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A multidisciplinary approach to the diagnosis and treatment of isolated 17,20-lyase deficiency: a case report

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ABSTRACT. Isolated 17,20-lyase deficiency is an extremely rare form of congenital adrenal hyperplasia caused by mutations in the *CYP17A1* gene, characterized by impaired androgen synthesis with preserved cortisol production. The clinical picture is often subtle, leading to delayed diagnosis. The description of clinical cases is crucial for understanding the features of this pathology. The article presents a detailed clinical description and analysis of the diagnostic pathway in a 13-year-old female patient with primary amenorrhea and delayed puberty. The examination included assessment of the hormonal profile, pelvic imaging, cytogenetic study, and molecular genetic Sanger sequencing of the *CYP17A1* gene with verification of findings in the parents. The patient with a 46,XX karyotype was found to have hypergonadotropic hypogonadism, cystic transformation of the ovaries, and an abnormally elevated level of 17-OH-progesterone. Genetic analysis revealed compound heterozygous pathogenic variants in the *CYP17A1* gene: c.1039C>T (inherited from the mother) and c.1319G>A (inherited from the father), confirming the diagnosis of isolated 17,20-lyase deficiency. Prescription of combined therapy with low doses of dexamethasone and estradiol valerate led to positive dynamics: the appearance and progression of secondary sexual characteristics, menarche, normalization of the hormonal profile, and regression of ovarian cysts. This case demonstrates the successful diagnosis of an extremely rare isolated form of 17,20-lyase deficiency, which requires a multidisciplinary approach and targeted genetic testing. The combination of pathogenic variants c.1039C>T and c.1319G>A in the *CYP17A1* gene leading to this form of congenital adrenal hyperplasia is described for the first time. Personalized therapy aimed at suppressing the ACTH-dependent pathway and replacing estrogen deficiency proved to be highly effective clinically and in laboratory findings, ensuring normal pubertal development.

KEYWORDS: *congenital adrenal hyperplasia, 17,20-lyase deficiency, CYP17A1, primary amenorrhea, delayed puberty, hypergonadotropic hypogonadism, compound heterozygous mutations, adolescents*

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Оригинальная статья

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Междисциплинарный подход к диагностике и лечению изолированного дефицита 17,20-лиазы: описание клинического случая

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РЕЗЮМЕ. Изолированный дефицит 17,20-лиазы — крайне редкая форма врожденной дисфункции коры надпочечников, обусловленная мутациями в гене *CYP17A1* и характеризующаяся нарушением синтеза андрогенов при сохранной продукции кортизола. Клиническая картина часто стерта, что приводит к поздней диагностике. Описание клинических случаев имеет ключевое значение для понимания особенностей течения этой патологии. В статье представлено подробное клиническое описание и анализ диагностического пути у пациентки 13 лет с первичной аменореей и задержкой полового развития. В рамках обследования выполнены оценка гормонального профиля, визуализация органов малого таза, цитогенетическое исследование и молекулярно-генетическое секвенирование гена *CYP17A1* с верификацией находок у родителей. У пациентки с кариотипом 46,XX выявлены гипергонадотропный гипогонадизм, кистозная трансформация яичников и аномально повышенный уровень 17-ОН-прогестерона. Генетический анализ выявил компаунд-гетерозиготные патогенные варианты в гене *CYP17A1*: c.1039C>T (унаследован от матери) и c.1319G>A (унаследован от отца), что подтвердило диагноз изолированного дефицита 17,20-лиазы. Назначение комбинированной терапии низкими дозами дексаметазона и эстрадиола валерата привело к положительной динамике: появлению и прогрессированию вторичных половых признаков, менархе, нормализации гормонального профиля и регрессу кист яичников. Представленное наблюдение демонстрирует успешную диагностику крайне редкой изолированной формы дефицита 17,20-лиазы, требующей междисциплинарного подхода и целенаправленного генетического тестирования. Впервые описана комбинация патогенных вариантов c.1039C>T и c.1319G>A в гене *CYP17A1*, приводящая к данной форме врожденной дисфункции коры надпочечников. Персонализированная терапия, направленная на подавление адренокортикотропного гормон-зависимого пути и замещение эстрогенового дефицита, доказала свою высокую клиническую и лабораторную эффективность, обеспечив нормальное пубертатное развитие.

КЛЮЧЕВЫЕ СЛОВА: *врожденная дисфункция коры надпочечников, дефицит 17,20-лиазы, CYP17A1, первичная аменорея, задержка полового развития, гипергонадотропный гипогонадизм, компаунд-гетерозиготные мутации, подростки*

Источник финансирования. Авторы заявляют об отсутствии внешнего финансирования при проведении исследования.

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INTRODUCTION

17,20-lyase deficiency is the rarest form of congenital adrenal hyperplasia (CAH), characterized by impaired androgen and cortisol synthesis accompanied by excessive mineralocorticoid production [1]. This deficiency is caused by mutations in the *CYP17A1* gene, which encodes the enzyme 17α-hydroxylase that also possesses 17,20-lyase activity. 17α-hydroxylase converts pregnenolone and progesterone into 17-hydroxypregnenolone and 17-hydroxyprogesterone, while 17,20-lyase subsequently converts 17-hydroxypregnenolone into dehydroepiandrosterone (DHEA) (Fig. 1) [2]. The molecular mechanisms underlying this dual catalytic activity and the imbalance between the two functions in different mutations continue to be actively investigated [3]. The dual catalytic activity of the enzyme explains the existence of different forms of CAH: combined 17α-hydroxylase and 17,20-lyase deficiency, as well as isolated 17,20-lyase deficiency. Notably, most mutations in the CYP17A1 gene result in the loss of both enzymatic functions [4, 5].

Among all forms of congenital adrenal hyperplasia, 17α-hydroxylase/17,20-lyase deficiency accounts for less than 1% of cases. Based on a review

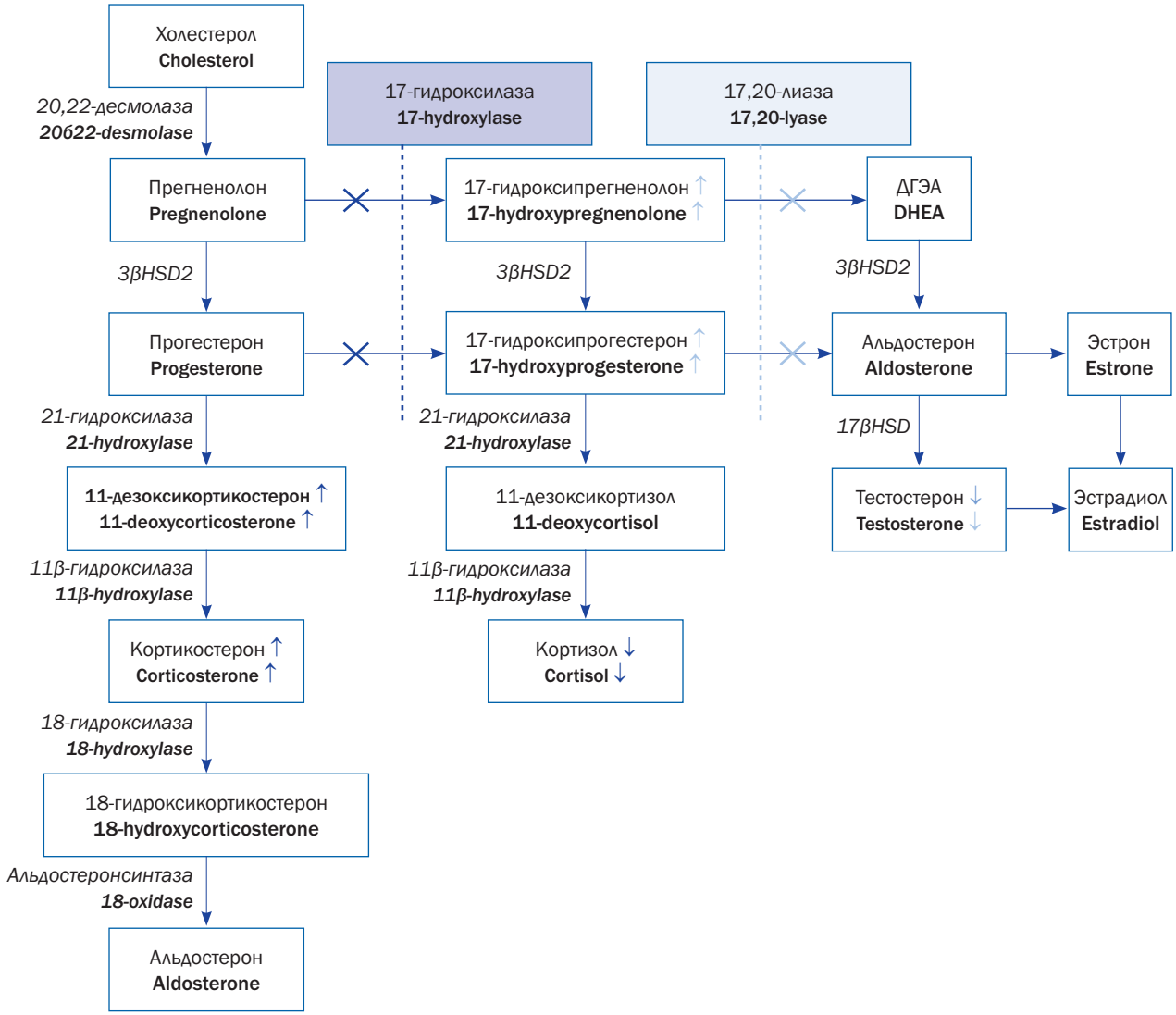

Fig. 1. Scheme of the main steroidogenesis pathways in the adrenal cortex and gonads: DHEA — dehydroepiandrosterone; 3βHSD2 — 3β-hydroxysteroiddehydrogenasetype 2; 17βHSD — 17β-hydroxysteroiddehydrogenase. Solid arrows indicate the direction of enzymatic reactions (adapted by [2])

Рис. 1. Схема основных путей стероидогенеза в коре надпочечников и гонадах: ДГЭА — дегидроэпиандростерон; 3βHSD2 — 3β-гидроксистероиддегидрогеназа типа 2; 17βHSD — 17β-гидроксистероиддегидрогеназа. Сплошные стрелки указывают направление ферментативных реакций (адаптировано по [2])

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of the literature, the prevalence of this deficiency remains uncertain due to the extreme rarity of the disorder and the limited number of clinical cases reported worldwide. To date, no more than 150 individuals with this condition have been described. These data refer primarily to the combined form of the enzyme deficiency, whereas the prevalence of isolated 17,20-lyase deficiency has not been established.

Herein, we present a clinical case of a patient with isolated 17,20-lyase deficiency confirmed by molecular genetic testing. The patient has a normal female karyotype (46,XX). We describe the diagnostic workup for delayed puberty and the observed response to hormone therapy.

CASE PRESENTATION

The patient was born from the second pregnancy and first term delivery. Her birth weight was 3500 g, length 51 cm, and Apgar scores were 8/9. At 7 months of age, she was diagnosed with hereditary spherocytosis (Minkowski–Chauffard syndrome). At 10 years of age, gallstones with clinical signs of cholestasis were detected. At 11 years of age, after prolonged conservative therapy had been ineffective, she underwent laparoscopic cholecystectomy and splenectomy. In the postoperative period, she developed recurrent adhesive disease, which led to a series of relaparotomies for adhesiolysis and abdominal drainage. At 12 years of age, she underwent appendectomy and ileostomy, followed by restoration of gastrointestinal patency and closure of the ileostomy. Only at the age of 13, when she presented with abdominal pain, ultrasound examination revealed uterine hypoplasia and cystic adnexal masses (up to 39 mm). Based on these findings, the girl was consulted by a gynecologist for the first time; there were no indications for surgical treatment, and endocrinological evaluation was recommended. Thus, at approximately 14 years of age, the child was first seen by a pediatric endocrinologist. In addition to abdominal pain, she reported the absence of menstruation and lack of hair growth in androgen-dependent areas. Family history revealed that the mother had menarche at 16 years of age and also had sparse axillary and pubic hair. Obtaining the paternal history was difficult because the parents lived separately.

At the time of examination by the pediatric endocrinologist, the patient had a proportional body habitus with evenly distributed subcutaneous fat. Notably, there was a complete absence of hair growth in androgen-dependent areas as well as on the shins and forearms. Tanner stage was B2, P1, with no menstruation (Me–); no axillary hair was observed, and the external genitalia were normally developed and of female type. Height was average at 163 cm (+0.41 SDS), and weight was appropriate for height at 57 kg (+0.71 SDS). Blood pressure was within the normal range for age.

Outpatient contrast-enhanced magnetic resonance imaging (MRI) of the pelvis was performed. It revealed a multicystic mass in the right ovary measuring 67×61×49 mm, with its medial border adjacent to the left ovary. The left ovary was normally positioned, measuring 28×22×16.8 mm, and contained follicles up to 18 mm within the stroma; no mass lesions were detected. The uterus was in *anteflexio* with no abnormalities; its dimensions were not specified.

Outpatient hormonal testing also demonstrated elevated levels of luteinizing hormone (LH) up to 26.0 mIU/mL and follicle-stimulating hormone (FSH) up to 13.9 mIU/mL. The estradiol level was normal (17 pg/mL). Given the finding of hypergonadotropic hypogonadism, further in-depth evaluation in a specialized endocrinology hospital was recommended.

Upon admission to the endocrinology department, serum electrolytes, liver enzymes, and glucose were within reference ranges. No evidence of thyroid pathology was found, and cortisol was normal. Androgen levels were as follows: 17-hydroxyprogesterone was elevated at 10.2 ng/mL, testosterone was elevated at 1.68 nmol/L. Dehydroepiandrosterone sulfate (DHEA-S) was normal at 0.8 μg/mL, and androstenedione was normal at 0.82 ng/mL. Pelvic ultrasound was performed. It revealed a uterine body length of 50 mm, width of 26.7 mm, and anteroposterior diameter of 12 mm. In place of the right ovary, a polycystic mass measuring 110×90×80 mm was observed. The left ovary measured 30×13×16.8 mm and contained small follicles. Due to the development of catarrhal symptoms, the evaluation was not completed.

One month later, the girl was readmitted, again presenting with abdominal pain. Repeat hormonal testing showed a decrease in androgen levels. Specifically, 17-hydroxyprogesterone was 6.87 ng/mL (previously 10.20 ng/mL), and androstenedione was 0.2 ng/mL (previously 0.82 ng/mL). DHEA-S was 0.03 μg/L (previously 0.8 μg/mL), and testo-

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sterone was 1.12 nmol/L (previously 1.68 nmol/L). LH and FSH levels remained elevated (23.06 and 13.5 mIU/mL, respectively), while estradiol was low at 21.5 pg/mL. Alpha-fetoprotein, beta human chorionic gonadotropin (β-hCG), and the tumor markers CA 19-9 and HE4 were within normal limits. CA 125 was 54 U/L, which decreased to 21 U/L one month later on follow up. Physical examination was unchanged, and sexual development remained the same. Follow up pelvic MRI, compared with previous imaging, showed marked progression of pathological findings. In the region of both ovaries, multilocular masses with heterogeneous (mucinous and hemorrhagic) contents and multiple septations were observed. The right mass measured 77×67×44 mm with a volume of 118 cm³, and the left mass measured 76×51×42 mm with a volume of 85 cm³. A significant amount of heterogeneous fluid was present around the masses. The uterus was hypoplastic, with dimensions including the cervix of 57×12×15 mm. Based on the MRI findings, bilateral ovarian biopsy and diagnostic vaginoscopy were performed. Histopathological examination of the biopsy specimens revealed a follicular cyst of the left ovary and a serous cyst of the right ovary.

The girl was consulted by a clinical geneticist. Given the delayed puberty, karyotyping was performed. Cytogenetic analysis revealed a normal female karyotype (46,XX). The genetic workup for the causes of delayed puberty then focused on monogenic etiologies. Polymerase chain reaction was used to screen for recurrent mutations in the *CYP21A2* gene to exclude the classic form of congenital adrenal hyperplasia (CAH). No variants in the *CYP21A2* gene were identified. A decision was made to rule out *CYP17A1* associated CAH. Sanger sequencing of the *CYP17A1* gene was performed, which identified two pathogenic variants in a heterozygous state: c.1039C>T and c.1319G>A. As a second step, the variants were confirmed by trio sequencing (the patient and her parents). The results confirmed biallelic involvement of the CYP17A1 gene in the patient. The c.1039C>T variant was inherited from the mother, and the c.1319G>C variant was inherited from the father.

Based on the comprehensive laboratory and instrumental findings, together with the clinical presentation, the girl was diagnosed with congenital adrenal hyperplasia due to isolated 17,20-lyase deficiency. The corresponding codes are OMIM #202110, ORPHA:90793, and ICD 10 E25.0.

Based on the above findings, the adolescent was started on replacement therapy with glucocorticoids (dexamethasone 0.25 mg/day) and estrogens (estradiol valerate 0.5 mg/day). On therapy, she developed hair growth on the labia and pubis, breast enlargement, and feminization of her body shape, with menarche occurring after 4 months.

Six months after starting therapy (at 14 years and 10 months of age), the girl was hospitalized in the endocrinology department for follow up assessment of treatment efficacy and safety. Positive changes were observed, including increased feminization of the body habitus with fat redistribution to the thighs and abdomen, breast enlargement, and the appearance of pubic hair (Fig. 2). Blood pressure and heart rate were within the normal range for age. Laboratory testing showed a decrease in LH to 5.63 mIU/mL (from 23.06 mIU/mL before therapy), while FSH remained elevated at 17.73 mIU/mL. Androgen levels also normalized. Specifically, 17-hydroxyprogesterone decreased to 2.24 ng/mL (from 6.87–10.2 ng/mL before therapy), and testosterone decreased to 0.1 ng/mL (from 1.12–1.68 ng/mL before therapy). DHEA S and androstenedione were unchanged, having previously been within reference ranges. Pelvic ultrasound demonstrated positive changes, including an increase in uterine size (length 50 mm, anteroposterior diameter 21.7 mm, width 28.7 mm, cervix 22×19 mm) and a reduction in ovarian volumes. The right ovary measured 49×23.4×23 mm with a volume of 13.8 cm³, and the left ovary measured 34×12.5×12.5 mm with a volume of 2.77 cm³. Given the positive clinical and laboratory response, the dexamethasone dose was maintained at 0.25 mg/day, while estradiol valerate was increased to 1.5 mg/day.

A subsequent laboratory follow up was performed another six months later (at 15 years and 4 months of age), i.e., one year after starting therapy. A height gain of 6 cm over the year was noted, while weight remained unchanged. Tanner stage was B4, P3 4, with menstruation present (Me+); no axillary hair growth was observed. FSH levels finally reached target values at 3.65 mIU/mL (previously 17.73 mIU/mL), and LH was 0.65 mIU/mL. Androgen levels remained low: DHEA S was 0.2 μmol/L, testosterone was 0.08 nmol/L, and 17 hydroxyprogesterone was 2.91 nmol/L. Pelvic ultrasound showed no adverse changes. No adjustment was made to hormone replacement therapy. The adolescent continued under regular follow up.

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Fig. 2. The patient's appearance during hormone replacement therapy (photos by the authors)

Рис. 2. Внешний вид пациентки на фоне заместительной гормональной терапии (фото авторов)

DISCUSSION

The most common clinical presentation of combined 17α-hydroxylase/17,20-lyase deficiency is a pubertal female patient with delayed puberty (primary amenorrhea and lack of secondary sexual characteristic development), hypertension, and hypokalemia [1, 6]. The classic triad of pronounced arterial hypertension has been repeatedly described in the literature [7]. Such children initially present to a pediatrician. However, the symptoms of arterial hypertension and hypokalemia are not consistently present [8–10] and occur mainly in cases of complete absence of 17α-hydroxylase activity [11]. Because of abdominal pain due to developing ovarian polycystosis, which may subsequently impair reproductive function [12], the next physician managing such a child is often a surgeon [13]. It is only after this that adolescent girls present to a pediatric gynecologist or endocrinologist, as illustrated by the present clinical case.

In our case, the patient presented with primary amenorrhea, lack of secondary sexual characteristic development, abdominal pain, and a history of multiple surgical interventions. At the same time, she had a complete absence of arterial hypertension and hypokalemia, which are characteristic of 17α-hydroxylase deficiency. This finding led us to suspect isolated 17,20-lyase deficiency.

Sanger sequencing of the *CYP17A1* gene revealed two pathogenic variants in a heterozygous state: c.1039C>T and c.1319G>A. The c.1039C>T variant in the *CYP17A1* gene is known to cause impaired interaction between cytochrome b5 and the 17α-hydroxylase/17,20-lyase enzyme, partially reducing its activity [14]. In this context, 17α hydroxylase activity is better preserved than 17,20-lyase activity [9,15]. Cases of compound heterozygous *CYP17A1* variants in 17α-hydroxylase/17,20-lyase deficiency have been reported in the literature, with the clinical presentation varying significantly depending on the mutation combination [10,16,17]. For example, Van Den et al. described two women with a 46,XY karyotype who carried compound heterozygous *CYP17A1* mutations [18]. S. Yamagata et al. reported a 67 year old patient with a homozygous c.1039C>T mutation in the *CYP17A1* gene and a normal female karyotype (46,XX) [9]. This patient had a muted clinical presentation of *CYP17A1* associated CAH, including developed secondary sexual characteristics, menstruation until menopause, hypertension, hypokalemia, and signs of hyperaldosteronism with suppressed renin activity. In our case, the c.1039C>T variant in the *CYP17A1* gene was inherited from the mother, who has sparse hair growth in hormone-dependent areas and relatively late menarche (at 16 years of age). The c.1319G>C variant was inherited from the father. Heterozygous carrier status of a *CYP17A1* mutation is hypothesized to lead to steroidogenesis insufficiency [15]. Our case is the first to report a combination of two heterozygous mutations, c.1039C>T and c.1319G>A, in the *CYP17A1* gene resulting in isolated 17,20-lyase deficiency in our patient.

CONCLUSION

This clinical case clearly demonstrates the effectiveness of a comprehensive approach to diagnosing rare forms of disorders of sex development. Targeted genetic testing played a key role by identifying two pathogenic variants in the *CYP17A1* gene in a compound heterozygous state in a patient with a 46,XX karyotype and primary amenorrhea. This is the first report of a combination of two heterozy-

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gous mutations, c.1039C>T and c.1319G>A, in the *CYP17A1* gene, which confirmed the diagnosis of isolated 17,20 lyase deficiency. Successful therapy combining suppression of ACTH dependent steroidogenesis with low dose dexamethasone and estrogen replacement therapy led to positive outcomes, including the onset of menstruation, development of female secondary sexual characteristics, and regression of cystic ovarian changes. These findings underscore the importance of personalized treatment and the need for lifelong multidisciplinary follow up of such patients.

ADDITIONAL INFORMATION

Authors' contribution. M.S. Ilchenko — collection and analysis of clinical data, writing the original draft (case description); S.A. Laptiev — performing and interpreting molecular genetic analysis, literature review; K.V. Skobeleva — conceptualization of the study, examination and treatment of the patient, manuscript editing; D.R. Tursumetova — literature review, writing the original draft, preparation of illustrations, bibliography design.

All authors read and approved the final version before publication.

Competing interests. The authors declare that they have no competing interests.

Consent for publication. Written consent was obtained from legal representatives of the patient for publication of relevant medical information within the manuscript.

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