



International Journal of Innovative Technologies in Social Science

e-ISSN: 2544-9435

Operating Publisher
SciFormat Publishing Inc.
ISNI: 0000 0005 1449 8214

2734 17 Avenue SW,
Calgary, Alberta, T3E0A7,
Canada
+15878858911
editorial-office@sciformat.ca

ARTICLE TITLE	PREGNANCY IN MUSCULAR DYSTROPHIES: A NARRATIVE REVIEW OF CLINICAL RISKS, PHYSICAL ACTIVITY, AND MATERNAL-FETAL OUTCOMES
----------------------	---

DOI	https://doi.org/10.31435/ijitss.3(51).2026.6609
------------	---

RECEIVED	24 June 2026
-----------------	--------------

ACCEPTED	09 September 2026
-----------------	-------------------

PUBLISHED	15 September 2026
------------------	-------------------

LICENSE



The article is licensed under a **Creative Commons Attribution 4.0 International License**.

© The author(s) 2026.

This article is published as open access under the Creative Commons Attribution 4.0 International License (CC BY 4.0), allowing the author to retain copyright. The CC BY 4.0 License permits the content to be copied, adapted, displayed, distributed, republished, or reused for any purpose, including adaptation and commercial use, as long as proper attribution is provided.

PREGNANCY IN MUSCULAR DYSTROPHIES: A NARRATIVE REVIEW OF CLINICAL RISKS, PHYSICAL ACTIVITY, AND MATERNAL-FETAL OUTCOMES

Maria Calusińska-Skowron (Corresponding Author, Email: calusinskamaria@gmail.com)
Municipal Hospital Complex in Chorzów, Chorzów, Poland
ORCID ID: 0009-0005-8749-0018

Klaudia Jeziorowska
Municipal Hospital Complex in Chorzów, Chorzów, Poland
ORCID ID: 0009-0008-2510-6991

Anna Kudłacik
Municipal Hospital Complex in Chorzów, Chorzów, Poland
ORCID ID: 0009-0009-9299-0593

Oktawian Kaczmarek
Municipal Hospital Complex in Chorzów, Chorzów, Poland
ORCID ID: 0009-0002-9766-5123

Karolina Baldys
WSB University, Collegium Medicum, Dąbrowa Górnicza, Poland
ORCID ID: 0009-0001-8764-6327

Magdalena Franków
Health Care Center in Olawa, Olawa, Poland
ORCID ID: 0009-0005-9979-0907

Kornel Adrian Jarosz
County Hospital Complex in Oleśnica, Oleśnica, Poland
ORCID ID: 0009-0004-2849-8872

Aleksandra Karolina Schmidt
Municipal Hospital Complex in Chorzów, Chorzów, Poland
ORCID ID: 0009-0003-4135-1245

Aleksandra Regina Żmuda
University Hospital in Kraków, Kraków, Poland
ORCID ID: 0009-0001-8655-5546

Julia Łukasik
University Hospital in Kraków, Kraków, Poland
ORCID ID: 0009-0008-0055-4296

ABSTRACT

Introduction and purpose: Muscular dystrophies are progressive, hereditary muscle disorders whose inheritance pattern and multisystem involvement complicate reproductive decisions, yet evidence on maternal and neonatal outcomes remains fragmented. This review summarises current literature on pregnancy in women with dystrophinopathies, myotonic dystrophy, FSHD, and LGMD, including the role of physical activity and rehabilitation.

Materials and methods: Literature was retrieved from PubMed, Scopus, and Google Scholar (January–June 2025), supplemented by reference list screening, and analysed narratively by subtype.

Review findings: Pregnancy in muscular dystrophy carries elevated but increasingly manageable risk depending on subtype. DM1 is linked to adverse neonatal outcomes via congenital myotonic dystrophy; dystrophinopathy carriers face cardiomyopathy and heart failure risk; FSHD and LGMD show increased instrumental delivery, falls, and weakness, supporting a role for individualised physiotherapy and respiratory training.

Conclusions: Reproductive care in muscular dystrophy benefits from a multidisciplinary approach with cardiac and respiratory monitoring, anaesthetic planning, genetic counselling, and rehabilitation support, though evidence remains limited to case reports and small cohorts.

KEYWORDS

Pregnancy, Muscular Dystrophies, Muscular Dystrophy, Duchenne, Myotonic Dystrophy, Physical Activity, Exercise, Rehabilitation

CITATION

Całusińska-Skowron, M., Jeziorowska, K., Kudłacik, A., Kaczmarek, O., Baldys, K., Franków, M., Jarosz, K. A., Schmidt, A. K., Żmuda, A. R., & Łukasik, J. (2026). Pregnancy in muscular dystrophies: A narrative review of clinical risks, physical activity, and maternal-fetal outcomes. *International Journal of Innovative Technologies in Social Science*, 3(51). [https://doi.org/10.31435/ijitss.3\(51\).2026.6609](https://doi.org/10.31435/ijitss.3(51).2026.6609)

COPYRIGHT

© The author(s) 2026. This article is published as open access under the **Creative Commons Attribution 4.0 International License (CC BY 4.0)**, allowing the author to retain copyright. The CC BY 4.0 License permits the content to be copied, adapted, displayed, distributed, republished, or reused for any purpose, including adaptation and commercial use, as long as proper attribution is provided.

1. Introduction

Muscular dystrophies are a subgroup of hereditary myopathies, characterised by progressive muscle degeneration caused by abnormalities in proteins essential for muscle fiber structure and function. Their pathophysiological mechanisms vary widely, ranging from complete absence or abnormal structure/function of a specific protein, as observed in Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD), to disturbances involving regulatory proteins affecting multiple genetic pathways, some of which remain to be fully understood, as in myotonic dystrophy and facioscapulohumeral muscular dystrophy (FSHD) [1]. In the general population frequency of muscular dystrophies is estimated at approximately 1 in 5,000 individuals [2]. Duchenne muscular dystrophy is the most common muscular dystrophy in children, with global birth prevalence estimated at 19.8 cases per 100,000 live male births [3], whereas in adults myotonic dystrophy is the most common, with an estimated prevalence of 9.99 cases per 100,000 individuals [4]. Muscular dystrophies can manifest in multiple organ systems, including muscular, cardiovascular, respiratory, neurological as well as gastrointestinal [2]. Due to this wide spectrum of symptoms and hereditary nature of these conditions, affected individuals are faced with many challenges in their reproductive health. Important factors to be considered are the impact of pregnancy on symptoms, possible complications during pregnancy and delivery, the role of physical activity and rehabilitation in maintaining function, along with neonatal outcomes [5].

The aim of this review is to summarize current evidence regarding pregnancy in women with muscular dystrophies, focusing on dystrophinopathies, myotonic dystrophy, FSHD, and LGMD, with particular emphasis on maternal, obstetric, and neonatal outcomes, as well as the place of physical activity and rehabilitation in their reproductive care.

Table 1. Characteristics of muscular dystrophies analyzed in the review

Condition	Genetic cause	Inheritance pattern	Typical age of onset	Major systemic involvement	Life expectancy
DMD/BMD	Dystrophin gene mutation	X-linked recessive	Childhood	Cardiac, respiratory	Reduced
DM1/DM2	CTG/CCTG repeat expansion	Autosomal dominant	Adolescence/Adulthood	Multisystemic	Variable*
FSHD	DUX4 derepression	Dominant/digenic	Adolescence/Adulthood	Musculoskeletal	Close to normal
LGMD	Variable	Dominant/recessive	Variable	Cardiac, respiratory	Variable

*Reduced mostly in CDM cases.

2. Dystrophinopathies

Dystrophinopathies are caused by mutations of the DMD gene, linked to the X-chromosome, characterised by a recessive inheritance pattern.

Boys affected by Duchenne muscular dystrophy (DMD) develop symptoms around the age of 2 years old; currently, with proper care, life expectancy is approximated between 20 and 40 years old [6]. The most common cause of death in DMD are cardiopulmonary complications, accounting for almost 80% [7]. Most commonly prolonged survival of individuals affected by DMD is linked to usage of corticosteroids and mechanical ventilation [8]. Becker's muscular dystrophy (BMD) is characterised by a later onset (around 8 years old) and slower progression; due to cardiac involvement the mean age of death was placed at 47.3 years; the life expectancy of individuals without significant cardiac complications may be placed near the average [9].

Aforementioned DMD and BMD mutations may appear de novo or be inherited from a carrier mother. Approximately two-thirds of DMD cases are estimated to be inherited, while de novo mutations appear more common in deletion-type variants than in duplications or small mutations [10]. In BMD, maternal carrier frequency is higher, with some studies reporting inheritance from carrier mothers in nearly 90% of cases [11]. Most heterozygous carriers of the mutation remain asymptomatic; those who develop symptoms are referred to in literature as "manifesting carriers". Possible manifestations in DMD include muscle damage (17% of all carriers), cardiomyopathy (8% of all carriers) [12] and rarely central nervous system involvement. Classical muscle symptoms in BMD carriers occur even less frequently than in DMD [13]. The inconsistency of manifestation in female carriers could be explained by different patterns of X-inactivation, perhaps overlapping with external factors such as aging [14].

Major concerns about pregnancy in female dystrophinopathies carriers (both symptomatic and asymptomatic) that we are going to elaborate on in this section are cardiomyopathy and newborn prognosis. The aftermath of pregnancy may also result in progression of symptoms, with at least one documented case of marked muscle damage progression after pregnancy [15], however currently it is not possible to estimate the likelihood of such events.

Pregnancy has been linked to development and progression of cardiomyopathy in the general population, due to several physiological adaptations that occur during gestation: increased heart rate and stroke volume, decreased systemic vascular resistance and structural remodeling of the heart muscle [16]. These changes raise concerns in regard to the impact of pregnancy on women with additional risk factors e.g. a genetic background of dystrophin mutations. Based on existing literature, only a few cases of such pregnancies have been reported. In most of them severe heart failure during the final weeks of pregnancy or postpartum was the first symptom of the disease, requiring pharmacological treatment and in one case the aid of a Ventricular Assist Device (VAD) [17,18,19]. Likewise there has been a case of a woman diagnosed as a manifesting carrier before pregnancy, whose cardiomyopathy did not worsen during pregnancy due to treatment and concluded with vaginal delivery of a healthy female newborn [20].

Once a female carrier of a dystrophinopathy (aware of her condition or not) becomes pregnant, the possible outcomes for the newborn are based on inherited chromosomes, with a 50% chance of transmitting

the pathogenic variant to a child, regardless of gender. Males who inherit the mutation are burdened with the prognosis of a symptomatic dystrophinopathy, whereas females may become carriers like their mothers, either asymptomatic or more rarely symptomatic. In very rare cases of Turner syndrome or Turner syndrome mosaicism coinciding with the presence of a pathogenic dystrophinopathy gene in females, symptoms akin to a classical manifestation found in males may develop [21].

It is crucial to inform and educate affected women who might find themselves contemplating their reproductive choices about those outcomes and available possibilities in genetic counselling.

Genetic testing for female carriers varies based on the presence of a known familial pathogenic variant or lack thereof. When the familial variant is known, testing is specific for this variant and whenever possible, compared with a control sample procured from a known carrier relative or an existing laboratory report. The actual testing methods can include a single probe or primer pair, MLPA, microsatellite analysis, high-density array CGH and even RNA analysis from a muscle biopsy. Without a known pathogenic variant and no available known affected relative it is recommended to run: deletions and duplications detections as well as small variants detection (Level 1 and Level 2 testing according to recent guidelines). When based on conclusions from these tests and further genetic counselling a diagnosed female carrier decides on a pregnancy, there are more possible diagnostic prospects to be considered: invasive prenatal diagnosis, non-invasive prenatal diagnosis and preimplantation genetic testing. In general, those methods should be limited to male pregnancies, unless there exist additional circumstances such as specific documented family genetic history [22]. A promising direction in genetic counselling for DMD carriers is CRISPR/Cas9 technology [23]. Its clinical use would be a potential breakthrough for the affected families, with hope for therapies specific to individual mutations in the future [24,25].

3. Myotonic dystrophy

Myotonic dystrophy (*dystrophia myotonica*, DM) is a subgroup of muscular dystrophies, characterised by the presence of myotonia, clinically defined as delay in muscle relaxation after contraction. DM is classified into two genetically and clinically distinct forms: DM1, also known as Steinert disease and DM2, also described as proximal myotonic myopathy. DM in general is the most common muscular dystrophy in Europeans and people of European ancestry [26].

Both DM1 and DM2 are caused by expansions of DNA tandem repeats, resulting in toxic RNA-mediated dysfunction. DM1 is caused by expansion of CTG repeats in the protein kinase (DMPK) gene on chromosome 19. DM2 is caused by expansion of CCTG repeats in the CNBP (ZNF9) gene on chromosome 3 [27]. Both are characterised by a dominant pattern of inheritance.

Clinical manifestations of DM include muscle damage, myotonia as well as a wide range of nonmusculoskeletal symptoms. Muscle weakness is progressive, and affects specific muscle groups depending on the type. Myotonia presents as painless stiffness of the muscles, and it is commonly provoked by cold, stress, or physical exertion. During neurological examination it may be visible as grip or percussion myotonia, and some patients additionally display the warm-up phenomenon, where repeated contraction temporarily relieves the stiffness [28,29]. Aside from muscular symptoms, DM is also a multisystem disorder. Clinical findings include cardiac arrhythmias, conduction irregularities, cardiomyopathy, reduced lung volume, cataracts, insulin resistance, GI tract issues, mental challenges, and even intellectual disabilities in congenital forms [30].

Important issues that need to be addressed in conversations on reproductive health in DM include the anticipation phenomenon, congenital myotonic dystrophy, and the influence of pregnancy and DM on each other.

The pathomechanism of both DM types lies in the increased number of tandem repeats in specific genes. The number of additional CTG repeats has been correlated with severity of DM1 symptoms. Clinical symptoms typically manifest in individuals with more than 50 CTG repeats. With transmission from generation to generation the number of repeats has a tendency to expand even more, and that anticipation tends to be greater in maternally inherited expansions [31]. Due to this phenomenon successive generations may experience more severe symptoms with an earlier onset. DM2 differs from DM1 in regard to anticipation. While the number of CCTG repeats considered pathogenic is established at 75 and more, there has been no clear correlation of expansion size and symptom severity [30].

Congenital myotonic dystrophy (CDM) is a subtype of DM1. The affected patients usually inherit more than 1000 CTG repeats, with 90% of cases being linked to maternal transmission [32]. Symptoms of CDM

vary depending on the phase of life. Prenatally the fetus may be small for gestational age (SGA), with reduced movements and polyhydramnios visible during an ultrasonography exam. After birth the newborn can present as hypotonic, with decreased reflexes, muscle weakness in distal muscles and neck, and a myopathic facial appearance with ptosis, bilateral facial weakness, and a tent-shaped mouth. A vast number of complications may occur, including visual impairment, respiratory distress, pulmonary hypoplasia, bronchopulmonary dysplasia, pneumothorax, recurrent infections, GI tract issues, feeding difficulties, cardiac defects and in the most severe cases premature death. Neonatal mortality rate in CDM is reported at up to 40% [33]. In early childhood affected children usually show partial functional improvement, with another onset of symptoms in early adulthood [34].

Pregnancy (and menstruation) may worsen symptoms of DM like mobility limitations, pain, myotonia and emotional issues due to fluctuating hormone levels [35]. Furthermore, DM1 is associated with an increased risk of miscarriage, ectopic pregnancies, placenta previa and pre-term labour (which is often attributed to congenital DM) [30, 36]. Likewise in women with DM2 a higher rate of urinary tract infections and pre-term labour have been observed [37]. Increased rates of pre-eclampsia and postpartum haemorrhage have been described in both DM subtypes, with pre-eclampsia occurring more frequently in DM2 [37,38].

4. FSHD (Facioscapulohumeral muscular dystrophy)

FSHD is a neuromuscular disorder divided genetically into 2 types, which are clinically homogeneous: FSHD1, caused by a deletion in the 4q chromosome, which results in derepression of DUX4 gene and FSHD2, which leads to the same result in a non-deletion mechanism. 95% of FSHD patients are diagnosed with type 1, which is characterised by a dominant inheritance pattern. FSHD2 is inherited in a digenic pattern, with 80% of affected individuals presenting with a mutation in a chromatin modifier gene, most commonly the SMCHD1 gene [39].

Aligned with its name, FSHD typically initially affects facial, scapular and humeral muscles, then distal lower limbs and later on in the disease's course affects other proximal and abdominal muscles [40]. Due to the epigenetic mechanism of FSHD, symptoms are highly variable between separate cases and also between generations of the same family, both in FSHD1 and FSHD2 [41]. Women tend to be clinically less affected, and are often diagnosed later on in life [42]. FSHD is a progressive illness, and about 20% of affected patients over the age of 50 are at least partial wheelchair users; targeted physical activity and mobility support may help preserve functional independence for longer, although life expectancy is generally not impacted [43].

The typical age of onset is between 15 and 30 years old, which can coincide with the reproductive years in many women's lives. According to a survey filled by a limited number of 38 participants, even 50% of those women were not aware of their diagnosis before pregnancy [44].

While fertility and the course of pregnancy as a whole do not seem to be significantly affected in FSHD [42], there are some concerns regarding the manifestation of the disease during pregnancy and the risks during delivery. Up to 24% of women experience increase in their symptoms, such as frequent falls, increased weakness or increased pain, and most of them do not improve postpartum, potentially making childcare more challenging; this is a pattern that reinforces the case for antenatal and postpartum physiotherapy input aimed at fall prevention and functional training for infant care tasks. If the abdominal and pelvic muscles are affected by FSHD, the second stage of labour may be inefficient, with the need for assisted instrumental delivery or a caesarean section [42,45]. The statistical significance of this observation was documented in a 2006 study, where the total rate of caesarean deliveries was reported at 23.8% (compared to 16.9% in the general population), and instrumental deliveries at 27% (compared to 11.6% in the general population) [44]. Due to this increased probability of an operative delivery, concerns for possible anaesthesia during the labour should be considered from the early stages of pregnancy, including planning for general vs regional anaesthesia and monitoring Forced Vital Capacity (FVC) over the course of the pregnancy, ideally paired with breathing-focused physiotherapy where respiratory muscle involvement is present [42].

Genetic testing as an aid in reproductive choices for FSHD patients is possible, though complex [46]. Prenatal diagnosis based on mapping the size of the D4Z4 repeat with the use of the Southern Blot method or LR-PCR is clinically used, preferably in the first trimester. Preimplantation genetic testing in FSHD1 remains challenging due to the complex genetic background of this condition and consequently risk of misdiagnosis. Advancements in this field are being made, in efforts to improve accuracy and clinical applicability of these approaches [47].

The complex genetic and epigenetic pathomechanisms of FSHD2 pose a scientific challenge in regard to credible genetic testing. Due to limited reliability of fetal risk assessment prenatal diagnosis and preimplantation genetic testing are currently clinically unavailable [41].

5. LGMD

Limb-girdle muscular dystrophies are a heterogeneous group of more than 30 conditions that initially affect proximal limb muscles causing their progressive weakness. Their patterns of inheritance may be dominant (LGMD-Ds) or recessive (LGMD-Rs) [48]. The spectrum of LGMD symptoms is wide and variable [49]. As the LGMD conditions progress they may involve most muscle groups, in advanced cases including the diaphragm or the myocardium. Some types, e.g. LGMD2I, are linked to a higher risk of respiratory muscle involvement, and consequently FVC reduction and respiratory failure [48].

One of the main concerns during pregnancy in LGMD cases is exacerbation of symptoms such as increased weakness (reported in almost 50%), imbalance resulting in frequent falls (57%) [50], hypertension and deterioration of cardiac function in certain types of LGMD (e.g. LGMD2I) [51]. Given the high reported rates of weakness and falls, individualised assessment for supervised strengthening, balance training, and assistive-device use across pregnancy may be a reasonable component of multidisciplinary care, though dedicated studies of exercise-based interventions in pregnant LGMD patients are lacking. The occurrence of operative deliveries seems to be increased, with one study reporting a rate of 35%, as well as a need for labour induction in 70% of cases [45, 52]. The course of pregnancy and newborn outcomes in LGMD patients may be unaffected, though due to variability between subtypes available data is limited and may not be representative across all LGMD subtypes [52].

Prenatal testing for LGMD is possible and accessible. If the pathogenic variant has already been found in the affected partner or their family, testing is targeted. Due to the recessive nature of many forms of LGMD, in certain regions with higher frequency of these disorders or higher rates of consanguinity it may also be advisable to test the other, healthy partner [53]. If it is established that both partners are carriers of a pathogenic variant, preimplantation genetic testing may be applied. Due to marked heterogeneity of LGMD, each case requires individualized reproductive counselling and management during pregnancy.

Table 2. Main maternal and obstetric complications reported in Muscular Dystrophies

Condition	Maternal complications	Obstetric complications	Delivery considerations
DMD/BMD	Cardiomyopathy, muscle weakness	HF, respiratory compromise	Increased caesarean risk
DM1/DM2	Myotonia, fatigue	Miscarriage, pre-term labour, postpartum haemorrhage	Anaesthetic caution
FSHD	Increased weakness, frequent falls	Operative delivery	Assisted vaginal delivery
LGMD	Increased weakness, respiratory decline	Labour induction, operative delivery	Respiratory monitoring

6. General delivery concerns in muscular dystrophies

Labour and vaginal delivery are physiologically complex, demanding processes. They require the coordination of pelvic muscles, abdominal muscles and myometrium in order to ensure effective labour progression. It creates a significant physiological burden for the mother, regardless of any underlying conditions and additional factors. Muscular dystrophies may negatively affect the birth process at every stage, and pose a significant physiological burden for the affected women. Muscle weakness may impair effective pushing during the second stage of labour and extend the duration of labour. Respiratory compromise may additionally contribute to the need for instrumental delivery or a caesarean section [38]. According to a study conducted in 2021, interviewing women affected by a variety of different neuromuscular disorders 70% of those patients wished to deliver vaginally, with 17% preferring caesarean section [50]. This report does not seem to diverge from the global preference for caesarean sections, generally stated below 20% [54,55]. Reported methods of birth in this study showed significantly higher rates of operative vaginal delivery (19%)

and planned caesarean section (20%) when compared to the general population (12% and 13% respectively) [50]. That discrepancy seems to be particularly pronounced in FSHD as described in its section [42]. Common reasons for emergency caesarean sections in all neuromuscular disorders include inability to push, lack of progression, fetal distress and abnormal fetal position [50].

7. Physical activity and rehabilitation considerations

Across the subtypes discussed above, muscle weakness, fatigue, impaired balance, and reduced respiratory reserve emerge as recurring themes affecting the course of pregnancy, the mechanics of the second stage of labour, and the postpartum transition to caregiving. These shared features suggest that structured physical activity and rehabilitation should be considered a component of multidisciplinary care, alongside cardiac, respiratory, obstetric, and genetic management, rather than an incidental concern.

In FSHD and LGMD in particular, high reported rates of falls and subjective weakness during pregnancy [44,50] point toward a role for individualised, supervised programmes addressing balance, core and pelvic girdle strength, and safe mobility strategies as pregnancy progresses and the centre of gravity shifts, consistent with broader evidence that structured physical activity favourably influences the course of labour and delivery outcomes in the general obstetric population [56]. Because a substantial proportion of women do not report improvement of these symptoms after delivery [42], physiotherapy during postpartum may also help prepare patients for the physical demands of infant care, including lifting, carrying, and feeding positions adapted to individual muscle strength. This strategy may be particularly relevant given that physical activity has also been associated with a reduced risk of postpartum depression in the general population [57], a consideration that may be amplified in women facing persistent physical limitations after delivery.

Where respiratory muscle involvement is present, as can occur in dystrophinopathy carriers, DM1/DM2, and selected LGMD subtypes, serial monitoring of Forced Vital Capacity should be implemented. Alongside this monitoring, respiratory physiotherapy or inspiratory muscle training may support ventilatory reserve during the physical exertion of labour and reduce the risk of postpartum respiratory decompensation [38,48].

Given the increased rates of instrumental and operative delivery reported across FSHD and LGMD cohorts [44,45,50,52], targeted pelvic floor and abdominal muscle assessment before and during pregnancy, informed by evidence supporting structured pelvic floor and trunk-stabilisation training across the peripartum period [58], could help clinicians anticipate difficulties with active pushing and create a delivery plan. Anaesthetic strategy should be decided with those factors in mind, and consistent with recommendations already made for anaesthetic planning in this population [42].

It should be emphasised that existing studies evaluating structured exercise or rehabilitation interventions specifically in pregnant women with muscular dystrophies remain scarce. The considerations above are extrapolated from broader reports of symptom burden in this population and from general principles of prenatal and postnatal physiotherapy, and should be interpreted as a rationale for future research rather than as an established set of guidelines.

8. Future perspectives

Evidence on reproductive health in muscular dystrophies remains limited. Most available studies are retrospective, drawn from small cohorts or single case reports, and many were conducted over 10 years ago. Prospective multicentre registries and international collaboration are needed to characterise maternal, obstetric, and neonatal outcomes by subtype and to support creation of evidence-based pregnancy management guidelines. Molecular diagnostics continue to broaden reproductive options. More sensitive sequencing has improved carrier detection, prenatal diagnosis, and preimplantation genetic testing, though accuracy remains a limiting factor in genetically complex conditions such as FSHD2 and several LGMD subtypes, where reliable fetal risk assessment is not yet established. Disease-modifying therapies, including exon skipping, other RNA-targeted approaches, and gene therapy, are altering the outlook for several subtypes. As survival and life quality of women affected by muscular dystrophies improve, more of them may pursue pregnancy, while the safety of available therapies during conception, gestation, and lactation remains largely unstudied and is a priority for future research. Likewise, prospective evaluation of structured physical activity, physiotherapy, and rehabilitation programmes tailored to pregnancy and the postpartum period in this population represents an important and currently unaddressed research priority.

Table 3. Selected studies reporting maternal, obstetric, and neonatal outcomes in muscular dystrophies

Authors	Condition	Study type	Key findings
Hoogerwaard et al. [12]	DMD/BMD carriers	Cohort study	Muscle symptoms in 17% and cardiomyopathy in 8% of carriers
Rudnik-Schöneborn & Zerres [36]	DM1	Cohort study	Increased miscarriage rate, pre-term labour, risk of CDM
Rudnik-Schöneborn et al. [37]	DM2	Cohort study	Increased rates of pre-eclampsia, UTIs, pre-term labour
Ciafaloni et al. [44]	FSHD	Survey study	Increased operative delivery
Libell et al. [52]	LGMD	Cohort study	Increased operative delivery rates
Moore et al. [50]	Various neuromuscular disorders	Questionnaire study	Higher operative delivery rates compared to general population

9. Conclusions

Pregnancy in women with muscular dystrophies is becoming increasingly manageable, particularly when supported by individualised genetic counselling and patient education. However outcomes vary substantially with disease subtype, molecular background, and the extent of systemic, especially cardiac and respiratory, involvement. Because these are rare diseases, available data is limited and is based largely on small cohorts and case reports; larger prospective studies are needed to predict maternal, obstetric, and neonatal outcomes more reliably.

As survival and quality of life in muscular dystrophy patients continue to improve, more women will face reproductive decisions. Coordinated care across neurology, obstetrics, cardiology, anaesthesia, physiotherapy and rehabilitation, and genetic counselling will be central to managing pregnancy safely in this population.

Author Contributions:

Conceptualization: Maria Całusińska-Skowron, Klaudia Jeziorowska.

Formal analysis: Oktawian Kaczmarek, Magdalena Franków, Aleksandra Karolina Schmidt, Kornel Adrian Jarosz.

Data curation: Aleksandra Regina Żmuda, Anna Kudłacik, Julia Łukasik, Karolina Bałdys.

Original draft preparation: Maria Całusińska-Skowron, Oktawian Kaczmarek, Magdalena Franków, Anna Kudłacik,

Review and editing: Aleksandra Karolina Schmidt, Kornel Adrian Jarosz, Aleksandra Regina Żmuda, Julia Łukasik.

Supervision: Klaudia Jeziorowska, Karolina Bałdys.

All authors have read and agreed to the published version of the manuscript.

Funding statement: This research received no external funding.

Conflicts of Interest: The authors declare no conflict of interest.

Declaration of the use of generative AI and AI-assisted technologies in the writing process: During the preparation of this work, the authors used ChatGPT for the purpose of assisting with grammar and readability. After using this tool, the authors reviewed and edited the content as needed and take full responsibility for the substantive content of the publication.

REFERENCES

1. Lovering, R. M., Porter, N. C., & Bloch, R. J. (2005). The muscular dystrophies: From genes to therapies. *Phys Ther*, 85(12), 1372–1388. Retrieved May 10, 2026, from <https://pmc.ncbi.nlm.nih.gov/articles/PMC4496952/>
2. LaPelusa, A., Asuncion, R. M. D., & Kentris, M. (2026). Muscular dystrophy. In *StatPearls*. StatPearls Publishing. Retrieved May 20, 2026, from <https://www.ncbi.nlm.nih.gov/books/NBK560582/>
3. Crisafulli, S., Sultana, J., Fontana, A., Salvo, F., Messina, S., & Trifirò, G. (2020). Global epidemiology of Duchenne muscular dystrophy: An updated systematic review and meta-analysis. *Orphanet J Rare Dis*, 15, 141. <https://doi.org/10.1186/s13023-020-01430-8>
4. Liao, Q., Zhang, Y., He, J., & Huang, K. (2022). Global prevalence of myotonic dystrophy: An updated systematic review and meta-analysis. *Neuroepidemiology*, 56(3), 163–173. <https://doi.org/10.1159/000524734>
5. Luglio, A., Maggi, E., Riviello, F. N., Conforti, A., Sorrentino, U., & Zuccarello, D. (2024). Hereditary neuromuscular disorders in reproductive medicine. *Genes (Basel)*, 15, 1409. <https://doi.org/10.3390/genes15111409>
6. Mercuri, E., Bonnemann, C. G., & Muntoni, F. (2019). Muscular dystrophies. *Lancet*, 394, 2025–2038. [https://doi.org/10.1016/S0140-6736\(19\)32910-1](https://doi.org/10.1016/S0140-6736(19)32910-1)
7. Wahlgren, L., Kroksmark, A. K., Tulinius, M., & Sofou, K. (2022). One in five patients with Duchenne muscular dystrophy dies from other causes than cardiac or respiratory failure. *Eur J Epidemiol*, 37(2), 147–156. <https://doi.org/10.1007/s10654-021-00819-4>
8. Huzarski, F., Marta, P. K., Ossolińska, A., Ferfecka, G. M., Pawelek, K. A., Stolarska, L., et al. (2025). Respiratory complications and management in Duchenne muscular dystrophy and amyotrophic lateral sclerosis. *J Educ Health Sport*, 78, 57513. <https://doi.org/10.12775/JEHS.2025.78.57513>
9. Bushby, K. M. & Gardner-Medwin, D. (1993). The clinical, genetic and dystrophin characteristics of Becker muscular dystrophy. i. natural history. *J Neurol*, 240(2), 98–104. <https://doi.org/10.1007/BF00858725>
10. Zimowski, J. G., Pawelec, M., Purzycka, J. K., et al. (2017). Deletions, not duplications or small mutations, are the predominant new mutations in the dystrophin gene. *J Hum Genet*, 62, 885–888. <https://doi.org/10.1038/jhg.2017.70>
11. Lee, T., Takeshima, Y., Kusunoki, N., et al. (2014). Differences in carrier frequency between mothers of Duchenne and Becker muscular dystrophy patients. *J Hum Genet*, 59, 46–50. <https://doi.org/10.1038/jhg.2013.119>
12. Hoogerwaard, E. M., Bakker, E., Ippel, P. F., Oosterwijk, J. C., Majoor-Krakauer, D. F., Leschot, N. J., et al. (1999). Signs and symptoms of Duchenne muscular dystrophy and Becker muscular dystrophy among carriers in the Netherlands: A cohort study. *Lancet*, 353(9170), 2116–2119. [https://doi.org/10.1016/S0140-6736\(98\)10028-4](https://doi.org/10.1016/S0140-6736(98)10028-4)
13. Glass, I. A., Nicholson, L. V., Watkiss, E., Johnson, M. A., Roberts, R. G., Abbs, S., et al. (1992). Investigation of a female manifesting Becker muscular dystrophy. *J Med Genet*, 29(8), 578–582. <https://doi.org/10.1136/jmg.29.8.578>
14. Adachi, K., Hashiguchi, S., Saito, M., Kashiwagi, S., Miyazaki, T., Kawai, H., et al. (2018). Detection and management of cardiomyopathy in female dystrophinopathy carriers. *J Neurol Sci*, 386, 74–80. <https://doi.org/10.1016/j.jns.2017.12.024>
15. Hori, H., Maeda, Y., Ishizaki, M., et al. (2015). A carrier with de novo mutation in the dystrophin gene whose myopathic symptoms became seriously progressive after pregnancy and delivery. *Muscle Nerve*, 52(5), 913–914. <https://doi.org/10.1002/mus.24744>
16. Schaufelberger, M. (2019). Cardiomyopathy and pregnancy. *Heart*, 105(20), 1543–1551. <https://doi.org/10.1136/heartjnl-2018-313476>
17. Gupta, A., Hazarika, A., Jain, K., Sikka, P., & Suri, V. (2017). An asymptomatic carrier of becker's muscular dystrophy with cardiomyopathy in pregnancy: Peripartum or not. *Int J Obstet Anesth*, 30, 77–78. <https://doi.org/10.1016/j.ijoa.2016.12.001>
18. Davies, J. E., Winokur, T. S., Aaron, M. F., Benza, R. L., Foley, B. A., & Holman, W. L. (2001). Cardiomyopathy in a carrier of duchenne's muscular dystrophy. *J Heart Lung Transplant*, 20(7), 781–784. [https://doi.org/10.1016/s1053-2498\(00\)00240-0](https://doi.org/10.1016/s1053-2498(00)00240-0)
19. Cheng, V. E. & Prior, D. L. (2013). Peripartum cardiomyopathy in a previously asymptomatic carrier of Duchenne muscular dystrophy. *Heart Lung Circ*, 22(8), 677–681. <https://doi.org/10.1016/j.hlc.2012.11.015>
20. Kinugawa, M., Ichinose, M., Matsui, H., Xiang, Z., Sayama, S., Toshimitsu, M., et al. (2025). Successful pregnancy outcome with preconception care in a symptomatic carrier of Duchenne muscular dystrophy: Case report and literature review. *Am J Med Genet A*, 197, e63926. <https://doi.org/10.1002/ajmg.a.63926>
21. Lamanna, B., Vinciguerra, M., Dellino, M., Cascella, G., Cazzato, G., Macorano, E., et al. (2022). Turner syndrome mosaicism 45,X/46,XY with genital ambiguity and Duchenne muscular dystrophy: Translational approach of a rare Italian case. *Int J Mol Sci*, 23, 14408. <https://doi.org/10.3390/ijms232214408>
22. Fratter, C., Dalgleish, R., Allen, S. K., et al. (2020). EMQN best practice guidelines for genetic testing in dystrophinopathies. *Eur J Hum Genet*, 28, 1141–1159. <https://doi.org/10.1038/s41431-020-0643-7>
23. Superson, M., Szymt, K., Wilk-Trytko, K., Krasnoborska, J., Samojedny, S., Szymańska, K., et al. (2024). Applications of gene modification technologies in the treatment of inherited diseases. *J Educ Health Sport*, 66, 50073. <https://doi.org/10.12775/JEHS.2024.66.002>

24. Ali, A., Rahman, M. Y., & Sheikh, D. (2024). The role of CRISPR/Cas9 in revolutionizing duchenne's muscular dystrophy treatment: Opportunities and obstacles. *Glob Med Genet*, 11(4), 349–357. <https://doi.org/10.1055/s-0044-1791803>
25. Michalska, A. T., Świda, M., Kubuj, C., Adaśko, G., Mazur, U., Bazarewicz, J., Siwik, M., Dmowski, D., Ogonowska, P., & Waszak, J. (2025). Duchenne muscular dystrophy - disease characterization and emergent genetic therapy - literature review. *Quality in Sport*, 43, 62338. <https://doi.org/10.12775/QS.2025.43.62338>
26. Thornton, C. A. (2014). Myotonic dystrophy. *Neurol Clin*, 32(3), 705–719. <https://doi.org/10.1016/j.ncl.2014.04.011>
27. Ranum, L. P., Rasmussen, P. F., Benzow, K. A., Koob, M. D., & Day, J. W. (1998). Genetic mapping of a second myotonic dystrophy locus. *Nat Genet*, 19(2), 196–198. <https://doi.org/10.1038/570>
28. Ashizawa, T., Gagnon, C., Groh, W. J., et al. (2018). Consensus-based care recommendations for adults with myotonic dystrophy type 1. *Neurol Clin Pract*, 8(6), 507–520. <https://doi.org/10.1212/CPJ.0000000000000531>
29. Schoser, B., Montagnese, F., Bassez, G., et al. (2019). Consensus-based care recommendations for adults with myotonic dystrophy type 2. *Neurol Clin Pract*, 9(4), 343–353. <https://doi.org/10.1212/CPJ.0000000000000645>
30. Vydra, D. G. & Rayi, A. (2026). Myotonic dystrophy. In *StatPearls*. StatPearls Publishing. Retrieved June 2, 2026, from <https://www.ncbi.nlm.nih.gov/books/NBK557446/>
31. Turner, C. & Hilton-Jones, D. (2010). The myotonic dystrophies: Diagnosis and management. *J Neurol Neurosurg Psychiatry*, 81(4), 358–367. <https://doi.org/10.1136/jnnp.2008.158261>
32. Lanni, S. & Pearson, C. E. (2019). Molecular genetics of congenital myotonic dystrophy. *Neurobiol Dis*, 132, 104533. <https://doi.org/10.1016/j.nbd.2019.104533>
33. Jain, A. & Al Khalili, Y. (2026). Congenital myotonic dystrophy. [updated 2023 feb 2]. In *StatPearls*. StatPearls Publishing. Retrieved April 15, 2026, from <https://www.ncbi.nlm.nih.gov/books/NBK560518/>
34. Johnson, N. E., Aldana, E. Z., Angeard, N., Ashizawa, T., Berggren, K. N., et al. (2019). Consensus-based care recommendations for congenital and childhood-onset myotonic dystrophy type 1. *Neurol Clin Pract*, 9(5), 443–454. <https://doi.org/10.1212/CPJ.0000000000000646>
35. Johnson, N. E., Hung, M., Nasser, E., Hagerman, K. A., Chen, W., Ciafaloni, E., et al. (2015). The impact of pregnancy on myotonic dystrophy: A registry-based study. *J Neuromuscul Dis*, 2(4), 447–452. <https://doi.org/10.3233/JND-150095>
36. Rudnik-Schöneborn, S. & Zerres, K. (2004). Outcome in pregnancies complicated by myotonic dystrophy: A study of 31 patients and review of the literature. *Eur J Obstet Gynecol Reprod Biol*, 114(1), 44–53. <https://doi.org/10.1016/j.ejogrb.2003.11.025>
37. Rudnik-Schöneborn, S., Schneider-Gold, C., Raabe, U., Kress, W., Zerres, K., & Schoser, B. G. (2006). Outcome and effect of pregnancy in myotonic dystrophy type 2. *Neurology*, 66(4), 579–580. <https://doi.org/10.1212/01.wnl.0000198227.91131.1e>
38. Zagorda, B., Camdessanché, J. P., & Féasson, L. (2021). Pregnancy and myopathies: Reciprocal impacts between pregnancy, delivery, and myopathies and their treatments: A clinical review. *Rev Neurol (Paris)*, 177(3), 225–234. <https://doi.org/10.1016/j.neurol.2020.09.014>
39. Statland, J. M. & Tawil, R. (2016). Facioscapulohumeral muscular dystrophy. *Continuum (Minneap Minn)*, 22(6), 1916–1931. <https://doi.org/10.1212/CON.0000000000000399>
40. Statland, J. & Tawil, R. (2014). Facioscapulohumeral muscular dystrophy. *Neurol Clin*, 32(3), 721–728. <https://doi.org/10.1016/j.ncl.2014.04.003>
41. Tawil, R. & van der Maarel, S. M. (2006). Facioscapulohumeral muscular dystrophy. *Muscle Nerve*, 34(1), 1–15. <https://doi.org/10.1002/mus.20522>
42. Vincenten, S. C. C., van der Stoep, N., Paulussen, A. D. C., Mul, K., Badrising, U. A., Kriek, M., et al. (2022). Facioscapulohumeral muscular dystrophy: Reproductive counseling, pregnancy, and delivery in a complex multigenetic disease. *Clin Genet*, 101, 149–160. <https://doi.org/10.1111/cge.14031>
43. McNiff, M. M., Hawkins, S., Haase, B., Bullivant, J., McIver, T., Mitelman, O., Emery, N., Tasca, G., Voermans, N., & Diaz-Manera, J. (2024). Facioscapulohumeral muscular dystrophy European patient survey: Assessing patient reported disease burden and preferences in clinical trial participation. *J Neuromuscul Dis*, 11(2), 459–472. <https://doi.org/10.3233/JND-230171>
44. Ciafaloni, E., Pressman, E. K., Loi, A. M., Smirnow, A. M., Guntrum, D. J., Dilek, N., et al. (2006). Pregnancy and birth outcomes in women with facioscapulohumeral muscular dystrophy. *Neurology*, 67, 1887–1889. <https://doi.org/10.1212/01.wnl.0000244471.05316.19>
45. Rudnik-Schöneborn, S., Glauner, B., Röhrig, D., & Zerres, K. (1997). Obstetric aspects in women with facioscapulohumeral muscular dystrophy, limb-girdle muscular dystrophy, and congenital myopathies. *Arch Neurol*, 54(7), 888–94. <https://doi.org/10.1001/archneur.1997.00550190076017>
46. Gatica, L. V. & Rosa, A. L. (2016). A complex interplay of genetic and epigenetic events leads to abnormal expression of the DUX4 gene in facioscapulohumeral muscular dystrophy. *Neuromuscul Disord*, 26(12), 844–852. <https://doi.org/10.1016/j.nmd.2016.09.015>

47. Giardina, E., Camaño, P., Burton-Jones, S., Ravenscroft, G., Henning, F., Magdinier, F., et al. (2024). Best practice guidelines on genetic diagnostics of facioscapulohumeral muscular dystrophy: Update of the 2012 guidelines. *Clin Genet*, 106(1), 13–26. <https://doi.org/10.1111/cge.14533>
48. Straub, V., Murphy, A., & Udd, B. (2018). 229th ENMC international workshop: Limb girdle muscular dystrophies—nomenclature and reformed classification, naarden, the Netherlands, 17-19 march 2017. *Neuromuscul Disord*, 28(8), 702–710. <https://doi.org/10.1016/j.nmd.2018.05.007>
49. Johnson, N. E. & Statland, J. M. (2022). The Limb-Girdle muscular dystrophies. *Continuum (Minneap Minn)*, 28(6), 1698–1714. <https://doi.org/10.1212/CON.0000000000001178>
50. Moore, U., Emmons, S. S., Rufibach, L., Straub, V., Diaz-Manera, J., & Guglieri, M. (2023). Patient reported pregnancy and birth outcomes in genetic neuromuscular diseases. *Neuromuscul Disord*, 33, 241–249. <https://doi.org/10.1016/j.nmd.2022.12.013>
51. von Guionneau, A., Burford, C., & Stone, S. (2019). Obstetric management of a woman with limb-girdle muscular dystrophy type 2i and dilated cardiomyopathy. *Obstet Med*, 12(4), 199–201. <https://doi.org/10.1177/1753495X18814622>
52. Libell, E. M., Bowdler, N. C., Stephan, C. M., Zimmerman, M. B., Gedlinske, A. M., & Mathews, K. D. (2021). The outcomes and experience of pregnancy in limb girdle muscular dystrophy type R9. *Muscle Nerve*, 63(6), 812–817. <https://doi.org/10.1002/mus.27184>
53. Mathur, P., Kaur, A., Vijay, U., Gupta, A., Agarwal, K., & Agrawal, L. (2024). Limb-girdle muscular dystrophies (LGMD): Clinical features, diagnosis and genetic variability through next generation sequencing. *Glob Med Genet*, 12(1), 100035. <https://doi.org/10.1016/j.gmg.2024.100035>
54. Mazzoni, A., Althabe, F., Liu, N. H., Bonotti, A. M., Gibbons, L., Sánchez, A. J., et al. (2011). Women's preference for caesarean section: A systematic review and meta-analysis of observational studies. *BJOG*, 118(4), 391–399. <https://doi.org/10.1111/j.1471-0528.2010.02793.x>
55. Tefera, M., Jemberu, L., Getachew, E., Andarge, G. A., Seid, K., Lakew, G., et al. (2025). Global preferences and determinant variables of caesarean delivery among pregnant women: A systematic review and meta-analysis. *Sultan Qaboos Univ Med J*, 25, 1166–1175. <https://doi.org/10.18295/2075-0528.2948>
56. Krysińska, A., Drzewiecka, A., Drzewiecki, A., Korczak, W., Gaszyńska, M., Madejska, M. A., Rudzka, A., Falkowski, W., Woźniak, W., & Magielski, J. (2025). The role of physical activity during pregnancy and its impact on the course of labour and labor outcomes - a review of literature. *Quality in Sport*, 41, 59997. <https://doi.org/10.12775/QS.2025.41.59997>
57. Sadowski, J., Dołęga, J., Zabawa, B., Łabuś, M., Krzykowski, K., Papież, Ł., Maciejczyk, T., Sieńko, A., Hudzińska, P., & Mól, P. (2024). Physical activity in pregnancy: A preventive measure against postpartum depression. *Quality in Sport*, 36, 56364. <https://doi.org/10.12775/QS.2024.36.56364>
58. Chawrylak, K., Kubas, M., Krzemińska, K., Kuszneruk, J., & Kłusek, M. (2024). Physical activity during postpartum period - an analysis of the latest studies and guidelines. *Quality in Sport*, 18, 53253. <https://doi.org/10.12775/QS.2024.18.53253>