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The problem of a child with short stature in the paediatrician's and family doctor's office

Problem dziecka z niedoborem wzrostu w gabinecie pediatry i lekarza rodzinnego

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Abstract

Height and weight measurements are an important element of every child's visit to the paediatrician's or family doctor's office. Short stature is defined as a height below the 3rd percentile for age and sex. It is important to identify children in the short stature group (3% of the population) whose short height is not due to familial factors and therefore requires treatment. Correct measurement of the child's height followed by its plotting onto a sex- and age-appropriate percentile chart is the first step to identify the problem of short stature in a child. This should be followed by determining the target height and growth rate. Therefore, regular updating of child's medical record book is of key importance. Diagnostic work-up is needed in children with height below the 3rd percentile or with height difference of more than 2 percentile channels compared to the mean parental height, as well as in children with excessively slow growth rate (indicated by progressive decrease of percentile height position).

Keywords: short stature, children, anthropometric measurements, diagnosis

Streszczenie

Pomiary wzrostu i masy ciała są istotnym elementem wizyty każdego dziecka w gabinecie pediatry czy lekarza rodzinnego. Niedobór wzrostu definiuje się jako wysokość ciała poniżej 3. percentyla w odniesieniu do danego wieku i płci. Ważne jest, aby w grupie, w której stwierdza się niedobór wzrostu (stanowiącej 3% populacji), zidentyfikować te dzieci, u których niski wzrost nie wynika z uwarunkowań rodzinnych i wymaga leczenia. Pierwszym krokiem pozwalającym na stwierdzenie u dziecka problemu niedoboru wzrostu jest prawidłowy pomiar wysokości ciała dziecka i naniesienie odpowiedniej wartości na siatce centylowej, właściwej dla płci i wieku. Następnie powinno się określić wzrost docelowy oraz ustalić dotychczasowe tempo wzrastania. Z tego względu tak istotne jest skrupulatne uzupełnianie książeczek zdrowia. Badań diagnostycznych wymagają dzieci z wysokością ciała poniżej 3. percentyla lub z wysokością ciała różniącą się o ponad 2 kanały centylowe w porównaniu ze średnim wzrostem rodziców, a także dzieci, których tempo wzrastania jest zbyt wolne (wskazuje na to postępujące obniżenie pozycji centylowej wysokości ciała).

Słowa kluczowe: niedobór wzrostu, dzieci, pomiary antropometryczne, diagnostyka

DEFINITION

Short stature is defined as a height below the 3rd percentile for age and sex. Percentile charts of the Department of Child and Adolescent Health of the Institute of Mother and Child, developed by I. Palczewska and Z. Niedźwiecka in 1999, are commonly used for the Polish population[1]. Their advantage is that they allow for an assessment of children from birth to 18 years of age.
Currently, medical record books include World Health Organization (WHO) growth charts for children up to 5 years of age[2] and OLA/OLAF charts, developed by the Children's Memorial Health Institute for children from 3 to 18 years of age[3]. Percentile charts developed by the Institute of Mother and Child based on WHO tables are also available[4]. Some charts use standard deviation (*SD*) rather than percentiles, with 3rd percentile corresponding to −1.88 *SD*[5].

MEASUREMENT METHODS

The diagnosis of short stature begins with a precise measurement of the child's height and plotting the obtained value onto age and sex-appropriate charts.
Correct measurement of body length in children up to 18 months of age should be performed in the supine position, using e.g. an Epstein's bench or a liberometer. The distance from the top of the head to the plantar plane of the feet perpendicular to the lower legs is measured.
In older children, the measurement is performed in a standing position, using a stadiometer or an anthropometer. In primary care practice office, it is possible to measure height using a tape stadiometer.
During the measurement, the child should stand barefoot, in an upright position, with heels, buttocks, shoulder blades and the back of the head adjacent to the plane of the measuring device or the wall. The head should be positioned in the Frankfurt plane (Fig. 1), where the line running through the upper edges of the external auditory openings and the lower edges of the eye sockets is parallel to the ground in the standing position and perpendicular when measured in a supine position. It is advisable to take three measurements and calculate the mean of the two most similar values during one visit. Measurements in a given child should be made at a similar time of day (preferably in the morning), using the same measuring device[6].
Regular anthropometric measurements plotted onto charts allow for early intervention in the case of:
- height below the 3rd percentile;
- low growth rate.

Special attention should be also paid to children whose height significantly deviates (>2 percentile channels) from the target height, calculated from the mean parental height according to the following formula:
- boys: [father's height + (mother's height + 13 cm)]/2;
- girls: [mother's height + (father's height – 13 cm)]/2.

The calculated target height is plotted onto a sex-appropriate percentile chart along the vertical line for the age of 18 years. 8.5 cm on either side of this calculated value represents the 3rd to 97th percentiles for the child's anticipated adult height[7].
An example of a percentile chart for a 9.5-year-old boy is shown in Fig. 2:
- height – 123 cm;
- mother's height – 164 cm, father's height – 184 cm (we add 13 cm to the mother's height on the percentile chart for boys);
- target height calculated from parental height – 180.5 cm.

Measurements plotted onto the percentile chart during each routine health check allow for the assessment of the child's growth rate, calculated on the basis of height increment during at least 6 months of follow-up per year (cm/year)[8].
Tab. 1 shows normal growth rate by age[8,9].
The minimum growth rate is −1.0 *SD*[9,10]:
- at 3 years – 7 cm/year;
- between 3 and 5 years – 6 cm/year;
- between 4 and 10 years – 5 cm/ year;
- in girls over 10 years of age and in boys over 12 years of age (before puberty) – 4 cm/year.

Lower growth rate should raise doctor's concerns.

CAUSES OF SHORT STATURE

Malnutrition is the leading cause of short stature worldwide. In developed countries, on the other hand, constitutional delay in growth and puberty (CDGP) (especially in boys) and familial short stature (FSS) (mainly in girls) are the most common aetiology[5,11].
In the case of children with short stature, it is very important to collect medical history, including:
- perinatal and obstetric history;
- development in infancy;
- chronic conditions;
- frequent infections;
- pharmacotherapy used;
- the curse of puberty in the child, parents and siblings.

Fig. 1. Frankfurt position of the head

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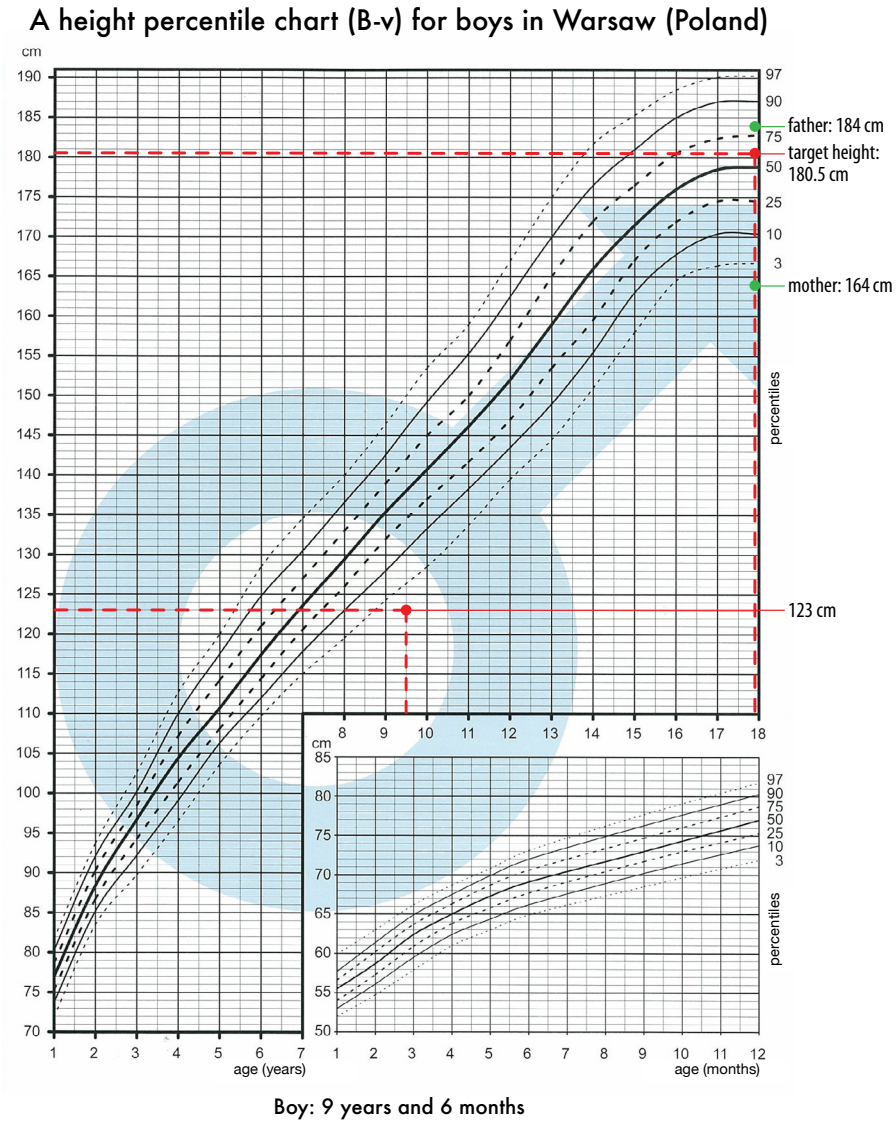

Fig. 2. An exemplary percentile chart for a 9.5-year-old boy (percentile chart developed by the Department of Child and Adolescent Health of the Institute of Mother and Child[1])

Children small for gestational age (SGA), i.e. with birth weight or length <–2.0 *SD*, require close monitoring[12]. Approximately 10–15% of these children will not reach the 3rd percentile of height (i.e. they will not experience the so-called catch-up growth) by the age of 4 years[13]. Importantly, these children can be qualified for treatment with growth hormone under a drug programme financed by the National Health Fund after the age of 4 years, when diagnostic workup is done and other causes of short stature are excluded.

The onset of puberty is an important factor determining the final height of a child. Girls and boys who start puberty before the age 8 and 9 years, respectively, or children with rapidly progressing puberty may initially be significantly taller than their peers, but they have a worse final growth prognosis due to their advanced bone age. These children require urgent endocrine evaluation and diagnosis in an endocrinology department (treatment of precocious and early puberty with poor growth prognosis).

Boys who did not start puberty until the age of 14 years, and girls who did not start puberty before the age of 13 years or who had not started menstruating by the age of 16 years are at the other end of the spectrum. Delayed puberty may run in families, result from hypothalamic-pituitary or gonadal insufficiency, and consequently lead to lower final height. It is important to collect careful family history on parental pubertal timing. Furthermore, girls should be consulted by a paediatric gynaecologist. In the event of symptoms such as headaches, dizziness, visual disturbances, and galactorrhoea, the patient should be urgently referred to the department of endocrinology[5].

Other causes of short stature may include any systemic diseases and mental problems. Urgent referral to an

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Age	Cm/year
0–12 months	About 25
2 years	12–13
3 years	About 8
4–5 years	6–7
From 6 years till puberty	5–6
Puberty	7–9 for girls
	8–11 for boys

Tab. 1. Normal growth rate[8,9]

endocrinologist is particularly necessary for children with growth retardation. The differential diagnosis should include[5]:

- endocrine causes (hypercortisolaemia in Cushing's syndrome, hypothyroidism, poorly controlled diabetes, poorly controlled diabetes insipidus, growth hormone deficiency – GHD);
- anaemia;
- chronic respiratory diseases (e.g. cystic fibrosis, uncontrolled asthma);
- severe congenital heart defects;
- chronic kidney disease;
- gastrointestinal diseases (celiac disease, liver disease, chronic bowel diseases such as Crohn's disease and ulcerative colitis);
- severe immunodeficiencies;
- emotional deprivation[14].

It should also be remembered that chronic use of glucocorticoids (GCs) may also lead to growth retardation. This applies to the use of both systemic and inhaled GCs. GCs inhibit the growth process, e.g. by suppressing the secretion of growth hormone and thyroid stimulating hormone (TSH). The growth of children with chronic diseases and after organ or bone marrow transplantation requiring long-term GC therapy should be monitored. GCs, even at replacement doses, may have a negative impact on the growth processes. It is recommended to use the lowest effective doses of GCs[15,16].

MEDICAL HISTORY AND PHYSICAL EXAMINATION

Tab. 2 shows data from medical history and physical examination of a child with short stature that can be used to guide proper management.

The assessment of dysmorphic features may direct the doctor to diagnose a genetic syndrome associated with short stature. In this case, it is recommended to refer the patient to a genetic clinic. Examples of such hereditary syndromes are shown in Tab. 3.

DIAGNOSTIC WORKUP

Basic tests in every child with short stature should include:

- complete blood count with differential to exclude anaemia;
- kidney and liver function parameters;
- glucose;
- thyroid hormone (TSH; free thyroxine, fT4).

All children are routinely screened for celiac disease based on the measurement of antibodies against tissue transglutaminase. Thyroid hormone testing can rule out hypothyroidism.

Medical history and physical examination data	Proposed management
Growth retardation, constipation, dry skin, brittle hair, delayed tooth eruption, delayed psychomotor development	Diagnosis for hypothyroidism
Growth retardation, excessive body weight, stretch marks, emotional lability	Diagnosis for hypercortisolism
Murmur over the heart, reduced exercise tolerance, reduced physical capacity	Cardiac diagnosis
Growth retardation and uncontrolled asthma	Reduce episodes of asthma exacerbations, use minimum effective doses of GCs
Constipation, diarrhoea, abdominal pain, anaemia, weight loss, blood in the stool	Diagnosis of gastrointestinal disorders
Oedema, reduced physical capacity, fatigue, loss of appetite	Diagnosis of kidney diseases
Growth retardation, visual disturbances, headaches, vomiting, dizziness	Urgent imaging diagnosis of the central nervous system

Tab. 2. Data from medical history and physical examination, and proposed management

Hereditary syndrome	Characteristic symptoms
Turner syndrome	Low hairline, webbed neck, deformed nails, oedema of the hands and feet, recurrent otitis media, cardiovascular defects (e.g. bicuspid aortic valve, aortic coarctation), urinary tract defects (e.g. horseshoe kidney)
Noonan syndrome	Heart defect (pulmonary stenosis), hypertelorism, wide, short neck
Trisomy 21	Intellectual disability, dysmorphic features (epicanthus, upward-slanting palpebral fissures, flat facial profile)
Prader–Willi syndrome	Reduced muscle tone, poor sucking reflex in infancy, uncontrolled appetite and lack of satiety from the age of 2 years
Skeletal dysplasia (achondroplasia)	Short limbs, frontal bossing, macrocephaly
Silver–Russell syndrome	Heterogeneous disorder: growth failure, relative macrocephaly, asymmetry in length of limbs or other body parts
Albright's hereditary osteodystrophy	Obesity, round face, shortened metacarpals (especially IV and V), intellectual disability

Tab. 3. Hereditary syndromes associated with short stature

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Primary care diagnosis	Specialist diagnosis
Complete blood count with differential	Anti-transglutaminase antibodies
Electrolytes	Cortisol
Glucose	Growth hormone (stimulation tests), IGF-1
TSH, fT4	Sex hormones
Hepatic enzymes	Karyotype assessment
Renal function parameters	Hand and wrist radiography to assess bone age
Abdominal US	CNS and pituitary MRI
CNS – central nervous system; **fT4** – free thyroxine; **IGF-1** – insulin-like growth factor 1; **MRI** – magnetic resonance imaging; **TSH** – thyroid stimulating hormone; **US** – ultrasonography.	

Tab. 4. Primary and specialist diagnosis

Item	FSS	CDGP
Parental height	Low	Medium (most often in the father)
Parental puberty	Typical timing	Often delayed (most often in the father)
Birth length	Normal or at the lower limit of normal	Normal
Growth rate	Normal	Slow
Bone age	Metrical	Delayed
Puberty onset	Typical timing	Delayed, extended puberty period
Growth rate during puberty	At the lower limit of normal	Slightly reduced, delayed growth spurt
Final height	Low	Usually normal

Tab. 5. Differential between familial short stature (FSS) and constitutional delay in growth and puberty (CDGP)[18–20]

Tab. 4 shows an exemplary set of tests proposed as part of primary health care and specialist care.

After excluding anaemia, hypothyroidism, celiac disease and other systemic causes, the endocrinologist refers the patient for diagnosis workup for growth hormone deficiency. As part of this workup, tests to assess the secretion of growth hormone are performed in the department of endocrinology [e.g. glucagon, clonidine, arginine, L-dopa and insulin (with the latter one rarely performed nowadays)][17]. Additionally, bone age is assessed based on a radiograph of the non-dominant hand and wrist, which allows to assess the biological age of the child[8]. It is helpful, among others, in the differentiation of short stature aetiology in the affected child with normal growth rate (differentiation of FSS and CDGP). Tab. 5 shows additional features differentiating between FSS and CDGP. In girls, karyotype testing should be performed to rule out Turner syndrome. In the case of confirmed growth hormone deficiency and before qualification for treatment with recombinant growth hormone in SGA children, diagnostic imaging must be performed to exclude organic lesions of the hypothalamic-pituitary region. Since the incidence of GHD is estimated at 1:4,000–1:10,000, it is one of the less common causes of short stature. However, it should be included in the differential diagnosis due to the possibility of effective treatment.

Over 5,000 children are treated for GHD in Poland. In addition to improving the final height, growth hormone treatment has a significant beneficial metabolic effect. The introduction of a new drug programme for adults and adolescents with severe growth hormone deficiency, financed by the National Health Fund, in 2020 allowed for continuing or implementing this treatment modality after completing growth-promoting therapy[5].

CONCLUSIONS

A family doctor plays a crucial role in the diagnosis of short stature in children. Correct measurement of the child's height and plotting the appropriate value onto sex and age-appropriate growth charts allow for the identification of children with short stature. The next step should be to define the target height and determine the current growth rate. Therefore, regular updating of child's medical record book is of key importance.

Collecting a thorough medical history from the parent or guardian is a key component of the child's visit to a family doctor's office. Perinatal and obstetric history is also important. When taking medical history, special attention should be paid to symptoms that may indicate increased intracranial pressure (morning nausea, vomiting, headaches or visual disturbances), which require urgent referral to a neurologist and brain imaging.

Short stature associated with overweight and obesity may be an alarming signal indicating possible secondary obesity. It is observed in children on GCs and children with Cushing's syndrome; if Cushing's syndrome is suspected, the child should be referred to the department of endocrinology.

Dysmorphic features and abnormal body proportions may additionally suggest the presence of hereditary syndromes. In addition to medical history and physical examination, diagnostic tests play an important role. The panel of the available primary care tests allows for the assessment of blood count, glucose, kidney, liver and thyroid function. If hypothyroidism is diagnosed, the child should be referred to the department of endocrinology.

Most children with short stature are healthy and only require monitoring of their growth rate. It is important to identify children whose short stature is not due to familial factors and requires treatment among these 3% of the population.

There are drug programmes reimbursed by the National Health Fund for growth hormone treatment in:

- children with GHD;
- children born too small for their gestational age;
- girls with Turner syndrome;
- children with Prader–Willi syndrome;
- children with chronic renal failure;
- severe growth hormone deficiency in adults and adolescents after completion of growth promoting therapy.

It should be remembered that a short stature child whose parents are short also requires endocrinological

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consultation and diagnostic workup due to the possibility of undiagnosed parental growth hormone deficiency.
Diagnosis should not be postponed, hoping that the child will "grow some more." However, no treatment is attempted after epiphyseal fusion. Systematic anthropometric measurements allow for quick intervention. In the case of failure to report for routine checkups, it is worth measuring children who visit the office, e.g. due to infection.
Also, the psychological aspect of short stature should not be forgotten. Short children are often treated as younger and experience problems with peer acceptance.

Conflict of interest

The authors do not declare any financial or personal links to other persons or organisations that could adversely affect the content of this publication or claim rights thereto.

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