Prader-Willi syndrome (PWS), a multisystem disorder, results in the absence of expression of paternal genes from chromosome 15q11.2-q13; it occurs with the prevalence of 1/10000-1/30000 in different populations. In real clinical practice PWS still remains a challenge for doctors, especially in resource-limited settings. In Belarus, PWS is underdiagnosed; its real rate is unknown.

**AIM AND METHODS**

We analyzed and described clinical course and care problems in 10 pediatric PWS patients (3M; 7F) aged 7,4±3.3 years (1,7-12,3), all have microdeletion of paternally inherited 15q11.2-q13 region. Mean follow-up time is 4,1±2,6 years (0,5-7,6). All patients are under follow-up and receive treatment with growth hormone (GH) in a specialized paediatric and adult's endocrinology center in Minsk.

**RESULTS**

**CHARACTERISTICS OF THE PATIENTS IN PERINATAL PERIOD**

- gestational intrauterine hypoxia – 10/10
- IUGR – 5/10; C section – 6/10
- severe muscle hypotonia as neonates – 10/10 needed feeding tubes as neonates – 10/10
- chiroptichordism in boys at birth – 3/3

Neurological/ clinical genetic exam as neonates – 10/10

**FEATURES OF THE PATIENTS UNDER FOLLOW-UP**

- facial dysmorphia, hypotonia, speech delay – 10/10
- overweight / obesity before 2 y.o. – 9/10
- GH treatment - 10/10
- mean time on GH - 1,9±1,2 (0,3-3,3) y.
- 1,9 y. GV with GH - 10,7±1,1 cm/ year
- central hypothyroidism – 5/10
- central adrenal insufficiency – 0/10
- day 7 and 7 night sleep apnea (clinically) – 3/10; more
- polysonomography – 1/10
- premature adrenarche - 3/7 girls
- metformin treatment – 2/10
- scoliosis – 4/10
- orchidopexy – 3/5 boys
- self-picking – 4/10
- psychiatric medication – 1/10

**REFERENCES**

5. Explain abstract design, methods, results and discussion of the genotype and peripheral sample, time to diagnosis and anthropometric data before commencement of recombinant human growth hormone treatment in Polish patients with Prader-Willi syndrome (diagnostic phase) 2002 May; 17:731-4

**CONCLUSIONS**

- Poor awareness and lack of knowledge about PWS in different paediatric specialists (neonatologists, general paediatricians, neurologists, even geneticists) leads to a delayed PWS diagnosis and postponed treatment / rehabilitation of the children. Education of medical professionals is mandatory.
- Severe perinatal muscular hypotonia at any age, feeding neonatal problems, chiroptichordism in boys since birth, as well as developmental delay and overweight / obesity early in life are strong indications for PWS genetic diagnosis.
- As PWS patients may have different endocrine problems and GH is a recognised treatment for children and adolescents, the early 1st visit to paediatric endocrinologist is beneficial for the patients.
- PWS is a condition that requires life-long medical and social patients' support, a unique multidisciplinary care system is required.
- The foundation of PWS families' organization in Belarus, with professional medical, legal, social support help would to attract more attention of the society to special needs of our patients, to facilitate access to diagnostic /treatment and improve quality of life in PWS.

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**P2-338 DIAGNOSTIC AND FOLLOW-UP PROBLEMS OF MEDICAL CARE FOR PRADER-WILLI SYNDROME CHILDREN IN RESOURCE-LIMITED SETTINGS**

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**INTRODUCTION**

At the age of 2 years (no GH) and 6,8 years (after 3,5 years of treatment with GH – an impressive improvement in growth, body composition, behavior, metabolic control, neuro- and speech development.)

**DESCRIPTION**

**Patient N.O.**

At the age of 13,8 years (no GH) and 12,6 years (after 9 months of treatment with small doses of GH – an improvement in body composition - 10 kg of weight loss, 6 cm plus in growth; easier eating and general behavioral control and better metabolic parameters)