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A clinical case of an *LHCGR* gene mutation leading to Leydig cell hypoplasia and disordered sex development

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ABSTRACT

Disorders of sex development (DSD) are increasingly discussed in modern medicine. Advances in diagnostic methods have led to an increased identification of individuals with these conditions. This study presents a rare case of a patient with DSD and describes the treatment strategy. A male infant, aged 11 months, was admitted to the urology department for inpatient treatment with a diagnosis of 46XY DSD accompanied by suspected urogenital sinus and vaginal aplasia. According to the medical history, the child's sex at birth could not be determined with certainty. Externally, the genitalia resembled female structures with incomplete feminization. Karyotyping confirmed a 46XY result. Physical examination in the pediatric hospital revealed that the external genitalia did not correspond to the karyotype. Bilateral, painless, and mobile gonad-like masses were palpated in the inguinal region. The urethral opening was located under a hypertrophied clitoris between hypoplastic labia minora. The labia majora exhibited transverse folds resembling a scrotum, and the vaginal introitus was absent. Whole-exome sequencing identified two mutations in the *LHCGR* gene. Diagnostic laparoscopy confirmed the absence of Müllerian and Wolffian duct remnants. A multidisciplinary medical council recommended that the parents raise the child as a female, and bilateral orchiectomy was performed. A feminizing genitoplasty is planned. This clinical case illustrates the management of a patient with DSD, emphasizing the key factors influencing treatment decisions. The availability of genetic testing in patients with DSD enables precise identification of the underlying cause, allowing for early adaptive surgical intervention, before the age of gender self-identification. Further analysis of these mutations and their clinical manifestations will contribute to improving the management of patients in this population.

Keywords: disorder of sex development; 46 XY; Leydig cell hypoplasia; *LHCGR*.

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Клинический случай мутации в гене *LHCGR*, приводящей к гипоплазии клеток Лейдига и нарушению формирования пола

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АННОТАЦИЯ

В современной медицине всё чаще поднимаются вопросы, связанные с нарушением формирования пола. Благодаря развитию современных методов диагностики выявление таких людей увеличивается. В этой статье дано описание редкого случая пациента с нарушением формирования пола и тактики его лечения. Пациент в возрасте 11 мес поступил на стационарное лечение в Урологическое отделение с диагнозом «нарушение формирования пола, 46XY. Урогенитальный синус? Аплазия влагалища?». Из анамнеза известно, что при рождении ребёнка возникли трудности с определением пола: внешне наружные половые органы напоминали женские с неполной феминизацией. При кариотипировании результат — 46XY. При физикальном осмотре в детском стационаре половые органы не соответствовали кариотипу. В паховой области с обеих сторон пальпировались объёмные образования, напоминающие гонады, безболезненные, подвижные. Уретра открывалась отверстием под гипертрофированным клитором между гипоплазированных малых половых губ. Большие половые губы имели поперечные складки по типу мошонки, вход во влагалище отсутствовал. Было проведено массовое параллельное секвенирование, в ходе которого выявили две мутации в гене *LHCGR*. Диагностическая лапароскопия показала отсутствие зачатков мюллерова или вольфова протоков. По заключению мультидисциплинарного консилиума родителям рекомендовано воспитание ребёнка в женском поле. В связи с чем провели операцию — двустороннюю орхиэктомию. Запланирована феминизирующая генитопластика. Данный клинический случай демонстрирует ведение пациента с нарушением формирования пола с акцентом на основных моментах, определяющих выбор тактики лечения. Имея возможность генетических исследований у пациентов с нарушениями пола, мы можем точно знать причину заболевания и приступить к «адаптивной» хирургии раньше — до возраста гендерной самоидентификации. Анализ данных мутаций и их клинических проявлений позволит лучше ориентироваться в ведении пациентов данной группы.

Ключевые слова: нарушение формирования пола; 46 XY; гипоплазия клеток Лейдига; *LHCGR*.

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BACKGROUND

Disorders of sex development (DSD) are increasingly discussed in modern medicine. Analysis of the scientific journal database PubMed showed a more than 2.5-fold increase in publications related to DSD in 2012–2022.

DSD is a congenital condition characterized by discrepancies in the biological sex system (i.e., genetic, gonadal, hormonal, and phenotypic sex), often leading to abnormal external genitalia development. In 2006, the European Society for Pediatric Endocrinology and Lawson Wilkins Pediatric Endocrine Society proposed a new DSD classification [1].

The classification includes three main groups: chromosomal DSD, 46 XY DSD, and 46 XX DSD. Individuals with 46 XY DSD present with ambiguous or female external genitalia caused by incomplete intrauterine masculinization and the presence or absence of Müllerian structures. The pathogenesis-based classification of 46 XY DSD includes several subgroups: gonadal development disorders, defects in androgen biosynthesis or action, and other sex-related pathologies (Fig. 1) [2].

To fully understand DSD, knowledge of human sexual development physiology is critical. Normal male development starts with the presence of the *SRY* gene on the Y chromosome, which initiates testicular differentiation from bipotential gonads [4]. Further male differentiation depends on hormonal signaling from placental and pituitary gonadotropins.

The anterior pituitary is stimulated by the hypothalamus to produce luteinizing hormone (LH) and follicle-stimulating hormone (FSH). FSH targets Sertoli cells in the testes. With the influence of this hormone, they begin to produce anti-Müllerian hormone (AMH), which inhibits the differentiation of female reproductive organs from the Müllerian duct. Moreover, LH targets Leydig cells, which are also located in the testes. With the influence of this hormone and chorionic gonadotropin from the placenta, active testosterone production begins [2]. LH later becomes the key factor in male development. It triggers various reactions leading to the development of male internal and external genitalia from the Wolffian duct and genital ridge.

Disruptions during this process can cause incomplete genital masculinization, resulting in a female-like phenotype in genotypic males. Such disorders may be caused by Leydig cell hypoplasia (agenesis), which is a genetic condition inherited in an autosomal recessive manner. It results from mutations in the *LHCGR* gene, which encodes the LH receptor in Leydig cells. The first reported case of this pathology was described in 1976. This genetic mutation is relatively rare, with an estimated prevalence of 1 in 1 million individuals [1].

The LHCGR receptor belongs to the rhodopsin-like family, which is a highly conserved superfamily of G protein-coupled receptors. This receptor is expressed by Leydig cells and obtains signals from LH and human chorionic gonadotropin (hCG). Upon activation, it stimulates several reactions leading to testosterone production. The *LHCGR* gene, located on chromosome 2p16.3, includes 11 exons [5]. A mutation in this gene results in LHCGR receptor dysfunction, leading to function loss. Thus, Leydig cells, which normally produce approximately 90% of the testosterone in the male body, become resistant to LH, eventually leading to hypoplasia [6].

Currently, most researchers distinguish two types of Leydig cell hypoplasia:

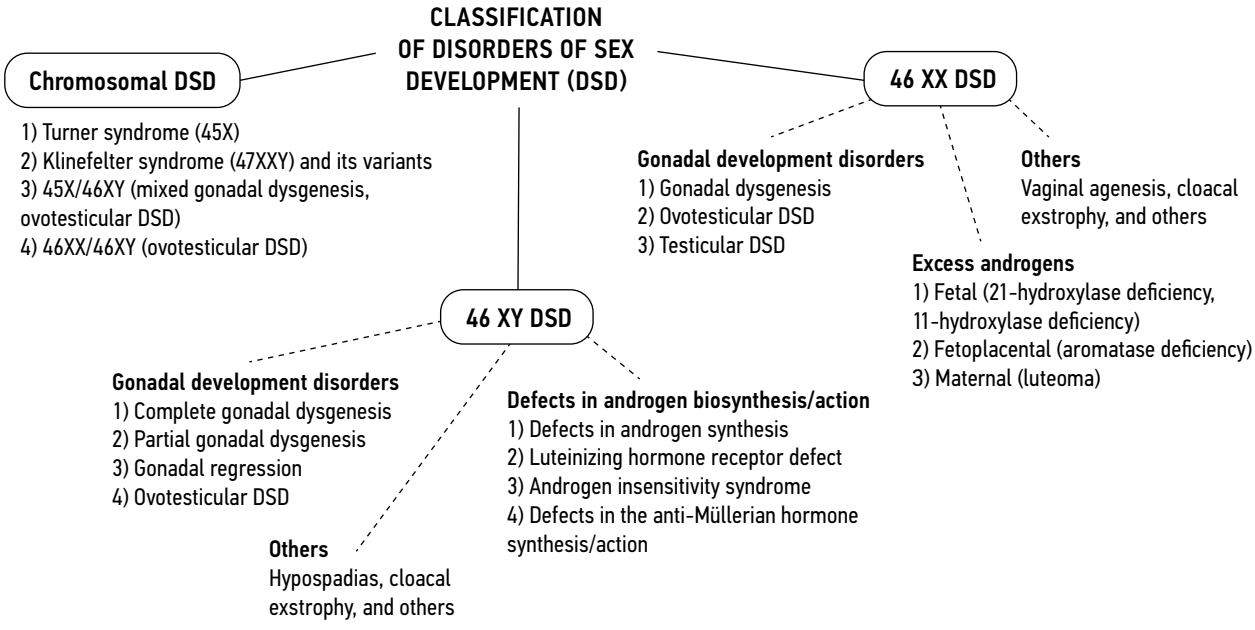

Fig. 1. Classification of disorders of sex development.

• Type 1 (severe form) is characterized by complete insensitivity of receptors to LH and hCG. This results in low blood testosterone levels and increased LH levels. The external genitalia are female-type, possibly with a blind-ending vagina, whereas the testes remain in the inguinal canals.

• Type 2 (milder form) is marked by partial resistance to LH. Clinically, it manifests as incomplete virilization, often with micropenis [7].

This study presents a clinical case of type 1 Leydig cell hypoplasia associated with a previously undescribed *LHCGR* gene mutation.

CASE DESCRIPTION

An 11-month-old patient, born to healthy parents, was admitted for inpatient examination and treatment because of abnormal external genitalia. The child was born from the fifth pregnancy after three full-term deliveries. No other children in the family had sex development abnormalities. During pregnancy, the mother smoked and had a respiratory viral infection in the second trimester. The delivery was complicated by tight umbilical cord entanglement at 40 weeks' gestation. The child was born full-term, weighing 3,484 g and measuring 54 cm in length. The Apgar scores were 8 at the first minute and 9 at the fifth minute.

At birth, determining the child's sex was challenging, as the external genitalia were abnormally developed: female-like genitalia with no vaginal opening, hypoplastic labia minora, enlarged clitoris, skin folds on the labia majora, and bilateral palpable round structures (~1 cm) in the inguinal area, near the labia majora, resembling inguinal hernias.

In the maternity hospital, a pelvic ultrasound was performed. The urinary bladder was normal; however, the uterus and other Müllerian duct derivatives were absent. Round structures, presumably testes, were found in the inguinal canals. On day 3 of life, the patient was transferred to the neonatal pathology department of a children's hospital for further evaluation. A comprehensive examination was conducted to assess the patient's somatic and neurological status, revealing the following comorbid conditions: grade 2 cerebral ischemia with hypertension syndrome, muscle dystonia syndrome, thymomegaly, urinary tract infection, patent foramen ovale, and false chord in the left ventricle. Karyotyping showed a male karyotype (46 XY). A multidisciplinary consultation was held, involving the patient's parents, a pediatrician, a urologist, a gynecologist, a surgeon, endocrinologists, and a geneticist. The passport sex of the child was determined to be female.

Following discharge, the child underwent stepwise examinations in the endocrinology department. Laboratory tests showed normal thyroid hormone levels and low testosterone levels. Subsequently, mass parallel sequencing was performed, revealing two mutations in the *LHCGR* gene. Exon 9 mutation, which has been described in the literature (HG38, chr2:48698650 G>T, c.831 C>A), was identified in a heterozygous state, resulting in the amino acid substitution p. Ser277Arg. In previous studies, this mutation has been reported as male precocious puberty. Computer modeling classified this mutation as an uncertain clinical significance. Exon 11 mutation, which has not been described in the literature (HG38, chr2:48687961A>T, c.1836T>A), was identified in a heterozygous state, leading to a stop codon formation and premature translation p. Tyr612Ter termination. An alternative variant c.1836T>G (p.Tyr612Ter) has been reported to be pathogenic in cases of pseudohermaphroditism. This variant is absent in the Genome Aggregation Database and could result in function loss of the corresponding gene copy. Based on the cumulative data, the exon 11 mutation was classified as a potential pathogenic variant.

After a comprehensive examination, determining the further treatment strategy for the patient was crucial. At age 11 months, the patient was admitted to the urology department, where a repeat ultrasound of the pelvis and inguinal areas was performed. The right testicle was 9.2 × 4.9 mm in size, and the left testicle was 9.0 × 4.7 mm, and the echostructure was normal, with clear, smooth contours. The epididymal head was not differentiated. Physical examination showed that the external genitalia resembled female anatomy: moderate clitoral head hypertrophy and a urethral opening located between the hypoplastic labia minora. No vaginal opening was noted, and the gonads were located in the inguinal areas and were painless, mobile, and appropriate for age (Fig. 2). In this case, using the Quigley and Prader scales to describe the external genitalia was challenging because these scales did not fully cover the observed anomalies.

Urethrocystoscopy was performed to exclude urinary and reproductive system abnormalities: the urethra accommodated a no. 9.5 cystoscope tube, with an unaltered urethral mucosa. The seminal colliculus was absent. The bladder neck had no visible anatomical abnormalities. The bladder contents were clear. The bladder mucosa was intact, and detrusor trabeculation was not pronounced. The ureteral orifices were located in their typical positions and were not dilated. The examination revealed no signs of urogenital sinus or similar anomalies. The absence of the seminal colliculus indicated the absence of a prostate.

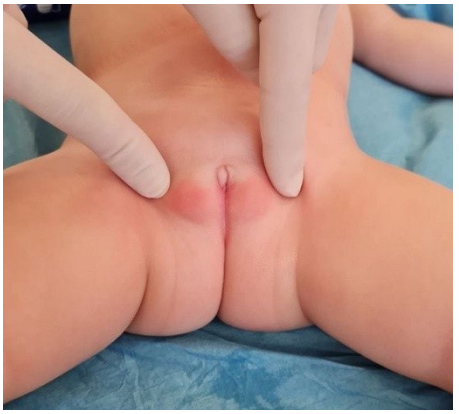

Fig. 2. The appearance of the patient's genitals.

Following these investigations, a multidisciplinary medical council was convened with the parents to determine the patient's further management. During this discussion, the parents, who were fully aware of the risks and nature of the child's pathology, chose to raise the child as female and provided consent for surgery. Laparoscopy was performed to confirm the absence of Müllerian or Wolffian duct remnants, with a possible orchiectomy. At the time of surgery, the child was 11 months old.

Diagnostic laparoscopy detected no uterus, prostate, or seminal vesicles. The surgical procedure concluded with bilateral orchiectomy via an inguinal approach. The excised tissue was sent for histological examination.

The histopathological report revealed scattered hypoplastic Leydig cells. The histological structure of the testes showed immature testicular tissue with a disrupted architecture, seminiferous tubules without lumens, and immature Sertoli cells. Leydig cells were extremely scarce and markedly hypoplastic (Leydig cell aplasia/hypoplasia). The postoperative course was uneventful. Reconstructive surgery for vaginal creation was planned for the future.

Leydig cell hypoplasia, similar to other conditions classified as DSD, is a complex pathology affecting the endocrine, reproductive, and urinary systems. Managing such patients requires a comprehensive approach, involving a wide range of qualified specialists and the use of various diagnostic methods. The Global Disorders of Sex Development Update since 2006: Perceptions, Approach, and Care presents three main approaches to managing patients with DSD: multidisciplinary, interdisciplinary, and transdisciplinary. The multidisciplinary approach involves simultaneous but independent contributions from team members representing two or more disciplines. In the interdisciplinary approach, specialists work together, each applying their discipline-specific knowledge while integrating their shared expertise to solve a common problem. The transdisciplinary approach entails specialists to work collaboratively to synthesize new treatment solutions by integrating expertise across multiple disciplines [8]. The present study employed a transdisciplinary approach to assess the risks of possible complications and predict the patient's future condition. This allowed determining the patient's legal sex early in childhood and initiate treatment—differing from the 2006 consensus recommendation for prolonged observation of DSD patients without specifying the legal sex. The patient was classified as female based on several key factors:

• Genetic sequencing, which identified two mutations in the *LHCGR* gene

• Low testosterone levels in laboratory tests, along with genetic findings and the absence of Wolffian duct derivatives in instrumental examinations, indicating a more severe pathology—Leydig cell hypoplasia type 1

• Poor prognosis regarding the testicular spermatogenic and hormonal function based on laboratory data

• The high complexity of performing masculinizing surgery in this anatomical variant of the external genitalia, along with the need for functional restoration and risk of postoperative complications

• Risk of malignancy in dysgenetic gonads

The final sex assignment was determined through a medical board discussion with the parents, who ultimately decided to raise the child as female.

Most DSDs are genetically determined; therefore, identifying the underlying genetic mutation improves diagnostic accuracy and classification. Genetic sequencing allows for decoding and detecting DNA alterations. In cases of reproductive system disorders, over 100 genes are analyzed. According to the 2023 European Society for Pediatric Urology (ESPU) data, genetic sequencing is becoming the "gold standard" in the diagnosis of DSD [9].

A growing perspective in medicine indicates that patients with DSD should determine their gender identity upon reaching puberty. However, this concept has its limitations and risks, as previously discussed. Owing to the high oncological risks and chosen female legal sex, the medical board opted for early orchiectomy as a safer course of action. Moreover, in 2017, the Parliamentary Assembly of the Council of Europe, ESPU, and other pediatric urology societies took a stand on DSD surgeries. Notably, limiting surgical interventions in children with DSD to emergency cases does not correspond to the World Health Organization's definition of health, which states that health is not merely the absence of disease but a state of complete physical, mental, and social well-being [9]. This issue is particularly relevant for children, as their body and identity develop over time. Establishing a clear sex identity early in life, along with psychological and physical well-being, reduces the need for multiple complex surgeries and facilitates better social adaptation.

Previously, congenital sex variations were classified only at the phenotypic and chromosomal levels, with correction performed through feminizing or masculinizing surgeries. However, gonadal management remained unclear. Currently, with modern genetic testing, the cause of sex development disorders can be accurately determined, and adaptive surgery can be performed earlier, before gender self-identification, thereby avoiding complications and psychological changes. Understanding the specific genetic defect in a patient enables discovering pathways for restoration or correction without losing valuable time and with greater confidence in our actions. Analyzing these mutations and their clinical manifestations enhances our ability to manage such patients effectively.

ADDITIONAL INFORMATION

Authors' contribution. G.B.R. — conceptualization, formal analysis, writing — original draft, visualization; A.K.Z. — validation, resources, investigation, writing — review and editing, supervision; M.R.S. — resources, writing — review and editing. Thereby, all authors made a substantial contribution to the conception of the work, acquisition, analysis, interpretation of data for the work, drafting and revising the work, final approval of the version to be published and agree to be accountable for all aspects of the work.

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Вклад авторов. Все авторы подтверждают соответствие своего авторства международным критериям ICMJE (все авторы внесли существенный вклад в разработку концепции, проведение исследования и подготовку статьи, прочли и одобрили финальную версию перед публикацией). Наибольший вклад распределён следующим образом: Г.Б.Р. — концептуализация, анализ, исследование, создание черновика, визуализация; З.А.К. — проверка, ресурсы, редактирование рукописи, общее руководство; Ш.М.Р. — ресурсы, редактирование рукописи.

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REFERENCES

1. Jahan S, Abul Hasanat M, Alam F, et al. Leydig cell hypoplasia: a unique paradox in the diagnosis of 46, XY disorders of sex development. *AACE Clinical Case Rep*. 2020;6(3):e117–e122. doi: 10.4158/ACCR-2019-0152

2. García-Acero M, Moreno O, Suárez F, Rojas A. Disorders of Sexual Development: Current Status and Progress in the Diagnostic Approach. *Curr Urol*. 2020;13:169–178. doi: 10.1159/000499274

3. Nikitina IL, Kudrjashova EK, Todieva AM, et al. K voprosu o svoevremennoj diagnostike narushenij formirovanija pola. *Lechaschi Vrach*. 2020;(3):17–21. (In Russ.) doi: 10.26295/os.2020.83.45.002

4. Estermann MA, Smith CA. Applying Single-Cell Analysis to Gonadogenesis and DSDs (Disorders/Differences of Sex Development). *Int J Mol*. Sci. 2020;21(18):6614. doi: 10.3390/ijms21186614

5. Yan M, Dilihuma J, Luo Y, et al. Novel Compound Heterozygous Variants in the LHCGR Gene in a Genetically Male Patient with Female External Genitalia. *J Clin Res Pediatr Endocrinol*. 2019;11(2):211–217. doi: 10.4274/jcrpe.galenos.2018.2018.0197

6. Hall JE, Guyton AG. *Medical Physiology according to Guyton and Hall*. 2nd ed. Kobrin VI, Galagudza MM, Umryukhin AE, editors. Moscow: Logosfera; 2018. 1328 p. (In Russ.)

7. Hassan HA, Essawi ML, Mekkawy MK, Mazen I. Novel mutations of the LHCGR gene in two families with 46,XY DSD causing Leydig cell hypoplasia I. *Hormones*. 2020;19(4):573–579. doi: 10.1007/s42000-020-00226-6

8. Lee PA, Nordenström A, Houk CP, et al. Global Disorders of Sex Development Update since 2006: Perceptions, Approach and Care. *Horm Res Paediatr*. 2016;85(3):158–180. doi: 10.1159/000442975

9. Radmayr C, Bogaert G, Burgu B, et al. *EAU Guidelines on paediatric urology*. European Association of Urology, 2023. 198 p.

СПИСОК ЛИТЕРАТУРЫ

1. Jahan S., Abul Hasanat M., Alam F., et al. Leydig cell hypoplasia: a unique paradox in the diagnosis of 46, XY disorders of sex development // AACE Clin Case Rep. 2020. Vol. 6, N. 3. P. e117–e122. doi: 10.4158/ACCR-2019-0152

2. García-Acero M., Moreno O., Suárez F., Rojas A. Disorders of Sexual Development: Current Status and Progress in the Diagnostic Approach // Curr Urol. 2020. Vol. 13. P. 169–178. doi: 10.1159/000499274

3. Никитина И.Л., Кудряшова Е.К., Тодиева А.М., и др. К вопросу о своевременной диагностике нарушений формирования пола // Лечащий врач. 2020. № 3. С. 17–21. doi: 10.26295/os.2020.83.45.002

4. Estermann M.A., Smith C.A. Applying Single-Cell Analysis to Gonadogenesis and DSDs (Disorders/Differences of Sex Development) // Int J Mol Sci. 2020. Vol. 21, N. 18. P. 6614. doi: 10.3390/ijms21186614

5. Yan M., Dilihuma J., Luo Y., et al. Novel Compound Heterozygous Variants in the LHCGR Gene in a Genetically Male Patient with Female External Genitalia // J Clin Res Pediatr Endocrinol. 2019. Vol. 11, N. 2. P. 211–217. doi: 10.4274/jcrpe.galenos.2018.2018.0197

6. Холл Дж.Э., Гайтон А.Г. Медицинская физиология по Гайтону и Холлу. 2-е изд. Под ред. Кобрина В.И., Галагудзы М.М., Умрюхина А.Е. Москва: Логосфера, 2018. 1328 с.

7. Hassan H.A., Essawi M.L., Mekkawy M.K., Mazen I. Novel mutations of the LHCGR gene in two families with 46,XY DSD causing Leydig cell hypoplasia I // Hormones. 2020. Vol. 19, N. 4. P. 573–579. doi: 10.1007/s42000-020-00226-6

8. Lee P.A., Nordenström A., Houk C.P., et al. Global Disorders of Sex Development Update since 2006: Perceptions, Approach and Care // Horm Res Paediatr. 2016. Vol. 85, N. 3. P. 158–180. doi: 10.1159/000442975

9. Radmayr C., Bogaert G., Burgu B., et al. EAU Guidelines on paediatric urology. European Association of Urology, 2023. 198 p.

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