Schimke immune-osseous dysplasia at the junction of specialties. A clinical case of disease diagnosis by allergologists and immunologists

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Abstract

Introduction. Schimke immuno-osseous dysplasia (SIOD) is an autosomal recessive, ultrarare disorder characterized by multisystem involvement accompanied by spondyloepiphyseal dysplasia of the skeleton, steroid-resistant proteinuric nephropathy leading to progressive loss of renal function, impaired immunity, and vascular damage caused by atherosclerosis. SIOD is caused by biallelic pathogenic variations in the SMARCAL1 gene.

The clinical manifestations of SIOD are very diverse: from a rapidly progressive disease in which children die in the first years of life, to milder forms in which they survive to adulthood. The correlation between genotype and phenotype is extremely weak, so it is impossible to predict either the clinical course or the outcome of the disease. For this reason, patients with this pathology can be seen by various specialists.

Case report. The publication presents a clinical case of a 4-year-old boy with immunological deficiency, developmental disorders, and skeletal anomalies, indicating in favor of SIOD.

Whole exome sequencing in the SMARCAL1 gene revealed mutation variants c.2542G>T (p.Glu848Ter); c.1682G>T (p.Arg g561Leu) in a heterozygous state.

Based on the results of the genetic study, and also taking into account that the disease is multisystemic, the child was examined by a nephrologist, orthopedist, endocrinologist and geneticist.

Conclusions of the nephrologist: glomerulopathy in Schimke syndrome: isolated proteinuria. Left calicectasis. Chronic kidney disease (CKD), stage 2. Glomerular filtration rate (Schwartz test) — 69.01 ml/min/1.73 m^2.

Endocrinologist's conclusion: syndromic short stature. Protein-energy malnutrition grade 2.

A telemedicine consultation was conducted with the Federal State Budgetary Institution National Medical Research Center for Pediatric Hematology and Oncology named after D. Rogachev, based on the results of which replacement therapy with intravenous or subcutaneous immunoglobulins was recommended, as well as hospitalization in the immunology department of this federal center.

Conclusion. This description is the first case of diagnostics of an ultrarare disease (1:1–3,000,000 live births) in Krasnodar region by regional specialists. In this patient, the course of the disease is characterized by a non-severe, non-progressive renal dysfunction, which gives reason to assume a milder form of the disease. Conducting replacement therapy with immunoglobulins makes it possible to improve the prognosis in this patient.

Keywords: Schimke's immune-osseous dysplasia, immunodeficiency, nephropathy, skeletal dysplasia, SMARCAL1 gene

Conflict of interest:
The authors declare no conflict of interest.

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Иммунокостная дисплазия Шимке на стыке специальностей. Клинический случай диагностики заболевания врачами — аллергологами-иммунологами

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Резюме

Введение. Иммунокостная дисплазия Шимке (ИКДШ) представляет собой аутосомно-рецессивное, крайне редкое заболевание, характеризующееся мультисистемным поражением, сопровождающимся спондилоэпифизарной дисплазией скелета, стероидрезистентной протеинурической нефропатией, приводящей к прогрессирующей потере функции почек, нарушением иммунитета, а также поражением сосудов, вызванным атеросклерозом. ИКДШ вызывается биаллельными патогенными вариациями в гене SMARCAL1.

Клинические проявления ИКДШ очень разнообразны: от быстро прогрессирующего заболевания, при котором дети умирают в первые годы жизни, до более легких форм, при которых они доживают до зрелого возраста. Корреляция между генотипом и фенотипом крайне слабая, поэтому невозможно предсказать ни клиническое течение, ни исход заболевания. По этой причине пациенты с данной патологией могут попасть на прием к различным узким специалистам.

Описание клинического случая. В публикации представлен клинический случай 4-летнего мальчика с иммунологическим дефицитом, нарушением развития, а также скелетными аномалиями, свидетельствующими в пользу ИКДШ.

При полноэкзомном секвенировании в гене SMARCAL1 обнаружены варианты мутаций c.2542G>T (p.Glu848Ter); c.1682G>T (p.Arg561Leu) в гетерозиготном состоянии.

По результатам полученного генетического исследования, а также учитывая, что заболевание носит мультисистемный характер, ребенок был осмотрен нефрологом, ортопедом, эндокринологом и генетиком.

Заключение врача-нефролога: гломерулопатия при синдроме Шимке: изолированная протеинурия. Каликоэктазия слева. Хроническая болезнь почек (ХБП), стадия 2. Скорость клубочковой фильтрации (проба Шварца) — 69,01 мл/мин/1,73 м^2.

Заключение врача-эндокринолога: синдромальная низкорослость. Белково-энергетическая недостаточность 2-й степени.

Проведена телемедицинская консультация с ФГБУ НМИЦ ДГОИ им. Д. Рогачева, по результату которой было рекомендовано проведение заместительной терапии внутривенными или подкожными иммуноглобулинами, а также госпитализация в отделение иммунологии данного федерального центра.

Заключение. Данное описание является первым случаем диагностики краевыми специалистами крайне редкого заболевания (1:1–3000000 живорожденных) в Краснодарском крае. У данного пациента течение заболевания характеризуется не тяжелым, не прогрессирующим нарушением почечной функции, что дает повод предположить более легкую форму заболевания. Проведение заместительной терапии иммуноглобулинами дает возможность улучшить прогноз у данного пациента.

Ключевые слова: иммунокостная дисплазия Шимке, иммунодефицит, нефропатия, дисплазия скелета, ген SMARCAL1

Конфликт интересов:
Авторы декларируют отсутствие явных и потенциальных конфликтов интересов, связанных с публикацией настоящей статьи.

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INTRODUCTION

Schimke immune-osseous dysplasia (SIOD), first described by Schimke et al. in 1971, is an autosomal recessive extremely rare multisystem disease with a prevalence of 1:1-3000000 live births [1, 2]. It is characterized by multisystemic lesions accompanied

by spondyloepiphyseal skeletal dysplasia, steroidresistant proteinuric nephropathy causing progressive loss of renal function and immune dysfunction [1, 3, 4], as well as vascular damage caused by atherosclerosis [5]. SIOD is caused by biallelic pathogenic variations in the SMARCAL1 gene, which encodes a protein belonging to the SWI/SNF family of proteins involved in chromatin remodeling and regulation of transcription of certain genes [6].

SIOD diagnosis is usually first suspected in a child with nephrotic proteinuria and disproportionate growth retardation. Most children develop terminal renal disease before the age of 10 years. The most common histologic findings on renal biopsy are focal segmental glomerulosclerosis or minimal change disease (7). There is no effective therapy for this renal disease, so patients with terminal renal failure are treated with renal replacement therapy. Some authors suggest kidney transplantation with an abbreviated immunosuppression protocol because of the risk of infection in the post-transplantation period [8].

This disease belongs to congenital immunity errors, a group of combined immunodeficiencies with various syndromal manifestations. Along with cellular immunity disorders, a number of patients also have humoral immunity disorders with disturbances in the number of different classes of immunoglobulins and the level of memory B-cells. Infections associated with both T-cell deficiency (low number of CD4+ (cluster of differentiation) T cells and/or with altered function) and humoral deficiency are usually the most frequent complication of the disease and the main cause of mortality [9].

Skeletal dysplasia is manifested by fetal delay, short stature, femoral head anomalies, flat ovoid vertebrae, and hypoplastic pelvis [10]. Other frequent clinical manifestations include hyperpigmented spots mainly on the trunk, low nose bridge, convex nasal tip, short trunk and neck, lumbar lordosis, protruding abdomen, unusual hair, and malocclusion of teeth, Hypothyroidism, anemia, hypertension, recurrent infections, arteriopathy, episodic cerebral ischemia, bone marrow failure and corneal opacities [5].

Thus, the clinical manifestations of SIOD are very diverse: from rapidly progressive disease, in which children die in the first years of life, to milder forms in which they survive to adulthood. The correlation between genotype and phenotype is extremely weak, so neither the clinical course nor the outcome of the disease can be predicted [10]. For this reason, patients with this pathology may be seen by different subspecialists.

The objective of this study is to describe a clinical case of diagnosis of the extremely rare disease SIOD by specialists of the Children's Regional Clinical Hospital (CRCH) in Krasnodar, Russia.

CLINICAL CASE

The boy's parents have given consent for information about the child to be used for research and publications.

Patient K. (4 years 1 month old) was referred by his local pediatrician to see an allergologist-immunologist at the Children's Diagnostic Center in Krasnodar with complaints of low weight and height gain, leukopenia, lymphopenia in the general blood count (GBC), frequent acute respiratory infections (ARI) during the cold season.

Past medical history data. Child from an unrelated marriage, from the 2nd pregnancy (1st pregnancy in 2014 froze). Delivery of the 1st premature at 28 weeks 6 days of gestation. Birth weight 650 g, height 33 cm, Apgar score 6-7 points. He was discharged at 2 months of life with the diagnosis: “Congenital pneumonia on the background of respiratory distress syndrome (RDS), severe course. Bronchopulmonary dysplasia, new form, moderate severity. Cerebral ischemia of the 2nd degree, oppression syndrome. Prematurity 28 weeks 6 days. Small fetal size for gestational age (3rd degree intrauterine hypotrophy). Hydronephrotic transformation of the right kidney. Cholestasis syndrome”.

Table 1. **Immunoglobulin levels in the patient with SIOD (author's table)**
Таблица 1. **Показатели иммуноглобулинов у пациента с ИКДШ (таблица автора)**

Parameters	Boy K.	Normal value
Immunoglobulin A, g/L	1,18	0,48–3,45
Immunoglobulin M, g/L	1,08	0,4–1,8
Immunoglobulin G, g/L	14,26	5–13
Alpha fetoprotein, IU/mL	2,29	0–5,8

Up to 3 years old he was observed at the Children's Diagnostic Center of CRCH by a pediatrician, nephrologist, neurologist, ophthalmologist, endocrinologist, orthopedist. Since the age of 2 years, he had complaints about periodic onset of chalazion on his eyes, received symptomatic therapy and surgical treatment. At the age of 3 he was consulted by immunologist and hematologist of CRCH, immunologic examination was performed, moderate absolute lymphopenia, moderate decrease in absolute concentration of all subpopulations of T-lymphocytes was revealed, the following diagnosis was made: "Recurrent chalazion. Secondary dysfunction of the immune system. Leukopenia, secondary lymphopenia". At the age of 3, he had an acute community-acquired right-sided pneumonia, without complications.

At the age of 3 years and 6 months, the child's mother visited an endocrinologist with complaints of low growth rate and poor weight gain. A diagnosis was made: "Chronic nutritional deficiency of the 2nd degree. Growth retardation. Subclinical hypothyroidism. Cryptorchidism". L-thyroxine replacement therapy was prescribed.

At the age of 4, he was consulted by an immunologist again. Parents complained about frequent ARI in the cold season, antibiotic therapy twice a year with duration of 5-7 days, changes in blood tests (leukopenia, lymphopenia), poor weight gain and growth.

On examination, weight 10 kg, height 90 cm. Preventive vaccinations in full, according to the national calendar.

The following results were obtained in the CBC: leukopenia ($4{,}78\times10^9$/L), lymphopenia ($1{,}38\times10^9$/L), thrombocytopenia (88×10^9/L).

The results of the child's immunologic examination are presented in Table 1 and Table 2.

Hand radiography in direct projection was performed: bone age corresponds to 2 years (Sadofieva classification was used).

The patient was diagnosed with "unspecified primary immunodeficiency".

A telemedicine consultation (TMC) was held with the Dmitry Rogachev National Medical Research Center for Pediatric Hematology, Oncology and Immunology (FSBI NMRC CGOI named after D. Rogachev). Recommended: full-exome sequencing of deoxyribonucleic acid (DNA).

The child was consulted by a geneticist of the medical and genetic consultation, additional examination was carried out: karyotype 46 XY, normal, male; sex chromatin — 0%, normal, male; blood phenylalanine 1.35 mg/dL (normal); blood thyroid hormone

Table 2. **Main immunological parameters of the patient with SIOD (author's table)**
Таблица 2. **Основные иммунологические показатели пациента с ИКДШ (таблица автора)**

Parameters	Boy K.	Normal value
$CD3^+$, %	33,2	62–69
$CD3^+$, $\times10^9$/л	0,46	1,8–3
$CD4^+$, %	18,1	30–40
$CD8^+$, %	10,3	25–35
$CD3^+/CD4^+$,$\times10^9$/л	0,25	1–1,8
$CD3^+/CD8^+$,$\times10^9$/л	0,14	0,8–1,5
$CD19^+$, %	43,3	21–28
$CD19^+$, $\times10^9$/л	0,6	0,7–13
TREC, $\times10^5$/leukocytes TREC,	127	470–4100
KREC, $\times10^5$/leukocytes KREC,	3174	780–7700

CD — cluster of differentiation; **TREC** — T-receptor excision circle; **KREC** — kappadele recombination excision circle.

2.5 μME/mL (normal); thin layer chromatography of blood amino acids (normal).

Molecular genetic study result: full-exome sequencing: variants c.2542G>T (p.Glu848Ter); c.1682G>T (p.Arg561Leu) were detected in the SMARCAL1 gene in heterozygous state. Validation of the identified variants was performed by Sanger direct automated sequencing (trio). Both variants are of parental origin and are in transposition.

Thus, based on the presence of immune dysfunction, emerging nephropathy, delayed and impaired bone growth, as well as the data of molecular genetic methods of research, the child was diagnosed with "primary immune deficiency (immune-osseous dysplasia Schimke) D84.8".

Based on the results of the genetic study obtained, given that the disease is multisystemic in nature, the child was examined by a nephrologist, orthopedist, endocrinologist and geneticist.

Nephrologist's conclusion: glomerulopathy in Schimke's syndrome: isolated proteinuria. Calicoectasia on the left. Chronic kidney disease (CKD), stage 2.

Results of biochemical blood analysis: urea 7.7 mmol/l; creatinine 73 μmol/l.

Results of the general urinalysis: protein in urine — 0.68 g/L.

Results of estimation of glomerular filtration rate (GFR): Schwartz test 69.01 ml/min/1,73 m^2; Schwartz-Lyon test 46.08 ml/min/1,73 m^2.

Endocrinologist's conclusion: syndromal stunting. Protein-energy deficiency of the 2nd degree.

A repeated TMC with FSBI NMRC CGOI named after D. Rogachev was conducted, which resulted in the following recommendation: replacement therapy with intravenous (IVIG) or subcutaneous immunoglobulin (SCIG), as well as hospitalization in the Department of Immunology of this federal center.

DISCUSSION

We present a clinical case of a 4-year-old boy with immunologic deficits, developmental disorders, and skeletal anomalies suggestive of SIOD. The exact etiology of SIOD is unclear, but mutations in the SMARCAL1 gene have been found in about 50-60% of patients with SIOD [11], which explains the genetic heterogeneity of the disease. Nevertheless, differences in the SMARCAL1 structure have been described as an explanation for different disease severity in patients with the same mutation [12].

SIOD shows phenotypic heterogeneity [13], and disease severity ranges from mild to severe. Patients with SIOD with severe phenotype usually die before the age of five years and are characterized by bone dysplasia, specific facial dysmorphism and T-cell deficiency caused by repeated infection and chromosome fragility [14]. On the other hand, compared to patients with severe SIOD, patients with the mild form have a slower progression of symptoms. Some may present without infections or sometimes clinically asymptomatic, with proteinuria undetectable in early childhood. Patients with mild SIOD usually live to 15 years of age, while some patients may live to 36 years of age [15]. Boerkoel [6] reported heterozygous mutations corresponding to a mild course of the disease. The clinical phenotype found in our patient is exactly as described by Boerkoel [6]: a patient of short stature, with kidney disease and lymphocytopenia without recurrent infections.

In the boy in our study, according to the immunogram data, there are changes in the T-cell population, while no changes were found in humoral immunity (level of CD19+ B-cells, serum immunoglobulins, KREC concentration). Meanwhile, according to the authors' data, SIOD is accompanied by both isolated T-cell deficiency [16, 17] and combined B- and T-cell deficiency [18].

CONSTRAINTS

Constraints of our case report included limited access to the patient's medical history prior to the patient's admission to our facility, which prevented us from obtaining a detailed history of the patient's condition.

CONCLUSION

This description is the first case of diagnosis by regional specialists of an extremely rare disease (1:1-3000000 live births) in the Krasnodar region. In this patient, the course of the disease is characterized by not severe, not progressive impairment of renal function, which suggests a milder form of the disease. Immunoglobulin replacement therapy can improve the prognosis of this patient.

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CONSENT FOR PUBLICATION

Written consent for publication of relevant medical information within the manuscript was obtained from the patients and patient's parents.

ИНФОРМИРОВАННОЕ СОГЛАСИЕ НА ПУБЛИКАЦИЮ

Пациенты и их законные представители добровольно подписали информированное согласие на публикацию персональной медицинской информации в обезличенной форме.