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BRIEF REPORT

Incidental finding of maternal sex chromosome aneuploidy from *DMD* carrier screening and single-nucleotide polymorphism (SNP)-based prenatal cell-free DNA screening

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Abstract

Prenatal cell-free DNA (cfDNA) screening and carrier screening (CS) may have incidental findings that have implications for maternal health outside the scope of the test. We investigated outcome information for individuals with both a *DMD* full gene deletion/duplication on CS and a suspected maternal X chromosome abnormality on SNP-based prenatal cfDNA screening. This retrospective analysis included de-identified data from pregnant individuals referred for CS and prenatal cfDNA screening at a single reference laboratory (9/2019–12/2021). Maternal karyotype and/or chromosomal microarray analysis results were requested from referring clinics for individuals with both *DMD* full gene deletion/duplication on CS and prenatal cfDNA screening results indicating potential maternal X chromosome aneuploidy. Of 333,814 individuals screened, 144 (1 in 2318) met study criteria, and for 84 (58.3%) we obtained information on whether diagnostic testing was received following these results. Of the 84 patients with follow-up information available, 34 (40.5%) received maternal diagnostic testing based on karyotype or chromosomal microarray analysis. At 97% ($n = 33$), the majority of patients with diagnostic testing had X chromosome aneuploidies, including trisomy X ($n = 22$, 64.7%), monosomy X mosaicism ($n = 8$, 24.2%), monosomy X ($n = 2$, 6.1%), and maternal X chromosome structural abnormality ($n = 1$, 2.9%). Our study supports a high likelihood of maternal sex chromosome abnormality in the presence of an inconclusive *DMD* result on CS and prenatal cfDNA screening suspicious for a maternal sex chromosome abnormality. Given the implications for maternal health, follow-up counseling, karyotype, and chromosomal microarray analysis may be recommended.

KEYWORDS
inconclusive DMD, monosomy X, NIPT, structural X chromosome variants, trisomy X, X-chromosome abnormality

1 | INTRODUCTION

Genetic testing can be used to assess the chances of having a child affected by specific genetic conditions and therefore has become a common component of both reproductive medicine and obstetrics. Reproductive carrier screening (CS) is used to help identify couples whose children might have an increased chance for inheriting an autosomal recessive or X-linked condition. In addition, noninvasive prenatal cell-free DNA (cfDNA) screening can be performed to identify the chances of chromosomal abnormalities in the pregnancy. Current American College of Medical Genetics and Genomics (ACMG) guidelines recommend that all patients should be offered prenatal cfDNA screening, over traditional screening, for trisomies 21, 18, and 13 as well as for sex chromosome abnormalities (Dungan et al., 2023). While the main purpose of reproductive genetic testing is to identify couples at risk for having children with a genetic condition, there has been increased recognition of incidental findings from reproductive genetic testing that have implications for maternal health, such as fertility, cardiovascular, or neurological issues (Leppig & Disteche, 2001).

Sex chromosome abnormalities are the most common chromosomal aneuploidy and occur in as many as 1 in 650 females (Jeon et al., 2012). Trisomy X syndrome occurs in ~1 in 1000 females (Otter et al., 2010), while Turner syndrome occurs in ~1 in 2000–2500 females (Martin-Giacalone et al., 2023). Sex chromosome abnormalities are, in some cases, identified as incidental findings during prenatal cfDNA screening (Lu et al., 2021). A recent report demonstrated that a maternal X chromosome abnormality was suspected in 1/1305 single-nucleotide polymorphism-based prenatal cfDNA tests (Martin et al., 2020). This incidental finding was confirmed by maternal chromosomal microarray analysis (CMA) with a positive predictive value of 94.3% (Martin et al., 2020). The Duchenne Muscular Dystrophy (*DMD*) gene is located on the X chromosome and is one of 113 genes that the ACMG recommends be included in CS panels (Gregg et al., 2021). Deletion or duplication of one or more exons in the *DMD* gene explains 65%–80% of dystrophinopathy cases (including DMD, Becker muscular dystrophy, and DMD-associated dilated cardiomyopathy), while sequence variants explain 20%–35% (Darras et al., 1993). Although a complete *DMD* gene deletion causing muscular dystrophy is uncommon, instances have been reported (Tuffery-Giraud et al., 2009). Copy number variant (CNV) analysis of the *DMD* gene by next-generation sequencing (NGS) (as performed in CS) sometimes identifies individuals with duplication or deletion of all *DMD* exons (Nallamilli et al., 2023). When this occurs on CS, an inconclusive result for *DMD* due to potential positivity for sex chromosome aneuploidy is typically reported, raising the possibility that an X chromosome aneuploidy may exist. In this scenario, subsequent or concurrent SNP-based prenatal cfDNA screening may provide insights clarifying the inconclusive result. Distinguishing these kinds of incidental findings from reproductive genetic testing can provide an avenue for earlier detection of sex chromosome abnormalities in women.

What is known about this topic?

Sex chromosome abnormalities are the most common chromosomal aneuploidy and are in some cases identified as incidental maternal DNA findings during prenatal cell-free DNA screening. Full gene duplications or deletions of the Duchenne Muscular Dystrophy (*DMD*) or surrounding genes identified through carrier screening by next-generation sequencing (NGS) may also indicate an X chromosome aneuploidy.

What this paper adds to the topic?

This study provides evidence to support that in the presence of full gene duplication or deletion of Duchenne Muscular Dystrophy (*DMD*) or surrounding genes on reproductive carrier screening, a prenatal cell-free DNA screening suspicious for a maternal sex chromosome abnormality can help elucidate the likelihood of a maternal sex chromosome abnormality. Our study highlights the importance of pursuing follow-up testing when an abnormal and/or inconclusive result is reported.

The objective of this study was to obtain outcome information for individuals who had undergone both CS (including *DMD* gene) and SNP-based prenatal cfDNA screening offered at a commercial laboratory in the United States and had results suspicious for a maternal X chromosome abnormality on both screening tests.

2 | MATERIALS AND METHODS

2.1 | Participants and study design

This retrospective analysis utilized de-identified data from pregnant individuals (with singleton or twin pregnancies) referred to a single reference laboratory for both prenatal cfDNA screening and reproductive CS that included the *DMD* gene, between 9/2019 and 12/2021. All test orders were received from OB/GYN or MFM clinics. The study cohort was restricted to individuals for whom both prenatal cfDNA screening results and CS indicated potential maternal X chromosome aneuploidy. Specifically, prenatal cfDNA was reported as "no results" for sex chromosomes due to a suspected maternal X chromosome finding and CS was reported as "inconclusive" for *DMD* due to a suspected full deletion or duplication of the *DMD* gene or surrounding genes on the X chromosome. While reports with such results recommend clinical follow-up and offer free appointments to review results with a lab genetic counselor, investigators were blinded to who utilized these services.

2.2 | Prenatal cfDNA screening

The single-nucleotide polymorphism (SNP)-based cfDNA screening method has been previously described (Pergament et al., 2014). Briefly, maternal and placental cfDNA were isolated from maternal plasma samples, amplified at specific loci, and sequenced using NGS (Pergament et al., 2014). Fetal fraction and chromosomal aneuploidies were analyzed using a proprietary SNP-based algorithm to determine the fetal copy number for chromosomes 13, 18, 21, X, and Y (Samango-Sprouse et al., 2016).

2.3 | Carrier screening (CS) for DMD

Genomic DNA from peripheral blood leukocytes was extracted. Sequence and copy number analyses for the *DMD* gene on CS panels were performed as previously described using hybrid-capture-based methodology followed by NGS (Westemeyer et al., 2020). Samples that were positive for single or multi-exonic *DMD* deletions or duplications were confirmed by multiplex-ligation dependent probe amplification. Samples showing duplications or deletions of all *DMD* exons (sometimes with additional regions on the X chromosome that are affected) were reported out as inconclusive due to being potentially positive for sex chromosome aneuploidy.

2.4 | Clinical follow-up

Clinical follow-up of individuals in the study cohort was performed via a faxed form and/or telephone contact with the office of the provider who ordered the tests. Clinics were contacted for information regarding maternal testing results, if any, including karyotype and/or CMA. In cases where maternal diagnostic testing was received, the clinics ordering the original screening arranged confirmatory karyotype and/or CMA. Research using these de-identified data has been deemed exempt from IRB review under the terms and conditions of Salus IRB #19040.

3 | RESULTS

During the study period, 333,814 individuals underwent both CS for the *DMD* gene and SNP-based prenatal cfDNA screening. Of these, 144 individuals (1 in 2318) had both an incidental maternal X chromosome finding on SNP-based cfDNA screening and *DMD* full gene deletion/duplication on CS, suggesting X chromosome aneuploidy, and were included in the study cohort (Figure 1).

Clinical follow-up was sought for all 144 cases and a response was obtained for 58.3% (84/144). Of these, 40.5% (34/84) had follow-up diagnostic testing with a karyotype or CMA, while the remaining 59.5% (50/84) had no further testing (Figure 1). Among the

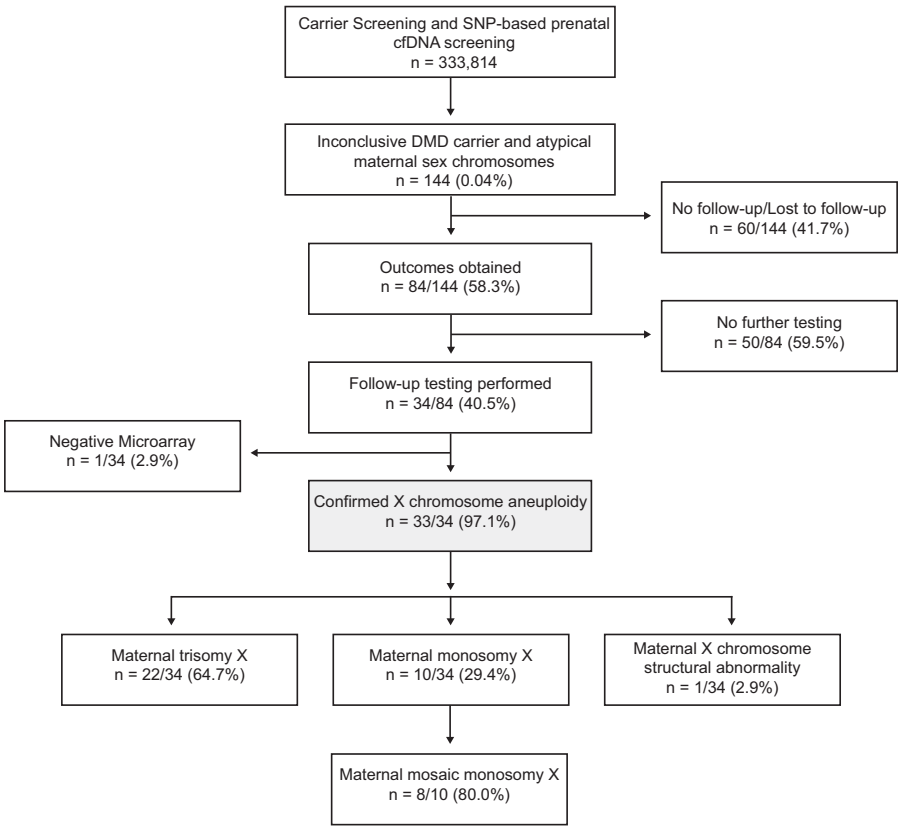

FIGURE 1 Flow diagram of participants included in the study cohort.

individuals with follow-up testing, X chromosome aneuploidy was confirmed in 97.1% (33/34): maternal trisomy X in 64.7% (22/34), maternal monosomy X in 29.4% (10/34), and maternal X chromosome structural abnormality in 2.9% (1/34) (Figure 1). Eighty percent (8/10) of individuals with monosomy X were reported as mosaic. Only one individual with follow-up diagnostic testing reported a negative CMA result.

4 | DISCUSSION

To our knowledge, this is the first case series reporting a patient cohort undergoing both prenatal cfDNA screening and CS with reported results of a suspected maternal X chromosome abnormality on prenatal cfDNA screening and *DMD* full gene deletion/duplication on CS. Herein, we report an occurrence rate of these results to be 1 in 2318. Of the cases where follow-up was obtained and diagnostic testing was performed, 97.1% had a confirmed maternal sex chromosome abnormality. Importantly, almost one-third had full or mosaic monosomy X and approximately two-thirds had trisomy X, all of which may have potential implications for maternal health including reproductive, cardiovascular, and metabolic conditions.

Although infertility is common in individuals with monosomy X, approximately 5%–6% of individuals with monosomy X achieve a spontaneous pregnancy (Dowlut-McElroy et al., 2022). A recent expert opinion piece by Dowlut-McElroy et al. on how best to handle incidental findings of sex chromosomal abnormalities from cfDNA screening proposed that women with >5% monosomy X mosaicism should be evaluated for Turner syndrome (Dowlut-McElroy et al., 2022). Evaluations may include an echocardiogram, as these individuals may be at risk for life-threatening medical complications such as aortic dissection, hypertension, premature ovarian insufficiency, and pregnancy-related complications (Dowlut-McElroy et al., 2022). However, individuals with mosaic monosomy X can exhibit variable expression with phenotypes that range from no symptoms at all to symptoms consistent with non-mosaic monosomy X, or Turner syndrome (Berglund et al., 2020). In a cohort of healthy non-pregnant individuals with confirmed mosaic 45,X/46,XX, the phenotypic manifestations of Turner syndrome were minimal (Tuke et al., 2019).

Individuals with trisomy X do not always have recognized or obvious health issues but can experience reduced fertility and various other health issues including developmental delays, urogenital malformations, epilepsy, and psychiatric disorders (Liu et al., 2024). A diagnosis of X chromosome aneuploidy may help explain a history of medical issues, as well as provide guidance for future management.

Patients with complex X chromosome abnormalities, such as the single case with a structural abnormality in this study, may also experience health issues like those with monosomy X or trisomy X depending on the regions of the X chromosome that are involved or interrupted (Leppig & Disteche, 2001). Follow-up with maternal diagnostic testing might therefore provide important information to guide clinical management. For follow-up studies, recommending both karyotype with extended cell counts and CMA based on the nature of the findings may be important (Hao et al., 2020).

Interestingly, most patients identified in our study had no additional follow-up testing. At this time, the reason for this is unclear. However, based on the available data, an inconclusive *DMD* result in the context of a suspected maternal chromosome abnormality on SNP-based testing indicates a high likelihood of a maternal X chromosome abnormality. In such cases, follow-up testing of the pregnant patients should be considered. Additional education for referring providers that encourages the discussion of the pros and cons of follow-up chromosome testing, and/or referral to genetic counseling, may be helpful; many patients may not understand the implications of having an inconclusive cfDNA screening or CS result indicating a previously undetected chromosome abnormality. Several qualitative studies have identified anxiety, confusion, and even decisional regret among individuals who receive inconclusive or unexpected prenatal screening results (Agatisa et al., 2015; Gammon et al., 2020). Post-test education and genetic counseling following an incidental maternal abnormality finding can help alleviate patient anxiety and increase understanding about the diagnosis, especially when expert guidance acknowledges the emotional weight of uncertain or unexpected results (Hartwig et al., 2019; Riggan et al., 2021). Patients who are educated about their genetic makeup may be better prepared if they receive inconclusive genetic testing results in future pregnancies. Additionally, individuals who have a known sex-chromosome abnormality should be counseled regarding the possibility of inconclusive results on CS for X-linked disorders such as DMD.

Although there were many cases that met study criteria, the study is limited by a smaller number of cases that had follow-up diagnostic testing. Therefore, further studies collecting follow-up information on more cases would be beneficial to fully elucidate these findings. There may also be ascertainment biases from restricting the cohort to individuals with both CS and prenatal cfDNA testing. Finally, while the capacity for SNP-based cfDNA testing to identify maternal sex chromosome abnormalities is a clear benefit to this type of testing, not all prenatal cfDNA testing has this ability, and extrapolation of the data herein to other prenatal cell-free DNA screening technologies may not be appropriate.

This study provides evidence to support that in the presence of an inconclusive *DMD* result on CS, a prenatal cfDNA screening suspicious for a maternal sex chromosome abnormality can help elucidate the likelihood of a maternal sex chromosome abnormality. Our study highlights the importance of pursuing follow-up testing when an abnormal and/or inconclusive result is reported.

AUTHOR CONTRIBUTIONS

Conceptualization: J.L., J.W., Y.W.; Formal data analysis: J.L., J.W., Y.W.; Investigation: J.L., J.W., Y.W.; Methodology: Y.W.; Writing – original draft: J.L., J.W., Y.W., J.B.O.; Writing – review and editing: J.L., J.W., G.G., E.M., E.R., Y.W., W.X., J.B.O., J.M.

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CONFLICT OF INTEREST STATEMENT

All authors are employees of Natera, Inc. with stocks or options to own stocks in the company.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

ETHICS STATEMENT

Human studies and informed consent: Research using these de-identified data has been deemed exempt from IRB review under the terms and conditions of Salus IRB number: 19040.

Animal studies: No animal studies were performed.

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