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PRADER-WILLI SYNDROME IN THE PRACTICE OF NEONATOLOGISTS

ABSTRACT

Since the late 80-ies of XX century, due to the expansion of laboratory capacity genetics, amount of new genetic disease has increased significantly. Clarification of the etiopathogenesis of the disease is of great importance to address the issue of disease prognosis, therapy and prognosis of the most important thing for parents about the possibility of the birth of their healthy baby. Therefore, caution is so important neonatologist, a thorough history and careful examination of the patient. The article is devoted and little-known wide range of medical problems - diagnosis and treatment of a rare genetic condition Prader-Willi syndrome in newborns admitted to the neonatal pathology and premature babies Perinatal Center №1 "RH №1-NCM" in the period from 2013 2016. We present our 4 cases of laboratory-confirmed Prader-Willi syndrome. On the first stage of nursing infants were suspected neurological conditions with muscular hypotonia, inhibition of unconditioned reflexes, ie, Neonatologist missed. The syndrome has characteristic symptoms, on the basis of which a neonatologist, pediatrician, neurologist, geneticist may be suspected in the newborn disease Prader-Willi. Flaccid syndrome child who manifested suppression of consciousness, faint, brief cry, difficulty in feeding, the oppression of the majority of congenital reflexes, diffuse muscle hypotonia, decreased spontaneous motor activity. The characteristic phenotype: light skin and hair, dolihotsefalicheskaya head shape, narrow high forehead, microgeny, gothic sky, bird-like face, palpebral boys, girls, almond-shaped eyes cryptorchidism boys, girls hypoplasia of the clitoris and the labia minora. The diagnosis was confirmed in all newborns tsitogenetichkim molecular analysis (nuc ish del (15) (q11.2q11.2) (SNRPN) [200]) and found a deletion in a gene SNRPN 100% of the interphase nuclei.

Keywords: newborn, Prader-Willi syndrome, a sluggish child, difficulty in feeding, a mutation.

INTRODUCTION

The basis of hereditary diseases are abnormalities (mutations) of hereditary information - chromosome, gene, and the mitochondrial. Hereditary diseases are numerous (there are more than 6000) and varied in manifestations. Differ hereditary and congenital. Genetic diseases are not always innate, they can occur at different ages: at birth, during childhood, even on the fifth, sixth, seventh decade of life. Some congenital diseases are not hereditary. In particular, some malformations may be associated with the action of harmful factors on the fetus during pregnancy, and the reason for their action is just that, and not damage the hereditary apparatus [1, 3].

One of the rare hereditary diseases is Prader-Willi syndrome, which is caused by the absence of the paternal copy of chromosome 15 site q11-13. In this section of chromosome 15 are the genes involved in the regulation of which genomic imprinting. The frequency - 1: 25000-10000 live births. The syndrome was first described in 1956 by scientists from Switzerland A. Prader, H.Villi and A. Labhart [2, 4]. It should be noted that with a conventional composition chromosomal karyotype studies reveal the pathology impossible. For this purpose special cytogenetic and molecular genetic methods. It turned out that the development of these diseases is associated with the new phenomena of genetic - genomic imprinting and uniparentalnoy Dis. Genomic imprinting - a different expression of the genetic material (homologous alleles) in the chromosomes depending on the paternal or maternal origin, ie, evidence of the influence of parents on the child's phenotype. Until now it was believed that the contribution to proyavlyaemost (expression) of genes of his father and mother are equal. The identified genomic imprinting has demonstrated the presence of selective expression of certain chromosomal loci according to their paternal or maternal origin. The exact cause uniparentalnoy (uniparental) Dis is not currently installed, but found that the inheritance of the two chromosomes is only one parent is a result of a series of genetic and biochemical disorders [1, 3, 4].

Clinically, this syndrome has certain phenotype, hypogonadism, the disease has two phases course: neonatal and up to 3 months of life sluggish baby syndrome with symptoms of a sluggish sucking reflex in an older age bulimia, which leads to obesity. With increasing child has delayed language skills, unnatural flexibility, intelligence decrease, inability to learning [3, 5].

After studying the forums Prader-Willi found that the biggest challenge is the diagnosis. According to the description of parents diagnosed with the data exposed children from 6 months to 7 years. Awareness of doctors neonatologists should be high, in time to submit to the genetics and molecular genetic analysis.

MATERIALS AND METHODS

Clinical observation of the 4 patients with Prader-Willi syndrome were in the department of pathology of newborn and premature babies №1 (OPNND №1) from 2013 to 2016 in the perinatal center of "RH №1-NCM".

RESULTS AND DISCUSSION

Clinical observations of patients in the neonatal period with Prader-Willi syndrome. All infants enrolled in the Department of newborns pathology №1 from central district hospitals, in prenatal diagnosis no features have been identified, and a woman gave birth in maternity hospitals 1 and 2 levels. The age of mothers was older than 30 years, the average age was 34 ± 3, 2, two Russian women, two of the Yakut nationality. Children from repeated pregnancies and childbirth. Three women were married and had one older healthy children. One was remarriage; older two children were from another marriage. In the analysis of obstetric and gynecological history revealed that all women have complications form abortion, miscarriages. The first stirring of the fetus 2 women felt at 22-23 weeks' gestation and data, women complained about the low mobility of the fetus. In 1 case, the pregnancy was in the breech position. 3 pregnant women gave birth by surgery, indications for surgery were the: pre-eclampsia, threatening asphyxia, breech presentation. Only one woman's pregnancy ended in premature birth. In terms of physical parameters one newborn had hypotrophy 2 extent. The average weight was 3250 ± 139 g, the average growth of 51 ± 2,5 cm, the average head circumference of 35 cm ± 1,9, average chest circumference 33 ± 1,7 cm. Two infants were born with a low Apgar score, the average was 6 ± 1.02 at the end of the first minute, 8 ± 1.03 at the end of the fifth minute. Analysis by gender revealed that boys were more (3), one girl. At 3 neonatal condition at birth is estimated as severe, leading syn-

drome doctors noticed muscle hypotonia and stigma dizembriogeneza. Most of the children were in need of intensive care: two of them in the early neonatal period was carried out artificial lung ventilation, all newborns carried out infusion therapy. During the 36 ± 3.6 days newborn needed nutrition through a tube. The lag in physical development was not due to nutrition through a tube.

All children sluggish baby syndrome manifested suppression of consciousness, sucking and swallowing reflexes are reduced, making it difficult for the feeding process, the oppression of the majority of congenital reflexes, weak, short-lived scream, diffuse muscle hypotonia, decreased spontaneous motor activity, decreased tissue turgor. The characteristic phenotype of both sexes: light skin and hair, dolihocephal head shape, narrow high forehead, microgeny, gothic sky, thin upper lip, low-set ears, cryptorchidism in boys, girls hypoplasia of the clitoris and the labia minora. Small differences by gender: boys was palpebral have almond-shaped girls. Those. as well as in Down syndrome children with Prader-Willi syndrome have a characteristic phenotype, physician neonatologist once is enough to see the look, to suspect the syndrome in the future.

Of the guide diagnosis mainly from sluggish lined baby syndrome. An example of the guide diagnoses: 1. Perinatal defeat of CNS, hypoxic genesis. Flaccid baby syndrome. Exclude spinal amyotrophy Verdniga-Hoffmann. Stigma of disembriogenesis.

2. Perinatal defeat of CNS, traumatic genesis. The syndrome of motor disorders. Stigma disembriogenesis. hip dysplasia - a child born in the breech position, subdural hematoma in posterior lamellar fossa, clinically and hip ultrasound was detected by brain MRT. The rest of the newborn on the results of laboratory and instrumental methods of concomitant somatic pathology detected.

One child exposed to this diagnosis when re-entering the 3 months of age in the psycho-neurological department for rehabilitation therapy, this patient was the first experience of doctors. Diagnosis subsequently had difficulty, Prader-Willi syndrome in newborns 3 was already clinically suspected and arrives at 20 days of life on the results of molecular cytogenetic analysis (nuc ish del (15) (q11.2q11.2) (SNRPN) [200]) is confirmed in medical genetic laboratory. SNRPN revealed a deletion of the gene in 100% of the interphase nuclei.

All newborns in mind dominance of muscular hypotonia and depression sucking reflex receiving massage, massage sucking muscles, physiotherapy. To activate the child used nootropic agents (piracetam). In the above background therapy 3 children discharged with a distinct sucking reflex, moved horn feeding expressed milk. There was positive changes in physical development

CONCLUSION

Clinical data and examples, we would like to expand the boundaries of knowledge physicians neonatologists, pediatricians, specialists neurologists and endocrinologists, as syndrome are diagnostic difficulties Prader-Willi. Promptly put the correct diagnosis and early treatment leads to a more optimistic forecast of the disease and important is the adoption of the child's parent diagnosis. Parent heard that this disease is genetic breakdown is not looking for somebody to blame for the child's illness. In all the above cases, the early neonatal period were diagnosed: spinal amyotrophy Verdniga-Hoffmann, natal trauma of the central nervous system, the prognosis of this group of diseases exclude the hope of a favorable outcome. When properly diagnosed with the child parents are advised to undergo genetic testing before planning further pregnancies, since there is a risk that the next child in the same parents born with Prader-Willi syndrome.

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IMPACT OF HEALTH AND SOCIAL FACTORS AND PERINATAL PATHOLOGY ON THE HEALTH AND QUALITY OF LIFE IN INFANTS

ABSTRACT

Currently, one of the features of the diseases of childhood is the growing prevalence of chronic physical and neuropsychiatric diseases that reduce quality of life. With the increasing incidence of neonatal morbidity the study of quality of life in children is very important. The article describes the medical, demographic and social aspects of neonatal morbidity, impact of perinatal factors on health and quality of life of children.

Keywords: newborns, premature babies, fertility, morbidity, perinatal pathology, quality of life.