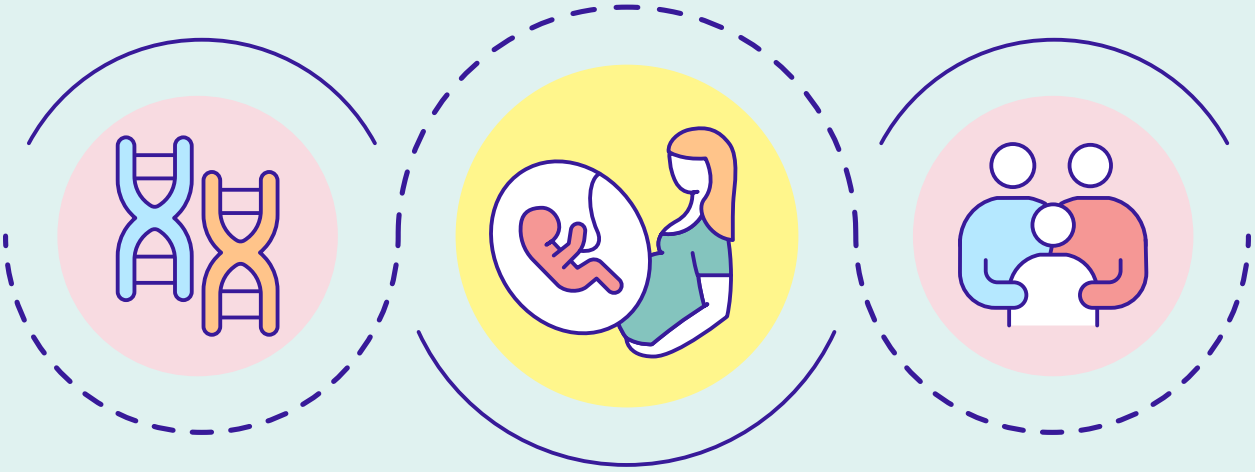
Clinical Recommendations *for* People of Pregnancy Potential with Myotonic Dystrophy



Myotonic Dystrophy FOUNDATION

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The Myotonic Dystrophy Foundation is the world's largest myotonic dystrophy-only patient advocacy organization. Our programs include funding critical research, providing comprehensive resources and support to affected individuals, advocating with government agencies to advance the drug development pipeline, increase research funding, and improve patient services.

Disclaimer: This guide was created to help clinicians navigate risks and care considerations for individuals living with myotonic dystrophy who are pregnant or considering becoming pregnant.

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Introduction

This resource is designed to provide clinicians with an overview of risks and care recommendations for individuals living with myotonic dystrophy (DM) who are pregnant or considering pregnancy. Because DM is a highly variable and multi-systemic disease, it is important to conduct individual risk assessments on a case-by-case basis. Additionally, due to the myriad manifestations and severities of DM, this document describes a variety of outcomes that may or may not be applicable to a patient.

Overall, people with DM of all phenotypes choose to become pregnant, and most are capable of having successful pregnancies. However, they are at increased risk for complications to the dyad (both pregnant person and fetus). Successful outcomes are more likely when pregnant patients are evaluated and cared for by a team of providers familiar with the potential risks and complications of DM and have a developed a plan with patients and their families.

Clinical guidelines are informed by clinical literature, and thus are subject to change. Currently, considerable gaps in the literature prevent understanding the differences between pregnancy outcomes among individuals with DM1 and DM2. Identified differences among pregnant people with DM2 (vs. DM1) include that DM2 is unlikely to result in congenital DM, as well a relative improvement of DM2 symptoms following delivery.

For all people living with DM that are considering pregnancy or reproduction, assessment and discussion with a genetic counselor to review prenatal testing and conception options is strongly recommended.

Patients with DM are at increased risk for miscarriage. Once pregnancy is achieved, physiological changes may worsen physical limitations and respiratory insufficiency, and can increase risk of sleep apnea and cardiac strain. Gestational diabetes is more common among individuals with DM. During delivery, there is an increased risk of preterm or protracted labor due to skeletal muscle weakness or smooth muscle weakness (uterus). Pregnant individuals with DM are also at risk for postpartum hemorrhage. After delivery, muscular weakness can contribute to difficulties with breastfeeding and infant care, and additional supports will be needed.

It is critical that clinical teams are aware of risks associated with pregnancy among individuals with DM, and that potential complications are discussed. Assessment of a patient's individual risk should occur prior to pregnancy in order to make the most informed reproductive health care decisions. Providers caring for pregnant people with DM should closely monitor changes in symptoms and onset of any new symptoms.

Pregnancy and Myotonic Dystrophy (DM): Risks and Recommendations

Risks to Pregnant Person	*Risks to Child*	*Recommendations*
PRECONCEPTION		
■ Increased need for reproductive technology and intervention[1]	■ Risk of inheritance of DM, CDM[1, 2]	■ Genetic counseling ■ Preconception counseling with maternal-fetal medicine. ■ Discussion with genetics regarding inheritance of DM, as well as potential risk for CDM[1, 2, 3] ■ Review or consultation with reproductive specialists regarding assistive technologies (if desired) to reduce the risk of DM inheritance thru IVF with PGT and PGD[1]
EARLY PREGNANCY		
■ Reduced fertility[1] ■ Ectopic pregnancy[2] ■ Miscarriage[1, 2, 7]	■ Risk of inheritance of DM, CDM[1, 2, 3] ■ Miscarriage[1, 2, 7]	■ Genetic counseling (if not already completed) ■ Establish care within a multidisciplinary team of health care providers[2] ■ Review risks for complications and provide appropriate support, referrals, and discussion of reproductive options as appropriate to patients care needs[2]
PREGNANCY		
■ Polyhydramnios (in patients who carry a child with CDM)[2] ■ Placenta previa[4] ■ Increased or aggravated myotonia[5] and symptom presentation[6, 7] ■ Pre-eclampsia[1] ■ Respiratory depression[12, 13]	If CDM: ■ Polyhydramnios with associated preterm labor and PPROM risks[2] ■ Fetal akinesia[8] ■ Hydrops fetalis[5] with associated later pregnancy loss ■ Complications related to previa, worsening DM in the pregnant person ■ Stillbirth[3, 5] ■ Birth defect (e.g., talipes)[1]	■ Ultrasound assessments of fetal anatomy, growth and fluid levels ■ Monitor for DM symptom progression[6] ■ For severe polyhydramnios: consideration of amnioreduction, delivery, or other therapies for polyhydramnios if respiratory compromise is present[9, 10] ■ If hydrops develops, monitor for associated compromise in pregnant person (Mirror Syndrome) ■ Multidisciplinary meeting and goals of care planning, including palliative care and pregnancy discussions for infants affected by CDM with associated poor prognosis ■ Anesthesia consultation ■ Predelivery/pre-admission multidisciplinary team planning including obstetrics, neurology, MFM, anesthesia, nursing and pediatrics (even if infant unaffected) to support safe delivery and healthy newborn transitions. ■ PT/OT consultation to prepare for ADLs of infant care and lactation/chest/breastfeeding

Risks to Pregnant Person	*Risks to Child*	*Recommendations*
LABOR & DELIVERY		
■ Premature onset of labor[1, 5] ■ Pre-eclampsia[1] ■ Prolonged labor and delivery due to uterine atony[1, 2] ■ Myotonia and muscle spasms[3, 11] ■ Respiratory depression[12, 13] ■ Cesarean delivery[2]	■ Placental abruption[1] ■ Complications due to polyhydramnios and uterine laxity including malposition/malpresentation, umbilical cord prolapse[14]	■ Know risks of utilizing anesthesia with DM patients ■ Multidisciplinary team support ■ Admission T&C given increased hemorrhage risk ■ Monitor closely for effective contractions, consider oxytocin supplementation[10] ■ If cesarean delivery is needed, monitor respiratory status carefully, avoid excessive sedation, provide pulmonary toilet and cough assist as needed
POSTPARTUM		
■ Hemorrhage/retained placenta due to uterine atony with associated risks for transfusion, compromise and hysterectomy[1, 4, 11, 13]	■ Birth defect (e.g., talipes) follow up as needed[1] ■ Respiratory failure or insufficiency and bronchic congestion[4] ■ Inability to swallow or suck, frequent choking[1] ■ Weakness and hypotonia[1] ■ Hydrops fetalis[5]	■ Post-partum support for ADLs of infant (if not affected) ■ NICU/Pediatric care provision as appropriate to infant condition ■ Review post-partum contraceptive options and care ■ Assure post-partum follow-up ■ Screen and refer for PPD/PPMD ■ Avoid early discharge to provide additional support ■ A postpartum management plan should be in place to accommodate any complications[10]

Glossary

Amnioreduction: A procedure to remove excess amniotic fluid from the uterus

Atonic uterus: A uterus unable to properly contract throughout labor and following delivery.

Fetal akinesia: Decreased fetal movement during pregnancy.

Hydrops fetalis: A condition in which large amounts of fluid collects in the tissues and organs of a baby, causing swelling (edema).

In vitro fertilization (IVF): A series of procedures in which mature eggs are retrieved from ovaries and fertilized by sperm outside of the uterus.

Maternal fetal medicine (MFM): A sub-specialty of obstetrics and gynecology focused on the diagnosis and treatment of complicated pregnant individuals and their fetuses.

Preimplantation genetic testing (PGT): A procedure in which cells are removed from the embryo for **preimplantation genetic diagnosis (PGD)**.

Polyhydramnios: An excessive amount of amniotic fluid in the uterus during pregnancy.

Other DM-related Resources

Anesthesia Guidelines
www.myotonic.org/anesthesia

Video: Beyond General Anesthesia: Opiates and Other Pain Meds
https://www.myotonic.org/digital-academy/beyond-general-anesthesia-opiates-and-other-pain-meds

Family Planning with Myotonic Dystrophy
https://www.myotonic.org/family-planning-dm

DM Clinical Care Recommendations, Resources, and Toolkits: www.myotonic.org/toolkits-publications

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We support and connect the myotonic dystrophy **community**.
We provide resources and advocate for **care**.
We accelerate research toward treatments and a **cure**.

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