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NEUROENDOCRINE DISORDERS IN PRADER-WILLI SYNDROME: FROM MOLECULAR MECHANISMS TO CLINICAL PRACTICE*

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Prader-Willi syndrome (PWS) is a rare genetic disorder belonging to the contiguous gene syndromes caused by abnormal DNA methylation in the Prader-Willi critical region (PWCR) in 15q11.2-q13 region [1]. The diagnosis and molecular cause can be made in the proband using simultaneous DNA methylation analysis and combined oligo-SNP microarray analysis (OSA). DNA methylation analysis detects exclusively maternal imprinting within the PWCR, while OSA allows identification of the molecular cause in patients with 15q11.2-q13 deletion, imprinting center deletion, as well as uniparental isodisomy and segmental isodisomy. In 2024–2025, the overall prevalence of this syndrome was 1 case per 10,000–30,000 population [2]. Between 2005 and 2021, the average birth incidence of Prader-Willi syndrome (PWS) in South Korea was 6.8 per 100,000 live births, with a notable increase observed after 2016. The median age at diagnosis was 1.0 year, and growth hormone therapy (GHT) was initiated at a median age of 2.0 years. Intensive care unit (ICU) admission was required for 25.5 % of patients, occurring predominantly during infancy. Intellectual disability and/or developmental delay were identified in 68.6 % of the cohort, while type 2 diabetes mellitus was prevalent in 15.1 % of cases [3]. In 2023, approximately 25,000 diagnosed prevalent cases of PWS were identified across the seven major markets (7MM), which include the United States, Germany, France, Italy, Spain, the United Kingdom, and Japan [4]. While PWS is officially recognized within the national registry of orphan diseases in Ukraine [5], substantiated epidemiological data concerning its prevalence and mortality indicators have not been identified in the available modern scientific publications. The global annual mortality rate among PWS patients is estimated at 1.25 %–3 %. To compare, in the general population this figure is about 1 %. The median age of death in the USA is 23–32 years [6].

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In Sweden, the average age of death is 42 years (increasing life expectancy due to active treatment of diabetes and cardiovascular complications) [7]. In France, this figure is 30 years (70 % of children die before the age of 2 years) [6]. In Japan, the highest mortality rate is at 35–40 years (the highest survival rate due to strict weight control and a developed support system) [8]. Unfortunately, in Ukraine there is no single state mortality registry specifically for the diagnosis of PWS, therefore reliable statistical data could not be found.

In 2025, the FDA approved the first drug to treat hyperphagia in PWS (VYKAT XR), which in future may significantly reduce mortality from gastrointestinal complications and obesity [9].

The drug activates ATP-sensitive potassium (KATP) channels in the hypothalamus, inhibiting the release of neuropeptide Y — one of the body's most potent orexigenic signals. This mechanism leads to attenuated hyperphagia, diminished food-related aggression, and decreased fat mass, which collectively correlate with a lower risk of aspiration, gastric rupture, type 2 diabetes, and morbid obesity [10].

PWS pathogenesis. Three main mechanisms are distinguished in PWS pathogenesis: genetic, molecular, and physiological [11].

The genetic mechanism is characterized by the lack of paternal genes expression in 15q11-q13, resulting in loss of function of SNORD116, MAGEL2, NDN genes. There are three genetic types of PWS:

1. Deletion of the paternal region in 15q11.2-q13 (65–75 % of cases). Microdeletion studies have confirmed that the loss of SNORD116 cluster is critical. Even if all other genes in the region are preserved, isolated loss of SNORD116 can lead to formation of full PWS phenotype. Deletions usually arise due to unequal crossing over during meiosis between breakpoints (BP1, BP2 and BP3) [11]. The patients with type 1 deletions (larger in size, from BP1 to BP3) often have more pronounced cognitive disorders and speech difficulties compared to type 2 (from BP2 to BP3) [12].
2. Maternal uniparental disomy (mUPD) (20–30 % of cases). Modern methods of prenatal diagnosis (NIPT) and mosaicism analysis have shown that mUPD most often occurs as a result of «trisomy rescue». First, a zygote with three chromosomes 15 is formed (due to non-disjunction in maternal meiosis), and then the paternal chromosome is randomly lost [13]. In mUPD, a «double dose» of genes expressed only from the maternal copy (e.g. UBE3A) is observed. This explains the lower risk of severe hyperphagia in early life, but the higher risk of psychotic disorders and autism spectrum disorders in adulthood [14, 15].
3. Imprinting center defects (1–3 % of cases). Both chromosomes 15 are present (one from the mother, the other from the father), but the paternal copy is «switched off» (has a maternal epigenetic marker). The role of epimutations has been detailed. It has been found that methylation disorders in the imprinting center (PWS-IC) lead to inactivation of the entire cluster of genes located below. Normally, the imprinting center «erases» the maternal mark during spermatogenesis. If this does not happen, the paternal chromosome behaves like the maternal one. This is the most difficult option for diagnosis, requiring specific DNA methylation analysis [16].

The molecular mechanism is realized by the disruption of RNA splicing and protein trafficking in cells, which leads to incorrect formation of neuronal connections in the hypothalamus. SNORD116 was found to be a small nucleolar non-coding RNA (SnoRNA) that regulates 2'-O-methylation of ribosomal RNA and affects splicing of other genes. Their deficiency leads to prohormone convertase-1 (PC-1) malfunction in the hypothalamus. As a result, the conversion of proinsulin to insulin and proopiomelanocortin (POMC) to satiety hormones is disrupted [11]. MAGEL2 gene, together with TRIM27 and USP7 proteins, forms MUST complex, which is responsible for retromer-mediated transport. MUST complex regulates the activity of WASH complex [17, 18]. WASH complex (Wiskott-Aldrich Syndrome Protein and SCAR Homolog) is a protein complex that acts as a «molecular architect» by forming an ac-

tin scaffold on the surface of endosomes, which ensures the return of receptors back to the cell membrane [18]. When MAGEL2 is absent, transmembrane proteins (such as leptin receptors LepR and oxytocin receptors OXTR) instead of returning to the membrane enter the lysosomes, where they are degraded [18].

The physiological mechanism is considered to be the result of molecular pathology leading to systemic hypothalamic dysfunctions: appetite dysregulation (hyperphagia), endocrine insufficiency — deficiency of growth hormone (GH), thyroid-stimulating hormone (TSH), sex hormones and autonomic disorders (apnea, thermoregulation disorders) [1, 19]. SNORD116 deficiency leads to impaired development of neurons in the arcuate nucleus (ARC) of the hypothalamus, namely POMC neurons responsible for satiety and AgRP neurons that stimulate appetite suffer [20]. Even with high levels of leptin, ARC neurons cannot adequately process this signal, which leads to uncontrolled hyperphagia [19]. SNORD116 also affects receptor expression, namely decrease in the expression of somatoliberin and creates a state of central resistance to its own growth stimuli [21]. MAGEL2 deficiency leads to a sharp decrease in the number of oxytocin-producing neurons in the paraventricular nucleus (PVN) [22]. From the PVN, oxytocin fibers project to the limbic system, where they regulate empathy, face recognition, and social memory. Some fibers also project to the brainstem (satiety block): to the Nucleus of the Solitary Tract (NTS), where oxytocin suppresses appetite [23]. MAGEL2 is actively expressed in the suprachiasmatic nucleus (SCN), the “biological clock”. Its absence disrupts sleep/wake rhythms, leading to daytime sleepiness and fragmented nighttime sleep [24]. Current clinical studies using magnetic resonance imaging (MRI) and functional magnetic resonance imaging (fMRI) have revealed some features of the brain architecture in PWS patients, namely, a decrease in the volume of gray matter in the prefrontal cortex and cingulate gyrus, the areas responsible for cognitive control and emotional processing, as well as an extension of the lateral brain ventricles. fMRI findings revealed excessive activation of the amygdala and ventral striatum (the so-called «reward centers») after eating [25]. Diffusion tensor imaging (DTI) detected the changes in the white matter of the brain of these patients, in particular, less ordering of white matter fibers, especially in the corpus callosum and fronto-limbic tracts, which is manifested by a decrease in fractional anisotropy [26, 27].

Endocrine homeostasis disorders. PWS is characterized by pronounced disorders of endocrine homeostasis. In childhood, patients are sensitive to insulin against the background of low basal insulin secretion, which later manifests as type 2 diabetes [28]. Almost 90 % of children and adults with PWS have growth hormone deficiency. Stimulation tests show a decrease in the blood somatotropin concentration and a consistently low level of insulin-like growth factor-1 (IGF-1). As a result, these patients are usually short, with signs of obesity, deficiency of muscle mass and bone mineral density [29]. The pathogenesis of growth hormone (GH) deficiency in PWS remains a matter of debate, mainly due to the prevalence of obesity that manifests in early childhood [30]. It is generally accepted that obese individuals without PWS tend to have lower GH secretion compared with those of normal body weight. Obesity is known to lead to changes in IGF, which results in lower availability of IGF-1 [31]. However, most studies show that total circulating IGF-1 levels in obese individuals are normal or even elevated [32]. In PWS patients, IGF-1 levels are often described as low, regardless of body mass index (BMI), indicating true growth hormone deficiency (GHD) [33].

A stimulation test with somatoliberin and simultaneous administration of pyridostigmine in obese children showed an increase in GH levels, which is explained by a decrease in induced somatostatinergic tone [34]. At the same time, children with PWS still showed a lower GH response and reduced serum IGF-1 concentrations, confirming the presence of true GHD [30].

Similar to other endocrine disorders in PWS, thyroid dysfunction is of central origin, namely, it develops due to dysregulation of the hypothalamic-pituitary-thyroid (HPT) axis. In PWS, the most common abnormality of thyroid function is central hypothyroidism, the prevalence

of which varies from 2 % to 72.2 % worldwide [30, 35]. Congenital hypothyroidism is more common in infants and young children with PWS, with the highest incidence in the 1- to 3-year-old age group and a gradual decline over time, probably due to transient dysfunction of the HPT axis [36, 37]. There is no evidence of a genetic association between the prevalence of central hypothyroidism in children with PWS [36], and no association between hypothyroidism and genotype has been found in adults with PWS [35]. In adult patients aged 35–38 years, regardless of gender, who received GH, compared with those who did not receive GH, the mean TSH level was lower at 2.25 ± 1.17 μIU/mL (compared to 2.80 ± 1.44 μIU/mL), and the free T4 level was significantly higher at 1.13 ± 0.70 ng/dL (compared to 1.03 ± 0.11 ng/dL) [38].

Impairment of the hypothalamic-pituitary-adrenal (HPA) axis with an adequate response to stressors, such as infections, is considered a potential cause of sudden and unexplained deaths in PWS [39]. Central adrenal insufficiency was diagnosed in 38 patients (8.5 % of the total study population), including 33 children [40]. About 46.7 % of the examined patients, according to the results of the synacthen simulation test, had the signs of HPA axis disorder, and low-dose hydrocortisone therapy in such patients significantly reduced the symptoms of chronic fatigue and muscle weakness, which were previously attributed only to the genetic features of PWS [41]. The main cause of these disorders is hypothalamic dysfunction, which leads to a deficiency of corticotropin-releasing hormone or adrenocorticotropic hormone. This, in turn, limits the body's ability to produce cortisol, especially during stress, infections, or surgery [42]. Premature activation of the adrenal cortex zona reticularise results in 30 % of children developing premature adrenarche (early appearance of pubic hair and change in sweat odor). This is thought to be related to dysregulation of IGF-1 and changes in androgen metabolism [1]. Most PWS patients receive therapy with recombinant human growth hormone, which has a positive effect on growth, body build, BMI, and potentially psychomotor development in children with this syndrome [43, 44].

The consequence of hypothalamic-pituitary disorders in PWS is hypogonadotropic hypogonadism. In this case, blood gonadotropins concentration, namely luteinizing and follicle-stimulating hormones, is reduced, which manifests by a deficiency of testosterone in men and estradiol in women [45]. In boys with PWS, in addition to the above, primary damage to the testicular tissue is possible, as evidenced by a decrease in the level of anti-Müllerian hormone and inhibin B [46].

CONCLUSIONS

1. Prader-Willi syndrome is a disorder of adjacent genes that develops due to the loss of expression of SNORD116 cluster and MAGEL2 gene. The molecular pathology is based on a deficiency of prohormone convertase 1 and a disorder of retromeric receptor transport, which underlies systemic hypothalamic dysfunction. Physiological disorders are primarily hypothalamic in origin and manifest as uncontrolled hyperphagia due to a defect in POMC neurons, disruption of circadian rhythms, thermoregulation, and oxytocin deficiency, which causes a characteristic behavioral phenotype.
2. The endocrine profile of Prader-Willi syndrome patients is characterized by polyglandular endocrinopathy: somatotropic insufficiency (90 % of cases); hypogonadotropic hypogonadism with a possible combination with primary gonadal damage; central hypothyroidism. Particular attention should be paid to latent adrenal insufficiency, which can cause sudden death in stressful situations.
3. Introduction of new diagnostic methods and approval of specific therapy for hyperphagia open up opportunities for a significant reduction in mortality and improvement of the quality of life of the patients. Creation of a single state registry and improvement of neonatal screening for early detection of the disease remain critically important for Ukraine.

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Prader-Willi syndrome (PWS) is a rare genetic disease belonging to adjacent genes syndromes, caused by abnormal DNA methylation in the Prader-Willi critical region (PWCR) in 15q11.2-q13 region. Three main mechanisms are distinguished in its pathogenesis: genetic, molecular and physiological. The endocrine profile of PWS patients is characterized by polyglandular endocrinopathy: somatotropic insufficiency (90 % of cases); hypogonadotropic hypogonadism with a possible combination with primary gonad lesions; central hypothyroidism. Special attention should be paid to latent adrenal insufficiency, which causes sudden death in stressful situations.

The article presents a review of the current literature on the overall prevalence, mortality, pathogenesis, neuroendocrine disorders, modern diagnostic and therapeutic options. A systematic search of literature sources was carried out in the databases Scopus, PubMed, Web of Science, Embase, the Cochrane Library, MedLine.

Keywords: Prader-Willi syndrome, SNORD116 RNA, pathogenesis, neuroendocrine system, review.

НЕЙРОЕНДОКРИННІ ПОШКОДЖЕННЯ ПРИ СИНДРОМІ ПРАДЕРА-ВІЛЛІ: ВІД МОЛЕКУЛЯРНИХ МЕХАНІЗМІВ ДО КЛІНІЧНОЇ ПРАКТИКИ

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Синдром Прадера-Віллі (СПВ) — це рідкісне генетичне захворювання, що належить до синдромів суміжних генів, зумовлене аномальним метилюванням ДНК у критичній ділянці Прадера-Віллі (PWCR) у локусі 15q11.2-q13. У патогенезі СПВ виділяють три основні механізми: генетичний, молекулярний та фізіологічний. Ендокринний профіль пацієнтів із СПВ характеризується полігландулярною ендокринопатією: соматотропна недостатність (у 90 % випадків); гіпогонадотропний гіпогонадизм із можливим поєднанням із первинним ураженням гонад; центральний гіпотиреоз. Особливу увагу слід приділяти прихованій надниркові недостатності, яка може бути причиною раптової смертності під час стресових ситуацій.

У статті представлено огляд сучасної літератури щодо загальної поширеності та смертності, патогенезу, нейроендокринних порушень, сучасних діагностичних та терапевтичних можливостей. Систематичний пошук джерел літератури проводили в базах Scopus, PubMed, Web of Science, Embase, The Cochrane Library, MedLine.

Ключові слова: синдром Прадера-Віллі, РНК SNORD116, патогенез, гіпоталамус, ендокринна система, огляд