P1-356

The «double diabetes» in adolescent with Prader-Willi syndrome patient.

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INTRODUCTION

Prader-Willi syndrome (PWS) is a complex, multisystem disorder that arises from lack of expression of paternally imprinted genes on chromosome 15q11-q13. Its major clinical features include neonatal hypotonia, short stature, developmental delay, hyperphagia, childhood onset obesity, hypothalamic endocrinopathy and characteristic appearance. It is known that due to severe obesity PWS patients are prone to develop type 2 diabetes mellitus (T2DM), while type 1 diabetes mellitus (T1DM) is extremely rare in this syndrome.

Case history:

We report on 14 years old boy, who was referred to our department due to uncontrolled glycemic status (blood glucose range 12-15 mmol/l). He was diagnosed to have diabetes mellitus at the age of 13 with classic manifestation (polyuria, polydipsia, fatigue) with ketonuria and hyperglycemia (blood glucose-28 mmol/l, HBA1C-8,1%). The insulin therapy was started. The boy received mixed insulin therapy (lispro, glargine) 27U/day (0,3 U/kg/day). The medical history revealed that his father had been ill with T2DM for 3 years and had retinopathy and neuropathy. The patient had neonatal muscle hypotonia, feeding problems, delayed development progress during childhood, orchidopexy at 1,5 years and progressive obesity with hyperphagia from 2 years of age. The boy was diagnosed with PWS at the age of 5.

On physical examination at 14-th y.o. the patient had abdominal obesity (87,6 kg, BMI SDS=+3,64) with acanthosis nigricans and characteristic clinical features, such as acromicria, dolichocephaly. The height was 146,9 cm (SDS=-1,8), Tanner stage1. Laboratory data showed high level of C-peptide 6,28 ng/ml (1,1-4,4),HBA1C- 9,9%. Dyslipidemia and non-alcoholic steatohepatitis were diagnosed. The standard liquid meal test showed the preserved C-peptide secretion (max 13,46 ng/ml) and high postprandial glucose (11,5 mmol/l). Taking in account the presence of diabetes mellitus in obese PWS patient with the clinical features of insulin resistance (acanthosis nigricans) and preserved C-peptide secretion the T2DM was suspected and Metformin therapy was initiated. During the 2-week of Metformin treatment (1500 mg/day) the insulin doses were able to be reduced to 4U/day (only Lispro) and glycemic profile was improved (blood glucose 5-7 mmol/l).To our surprise the IA-2A and ZnT8A levels were positive (32U/ml and 199,4 U/ml, respectively). So, the boy was diagnosed with T1DM. We didn't find the diabetes mellitus complications during examination.

CONCLUSION

To our knowledge this is the first report of combined presentation of features both type 1 and 2 diabetes in PWS, so called «double diabetes». Further long-term evaluation of this patient will show the particularities of the T1DM development in PWS patient with obesity and insulin resistance.

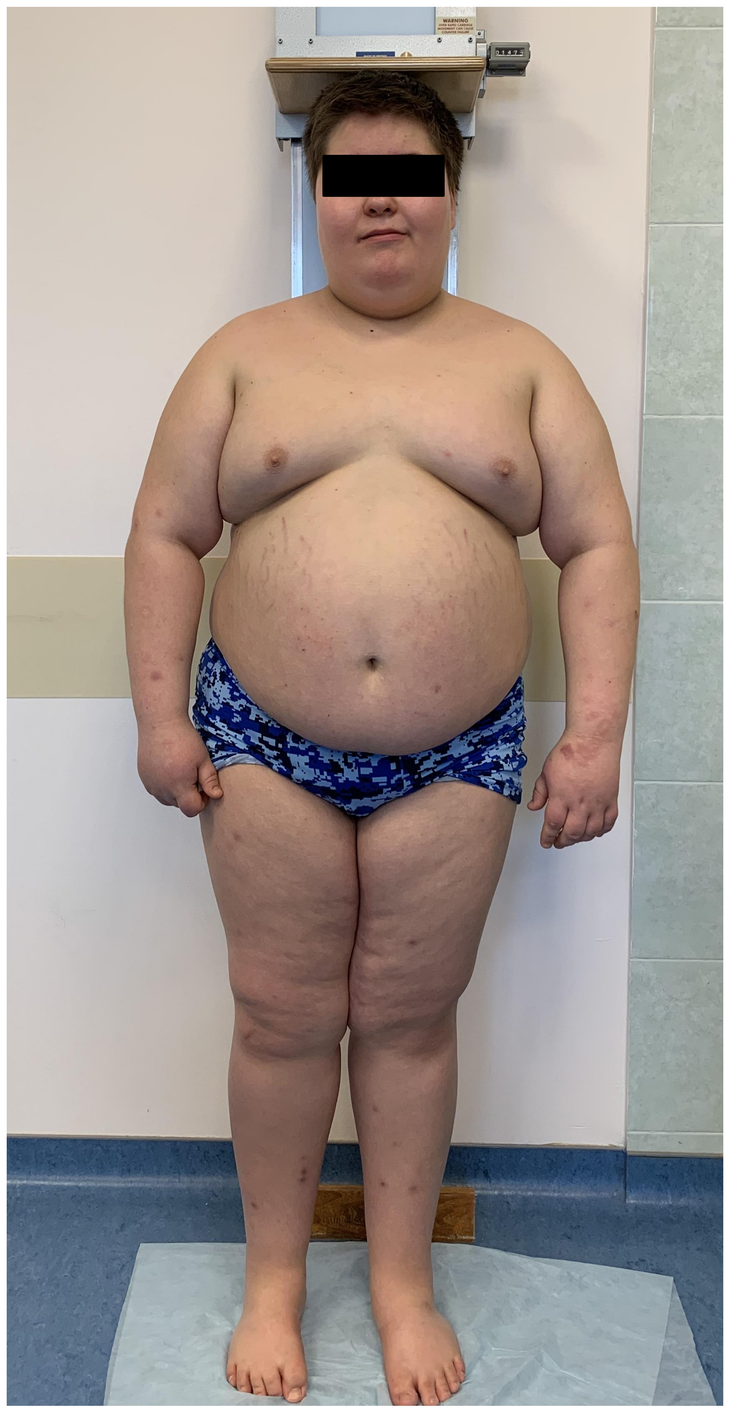

Figure 1. A 14 year old boy with PWS

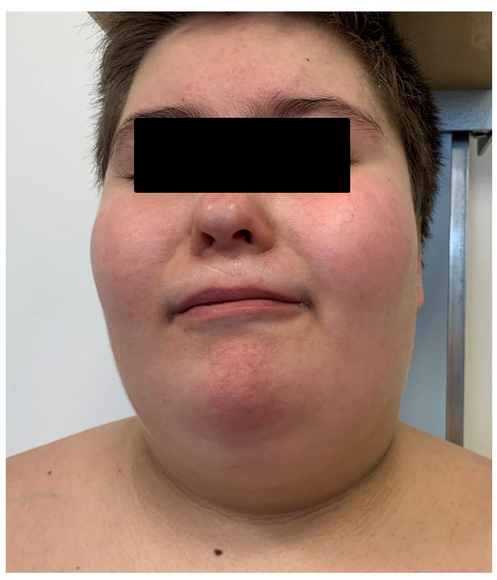

Figure 2. Acanthosis nigricans in a boy with PWS

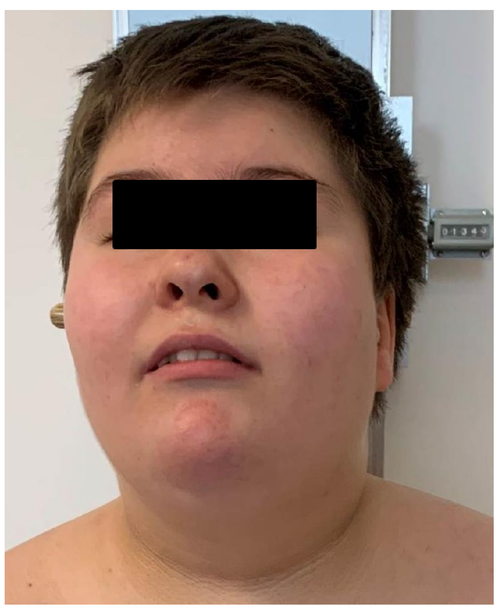

Figure 3. Less severe acanthosis nigricans after 12 days on Metformin treatment