

Nonclassical manifestation of Prader-Willi syndrome patient.

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INTRODUCTION

Prader-Willi syndrome (PWS) is a complex, multisystem disorder and is the most frequent cause of syndromic obesity that arises from lack of expression of paternally inherited imprinted genes on chromosome 15q11-q13. Its major clinical features include neonatal hypotonia, short stature, developmental delay, behavioral abnormalities, hyperphagia, severe obesity, hypothalamic hypogonadism, and characteristic appearance. However «nonclassical forms» of this syndrome exist.

RESULTS

We report on 16 years old girl, who was referred to an endocrinologist due to amenorrhea, overweight and a short stature.

When she was born, she presented neonatal muscle hypotonia, feeding problems. Her development progress was slightly delayed during childhood and she had learning difficulties at school. The girl didn't have the excessive appetite and her weight parameters were normal until 15 years. Growth retardation appeared at 14 years and at 15 - excessive weight gain. Fig.2

Physical examination revealed: height 145,8 cm (SDS=-2,73), weight 59 kg, BMI=27,75 kg/m² (SDS=+1,92), acromicria without facial dysmorphic features, Tanner stage 3, primary amenorrhea. Fig.1

Laboratory data showed IGF-1 level of 165,8 ng/ml, LG level of 0,4 IU/l, FSG 3,23 IU/l, estradiol level was 86,7 pmol/l. Gonadotrophin releasing hormone stimulation test revealed mild hypogonadotropic hypogonadism (LH peak 4,4 IU/l, FSH peak 11,8 IU/l). During GH provocative testing with the insulin GH deficiency was excluded. Dual-energy x-ray absorptiometry revealed osteopenia. Diagnosis of Prader-Willi syndrome was suspected and genetically confirmed by methylation analysis of SNRPN. The girl was placed on sex hormone replacement therapy.

CONCLUSION

These data allow to suggest the variable strengths of various PWS characteristics and our findings also raise questions about more theoretical issues.

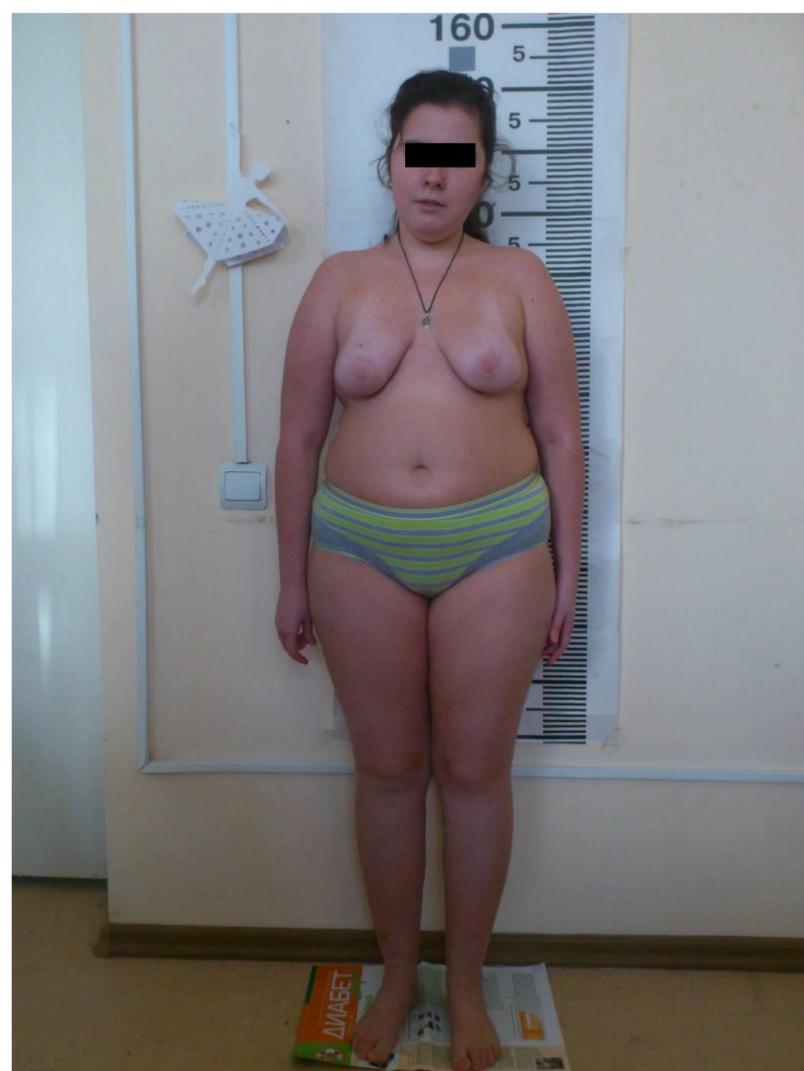


Figure 1. A 16 year old girl with PWS

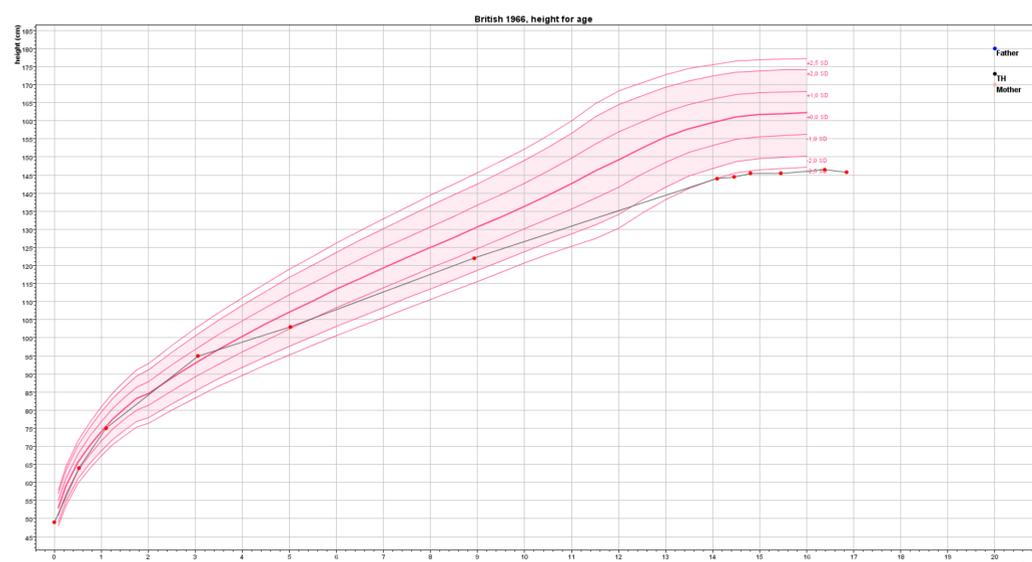


Figure 2. Growth chart of a girl with PWS

