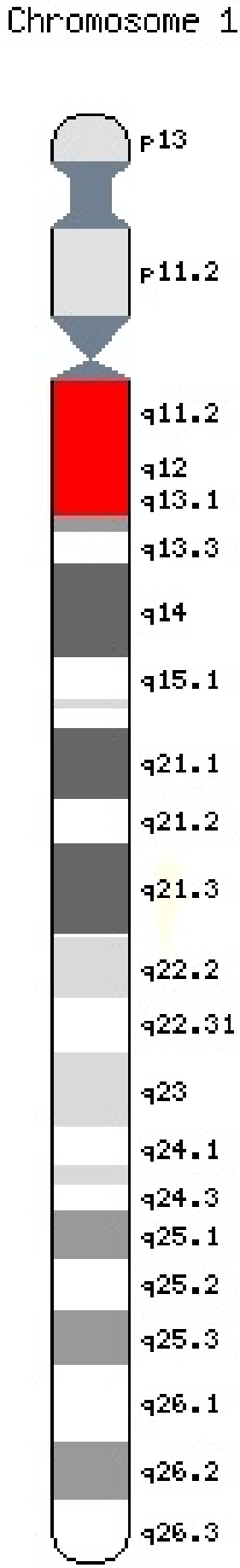
Prader-Willi syndrome

Template:Infobox - genetic disease **Prader–Willi syndrome** (PWS) is a genetically determined disease classified among microdeletion syndromes. The manifestations of PWS are caused by *hypothalamic dysfunction* and vary depending on the patient's age, the disease is characterized mainly by an uncontrollable appetite, short stature, hypogonadism and mild mental retardation. The prevalence is the same in girls and boys.

> The disease was described in 1956 by Swiss doctors - pediatrician and endocrinologist **Andrea Prader'*, internist Alexis Labhart and pediatrician* Heinrich Willi.**[1]

Etiology



The cause at the molecular level is the loss of gene expression in a critical region at

15. of chromosome (15q11−13) originating **from father** (paternal chromosome). The reason may be:

- **microdeletion** in critical region - 70% of cases;
- **uniparental disomy** critical regions (when both present copies of the segment will be of maternal origin) – 25% of cases;
- other damage to the given genes (unbalanced translocations; mutations in the imprinting center)[2][3]

The result is a malfunction of the hypothalamus, which, among other things, regulates feelings of hunger and thirst and releases hormones that affect growth and sexual development.[3] Loss of gene expression in the same region of the 15th chromosome - but of **maternal origin'** - is the molecular cause of **Angelman syndrome'**. The genetic

changes in this region of human chromosome 15 can thus clearly demonstrate the importance of *[Gene imprinting and human pathology|genomic imprinting]] for the development of human diseases.*

Clinical picture

Newborns and infants

- significantly reduced muscle tone (*hypotonia*)
- craniofacial dysmorphia – almond-shaped eye slits, narrowed head in the temple area, thin upper lip
- failure to thrive – partly due to poor sucking reflex
- strabismus
- fatigue, apathy, poor response to stimulation, weak crying[3]

Toddlers and Preschoolers

- unmanageable *craving for food* followed by *obesity* - the cause is a high level of *ghrelin* (orexigenic effects)
- hypogonadotropic *hypogonadism* - insufficient production of GnRH by the hypothalamus is the cause of reduced production of sex hormones and reduced fertility, secondary sexual characteristics are poorly developed
- **small stature'** (around 150 cm), less muscle, short arms and legs
- learning problems
- delayed motor development
- delayed speech development and poor articulation
- behavior disorders - stubbornness and tantrums, usually related to food
- sleep cycle disorder
- scoliosis[3]

Complications

Complications of obesity

- type 2 diabetes mellitus
- cardiovascular disease, myocardial infarction
- sleep apnea syndrome[3]

Complications of hypogonadism

- infertility
- osteoporosis[3]

Treatment

There is no causal treatment. Children with PWS require comprehensive care:

- nutrition – in infancy, high-calorie formulas, later, on the contrary, a low-calorie diet,
- substitution of '*growth hormone,*
- substitution of **sex hormones,**
- surgical treatment - metabolic surgery - "gastric bypass"
- rehabilitation, pedagogical-psychological care, speech therapy care, etc.[3]

Links

Related Articles

- Gene imprinting and human pathologies • Gene imprinting • Uniparental disomy
- Angelman syndrome
- Microdeletion syndromes

Použitá literatura

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Reference

1. http://www.whonamedit.com /synd.cfm/1836.html
2. https://emedicine.medscape.com/article/947954-clinical
3.

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Kategorie:Genetika Kategorie:Pediatrie