Johansson-Blizzard Syndrome with Pan-hypopituitrism, Type 2 Diabetes and Pancreatic insufficiency : Effect of Treatment

Ashraf T Soliman*, Fawzia Alyafie*

Department of Pediatrics , Alexandria Children's Hospital *and Hamad Medical Center, PO Box: 3050, Doha, Qatar

Introduction

Johanson-Blizzard syndrome (JBS) is a disease characterized by poor growth, variable dysmorphic features, including aplasia or hypoplasia of the nasal alae, sensori-neural deafness, abnormal hair patterns or scalp defects, and oligodontia. Other features of JBS include pan-hypopituitarism, imperforate anus, and PI. Pancreatic endocrine insufficiency (PI)

Case study

AM is a 10 years- old boy, born at term with aplasia of the alae nasi and congenital sensori-neural deafness, severe prenatal growth retardation and mental retardation.

He presented at the age of 1.5 year with severe post-natal growth retardation and PI. He was started on Pancrex.

Endocrine tests showed low TSH and FT4 and cortisol deficiency.

He was started on L-thyroxine and hydrocortisone. In spite of his weight gain his linear growth was slow.

At 2 years his peak GH responses to glucagon and clonidine stimulation tests were 3 and 5 ng/ml respectively with low IGF-I.

MRI head showed hypoplastic pituitary gland. GH therapy was started and at that time he was diagnosed genetically with JBS.

At 7 years he attained complete catch up growth (Height at 50th centile for age) At 10 years of age his random blood glucose (RBS) was noticed to be abnormal 8.5 mmol/L.

OGTT showed fasting hyperglycemia and post prandial hyperglycemia (table 1)

He did not have acanthosis or goiter and the other systemic examination showed no new findings. He was prepubertal.

Oral glucose tolerance test with insulin measurement showed hyperglycemia and high insulin secretion (insulin resistance).

Results

The diagnosis of Type 2 DM was given. Oral metformin 500mg twice daily was prescribed with excellent response to the treatment (table).

Table 1	Fasting	2 hr prandial
Glucose (mmol/L)	7.1	17.4
Insulin (µunit/ml)	19	48
Cpeptide (ng/ml)	3	5.88

After a year on treatment his HbA1C = 5.9 %

Discussion

DM is a rare complication of JBS with only 3 cases reported in the literature, 2 of which had history of pancreatic exocrine insufficiency and found to have insulin deficiency.
The third case had DM with insulin resistance without pancreatic exocrine insufficiency.
This is the first case with pan-hypopituitarism, type 2 DM in a pancreatic insufficient JBS patient.

Conclusion

In this patient with JBS and type 2 DM glycemic control was successfully achieved with oral Metformin.

55ESPE 261--P1
Diabetes
ashraf soliman
DOI: 10.3252/pso.eu.55ESPE.2016
Poster presented at: ESPE 2016 PARIS 10-13 SEPTEMBER
PosterSessionOnline.com