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The Role of Growth Hormone Therapy in Managing Pediatric Hereditary Disorders

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Abstract: Growth hormone (GH) deficiency is the most common pituitary hormone deficiency in children and can occur in isolation or in combination with other pituitary hormone deficiencies. In typical cases, growth hormone deficiency in children is characterized by an abnormally low growth rate and short stature while maintaining normal body proportions. Diagnostics include measuring pituitary hormone levels and CT or MRI to detect structural abnormalities of the pituitary gland or brain tumors. Treatment usually includes hormone replacement therapy and removal of the tumor if it is causing the disorder.

Keywords: Growth hormone, panhypopituitarism, GH-releasing hormone receptor, hereditary genetic causes.

Relevance. There is both an isolated deficiency of growth hormone, and associated with panhypopituitarism. In either case, GH deficiency can be acquired or congenital (including due to hereditary genetic causes). In rare cases, there is an abnormality of the GH receptors, which causes insensitivity to the hormone at normal levels in the blood.

Isolated GH deficiency occurs with a frequency of 1/4000 to 1/10000. As a rule, it is idiopathic, but about 25% of cases have an established etiology. Congenital causes include abnormalities of the GH-releasing hormone receptor and GH1 genes, as well as some malformations of the central nervous system. Acquired causes include therapeutic radiation to the central nervous system (high doses of radiation can cause panhypopituitarism), meningitis, histiocytosis, and traumatic brain injury. Preventive or therapeutic irradiation of the spine further reduces the growth potential of the vertebrae and threatens to further reduce the final growth.

Panhypopituitarism can have genetic causes, such as hereditary or sporadic mutations that affect cells in the pituitary gland. In such cases, pathologies of other organs and systems are also encountered, in particular, midline facial defects such as cleft palate or septooptic dysplasia (which includes septum atresia, optic atrophy and hypopituitarism). Panhypopituitarism can also result from a variety of lesions that affect the hypothalamus (disrupting the secretion of releasing hormones) or the pituitary gland, examples include tumors (eg, most commonly craniopharyngioma), infections (eg, tuberculosis, toxoplasmosis, meningitis), and infiltrative processes. The combination of lytic damage to the bones of the skeleton or skull with diabetes insipidus indicates Langerhans cell histiocytosis.

About 3% of children have severe growth retardation. At the same time, growth hormone deficiency as a cause of short stature is detected in no more than 8.5% of them. In other children, the most often revealed constitutional features of growth and development, less often - a deficiency of other anabolic hormones, severe somatic diseases, genetic and chromosomal diseases.

Research objective: The objective of this study is to analyze the role and efficacy of growth hormone (GH) therapy in managing pediatric hereditary disorders that result in short stature and growth retardation. The paper aims to explore both isolated GH deficiencies and conditions associated with panhypopituitarism, as well as the impact of congenital and acquired causes on physical development. A particular focus is given to the genetic and endocrine factors affecting linear growth in children.

Materials and Methods. This study is based on a comprehensive literature review and clinical observations regarding pediatric patients with growth hormone deficiency. Data was gathered from academic sources including medical journals, textbooks, and clinical guidelines focusing on endocrinology. The diagnostic criteria reviewed include serum GH levels, imaging techniques (MRI/CT) to detect pituitary abnormalities, and genetic testing for mutations affecting GH pathways. In addition, case data on treatment responses to hormone replacement therapy were assessed to understand the effectiveness and limitations of GH treatment in children with hereditary short stature.

Stunting can be caused by two fundamentally different problems. The first is when the child is stunted due to an existing disease. Then treatment is needed, which, quite possibly, will really allow him to grow up and maintain health. Which of these factors affect growth? Infectious diseases, heart defects, chronic bone diseases, etc. cause various disorders in the body and retard its growth.

Diseases of the endocrine glands, such as the pituitary gland, the thyroid gland, and the adrenal glands, have a particularly great influence. That is why growth retardation should be the reason for contacting an endocrinologist. The most severe disorders of growth processes are observed in pathology of the endocrine system. It is known that almost all hormones are directly or permissively involved in growth processes.

Human growth is genetically programmed. It is carried out by somatotropin. It is called growth hormone because in children and adolescents, as well as young people with not yet closed growth zones in the bones, it causes a pronounced acceleration of linear (in length) growth, mainly due to the growth of long tubular bones of the extremities. Growth hormone secretion gradually decreases with age. It is minimal in the elderly and the elderly, maximum in adolescents during the period of intense linear growth and puberty. In adults, a pathological increase in the level of growth hormone or prolonged administration of exogenous growth hormone at doses characteristic of a growing organism leads to thickening of the bones and coarsening of facial features. If the secretion of this hormone stops prematurely, then growth stops. This can be for various reasons: injury. diseases, etc. Growth retardation (nanism) caused by a deficiency of growth hormone is one of the urgent problems of the whole society. The disease manifests itself in a significant slowdown in physical development, as a result, children who do not receive appropriate treatment cannot reach the usual average height of an adult over the years and are doomed to remain small throughout their lives, in fact - disabled.

In most cases, growth retardation is due to hereditary factors. Children of short parents with the so-called family delayed maturation (puberty begins later than usual) grow poorly. Usually, this requires only the supervision of a doctor, but sometimes in these cases the help of an endocrinologist may be required: with age, such children, due to slower growth or late onset of puberty, may not reach their maximum possible growth potential inherent in the genes. Currently, with a timely visit to a doctor, this can be avoided and even slightly improved growth rates. In some families, there is a low growth in both parents, then the growth retardation in a child in the absence of signs of any pathology is just a constitutional feature. With such a short stature, no special treatment is required, however, general strengthening measures will help the body realize its existing potential. In the first years of life, growth retardation can occur due to malnutrition, impaired intestinal absorption. If the child is lagging behind in growth from peers, and the reason for the lag is unclear, be sure with the help of a doctor to exclude the latent course of celiac disease in the child - intolerance to gluten (gluten) - a protein found in wheat and some other cereals. Depending on the cause of growth retardation, they are divided: endocrine - associated with a disease of the endocrine glands (for example, underdevelopment or lack of development of the pituitary gland (area of the brain), hypothyroidism (decreased levels of thyroid hormones), etc.); non-endocrine - not associated with endocrine gland disease.

Conclusion. Growth hormone deficiency, whether isolated or associated with panhypopituitarism, remains a significant cause of growth retardation in children. Early diagnosis and timely initiation of GH therapy can greatly improve physical development and help affected children reach a height closer to their genetic potential. However, identifying the exact etiology—especially in hereditary cases—is

crucial for choosing an appropriate treatment strategy. Continued endocrinological supervision and patient-specific approaches are essential to improve quality of life and reduce long-term disability in these children.

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