Campus Bio-Medico University Hospital of Rome.

Patients A total of 56 children, aged between 5 and 15 years and presenting NE and SDB, were enrolled.

Interventions After clinical evaluation, children were randomized in two different treatment groups: group 1, desmopressin with or without oxybutynin, dietary and lifestyle recommendations and myofunctional therapy; group 2, desmopressin with or without oxybutynin, and dietary and lifestyle recommendations.

Additionally, group 1 was provided with the modified pediatric sleep questionnaire (PSQ-22) to describe the severity of patient sleep disturbances. All patients were treated for 3 months.

Main outcome measures To evaluate if an effective and non-invasive treatment of SDB may decrease the frequency of NE in those children presenting both the abovementioned clinical conditions.

Results After 3 months of therapy and 3 months of follow-up after drugs withdrawal, the response rate distribution was 79,2% for the group 1 and 64,3% for the group 2. The PSQ-22 was positive for 66,7% and negative for the remaining 33,3%. The results were not statistically significant ($p > 0,05$).

Conclusions This study evaluated the effectiveness of a combined approach in treatment of NE, in order to reflect its multifactorial aetiology. A higher response rate was achieved by the group treated with the combination of pharmacological and myofunctional therapy. However, the results were not statistically significant and likely this was due to the small size of the analyzed sample. All above considered, further and more extensive studies are needed.