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PRADER-WILLI SYNDROME, DIAGNOSTICS AND CURRENCY FEATURES

Abstract. *Five boys with Prader-Willi syndrome were examined at the endocrinologist by 2016. All children had minimal diagnostic signs of the syndrome, namely: muscular hypotension, hypogonadism, obesity, mental retardation of varying severity, small hands and feet. In two children there was a disruption of glucose tolerance. All patients had the manifestation of hypergonadotropic hypogonadism. If a minimal diagnostic criteria are found in the newborn, a genetic analysis is necessary.*
Key words*: Prader-Willi syndrome, obesity, children, hypogonadism.*

Introduction. The Prader-Willi syndrome (PWS) was first described by the Swiss pediatricians A. Prader and H. Willi in 1956. Its frequency in the world is 1 per 25,000-10,000 newborns.

The Prader-Willi syndrome arises from the deletion of the parent copy of the imprinted SNRPN gene of the small nuclear ribonucleoprotein N polypeptide and the necdin gene, which is adjacent to the mRNA clusters SNORD64, SNORD107, SNORD108 and two copies of SNORD 109, 29 copies of SNORD116 (HBII-85) and 48 copies SNORD115 (HBII-52). They are located on the 15th chromosome in the region 15q11.2-q13. This is the so-called PWS / AS region, which can be lost as a result of the action of one of several genetic mechanisms in most cases as a result of mutations. There are other more rare mechanisms of development of this syndrome: maternal isodisomy, that is, when both chromosomes 15 are obtained from the mother, random mutations, chromosomal translocations and gene deletions [5, 7].

The risk of the birth of a sick child in a family where there is already one patient is completely dependent on the genetic mechanism that caused the disorder. The probability of a sick child's birth is less than 1% if he has a gene deletion or isodisomy, but if the child has a mutation of the region, which is characterized by the phenomenon of imprinting, then the risk rises to 50%, in the case of chromosomal translocations, the occurrence of the disease can be predicted in the next child in 25%. For the diagnosis of all known mechanisms, prenatal testing should be used [1].

Children with Prader-Willi syndrome usually are born full-term with insignificant intrauterine hypotrophy, often in asphyxia, 10-40% previa gluteus. There are two phases of the syndrome [2]. The first is inherent in children of 12-18 months. Immediately after birth, severe muscle hypotension is noted. The symptom is very pronounced, children do not make spontaneous movements and can not suck. As a result, the formation of static and locomotor functions is sharply delayed: they do not hold their heads, they do not sit. The physiological reflexes of the newborns are decreasing or absent: Moro's reflex, step reflex, and Bauer's response. There is also a tendency to hypothermia. There are other anomalies: a high, narrow forehead; almond-shaped incision of the eye slits with thin, drooping eyelids; skin and hair are lighter than all other family members have, hypopigmentation of the iris (in 75% of cases); microdontia, hypoplasia of the cartilages of the auricles, scoliosis, ectropion (eyelid eversion), glaucoma. The second phase of the disease develops in a few weeks, months or until the end of the first - the beginning of the second year of life. Hypotension gradually decreases and bulimia develops: the child constantly experiences hunger, actively searches for food and, as a result, obesity develops.

Subcutaneous fatty tissue is distributed unevenly, most of it on the trunk and proximal parts of the limbs - hips, shoulders. The feet and hands are disproportionately small. This phase begins to attract attention to mental retardation [3]. Hypogonadism is also characteristic. Typically, patients also have a deficiency in FSH and LH secretion, which can lead to a delay in puberty and underdevelopment of the sex glands [4]. In boys, hypoplasia of the penis is observed, in girls hypoplasia of large and small labia and in 50% of cases - the uterus. Further development of diabetes is characteristic. In the blood biochemistry there are no abnormalities [6].

Differential diagnosis is performed with other syndromes accompanied by severe muscle hypotension, delayed psychomotor development (myopathy, spinal amyotrophy, Opitt-Frías syndrome) with obesity syndromes (Lawrence-Moon-Barde-Biddle, Alström, Cohen syndrome, adiposogenital dystrophy, etc.) 8].

Objective of the study was to analyze the course of the Prader-Willi syndrome in children of the Chernivtsi region.

Material and methods. Five children with Prader-Willi syndrome were examined, who were on the endocrinologist's monitoring in 2016.

Results of the study and their discussion. All children with this disease were male. Up to a year, the diagnosis is made for two children (40%), up to 4 years also for two, and one child for 14 years. All children had minimal diagnostic signs of the syndrome, namely: muscle hypotension, hypogonadism, obesity, mental retardation of varying severity, small hands and feet.

General clinical tests showed no abnormalities. Two children had a disruption of glucose tolerance, which was proved with a glucose tolerant test. Biochemical blood test did not show any other abnormalities.

Three children had a true bilateral cryptorchidism. So, as in all patients hypogonadism was clinically diagnosed, in all children the level of sex hormones was studied.

The level of hormones in children was: FSH - 14.04 mlOd / ml, testosterone 1.1 ng / ml, LH - 12.6Mod / ml, that is, hypergonadotropic hypogonadism was detected.

All children underwent ultrasound of the thyroid gland. Also, children with suspected autoimmune thyroiditis were assessed for TSH, T3 and T4 levels and the detection of antibodies to thyroid peroxidase. Two children confirmed the diagnosis of "autoimmune thyroiditis, hypertrophic form, euthyroidism."

Two children had short-sightedness and one child suffered from spinal amyotrophy of Wernig-Hoffmann.

All patients received the following treatment: massage, exercise therapy, monitoring changes in the musculoskeletal system, correction of nutrition and cognitive abnormalities, treatment of endocrinological pathology, surgical interventions.

Conclusions. If a child has a low weight and height in the case of full term pregnancy at birth; previa gluteus, some microanomalies of development; pronounced hypotension of the muscles, reduced pigmentation of the skin, iris of the eyes and hair, it is recommended to perform molecular-genetic testing by the FISH method to detect the microdeletion of the 15th chromosome.

Prospects for further research. To study the features of genetic disorders in children of the Chernivtsi region with the Prader-Willi syndrome.

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