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Presentation of a Case of XX Male Syndrome (de la Chapelle Syndrome)

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ABSTRACT

Introduction: 46,XX testicular disorder/difference of sex development (46,XX testicular DSD), also known as De la Chapelle syndrome, is a rare disorder of sex development characterized by testicular development and a male phenotype despite a 46,XX karyotype. In most cases, translocation of the **SRY** gene from the Y chromosome to the X chromosome plays a central role in the pathogenesis of the condition. Discordance between fetal sex predicted by cell-free fetal DNA (cfDNA) screening and ultrasonographic findings may represent one of the earliest clues to this disorder.
Case Report: A 35-year-old woman with a history of three miscarriages and one successful delivery was followed during her fifth pregnancy. Second-trimester screening indicated a risk of 1 in 1,700 for Down syndrome. Anomaly ultrasound revealed a left choroid plexus cyst and mild bilateral pyelectasis. Cell-free DNA (cfDNA) screening, with a fetal fraction of 8.82%, indicated a low risk for the chromosomal abnormalities assessed but reported the fetal sex as female. In contrast, the fetal sex was reported as male on multiple subsequent ultrasound examinations, and a detailed ultrasonographic examination demonstrated the presence of testes. Amniocentesis was recommended; however, the parents declined the procedure. The infant was delivered by cesarean section at an appropriate gestational age, with Apgar scores of 10 and 9 at 1 and 5 minutes, respectively. Physical examination revealed a completely male phenotype. Postnatal karyotyping demonstrated a **46,XX** karyotype, raising the possibility of 46,XX testicular DSD. Subsequently, SRY gene testing was performed to investigate the molecular basis underlying the condition, and the result was positive.
Conclusion: This case highlights the importance of recognizing discordance between cfDNA screening results and ultrasonographic findings. In such circumstances, careful ultrasonographic evaluation, genetic counseling, and cytogenetic and molecular investigations may play important roles in the diagnosis of DSD and in planning appropriate postnatal follow-up.

Keywords: 46,XX testicular DSD; de la Chapelle Syndrome; cfDNA; Karyotype; SRY; Disorder of Sex Development; Prenatal Screening.

Introduction

Human sex development is a complex, multistep process that begins at fertilization and results from the coordinated interaction of chromosomal sex, gonadal determination, differentiation of the internal and external genitalia, and the precise regulation of a network of genetic and hormonal factors [1]. Under typical conditions, a 46,XX chromosomal complement is associated with ovarian differentiation and a female phenotype, whereas a 46,XY chromosomal complement is associated with testicular differentiation and a male phenotype. However, disruption at any stage of this process can lead to discordance between chromosomal, gonadal, and anatomical sex, collectively referred to as disorders or differences of sex development (DSD) [1,2]. These conditions encompass a broad clinical and genetic spectrum and may present at birth with atypical or ambiguous genitalia or remain undiagnosed until puberty or adulthood, particularly in the context of gonadal dysfunction, hypogonadism, or infertility [3].

Among the various forms of DSD, conditions associated with a 46,XX karyotype are of particular clinical and biological interest. Depending on the underlying genetic mechanism and the developmental pathway affected, gonadal differentiation in these individuals may result in ovarian tissue, testicular tissue, or a combination of both [4]. One of the rarest and biologically distinctive forms within this group is 46,XX testicular DSD, historically referred to as XX male syndrome or De la Chapelle syndrome. In this condition, individuals have two X chromosomes, yet, contrary to the expected developmental pathway, the gonads differentiate toward testicular development, and in many cases, a male external phenotype develops [5]. The estimated prevalence of this condition is approximately 1 in 20,000 male births, and a substantial proportion of affected individuals are diagnosed later in life during infertility evaluations, largely due to absent or severely impaired spermatogenesis [6].

Testicular development in the embryo is regulated by a complex network of genes involved in gonadal sex determination, among which SRY (Sex-Determining Region of Y) plays a central role. This gene, which is normally located on the short arm of the Y chromosome, initiates downstream pathways, including SOX9, thereby promoting differentiation of the bipotential gonad toward testicular development [7]. In the majority of cases of 46,XX testicular DSD, the SRY gene can be detected in individuals with a 46,XX karyotype. In these cases, translocation of a portion of the Y chromosome containing SRY to one of the X chromosomes, typically during abnormal recombination in paternal meiosis, can activate the testicular differentiation pathway in an embryo lacking an intact Y chromosome [8,9]. However, not all cases of 46,XX testicular DSD are SRY-dependent. In SRY-negative cases, genetic alterations affecting testicular differentiation pathways, including variants involving SOX9, SOX3, NR5A1, and WT1, may contribute to the development of the phenotype. Therefore, a 46,XX karyotype alone is insufficient to determine the underlying molecular mechanism of this disorder, and additional molecular investigations may facilitate more precise patient classification [10,11].

Clinically, 46,XX testicular DSD is often associated with a normal or near-normal male phenotype at birth and may therefore remain undetected during the initial neonatal examination. In many affected individuals, the diagnosis is established after puberty following the development of small testes, gynecomastia, hypogonadism, or infertility. In contrast, some patients may be identified at birth because of varying degrees of genital atypia or ambiguity. This phenotypic variability indicates that the relationship between chromosomal sex, gonadal differentiation, and external genital appearance is not always simple or direct. In some cases, the first indication of the disorder may emerge not from clinical examination but from genetic testing or fetal imaging during pregnancy [11,12].

With advances in fetal diagnostic technologies, the analysis of cell-free fetal DNA (cfDNA) circulating in maternal blood has become one of the most important tools for noninvasive prenatal screening. cfDNA is widely used to screen for common aneuploidies and can also provide information regarding sex chromosomes and fetal sex prediction. Despite its high accuracy, cfDNA remains a screening test and should not be considered equivalent to a definitive diagnostic procedure such as amniocentesis or chorionic villus sampling. Therefore, cfDNA results should be interpreted in conjunction with clinical information, ultrasonographic findings, and, when unexpected findings arise, the results of diagnostic testing [13,14].

One of the important challenges associated with the use of cfDNA is fetal sex discordance, in which the sex predicted by cfDNA differs from the sex observed on ultrasound or determined after birth. This discordance may result from various factors, including technical errors, differences between placental and fetal DNA, mosaicism, sex chromosome abnormalities, or disorders of sex development [15]. Studies have shown that with the increasing use of cfDNA screening, cases of fetal sex discordance are being recognized earlier during pregnancy, and such discordance may represent the first indication of an underlying genetic or chromosomal abnormality or a disorder affecting the pathway of sex development. Therefore, discordant fetal sex results between cfDNA and ultrasonography should not be attributed solely to laboratory or imaging error and warrant systematic evaluation and specialized genetic counseling [16,17].

In recent years, several reports have described the prenatal diagnosis of 46,XX testicular DSD following

unexpected findings on noninvasive prenatal testing (NIPT) or cell-free DNA (cfDNA) screening. For example, one case report described an abnormal NIPT result and a discrepancy between the fetal karyotype and ultrasonographic findings that ultimately led to the diagnosis of 46,XX male syndrome. In another report, 46,XX testicular DSD was identified prenatally, and the presence of SRY on the X chromosome was confirmed by amniocentesis. These reports indicate that integrating information from noninvasive screening, ultrasonography, and diagnostic testing may facilitate the prenatal identification of some rare forms of DSD [18,19].

Nevertheless, discordance between the fetal sex reported by cfDNA and the sex observed on ultrasonography remains an important diagnostic challenge in maternal–fetal medicine, as it encompasses a broad range of potential causes. Differentiating between technical or laboratory errors, genetic differences between placental and fetal tissues, mosaicism, and DSD requires careful and multidisciplinary evaluation. Early recognition of such conditions may, in turn, allow appropriate planning of further investigations, genetic counseling, family preparation, and postnatal follow-up. Clinical guidelines also emphasize that when unexpected findings or ultrasound abnormalities are present, genetic counseling and, when indicated, diagnostic testing should be considered alongside cfDNA screening [20,21].

From this perspective, reporting cases characterized by discordance among cfDNA findings, ultrasonographic observations, and postnatal chromosomal status may increase clinicians' awareness of one of the rare causes of fetal sex discordance. The present case is noteworthy because, during a single pregnancy, cfDNA screening predicted a female fetus, whereas multiple ultrasound examinations and a detailed specialized scan indicated a male phenotype and the presence of fetal testes. Following birth, the newborn had a male external phenotype, while postnatal karyotyping revealed a 46,XX chromosomal complement. Taken together, these findings led to a diagnosis of 46,XX testicular DSD and referral of the newborn for genetic evaluation and multidisciplinary follow-up. Therefore, the aim of this case report is to describe a rare case of 46,XX testicular DSD presenting as prenatal discordance between cfDNA and ultrasonographic findings, to highlight the diagnostic significance of such discordance, and to emphasize the importance of a multidisciplinary approach in the evaluation and management of similar cases.

Case Presentation

A 35-year-old pregnant woman (gravida 5, para 1), with a history of one successful delivery resulting in a healthy female infant, was followed during her fifth pregnancy at Saraam Super-Specialty Hospital, Tehran, Iran. Her previous pregnancy losses occurred between 2017 and 2019 and were managed by curettage. Following evaluation for recurrent pregnancy loss, she underwent lymphocyte immunotherapy and was treated with aspirin and enoxaparin. Her fourth pregnancy, in 2021, resulted in the delivery of a healthy infant. The patient had a history of hypothyroidism and was receiving appropriate thyroid hormone replacement therapy. Before the current pregnancy, routine preconception evaluations were performed, and her medication dose was adjusted because of an elevated thyroid-stimulating hormone (TSH) level. During the current pregnancy, aspirin, folic acid, and vitamins B6 and B12 were continued. A booster dose of lymphocyte immunotherapy was also administered, after which she was permitted to attempt conception. First-trimester screening results were reported as normal. During the pregnancy, cervical shortening was diagnosed, and a cervical cerclage was performed.

Second-trimester screening indicated a low risk of Down syndrome (1 in 1,700). A detailed anomaly scan revealed a left choroid plexus cyst and mild bilateral renal pelvic dilatation (pyelectasis). Because cfDNA screening had not previously been performed, noninvasive prenatal testing (NIPT) was requested. The cfDNA test showed a fetal fraction of 8.82% and indicated a low risk for the chromosomal abnormalities assessed. However, an unexpected finding was reported regarding fetal sex: cfDNA screening predicted a female fetus, whereas the fetal sex had consistently been identified as male on previous ultrasound examinations. Because of this discordance between the cfDNA result and the ultrasonographic findings, further evaluation was recommended. Amniocentesis was proposed to clarify the fetal chromosomal status; however, the parents declined the procedure. Therefore, a repeat detailed ultrasound examination was performed. On specialized ultrasonography, the fetal testes were visualized, supporting a male phenotypic sex despite the female sex predicted by cfDNA screening.

The pregnancy progressed to an appropriate gestational age, and the infant was delivered by cesarean section. Apgar scores were 10 and 9 at the first and fifth minutes, respectively. Physical examination immediately after birth revealed a completely male external genital phenotype. Given the prenatal sex discordance and the discrepancy between the phenotypic sex and the cfDNA result, postnatal cytogenetic evaluation was performed. Karyotyping revealed a 46,XX chromosomal complement.

Based on the combination of a male phenotype, prenatal visualization of the testes, and a 46,XX karyotype, a provisional diagnosis of 46,XX testicular disorder/difference of sex development (46,XX testicular DSD), also known as De la Chapelle syndrome, was considered. The infant was subsequently referred for genetic counseling and

further molecular evaluation. According to the genetic counselor, the most likely underlying mechanism was translocation of the SRY gene from the Y chromosome to the X chromosome, resulting in activation of the testicular differentiation pathway despite the absence of an intact Y chromosome. Subsequently, SRY gene testing was performed to investigate the molecular basis underlying the condition, and the result was positive.

The family received genetic counseling regarding the diagnosis, the need for multidisciplinary follow-up, and potential issues that might arise during childhood and puberty. The infant was subsequently followed in the pediatric clinic, with ongoing assessment and coordination between genetic care and specialized pediatric services.

Discussion

46,XX testicular disorder/difference of sex development (46,XX testicular DSD), classically known as De la Chapelle syndrome, is one of the rarest forms of disorders of sex development and is characterized by testicular development and varying degrees of a male phenotype despite a 46,XX karyotype [22,23]. This condition is of particular clinical importance because, in many affected individuals, the external male phenotype at birth is normal or nearly normal, and the diagnosis may be delayed until adolescence or adulthood during evaluations for infertility, hypogonadism, or other forms of gonadal dysfunction. The prevalence of this condition has been estimated at approximately 1 in 20,000–25,000 male births [10]. In the present case, unlike the typical pattern of delayed diagnosis, the first clue to an underlying disorder of sex development emerged during the fetal period following discordance between the cfDNA result and ultrasonographic findings.

At the molecular level, the most well-established mechanism underlying 46,XX testicular DSD is the translocation of a segment of the Y chromosome containing the SRY gene to one of the X chromosomes. By activating the testicular differentiation pathway, particularly through the molecular network involving SOX9, SRY can direct the fate of the bipotential gonad toward testicular differentiation even in the absence of an intact Y chromosome. This translocation typically occurs during abnormal recombination in paternal meiosis. Consequently, the presence of a 46,XX karyotype does not necessarily imply the complete absence of Y-chromosome-derived genetic material. Conversely, in a subset of patients in whom SRY is not detected, alterations in downstream sex-determining genes and pathways, including SOX9, SOX3, NR5A1, and other factors involved in gonadal differentiation, may contribute to the phenotype [10,24]. Therefore, in the present case, SRY testing and, if negative, broader molecular investigations are important for determining the underlying mechanism.

One of the most important features of the present case was the persistent discordance between the fetal sex reported by cfDNA and the sex observed on ultrasonography. Cell-free DNA in maternal blood is derived predominantly from the placenta; therefore, the genetic information obtained from cfDNA does not necessarily represent a direct analysis of fetal cells. Although cfDNA is among the most accurate noninvasive screening methods for common aneuploidies, it is not a definitive diagnostic test and may be influenced by both biological and technical factors [25]. Accordingly, cfDNA results should be interpreted in the context of clinical and ultrasonographic findings, and additional diagnostic evaluation should be considered when discordance is identified.

In 46,XX testicular DSD, one possible explanation for this discordance is the presence of a small Y-chromosomal segment containing SRY on an X chromosome, while the amount of Y-specific DNA in cfDNA may be insufficient to exceed the detection threshold of the testing method. Under such circumstances, the fetus has a 46,XX karyotype, and cfDNA may therefore report a female fetal sex in the absence of a detectable Y-chromosome signal, whereas the presence of translocated SRY in the fetal genome may be sufficient to induce testicular differentiation and a male phenotype. This mechanism has previously been described as a potential cause of discordance between NIPT results and phenotypic sex or fetal karyotype [26]. Therefore, in the present case, the female result reported by cfDNA should not, by itself, be interpreted as evidence against a male phenotypic sex; rather, it should be considered in the context of the biological limitations of the method and the possibility of a small Y-chromosomal segment or translocated SRY sequence.

The ultrasonographic findings in this case are also of particular importance. Fetal sex was reported as male on multiple ultrasound examinations, and the visualization of the testes on specialized ultrasonography provided additional evidence supporting male gonadal differentiation. These findings are consistent with previous reports of 46,XX testicular DSD in which discordance between the fetal sex reported by NIPT and ultrasonographic findings prompted further investigations and ultimately led to the diagnosis of a disorder of sex development [26,27]. In a report published in 2023, 46,XX testicular DSD was identified following an unexpected NIPT finding, and amniocentesis demonstrated the presence of SRY on the X chromosome [27]. Similarly, a 2026 case report described an infant with a completely male phenotype in whom NIPT predicted a female fetus, while postnatal investigations demonstrated the presence of Y-chromosomal material and SRY [28]. Collectively, these findings suggest that fetal sex discordance between cfDNA screening and ultrasonography, although uncommon, may represent

the first indication of an underlying disorder of sex development.

However, discordance between cfDNA and ultrasonography has multiple potential causes, and 46,XX testicular DSD represents only one of the differential diagnoses. Laboratory or sampling errors, differences between placental and fetal DNA, confined placental mosaicism, structural abnormalities of the sex chromosomes, cryptic or partial Y-chromosomal material, and other forms of DSD may all present with a similar pattern. Therefore, when such discordance is encountered, the genetic or gonadal sex of the fetus should not be determined solely on the basis of either testing modality. Detailed ultrasonographic assessment, review of the screening results, genetic counseling, and, when accepted by the parents, diagnostic testing such as amniocentesis can help distinguish among the possible underlying causes. Current guidance from the American College of Obstetricians and Gynecologists (ACOG) emphasizes that cfDNA is a screening test rather than a diagnostic test; when unexpected or abnormal findings are identified, further evaluation and, when appropriate, diagnostic testing should be considered [15,29].

In the present case, amniocentesis was recommended to evaluate the fetal chromosomal status; however, the parents declined the procedure. Consequently, definitive chromosomal diagnosis was deferred until after birth. Although the lack of prenatal diagnostic testing represents a limitation in the prenatal evaluation, serial ultrasonographic follow-up and visualization of the testes, together with the male phenotype after birth, provided sufficient rationale for postnatal karyotyping. The finding of a 46,XX karyotype in association with a completely male phenotype and prenatal visualization of the testes strongly raised the possibility of 46,XX testicular DSD. However, because the SRY test result was not yet available at the time of this report, the precise molecular mechanism cannot be considered definitive. This point should be taken into account when interpreting the case, and the final molecular diagnosis should be established after determination of the SRY status and, if necessary, analysis of other genes involved in the testicular differentiation pathway.

Early diagnosis of this condition is clinically important, even in an infant with a completely male external phenotype. Individuals with 46,XX testicular DSD are typically infertile because of severe impairment or complete absence of spermatogenesis and may subsequently develop hypogonadism, delayed or impaired puberty, gynecomastia, or other forms of gonadal dysfunction. Therefore, diagnosis during infancy provides an opportunity to establish a long-term follow-up plan. Such follow-up may include monitoring of growth and development, assessment of hormonal status and gonadal function at appropriate stages, monitoring of pubertal development, and evaluation of the testes. Early diagnosis also provides an appropriate opportunity for gradual, age-appropriate counseling of the family regarding the nature of the condition, potential long-term implications, and issues related to future fertility.

From a clinical management perspective, this case underscores the importance of a multidisciplinary approach to DSD. Management of these conditions is not limited to establishing the karyotype and may require collaboration among maternal–fetal medicine specialists, obstetricians and gynecologists, pediatricians, pediatric endocrinologists, medical geneticists, pediatric urologists, and, later in life, endocrinology and reproductive medicine specialists. The goal of this approach is not only to determine the underlying genetic mechanism but also to provide individualized care addressing gonadal status, growth, pubertal development, psychosocial well-being, and future fertility. In the present case, referral for genetic counseling and pediatric follow-up after identification of the 46,XX karyotype represented an important step toward establishing such a coordinated care pathway.

Another noteworthy aspect of this case is the potential role of ultrasonography when an unexpected cfDNA result is encountered. Although cfDNA performs very well in screening for common aneuploidies, information regarding fetal sex should also be interpreted within the context of the screening nature of the test. When the cfDNA result conflicts with repeated ultrasonographic findings, repeat or specialized ultrasonographic evaluation may provide valuable information regarding the fetal phenotype. In the present case, visualization of the testes on specialized ultrasonography was particularly informative, as it demonstrated that the cfDNA result alone could not fully explain the fetal sex-developmental status.

This report also has several limitations that should be acknowledged. First, molecular confirmation of the condition had not been completed at the time of reporting; therefore, the SRY status and its potential genomic location in the patient had not yet been established. Second, because amniocentesis was not performed, direct assessment of the fetal chromosomal status during pregnancy was not possible, and the 46,XX karyotype was established only after birth. Third, information regarding postnatal hormonal evaluation, postnatal imaging of the gonads, and analysis of other genes involved in the sex-development pathway was limited at the time of this report. Therefore, although the clinical and cytogenetic findings strongly support a diagnosis of 46,XX testicular DSD, determination of the precise genetic mechanism and complete characterization of the patient's phenotype will require longer-term follow-up and additional molecular investigations.

Overall, this case demonstrates that discordance between fetal sex predicted by cfDNA and sex observed on ultrasonography may represent more than

a simple discrepancy between screening results and, in rare cases, may provide the first clue to an underlying DSD. Integrating cfDNA results, detailed ultrasonographic assessment, karyotyping, and molecular testing can facilitate the diagnostic evaluation of these patients. In the present case, the female sex reported by cfDNA, together with repeated ultrasonographic evidence of a male fetus, birth of an infant with a completely male phenotype, and subsequent identification of a 46,XX karyotype, led to the consideration of 46,XX testicular DSD. This experience underscores the importance of integrated interpretation of screening and imaging findings, as well as the value of genetic counseling and multidisciplinary follow-up in cases of fetal sex discordance.

Conclusion

Discordance between fetal sex reported by cfDNA screening and ultrasonographic findings may, although rarely, represent the first indication of a disorder of sex development such as 46,XX testicular DSD (De la Chapelle syndrome). This case demonstrates that, despite the high accuracy of cfDNA screening for common chromosomal abnormalities, its results should be interpreted in conjunction with clinical and ultrasonographic findings and within the context of its screening nature. When persistent discordance is identified, detailed ultrasonographic assessment, genetic counseling, and, when feasible, cytogenetic and molecular diagnostic testing can facilitate accurate diagnosis. Early recognition of DSD allows appropriate planning of long-term follow-up, assessment of gonadal and hormonal function, and age-appropriate counseling for the family. Therefore, an integrated and multidisciplinary approach to fetal sex discordance may play an important role in the timely diagnosis and management of rare forms of DSD.

Ethical Considerations

In conducting this research, all ethical principles in medical and biological research were observed in accordance with the Declaration of Helsinki, and the rights, dignity, and confidentiality of the participants were protected.

Conflict of Interests

There was no conflict of interest in this study.

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