OBJECTIVE: Although the conventional prevalence of myotonic dystrophy is 1:8,000, the prevalence in Korean population was recently reported as 1:1,245. With higher domestic result than expected, we aimed to investigate the clinical characteristics of pregnancies complicated by congenital myotonic dystrophy in our institution.

METHODS: We have reviewed 11 paired cases of neonates diagnosed with congenital myotonic dystrophy and their mothers between July 2004 and May 2014, with clinical features including maternal history of infertility, prenatal ultrasonographic findings, and neonatal outcomes. Cytosine-thymine-guanine (CTG) repeat expansion in the myotonic dystrophy protein kinase gene of both neonates and their mothers was also examined.

RESULTS: None of mother was aware of their myotonic dystrophy traits before pregnancy. History of infertility followed by assisted reproductive technology accounted for 57.1% (4/7). Distinctive prenatal ultrasonographic finding was severe idiopathic polyhydramnios (66.7%, 4/6) with median amniotic fluid index of 43 (range, 37 to 66). In 37.5% (3/8) cases, decreased fetal movement was evident during prenatal ultrasound examination. For neonatal outcomes, more than half (6/11) were complicated with preterm birth and the proportion of 1-minute Apgar score <4 and 5-minute Apgar score <7 was 44.4% (4/9) and 66.7% (6/9), respectively. Most of neonates were admitted to the neonatal intensive care unit (9/10) because of hypotonia with respiratory problems and there was one infant death. Median number of cytosine-thymine-guanine repeats in mothers and neonates was 400 (range, 166 to 1,000) and 1,300 (range, 700 to 2,000), respectively.

CONCLUSION: Our data suggest that severe idiopathic polyhydramnios with decreased fetal movement in pregnant women, especially with a history of infertility, requires differential diagnosis of congenital myotonic dystrophy.
2. **Prenatal, Neonatal, and Early Childhood Features in Congenital Myotonic Dystrophy.**

**Author(s):** Zapata-Aldana, Eugenio; Ceballos-Sáenz, Delia; Hicks, Rhiannon; Campbell, Craig  
**Source:** Journal of neuromuscular diseases; 2018; vol. 5 (no. 3); p. 331-340  
**Publication Date:** 2018  
**Publication Type(s):** Journal Article  
**PubMedID:** 30010141  
**Abstract:** BACKGROUND Congenital myotonic dystrophy (CDM) is the neonatal onset and most severe presentation of Myotonic Dystrophy type 1. Since it first description, perinatal complications have been detailed including prolonged hospital stay, respiratory and feeding therapy during the neonatal period, although long-term complications are less documented. OBJECTIVE Present a prospective cohort of CDM and compare it to the literature of other CDM case series, to adequately describe and contrast the prenatal, neonatal and infancy features of CDM. METHODS A 5-year cohort of CDM eligible cases was conducted via the Canadian Pediatric Surveillance Program. 38 patients met the inclusion criteria. Comparison to other CDM case series published in the literature between 1992 and 2016 about perinatal and infancy morbidity. RESULT From a total of 118 cases, the most frequent features were Polyhydramnios (58%), feeding therapy (77%), intubation and ventilation (58%); neonatal death was reported in 16% of the cases; the most frequent long-term morbidity were respiratory tract infections. CONCLUSIONS We performed a detailed description of the main perinatal features of CDM and precise documentation of the mortality and morbidity during the first five years of life. This is an essential step in the knowledge of the natural history of CDM.  
**Database:** Medline
3. Combined spinal-epidural and local anesthetic infiltration for cesarean delivery in a patient with myotonic dystrophy and severe cardiopulmonary compromise

**Author(s):** Cai Y.; Scott C.; Anwar A.; James P.; Warrick A.; Vanderhoef K.; Diachun C.

**Source:** Regional Anesthesia and Pain Medicine; Oct 2018; vol. 43 (no. 7)

**Publication Date:** Oct 2018

**Publication Type(s):** Conference Abstract

**Abstract:** Background and Aims: Myotonic dystrophy (MD) is a rare disorder characterized by progressive myopathy and myotonia. Systemic complications may include cardiomyopathy and difficulty weaning from mechanical ventilation (MV). Discussion of obstetrical cases in the context of these comorbidities are sparse, and to our knowledge, this is the first case of regional anesthesia for full-term delivery in a parturient with MD and concomitant cardiopulmonary compromise. Methods: Case report. Results: A 27-year-old G3P0020 at 35w2d with MD and a history of prolonged tracheostomy was admitted for progressive hypoxia requiring supplemental oxygen at 25 weeks gestation. Hospital workup revealed periodic episodes of non-sustained ventricular tachycardia and severe right ventricular enlargement with septal wall flattening during both systole and diastole. Caesarean section was scheduled for 36w2d. Prior to surgery, arterial line and defibrillation pads were placed. A combined spinal-epidural (CSE) was placed at L3-4 without intrathecal drug administration. Due to incomplete sensory deficit, local infiltration using lidocaine was used on skin and uterine incision regions. A viable male with APGAR scores of 3 at 1 minute and 7 at 5 minutes was delivered at 33 minutes after skin incision. Hemodynamic stability was maintained with intravenous epinephrine and vasopressin infusions, and spontaneous ventilation was preserved throughout. Conclusions: Given the risk of prolonged MV, neuraxial anesthesia was critical to maintaining spontaneous respiration. Epidural or intrathecal opioid was avoided due to risk of apnea, and intrathecal anesthetic avoided due to sympathectomy. A CSE without intrathecal dosing may provide inadequate anesthesia and can be supplemented with local infiltration of lidocaine.

**Database:** EMBASE

4. Abnormally invasive placentation in a woman with congenital myotonic dystrophy.

**Author(s):** Dorcier, Lise-Marie; Coatleven, Frédéric; Madar, Hugo; Sentilhes, Loïc

**Source:** International journal of gynaecology and obstetrics: the official organ of the International Federation of Gynaecology and Obstetrics; Mar 2018; vol. 140 (no. 3); p. 376-377

**Publication Date:** Mar 2018

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 29072789

Available at International journal of gynaecology and obstetrics: the official organ of the International Federation of Gynaecology and Obstetrics - from Wiley Online Library Science, Technology and Medicine Collection 2017

**Database:** Medline
5. An unusual case of placenta percreta in a patient with myotonic dystrophy

**Author(s):** Levin G.; Zigron R.; Matan L.; Haj Yahya R.; Rottenstreich A.

**Source:** European Journal of Obstetrics Gynecology and Reproductive Biology; Feb 2018; vol. 221 ; p. 206-207

**Publication Date:** Feb 2018

**Publication Type(s):** Letter

**PubMedID:** 29306564

**Database:** EMBASE

6. Preimplantation Genetic Diagnosis for Myotonic Dystrophy Type 1 and Analysis of the Effect of the Disease on the Reproductive Outcome of the Affected Female Patients.

**Author(s):** Fernández, Raquel María; Lozano-Arana, María Dolores; Sánchez, Beatriz; Peciña, Ana; García-Lozano, Juan Carlos; Borrego, Salud; Antiñolo, Guillermo

**Source:** BioMed research international; 2017; vol. 2017 ; p. 9165363

**Publication Date:** 2017

**Publication Type(s):** Journal Article

**PubMedID:** 29349085

Available at BioMed research international - from Europe PubMed Central - Open Access

**Abstract:** Myotonic dystrophy type 1 (DM1) is the most common adult muscular dystrophy and presents an autosomal dominant inheritance. A reproductive option for the families affected is preimplantation genetic diagnosis (PGD). One limitation of this option is the nonoptimal response to ovarian stimulation of the women with DM1, although controversial results exist regarding this subject. In this study, we have analyzed the results of the PGD program applied to DM1 at our institution. A total of 35 couples have been included in our program since 2010, and 59 cycles have been performed. The percentage of transfers per cycle was 64.4% and the live birth rate per cycle was 18.6%. Interestingly, statistically significant differences were observed for the clinical results in the group of couples with an affected female versus the group with an affected male or versus a group of couples with different referral reasons. Specifically, both the percentage of mature oocytes out of the total oocytes retrieved and the percentage of fertilization were considerably lower in the group of DM1 females. Our findings would suggest the possibility of achieving less favourable PGD outcomes in women with DM1 in comparison with other pathologies, although the underlying mechanism remains unknown.

**Database:** Medline
7. Developmental Milestones and Quality of Life Assessment in a Congenital Myotonic Dystrophy Cohort.

Author(s): Prasad, Madhavi; Hicks, Rhiannon; MacKay, Melissa; Nguyen, Cam-Tu; Campbell, Craig

Source: Journal of neuromuscular diseases; Aug 2016; vol. 3 (no. 3); p. 405-412

Publication Date: Aug 2016

Publication Type(s): Research Support, Non-u.s. Gov't Journal Article

PubMedID: 27854230

Abstract: BACKGROUND Congenital myotonic dystrophy (CDM) is a neuromuscular disorder caused by a CTG triplet repeat expansion in the DMPK gene. In addition to the expected motor delay, affected children often have significant developmental disability in language and cognitive realms, which ultimately impacts on quality of life. OBJECTIVE In a prospective cohort of children with CDM to 1) present the profile of language and motor developmental milestones, and 2) describe their early childhood health related quality of life (HRQOL). METHODSA five year cohort study of eligible incident cases of CDM was performed via the Canadian Pediatric Surveillance Program (CPSP). Consenting subjects were then followed from infancy in a prospective cohort study. Caregivers were contacted every 3 months for the first year of life, and then twice yearly in order to obtain data concerning language skills, motor development and parent proxy HRQOL from the PedsQL and Infant and Toddler Quality of life (ITQOL) questionnaires. RESULTS Milestones were achieved at later ages in patients when compared to healthy children. Girls appeared to be more delayed than boys in both language and motor skills. HRQOL scores remained stable in this cohort, for both the PedsQL and ITQOL. CONCLUSIONS Understanding developmental milestones and quality of life are important parameters when judging a child’s overall health. For CDM patients delineating developmental milestones and QOL have important clinical care and research implications.

Database: Medline


Author(s): Johnson, Nicholas E; Butterfield, Russell; Berggren, Kiera; Hung, Man; Chen, Wei; DiBella, Deanna; Dixon, Melissa; Hayes, Heather; Pucillo, Evan; Bounsanga, Jerry; Heatwole, Chad; Campbell, Craig

Source: Neurology; Jul 2016; vol. 87 (no. 2); p. 160-167

Publication Date: Jul 2016

Publication Type(s): Journal Article

PubMedID: 27306634

Available at Neurology - from Unpaywall

Abstract: OBJECTIVE Herein, we describe the disease burden and age-related changes of congenital-onset myotonic dystrophy (CDM) in childhood. METHODS Children with CDM and age-matched controls aged 0 to 13 years were enrolled. Participants were divided into cohorts based on the following age groups: 0-2, 3-6, and 7-13 years. Each cohort received age-appropriate evaluations including functional testing, oral facial strength testing, neuropsychological testing, quality-of-life measurements, and ECG. Independent-samples t test or Wilcoxon 2-sample test was used to compare the differences between children with CDM and controls. Probability values less than 0.05 are reported as significant. RESULTS Forty-one participants with CDM and 29 healthy controls were enrolled. The 6-minute walk was significantly different between CDM (258.3 m [SD 176.0]) and
The mean lip force strength was significantly different in CDM (2.1 N [SD 2.8]) compared to control participants (17.8 N [SD 7.6]). In participants with CDM, the mean IQ (65.8; SD 18.4) was 3 SDs below the mean compared to standardized norms. Measurements of grip strength, sleep quality, and quality of life were also significantly different. Strength measures (oral facial strength, grip strength, and 6-minute walk) correlated with each other but not with participant IQ.

**CONCLUSION**

This work identifies important phenotypes associated with CDM during childhood. Several measures of strength and function were significantly different between participants with CDM and controls and may be useful during future therapeutic trials.

**Database:** Medline

---

**9. Preimplantation genetic diagnosis for female carriers of myotonic dystrophy type 1**

**Author(s):** Hehr A.; Eichhammer L.; Hehr U.; Seifert B.; Paulmann B.; Seifert D.; Gasner C.

**Source:** Medizinische Genetik; Mar 2016; vol. 28 (no. 1); p. 109-110

**Publication Date:** Mar 2016

**Publication Type(s):** Conference Abstract

**Available at Medizinische Genetik - from SpringerLink**

**Abstract:**

Myotonic dystrophy type 1 is one of the major indications for preimplantation genetic diagnosis (PGD) for monogenic inherited disorders worldwide. Transmission of the unstable extended CTG trinucleotide repeat within the 3'-untranslated region of the DMPK gene from the prospective carrier mother on average results in a further expansion by about 950 CTG units with congenital and severe clinical manifestation in her offspring (Martorell et al., 2007). While the prospective mother herself may be clinically healthy or only mildly affected, her further health and clinical course may be compromised by any pregnancy and delivery, but also by an assisted reproduction required for PGD. Furthermore, preliminary data suggest a reduced PGD success rate compared to PGD for other monogenic inherited disorders (Srebnik et al., 2014), demanding thorough pre-PGD assessment and counselling of each couple regarding their individual risks and chances to deliver a healthy child. In the unique setting of the German embryo protection law we performed at our center in Regensburg polar body diagnosis (PBD) to 9 female DMPK carriers. Transfer of 38 embryos in 22 of 24 PBD treatment cycles (91,7% PBD cycles with embryo transfer) resulted in 4 clinical pregnancies (18,2% pregnancy rate/transfer cycle), 2 missed abortions and delivery of 2 children. However, 59 PBD cycles for the other 3 main indications in our lab (CFTR 31, FraX premutation 16, FraX full mutation 12 cycles) resulted in higher pregnancy rates per cycle with embryo transfer (ET) of 45,8% (CFTR), 23,1% (FraX premutation) and 55,5% (FraX full mutation) and birth/ongoing pregnancies per PBD cycle to ET of 41,6% (CFTR), 15,3% (FraX premutation) and 33,3% (FraX full mutation) respectively, confirming an important impact of the underlying genetic condition on PGD outcome. Current functional data suggest RNA toxicity as a major pathomechanism underlying the multisystemic clinical manifestations in myotonic dystrophy type 1 including a diminished ovarian reserve. However, preliminary data from our 24 PBD treatment cycles formyotonic dystrophy type 1 did show a similar number of retrieved (9,7 vs. 10,8 overall cohort), mature (9,1 vs. 10,6) or fertilized oocytes (6,0 vs. 6,7) per PBD cycle at a median female age of 33,19 years (34,67 years overall cohort). We did, however, observe a reduced implantation rate of 13,1% vs. 17,5% per transferred embryo and birth rate of 8,3% vs. 20,9% per PBD cycle compared to our overall cohort, correlated with decreased Anti-Mueller-Hormon levels. In an ongoing study we currently address potential predictive biomarkers for favourable PGDoutcome in order to improve individual pre-PGD counselling of female carriers interested in PGD. Moreover, important PGD outcome parameters after PBD and trophectoderm biopsy for Myotonic dystrophy type 1 will be assessed and compared in order to identify PGD treatment conditions, most likely to result in livebirth with a minimal number of oocyte retrieval cycles.
10. The Impact of Pregnancy on Myotonic Dystrophy: A Registry-Based Study

**Author(s):** Johnson N.E.; Hung M.; Chen W.; Nasser E.; Hagerman K.A.; Ciafaloni E.; Heatwole C.R.

**Source:** Journal of Neuromuscular Diseases; 2015; vol. 2 (no. 4); p. 447-452

**Publication Date:** 2015

**Publication Type(s):** Article

Available at [Journal of Neuromuscular Diseases](https://www.journals.org) - from Unpaywall

**Abstract:**

**Background:** The rate of symptom progression during pregnancy in myotonic dystrophy (DM) is not currently known. Further, there is little data regarding the rate of pregnancy complications and neonatal outcomes in DM. **Objective:** This study assesses symptom progression and complication rates during pregnancy in women with DM. **Methods:** DM women completed surveys regarding their prior pregnancies. Participants identified complications during their pregnancies and completed the Myotonic Dystrophy Health Index-Short Form (MDHI-SF) to measure their disease burden and identify the severity of select symptoms six-months prior to, during, and six-months after their first pregnancy. **Results:** 152 women with DM reported on 375 pregnancies. Among these pregnancies, there was a 32.5% miscarriage rate. Some complications were common including: pre-Term labor (27.8%), pre-eclampsia (10.4%), and peripartum hemorrhage (13.9%). Participants’ perception of their mobility and ability to perform activities, as measured by the MDHI-SF, worsened during pregnancy and did not recover during the post-partum period. **Discussion:** Miscarriage, maternal disease progression during pregnancy, and other pregnancy related complications may occur in DM. Women with DM should be counseled on these potential risks prior to considering pregnancy.

**Database:** EMBASE

11. Myotonic dystrophy and the importance of the multidisciplinary team in antenatal care

**Author(s):** Lovell R.; Cameron H.

**Source:** BJOG: An International Journal of Obstetrics and Gynaecology; Nov 2014; vol. 121 ; p. 21

**Publication Date:** Nov 2014

**Publication Type(s):** Conference Abstract

Available at [BJOG: An International Journal of Obstetrics and Gynaecology](https://www.bjog.org) - from Wiley Online Library Science, Technology and Medicine Collection 2017

Available at [BJOG: An International Journal of Obstetrics and Gynaecology](https://www.bjog.org) - from Unpaywall

**Abstract:**

**Background:** Myotonic dystrophy (MD) is an autosomal dominant disorder, with a prevalence of 1 in 8000. Features include progressive muscle weakness and myotonia, cardiac conduction abnormalities and IBS-like symptoms, due to smooth muscle involvement. It is a disorder of varying severity; milder cases may only be diagnosed following the onset of symptoms during pregnancy or the birth of a baby with CMD, a more severe early-onset form of the disease. MD is associated with an increased risk of fetal loss, preterm delivery, polyhydramnios (due to reduced swallowing by an affected fetus), abnormal placentation, dysfunctional uterine contractions and therefore abnormal labour, and postpartum haemorrhage (due to atony or retained placenta). The fetus has a 50% chance of being affected, and 10-63% will have CMD. **Case:** A 31-year-old P1 booked at 7+ weeks. Her first child was born by emergency caesarean section (CS) under general anaesthetic (GA) at 27 weeks of gestation, but sadly died at 4 weeks from respiratory failure due to congenital myotonic dystrophy (CMD). The woman had a complicated recovery from GA. She was then found to...
have type 1 MD and first degree heart block. At booking, she reported mild muscle weakness and IBS-like symptoms. She was referred to fetal medicine where chorionic villus sampling was performed, with a normal result. She was subsequently found to have a major placenta praevia. She had an anaesthetic review and a plan was made for delivery by CS under spinal anaesthetic around 32-34 weeks gestation. Conclusion: Antenatal care needs to be carefully coordinated between a multidisciplinary team, involving the woman's neurologist, fetal medicine, genetics and neonatology. Muscle symptoms can worsen during pregnancy, and require input from physiotherapists and occupational therapists. An antenatal anaesthetic review should be sought as GA should be avoided (respiratory muscle weakness) and care taken with analgesics.

Database: EMBASE

12. Myotonic dystrophy: Diagnosis, management and new therapies

Author(s): Turner C.; Hilton-Jones D.

Source: Current Opinion in Neurology; Oct 2014; vol. 27 (no. 5); p. 599-606

Publication Date: Oct 2014

Publication Type(s): Review

PubMedID: 25121518

Available at Current Opinion in Neurology - from Ovid (Journals @ Ovid) - Remote Access

Abstract: Purpose of review: Myotonic dystrophies type 1 and type 2 are progressive multisystem genetic disorders with clinical and genetic features in common. Myotonic dystrophy type 1 is the most prevalent muscular dystrophy in adults and has a wide phenotypic spectrum. The average age of death in myotonic dystrophy type 1 is in the fifth decade. In comparison, myotonic dystrophy type 2 tends to cause a milder phenotype with later onset of symptoms and is less common than myotonic dystrophy type 1. Historically, patients with myotonic dystrophy type 1 have not received the medical and social input they need to maximize their quality and quantity of life. This review describes the improved understanding in the molecular and clinical features of myotonic dystrophy type 1 as well as the screening of clinical complications and their management. We will also discuss new potential genetic treatments. Recent findings: An active approach to screening and management of myotonic dystrophies type 1 and type 2 requires a multidisciplinary medical, rehabilitative and social team. This process will probably improve morbidity and mortality for patients. Genetic treatments have been successfully used in in-vitro and animal models to reverse the physiological, histopathological and transcriptomic features. Summary: Molecular therapeutics for myotonic dystrophy will probably bridge the translational gap between bench and bedside in the near future. There will still be a requirement for clinical screening of patients with myotonic dystrophy with proactive and systematic management of complications. Copyright © Lippincott Williams & Wilkins. Unauthorized reproduction of this article is prohibited.

Database: EMBASE
13. New insights about the incidence, multisystem manifestations, and care of patients with congenital myotonic dystrophy

**Author(s):** Hilbert J.E.; Johnson N.E.; Moxley III R.T.

**Source:** Journal of Pediatrics; 2013; vol. 163 (no. 1); p. 12-14

**Publication Date:** 2013

**Publication Type(s):** Editorial

**PubMedID:** 23507025

Available at The Journal of Pediatrics - from Patricia Bowen Library & Knowledge Service West Middlesex University Hospital NHS Trust (lib302631) Local Print Collection [location] : Patricia Bowen Library and Knowledge Service West Middlesex university Hospital.

**Database:** EMBASE

14. A postnatal diagnosis of myotonic dystrophy in both the mother and baby: Case report and literature review

**Author(s):** Boyes Z.; Swain V.; Cunningham S.

**Source:** BJOG: An International Journal of Obstetrics and Gynaecology; Jun 2013; vol. 120 ; p. 122

**Publication Date:** Jun 2013

**Publication Type(s):** Conference Abstract

Available at BJOG: An International Journal of Obstetrics and Gynaecology - from Wiley Online Library Science , Technology and Medicine Collection 2017

Available at BJOG: An International Journal of Obstetrics and Gynaecology - from Unpaywall

**Abstract:** Case A 23-year-old female presented to A and E department feeling generally unwell with high grade pyrexia. On assessment a concealed pregnancy approximately 20 weeks of gestation was discovered. A diagnosis of septic shock and type 2 respiratory failure due to pyelonephritis and severe community acquired pneumonia was made. The patient was managed in ITU for an extended period of 3 weeks due to severe respiratory compromise and suffered a respiratory arrest and brief asystolic cardiac arrest. The pregnancy was dated at 24 weeks of gestation and the remaining antenatal period was complicated by recurrent urinary tract infections and pyelonephritis, requiring multiple admissions to the antenatal ward. At 29 weeks of gestation, ultrasound scan showed polyhydramnios with AFI 36.2 cm and dilated fetal stomach. The pregnancy progressed to 33 weeks 6 days when spontaneous rupture of membranes occurred with significant meconium and fetal bradycardia. Emergency caesarean section resulted in the live birth of a 2050 g male infant with Apgar’s 3 at 1 min and 6 at 5 min. The baby was ventilated from birth but unfortunately died in the third week of life due to respiratory complications. Cytogenetic analysis revealed a diagnosis of myotonic dystrophy type 1 (DM1) in the baby and the diagnosis was subsequently confirmed in the mother. Discussion DM1 is an autosomal dominant disorder caused by CTG triple repeat expansion in the dystroph myotonica protein kinase (DMPK) gene. Severity is related to the number of repeats. It may present as a severe, often lethal form at birth causing severe respiratory difficulty or milder childhood form with progressive muscle weakness and cognitive dysfunction. The condition displays anticipation whereby successive generations are more severely affected as demonstrated by our case. The severe congenital form occurs from extreme expansion of the triplet repeat due to maternal rather than paternal transmission. In cases such as ours where the mother was affected mildly, the condition can remain unidentified until it is inherited by the fetus in the severe form. Antenatal diagnosis is challenging when there is no history of the condition but ultrasound findings of talipes, polyhydramnios and tented mouth should prompt suspicion. For pregnant patients with
myotonic dystrophy, there is an increased risk of recurrent urinary tract infection, respiratory difficulties and preterm labour requiring close monitoring throughout the pregnancy.

Database: EMBASE

15. Steinert's disease and pregnancy

Author(s): Ganta S.; RaviMohan V.


Publication Date: Jun 2013

Publication Type(s): Conference Abstract

Abstract: Background Steinert's disease or Myotonic dystrophy is a progressive systemic disease with autosomal dominant inheritance, incidence being 2.5-5.5/100 000. Pregnancy associated with myotonic dystrophy is rare. Based on the presentation they are classified into 4 types. The 'mild type' is characterised by cataract and mild weakness affecting elderly patients. 'Adult type' starts after puberty and progresses slowly with cardio-vascular, gastrointestinal and respiratory manifestations. 'Childhood type' occurs from 1 to 10 years of age and is characterised by hypotonia, learning difficulties and limited motor skills. 'Congenital DM' is the severest form with symptoms of generalised muscular hypoplasia and mental retardation. About 75% of these babies die within the first year. The myotonic dystrophy gene is located on the long arm of chromosome 19, band 13q contains an unstable trinucleotide - CTG. The severity of the presentation depends on the repetition of CTG leading to DNA instability. Case Herein we present a case of a 30-year-old P0 + 2 known to have adult myotonic dystrophy. Despite extensive counselling declined CVS to assess fetal inheritance of MD. We planned for serial growth scan from 28 weeks, MDT included anaesthetic, paediatric involvement. The 20 week anomaly scans showed bilateral talipes. Growth scan revealed growth above 90th centile and polyhydramnios. Polyhydramnios and bilateral talipes increased the clinical suspicion of fetal involvement. Outcome This case highlighted the complications that may arise during pregnancy, delivery, including anaesthetic problems, and in the neonatal period. During pregnancy polyhydramnios can be a first sign of the disease leading to premature labour. She presented at 31 weeks with symptoms of preterm labour and received steroids and tocolysis, was later admitted at 35 weeks in spontaneous preterm labour. The first stage of labour was prolonged needing syntocinon augmentation. The second stage required assisted delivery due to poor voluntary effort secondary to muscular dysfunction. Third stage of labour was complicated by PPH due to inadequate uterine contraction known to be unresponsive to oxytocin. Despite the recommendation to avoid opiate analgesia lack of epidural services mandated the use of opiates. The affected neonate showed classic anticipation with a severe form of disease. Prognosis depended on gestational age (>35 weeks) and length of mechanical ventilation (<21 days). Sadly the baby was weaned off the ventilator by Day 50.

Database: EMBASE

**Authors:** Kamsteeg, Erik-Jan; Kress, Wolfram; Catalli, Claudio; Hertz, Jens M; Witsch-Baumgartner, Martina; Buckley, Michael F; van Engelen, Baziel G M; Schwartz, Marianne; Scheffer, Hans

**Source:** European journal of human genetics : EJHG; Dec 2012; vol. 20 (no. 12); p. 1203-1208

**Publication Date:** Dec 2012

**Publication Type(s):** Research Support, Non-u.s. Gov't Journal Article

**PubMedID:** 22643181

**Abstract:** Myotonic dystrophy is an autosomal dominant, multisystem disorder that is characterized by myotonic myopathy. The symptoms and severity of myotonic dystrophy type 1 (DM1) ranges from severe and congenital forms, which frequently result in death because of respiratory deficiency, through to late-onset baldness and cataract. In adult patients, cardiac conduction abnormalities may occur and cause a shorter life span. In subsequent generations, the symptoms in DM1 may present at an earlier age and have a more severe course (anticipation). In myotonic dystrophy type 2 (DM2), no anticipation is described, but cardiac conduction abnormalities as in DM1 are observed and patients with DM2 additionally have muscle pain and stiffness. Both DM1 and DM2 are caused by unstable DNA repeats in untranslated regions of different genes: A (CTG)n repeat in the 3'-UTR of the DMPK gene and a (CCTG)n repeat in intron 1 of the CNBP (formerly ZNF9) gene, respectively. The length of the (CTG)n repeat expansion in DM1 correlates with disease severity and age of onset. Nevertheless, these repeat sizes have limited predictive values on individual bases. Because of the disease characteristics in DM1 and DM2, appropriate molecular testing and reporting is very important for the optimal counseling in myotonic dystrophy. Here, we describe best practice guidelines for clinical molecular genetic analysis and reporting in DM1 and DM2, including presymptomatic and prenatal testing.

**Database:** Medline

17. A myotonic dystrophy 1 patient complicated with placental adherence after miscarriage of one dichorionic diamniotic twin following her tenth in vitro fertilization and embryo transfer.

**Authors:** Endo, Toshiaki; Baba, Tsuyoshi; Sugio, Asuka; Morishita, Miyuki; Takahashi, Madoka; Akashi, Yushi; Ishioka, Shinichi; Tachi, Nobutada; Imai, Tomihiro; Tamakawa, Mitsuharu; Saito, Tsuyoshi

**Source:** Archives of gynecology and obstetrics; Dec 2012; vol. 286 (no. 6); p. 1605-1608

**Publication Date:** Dec 2012

**Publication Type(s):** Letter Case Reports

**PubMedID:** 23011730

**Abstract:** Myotonic dystrophy type 1 (DM1) is an autosomal dominant, multisystem disorder that is characterized by myotonic myopathy. The symptoms and severity of DM1 range from severe and congenital forms, which frequently result in death because of respiratory deficiency, through to late-onset baldness and cataract. In adult patients, cardiac conduction abnormalities may occur and cause a shorter life span. In subsequent generations, the symptoms in DM1 may present at an earlier age and have a more severe course (anticipation). In DM1, no anticipation is described, but cardiac conduction abnormalities as in DM1 are observed and patients with DM2 additionally have muscle pain and stiffness. Both DM1 and DM2 are caused by unstable DNA repeats in untranslated regions of different genes: A (CTG)n repeat in the 3'-UTR of the DMPK gene and a (CCTG)n repeat in intron 1 of the CNBP (formerly ZNF9) gene, respectively. The length of the (CTG)n repeat expansion in DM1 correlates with disease severity and age of onset. Nevertheless, these repeat sizes have limited predictive values on individual bases. Because of the disease characteristics in DM1 and DM2, appropriate molecular testing and reporting is very important for the optimal counseling in myotonic dystrophy. Here, we describe best practice guidelines for clinical molecular genetic analysis and reporting in DM1 and DM2, including presymptomatic and prenatal testing.

**Database:** Medline

Author(s): Awater, Carina; Zerres, Klaus; Rudnik-Schöneborn, Sabine

Source: European journal of obstetrics, gynecology, and reproductive biology; Jun 2012; vol. 162 (no. 2); p. 153-159

Publication Date: Jun 2012

Publication Type(s): Comparative Study Journal Article

PubMedID: 22459654

Abstract: OBJECTIVE Information about pregnancy and delivery in hereditary neuromuscular disorders (NMD) is limited and largely restricted to small case series and single case reports. Further data of obstetric histories in clinically and genetically defined subgroups are required.

STUDY DESIGN We reviewed the obstetric histories of 178 patients with myotonic dystrophy type 1 (DM1) and 2 (DM2), Charcot-Marie-Tooth disease (CMT), spinal muscular atrophy (SMA), limb-girdle muscular dystrophy (LGMD), facioscapulohumeral muscular dystrophy (FSHD), and congenital myopathy (CM) by means of questionnaires and medical reports. Patients were recruited in the period 1992-2010 after they had at least completed one pregnancy. A total of 380 pregnancies resulting in 315 children were documented.

RESULTS Compared to the normal German population, the number of miscarriages and hypertensive diseases in pregnancy was not increased in the cohort. Patients with NMD delivered more frequently by vaginal operations (8.9-18.2%) and by cesarean births with significantly high rates in DM1 (36.7%) and SMA (42.4%). Preterm deliveries were recorded in 30.7% of DM1, 12.6% of DM2, and 29.4% of SMA gestations. Abnormal fetal presentation occurred significantly more frequently in DM1 (34.6%) and LGMD (26.7%) deliveries and was a feature of chairbound patients. Considering a possible influence of pregnancy on the disease course, about half of LGMD, one-third of SMA, and one fifth of CMT patients reported a deterioration of symptoms in pregnancy. Neonatal outcome was favorable in all NMD but DM1, where infantile morbidity and mortality is often but not exclusively related to congenitally affected children.

CONCLUSION Our data are important for obstetric care and genetic counseling of women with NMD who are contemplating pregnancy.

Database: Medline

Author(s): Owen, P M; Chu, C

Source: Anaesthesia and intensive care; Mar 2011; vol. 39 (no. 2); p. 293-298

Publication Date: Mar 2011

Publication Type(s): Case Reports Journal Article

PubMedID: 21485681

Available at Anaesthesia and intensive care - from ProQuest (Hospital Premium Collection) - NHS Version

Abstract:A 21-year-old primiparous patient with subclinical myotonic dystrophy presented at a tertiary hospital at 38 weeks gestation in active labour, having previously been reviewed in the high-risk pregnancy clinic. A non-reassuring cardiotocogram and raised foetal scalp lactate necessitated an emergency caesarean section. On extubation following an otherwise unremarkable general anaesthetic, the patient required prompt re-intubation due to respiratory compromise, followed by a more gradual period of weaning from positive pressure ventilation. This review explores the implications of myotonic dystrophy for anaesthesia, discusses its multi-system involvement and highlights the difficulties in identifying at-risk patients in the perioperative setting.

Database: Medline

20. The reproductive outcome of female patients with myotonic dystrophy type 1 (DM1) undergoing PGD is not affected by the size of the expanded CTG repeat tract.

Author(s): Verpoest, Willem; Seneca, Sara; De Rademaeker, Marjan; Sermon, Karen; De Rycke, Martine; De Vos, Michel; Haentjens, Patrick; Devroey, Paul; Liebaers, Ingeborg

Source: Journal of assisted reproduction and genetics; Jun 2010; vol. 27 (no. 6); p. 327-333

Publication Date: Jun 2010

Publication Type(s): Journal Article

PubMedID: 20221684

Available at Journal of assisted reproduction and genetics - from ProQuest (Hospital Premium Collection) - NHS Version
Available at Journal of assisted reproduction and genetics - from Europe PubMed Central - Open Access
Available at Journal of assisted reproduction and genetics - from SpringerLink

Abstract: PURPOSE This study aims to analyze the relationship between trinucleotide repeat length and reproductive outcome in a large cohort of DM1 patients undergoing ICSI and PGD. METHODS Prospective cohort study. The effect of trinucleotide repeat length on reproductive outcome per patient was analyzed using bivariate analysis (T-test) and multivariate analysis using Kaplan-Meier and Cox regression analysis. RESULTS Between 1995 and 2005, 205 cycles of ICSI and PGD were carried out for DM1 in 78 couples. The number of trinucleotide repeats does not have an influence on reproductive outcome when adjusted for age, BMI, basal FSH values, parity, infertility status and male or female affected. Cox regression analysis indicates that cumulative live birth rate is not influenced by the number of trinucleotide repeats. The only factor with a significant effect is age (p < 0.05). CONCLUSION There is no evidence of an effect of trinucleotide repeat length on reproductive outcome in patients undergoing ICSI and PGD.

Database: Medline

**Author(s):** Iacoponi S.; Cuerva M.; Paredes B.; De La Calle M.; Rodriguez R.; Gonzalez A.

**Source:** Journal of Maternal-Fetal and Neonatal Medicine; May 2010; vol. 23 ; p. 195

**Publication Date:** May 2010

**Publication Type(s):** Conference Abstract

**Abstract:** Clinical Cases or Summary Results: Steinert's disease is a genetic condition, which is inherited in an autosomal dominant pattern, result from expansion of CTG trinucleotide repeat gene (chr19q), encoding a putative protein kinase. It is a severe form of muscular dystrophy marked by generalized weakness and muscular wasting. The onset can be any time from birth to middle age. The complications during pregnancy are miscarriage, premature labor, hydramnios, atonic postpartum hemorrhage, difficulties during delivery, anaesthetic accidents. We report the case of a healthy 37 years old pregnant, with an ordinary family anamnesis. She was diagnosticated of gestational diabetes and inespecific polyhydramnios during the 30th week of pregnancy. Due to labor contractions and metrorrhagia in the 35th week she came to our emergency department. She underwent a cesarean section delivery of a male baby, who suffered severe breathing difficulties and generalized myotonia. Afterwards, the baby was diagnosticated with Steinert's congenital disease. Following genetic analysis of the mother revealed that she also suffers Steinert's disease.

The diagnosis of the congenital Steinert's disease is really difficult, when the parents are unaware of the disease. Our objective is to emphasize in the importance of a good anamnesis and the characters that can be found out by ultrasound (hydramnios, reduction of fetal tone and active movements, micrognathia, arthrogryposis) that can bring us at least to a prenatal suspicion.

**Database:** EMBASE

---

22. Myotonic dystrophy and pregnancy.

**Author(s):** Khan, Zainab Ashraf; Khan, Shahid Aziz Anwer

**Source:** JPMA. The Journal of the Pakistan Medical Association; Oct 2009; vol. 59 (no. 10); p. 717-719

**Publication Date:** Oct 2009

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 19813690

**Abstract:** Myotonic dystrophy is the most common neuromuscular disease in adults with a prevalence of 2.4-5.5 per 100,000. Here we describe two cases of DM and discuss their obstetric complications. Our first case concerns a 39 year old multipara whose pregnancies were complicated by recurrent abdominal pain, polyhydramnios and post partum haemorrhage which was attributed to DM. In our second case we discuss the management of a 27 year old woman with dichorionic, diamniotic twins. Chorionic Villous Sampling at 11 weeks revealed one of the fetuses, a male; to be afflicted by DM. Selective termination of the affected twin was performed. Unfortunately, she developed severe oligohydramnios and chronic liquor leak. This resulted in the intra-uterine death of the second twin 5 days later. Our cases highlight the importance of prenatal diagnosis and prompt genetic counselling. A multidisciplinary team approach is required in the management of such high risk cases.

**Database:** Medline
23. Polar body biopsy for Curschmann-Steinert disease and successful pregnancy following embryo vitrification.

**Author(s):** Macas, Ervin; Mátyás, Gábor; Reuge, Philippe; Berger, Wolfgang; Imthurn, Bruno

**Source:** Reproductive biomedicine online; Jun 2009; vol. 18 (no. 6); p. 815-820

**Publication Date:** Jun 2009

**Publication Type(s):** Journal Article

**PubMedID:** 19490786

**Abstract:** This report describes the first successful case of preimplantation genetic diagnosis (PGD) for myotonic dystrophy type Curschmann-Steinert (DM1) using polar body biopsy with vitrification. A 39-year-old woman with expansion of a CTG trinucleotide repeat in the DMPK gene was included into the study centre's PGD programme. After intracytoplasmic sperm injection, a total of 13 fertilized oocytes were successfully biopsied for the first and second polar body. Nested multiplex polymerase chain reaction was used to amplify the CTG repeat region in DMPK along with two linked polymorphic markers. Six pronuclear stage (PN) oocytes were diagnosed as unaffected and four as affected by the CTG expansion, while analysis of the remaining PN oocytes was inconclusive. Three normal PN oocytes were left in culture to develop to cleavage-stage embryos and the remaining three were vitrified by applying the Cryotop method. On the following day, only one embryo was transferred into the patient's uterus and the remaining two were vitrified because of the progressive threat of ovarian hyperstimulation syndrome. Since the fresh cycle did not result in a pregnancy, 6 months later the two vitrified cleavage-stage embryos were warmed and transferred back to the patient. A clinical pregnancy was established and a healthy boy was born following Caesarean section in week 39 of gestation.

**Database:** Medline

24. Respiratory compromise after MgSO4 therapy for preterm labor in a woman with myotonic dystrophy: a case report.

**Author(s):** Catanzarite, Val; Gambling, David; Bird, Lynne M; Honold, Jose; Perkins, Erik

**Source:** The Journal of reproductive medicine; Mar 2008; vol. 53 (no. 3); p. 220-222

**Publication Date:** Mar 2008

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 18441730

**Abstract:** BACKGROUND: MgSO4 is widely used for tocolysis. Serious complications are rare as long as dosing is carefully monitored. Adverse effects in myotonic dystrophy have not been previously described. CASE: A 35-year-old woman, gravida 1, para 0, was hospitalized with suspected mild myotonic dystrophy, polyhydramnios and preterm labor at 33 weeks. MgSO4 infusion rapidly resulted in respiratory compromise. Muscular strength returned to baseline after the infusion was stopped. Mother and infant proved to have myotonic dystrophy. CONCLUSION: The choice of tocolytic medication in maternal myotonic dystrophy is problematic. Beta-2 sympathomimetics have been reported to precipitate myotonia. This case illustrates the potential for MgSO4 to cause respiratory embarrassment. Indomethacin may be the tocolytic of choice in myotonic dystrophy.

**Database:** Medline
25. Preimplantation genetic diagnosis for myotonic dystrophy type 1 in the UK

**Author(s):** Kakourou G.; Dhanjal S.; Mamas T.; Fordham K.; Delhanty J.D.A.; Harper J.C.; SenGupta S.B.; Gotts S.; Doshi A.; Serhal P.; Ranieri D.M.

**Source:** Neuromuscular Disorders; Feb 2008; vol. 18 (no. 2); p. 131-136

**Publication Date:** Feb 2008

**Publication Type(s):** Article

**PubMedID:** 18053720

**Abstract:** Myotonic dystrophy type 1 (DM1) is a dominant multisystemic disorder caused by expansion of a trinucleotide repeat in a non-coding region of DMPK. Prenatal diagnosis (PND) is available; however, the decision to terminate affected pregnancies is difficult as the extent of disability is hard to predict from the size of the expansion. In preimplantation genetic diagnosis (PGD) genetic analysis is carried out before the establishment of pregnancy. This paper reviews the largest number of cycles of PGD for DM1 in the UK indicating that PGD is a practical option for affected couples. © 2007 Elsevier B.V. All rights reserved.

**Database:** EMBASE

26. Myotonic dystrophy with pregnancy

**Author(s):** Radhika A.G.; Vaid N.B.; Radhakrishnan G.; Arora M.; Grover A.

**Source:** Journal of the Indian Medical Association; May 2007; vol. 105 (no. 5); p. 269-270

**Publication Date:** May 2007

**Publication Type(s):** Article

**PubMedID:** 17915797

**Abstract:** Myotonic dystrophy is a rare heredodegenerative muscular disorder in which pregnancy is unusual. Because of the autosomal dominant inheritance of the disease, 50% of children of an affected parent may have the disease; 20% of them are asymptomatic at birth. Foetal involvement may be manifested by polyhydramnios, arthrogryposis multiplex in utero, respiratory difficulties, and floppiness at birth. A case of myotonic dystrophy with pregnancy is presented here.

**Database:** EMBASE

Author(s): Zaki, M; Boyd, P A; Impey, L; Roberts, A; Chamberlain, P

Source: Ultrasound in obstetrics & gynecology : the official journal of the International Society of Ultrasound in Obstetrics and Gynecology; Mar 2007; vol. 29 (no. 3); p. 284-288

Publication Date: Mar 2007

Publication Type(s): Journal Article

PubMedID: 17238150

Abstract: OBJECTIVEThe objective of this study was to assess the maternal and prenatal ultrasound findings and outcome in pregnancies complicated by congenital myotonic dystrophy Type 1 (DM1). METHODSA retrospective chart review of all patients with a diagnosis of DM1 and pregnancy presenting to the Oxford Radcliffe Hospital between 1990 and 2004 was undertaken. Obstetric case notes were reviewed and details of all pregnancies obtained. This included data on prenatal diagnostic tests and obstetric ultrasound scans performed as well as pregnancy complications and pregnancy outcome. Maternal and fetal CTG expansion size was also recorded where available. Maternal genetic case notes were reviewed for details of maternal grip myotonia. RESULTSSixty pregnancies among 26 couples in which one of the parents was a carrier of DM1 were identified during the study period. These resulted in 36 (60%) pregnancies affected by congenital DM1 and 19 (31.7%) unaffected pregnancies. There were four miscarriages and one termination of pregnancy for non-medical reasons. Nineteen of the 36 affected pregnancies ended in termination following the antenatal diagnosis of congenital DM1 by either chorionic villus sampling (CVS) or amniocentesis. In the remaining 17 affected pregnancies (16 singleton and one twin) there was one miscarriage of an affected fetus with co-existing Down syndrome and eight perinatal deaths. The principal cause of perinatal death was respiratory failure in the early neonatal period. Antenatally noted clinical/sonographic abnormalities in these pregnancies included polyhydramnios (100%), talipes (26.6%) and borderline ventriculomegaly (13.3%). Uni- or bilateral talipes was noted at delivery in 10 of 16 (62.5%) neonates. Maternal grip myotonia was present in all but one of these cases. CONCLUSIONThe antenatal findings of polyhydramnios and talipes should prompt a search for maternal grip myotonia. If present, definitive testing for congenital DM1 should be considered.

Database: Medline

28. Myotonic dystrophy in pregnancy 'a salutary tale'.

Author(s): Sayed, A T; Moran, P A

Source: Journal of obstetrics and gynaecology : the journal of the Institute of Obstetrics and Gynaecology; Apr 2006; vol. 26 (no. 3); p. 258-260

Publication Date: Apr 2006

Publication Type(s): Case Reports Journal Article

PubMedID: 16698637

Database: Medline
29. Congenital myotonic dystrophy.

Author(s): Upadhyay, K; Thomson, A; Luckas, M J M

Source: Fetal diagnosis and therapy; 2005; vol. 20 (no. 6); p. 512-514

Publication Date: 2005

Publication Type(s): Case Reports Journal Article

PubMedID: 16260886

Available at Fetal diagnosis and therapy - from ProQuest (Hospital Premium Collection) - NHS Version

Abstract: We describe a case of severe congenital myotonic dystrophy (CDM). A 38-year-old primigravida, who was known to suffer from mild myotonic dystrophy (DM), conceived spontaneously and booked for confinement at 11 weeks in our unit. The couple had been fully counseled about the risks of transmission of this condition to their offspring before embarking on this pregnancy. Despite being fully aware of the risks, they declined prenatal diagnosis. The pregnancy was monitored by serial ultrasound scans. The diagnosis of CDM was suspected by ultrasound markers of borderline ventriculomegaly, polyhydramnios, and reduced fetal movements. The pregnancy ended prematurely at 33 weeks in an emergency caesarean section because of severe fetal compromise. The neonate died almost immediately after birth. The genetic analysis of cord blood confirmed severe DM. This case highlights the importance of ultrasound markers for the diagnosis of CDM in the absence of definitive prenatal diagnosis.

Database: Medline


Author(s): Jayawant, Sandeep; Sinha, Rajiv

Source: Indian pediatrics; Jul 2004; vol. 41 (no. 7); p. 746-747

Publication Date: Jul 2004

Publication Type(s): Letter Case Reports

PubMedID: 15297697

Database: Medline
31. Myotonic dystrophy—no evidence for preferential transmission of the mutated allele: a prenatal analysis.

**Author(s):** Zunz, Eran; Abeliovich, Dvorah; Halpern, Gabrielle J; Magal, Nurit; Shohat, Mordechai

**Source:** American journal of medical genetics. Part A; May 2004; vol. 127

**Publication Date:** May 2004

**Publication Type(s):** Journal Article

**PubMedID:** 15103717


**Abstract:** Myotonic dystrophy is the commonest autosomal dominant type of muscular dystrophy in adults. It is one of the trinucleotide repeat expansion disorders, and its severity correlates with the number of CTG repeats in the myotonic dystrophy gene. It has been suggested that myotonic dystrophy exhibits the phenomenon of preferential transmission of the larger mutated alleles that has been described in other trinucleotide repeat disorders. Several authors have reported that the frequency of transmission of the mutated alleles is higher than 50%—a finding that, if true, does not comply with the Mendelian laws of segregation. However, these studies were based on data from the analysis of pedigrees with ascertainment bias. In our study, we determined the frequency of transmission of mutated alleles using data from prenatal molecular studies, which are not subject to ascertainment bias. This is the first study to examine the segregation of the mutated alleles in myotonic dystrophy in pregnancy. Eighty-three fetuses were examined, 30 of 62 mothers (48.38%) and 8 of 21 fathers (38.09%) transmitted the mutated allele, giving an overall transmission rate of 45.78%. We found no evidence of statistically significant deviation of the frequency of transmission of the mutated alleles from the 50% expected in autosomal dominant disorders. This study, unlike previous ones, excludes preferential transmission in myotonic dystrophy, a finding that may be attributable to the lack of correction for ascertainment bias in previous studies and to the use of prenatal data in this study.

**Database:** Medline

32. Outcome in pregnancies complicated by myotonic dystrophy: a study of 31 patients and review of the literature.

**Author(s):** Rudnik-Schöneborn, Sabine; Zerres, Klaus

**Source:** European journal of obstetrics, gynecology, and reproductive biology; May 2004; vol. 114 (no. 1); p. 44-53

**Publication Date:** May 2004

**Publication Type(s):** Case Reports Journal Article Review

**PubMedID:** 15099870

**Abstract:** OBJECTIVES Myotonic dystrophy can be associated with increased obstetric risks, but the maternal contribution for gestational outcome is difficult to establish considering the varying degrees of severity and the influence of fetal factors. STUDY DESIGN We analyzed the pregnancy course and outcome of 31 women with classic myotonic dystrophy, who delivered a total of 66 children. In addition, 93 gestations from the literature were reviewed. RESULTS As most patients were not aware of their diagnosis at reproductive age, often the first indication of the maternal disease was a severely affected child (39%). Miscarriages and pre-eclampsia did not increase. Ectopic pregnancies occurred in 4%, placenta previa in 9% of gestations, while postpartum hemorrhage due to uterine atonia was only reported twice. Severe urinary tract infections were reported for 19% of
the patients, but were only rarely encountered in the literature. Preterm labor, before 34 weeks, occurred in 19% of gestations and was often, but not exclusively attributed to congenitally affected fetuses in contrast to polyhydramnios (17%). Labor abnormalities of all three stages were frequent, increasing the number of operative deliveries (cesarean section rate 36%). Perinatal mortality was 15% and mainly related to congenitally affected children. CONCLUSION: The risk for obstetric complications and urinary tract infections increases for pregnant patients with myotonic dystrophy. They need constant obstetric monitoring. It is hoped that a better awareness of the clinical picture might help to improve gestational outcome in myotonic dystrophy.

**Database:** Medline

---

**33. Torsion of a non-gravid leiomyomatous uterus in a patient with myotonic dystrophy complaining of acute urinary retention: anaesthetic management for total abdominal hysterectomy.**

**Author(s):** Varras, M; Polyzos, D; Alexopoulos, Ch; Pappa, P; Akrivis, Ch

**Source:** Clinical and experimental obstetrics & gynecology; 2003; vol. 30 (no. 2-3); p. 147-150

**Publication Date:** 2003

**Publication Type(s):** Case Reports Journal Article Review

**PubMedID:** 12854863

**Abstract:** Torsion of a pregnant uterus is rare, but torsion of a non-pregnant uterus is extremely rare. Abdominal pain is the major symptom. Other symptoms include vaginal bleeding, urinary tract symptoms and gastro-intestinal manifestations. We present a case of a 37-year-old white nullipara who presented at the emergency room with acute urinary retention. Medical history revealed that the patient carried the disease of myotonic dystrophy, which was diagnosed two years before. Physical examination revealed a tender, distended bladder, which was easily catheterized, draining 900 ml of clear urine. The abdomen was soft with no muscle guarding or rebound tenderness. A palpable large dense mass occupying the cul-de-sac was found during bimanual examination. Abdominal ultrasound examination revealed a large intramural leiomyoma approximately 10 cm in diameter, in the posterior wall of the uterus, which repelled the bladder. In neurological examination the muscular tone and reflexes were reduced in the lower extremities. Myotonic phenomenon was not found. The patient was thought to suffer from myotonic dystrophy and therefore the possibilities for pulmonary and cardiac complications or malignant hyperthermia had to be kept in mind during the anaesthetic management. The patient underwent an exploratory laparotomy and the uterus was found to have undergone a 60 degrees rotation along the corpus and the cervix uteri transition line. Total abdominal hysterectomy with bilateral salpingo-oophorectomy was performed. The intra- and postoperative course of the patient was uneventful. In conclusion, in this patient the uterine pathology (large leiomyoma) in combination with the disease of myotonic dystrophy seemed to be the predisposing factors for the torsion of the non-pregnant uterus. Also, the anaesthetic implications for total abdominal hysterectomy in myotonic dystrophy are discussed and the international literature is reviewed.

**Database:** Medline
34. Recurrent myotonic crisis in a pregnant woman with myotonic dystrophy

Author(s): Benito-Leon J.; Aguilar-Galan E.V.

Source: European Journal of Obstetrics Gynecology and Reproductive Biology; 2001; vol. 95 (no. 2); p. 181

Publication Date: 2001
Publication Type(s): Article
PubMedID: 11301165
Database: EMBASE

35. Myotonic dystrophy in pregnancy

Author(s): Keriakos R.; Aziz N.; Sidra L.

Source: Journal of Obstetrics and Gynaecology; 1999; vol. 19 (no. 1); p. 71-73

Publication Date: 1999
Publication Type(s): Article

Available at Journal of Obstetrics and Gynaecology - from ProQuest (Hospital Premium Collection) - NHS Version
Database: EMBASE

36. Combined maternal and congenital myotonic dystrophy managed by a multidisciplinary team.

Author(s): Atlas, I; Smolin, A

Source: European journal of obstetrics, gynecology, and reproductive biology; Dec 1999; vol. 87 (no. 2); p. 175-178

Publication Date: Dec 1999
Publication Type(s): Case Reports Journal Article
PubMedID: 10597970

Abstract: Myotonic dystrophy is a rare autosomal dominant degenerative neuromuscular and neuroendocrine disease. Pregnancy can aggravate the maternal disease. Obstetrical complications include stillbirth, premature labor, polyhydramnion, abnormal presentation, prolonged labor, increased operative delivery, postpartum hemorrhages and anesthetic accidents. If the fetus is affected severe neonatal morbidity and mortality with arthrogryposis and mental retardation is common. We present a case where the family chose continuation of pregnancy with a known diagnosis of maternal and severe fetal myotonic dystrophy. A multidisciplinary team was used in the management of pregnancy and counseling the patient.

Database: Medline
37. Antenatal and preoperative genetic and clinical assessment in myotonic dystrophy.

Author(s): Boyle, R

Source: Anaesthesia and intensive care; Jun 1999; vol. 27 (no. 3); p. 301-306

Publication Date: Jun 1999

Publication Type(s): Case Reports Journal Article

PubMedID: 10389568

Available at Anaesthesia and intensive care - from ProQuest (Hospital Premium Collection) - NHS Version

Abstract: The antenatal investigation of an obstetric patient with a history of myotonia is described. The smooth and striated muscle dysfunction in myotonic dystrophy renders these patients, as a group, liable to surgical correction and exposure to anaesthesia. A caesarean section is reported to illustrate the preferred timing of diagnosis and peripartum management. While regional anaesthesia is preferred, myotonic dystrophy is not a contraindication to general anaesthesia, provided risks are anticipated and steps taken to minimize complications.

Database: Medline

38. Increased risk for abnormal placentation in women affected by myotonic dystrophy.

Author(s): Rudnik-Schöneborn, S; Röhrig, D; Zerres, K

Source: Journal of perinatal medicine; 1998; vol. 26 (no. 3); p. 192-195

Publication Date: 1998

Publication Type(s): Case Reports Journal Article

PubMedID: 9773377

Abstract: The obstetric histories of 26 women with myotonic dystrophy, who had a total of 66 live births, were reviewed by means of questionnaires and medical reports. Six patients (23%) each had one pregnancy complicated by placenta praevia (affecting 9% of all completed pregnancies), pointing toward a susceptibility to abnormal placentation in this disorder. As involvement of the genitourinary tract is common in myotonic dystrophy, an increased risk for placenta praevia has to be considered in the antenatal care of these patients.

Database: Medline

Author(s): Rudnik-Schöneborn, S; Nicholson, G A; Morgan, G; Röhrig, D; Zerres, K

Source: American journal of medical genetics; Dec 1998; vol. 80 (no. 4); p. 314-321

Publication Date: Dec 1998

Publication Type(s): Journal Article

PubMedID: 9856556

Available at American journal of medical genetics - from Wiley Online Library Science, Technology and Medicine Collection 2017

Abstract: The obstetric histories of 26 women with myotonic dystrophy (DM), who had a total of 67 gestations, were reviewed retrospectively comparing gestations with affected (DM-fetuses) and unaffected fetuses (UA-fetuses). Second, the influence of gestation on the disease course and the personal attitude towards family planning in DM was assessed. Miscarriages and terminations occurred in 11 pregnancies. Of the 56 infants carried to term, 29 had or most likely had inherited the gene for DM from their affected mothers at the time of investigation; 18 (61%) in this series were affected by the congenital form of DM. Perinatal loss rate was 11% and associated with congenital DM. The rate of obstetric complications was significantly increased in all women. However, preterm labor was a major problem in gestations with DM-fetuses (55 vs. 20%), as was polyhydramnios (21% vs. none). While forceps deliveries or vacuum extractions were required in 21% of deliveries with DM-fetuses and only 5% of UA-fetuses, the frequency of Cesarean sections was similar in both groups (24 and 25%). Obstetric problems were inversely correlated with age at onset of maternal DM, while no effect of age at delivery or birth order on gestational outcome was seen. DNA analysis confirmed the diagnosis in 19 patients by the presence of enlarged CTG repeats (EcoRI-expansions) on chromosome 19. Of the 17 patients whose CTG repeat length was known, 59% were classified as E2 (corresponding to 500-1000 repeats), 24% as E1 (1500 repeats) were seen in three patients (17%). Obstetric complications or congenitally affected children occurred in all maternal phenotypes and CTG repeat classes. Eight (31%) patients experienced a worsening of symptoms that was temporary, weight related in three cases, and persistent in five. With the exception of three patients, most new mothers were able to care for their families. To conclude, pregnant women with DM need constant obstetric monitoring and should be advised to deliver in centres with perinatal facilities.

Database: Medline
40. Myotonic dystrophy is a significant cause of idiopathic polyhydramnios.

Author(s): Esplin, M S; Hallam, S; Farrington, P F; Nelson, L; Byrne, J; Ward, K

Source: American journal of obstetrics and gynecology; Oct 1998; vol. 179 (no. 4); p. 974-977

Publication Date: Oct 1998

Publication Type(s): Case Reports Journal Article

PubMedID: 9790382

Abstract: OBJECTIVE: Myotonic dystrophy, the most common form of muscular dystrophy seen in pregnant women, may be a significant cause of middle trimester polyhydramnios. Our purpose was to determine the prevalence of myotonic dystrophy in women with idiopathic polyhydramnios and to characterize the ultrasonographic findings associated with cases.

STUDY DESIGN: We examined the cases of 67 patients who were delivered of infants at the University of Utah between 1992 and 1996 with a diagnosis of idiopathic polyhydramnios (amniotic fluid index >25). Women with diabetes mellitus, hydrops, or fetal anomalies known to cause polyhydramnios were excluded from the study. Amniotic fluid samples or cord blood samples were obtained from 41 patients, and polymerase chain reaction amplification and Southern blot analysis were performed to detect the presence of the myotonic dystrophy mutation. Ultrasonographic findings, prenatal course, and neonatal outcomes were reviewed in all cases.

RESULTS: Four of the 41 patients tested had the myotonic dystrophy mutation, yielding a prevalence in our population of 9.7%. Three of the 4 patients reported a family history of myotonic dystrophy. Ultrasonographic findings associated with a positive result included abnormal posturing of extremities (3/4) and unilateral clubbed foot (3/4). No other structural or growth abnormalities were seen. Two of the patients were delivered before term, 1 at 26 weeks and 1 at 32 weeks. Three of the 4 infants were severely affected, necessitating admission to the intensive care unit, and 1 died on day 11 after birth. One infant, whose myotonic dystrophy mutation consisted of between 800 and 900 triplet repeats, did not require admission to the intensive care unit.

CONCLUSION: Myotonic dystrophy may be seen as idiopathic polyhydramnios and should be considered as part of the differential diagnosis in these cases. Women with a familial history of myotonic dystrophy or ultrasonographic evidence of hypotonia, including positional abnormalities of the extremities, should be offered deoxyribonucleic acid testing for the myotonic dystrophy mutation.

Database: Medline

41. Polyhydramnios: an association with congenital myotonic dystrophy.

Author(s): Schild, R L; Plath, H; Hofstatter, C; Brenner, R; Mann, E; Mundegar, R R; Steinbach, P; Hansmann, M

Source: Journal of obstetrics and gynaecology : the journal of the Institute of Obstetrics and Gynaecology; Sep 1998; vol. 18 (no. 5); p. 484-485

Publication Date: Sep 1998

Publication Type(s): Journal Article

PubMedID: 15512152

Available at Journal of obstetrics and gynaecology : the journal of the Institute of Obstetrics and Gynaecology - from ProQuest (Hospital Premium Collection) - NHS Version

Database: Medline
42. Prenatal diagnosis of congenital myotonic dystrophy and counseling of the pregnant mother: case report and literature review.

Author(s): Geifman-Holtzman, O; Fay, K

Source: American journal of medical genetics; Jul 1998; vol. 78 (no. 3); p. 250-253

Publication Date: Jul 1998

Publication Type(s): Case Reports Journal Article Review

PubMedID: 9677060

Available at American journal of medical genetics - from Wiley Online Library Science, Technology and Medicine Collection 2017

Abstract: The molecular basis of the myotonic dystrophy (MD) kinase gene is expansion of the CTG repeat at the 3'-untranslated region of the MD gene. Variability of the CTG repeat size in different tissues of affected individuals has been demonstrated. The objective of this report was to examine and review the feasibility of prenatal diagnosis of congenital myotonic dystrophy (CMD) in pregnant women with MD using CTG repeat sizes in amniocytes or villi. We present a case of a pregnant woman with MD who underwent prenatal diagnosis of MD using amniocytes. The repeat size in the amniocytes was smaller than the repeat size in the maternal leukocytes and smaller than the repeat size in the infant blood. The infant had CMD. We also reviewed the literature for reports on MD cases that were prenatally tested for CTG repeat size using amniocytes or chorionic villi. Data were tabulated based on the number of maternal CTG repeats, prenatal procedure [amniocentesis or chorionic villus sampling (CVS)], CTG repeat size in fetal tissue, fetal/infant blood, and pregnancy outcome. Twenty-seven pregnancies at risk for MD that underwent prenatal diagnosis were reported. Eleven (40.7%) of the 27 pregnancies underwent amniocentesis, and 16 (59.3%) underwent CVS. Fourteen patients (61%) demonstrated an increase in CTG repeat size in the amniocytes or villi compared with the maternal repeat size. Nine (33%) of the 27 pregnancies were terminated because of CMD risk. The outcomes of 11 (40.7%) pregnancies were consistent with CMD. CMD was diagnosed in fetuses demonstrating expansion or contraction of the CTG mutation in the amniocytes. Prenatal diagnosis of MD is possible by using mutation analysis on maternal and fetal DNA and detection of the CTG repeat expansion. Prenatal diagnosis of CMD is more complex. The possible lack of correlation between CTG repeat size in amniocytes, villi, and other fetal tissues is a potential limitation in prenatal diagnosis and counseling of CMD using CTG repeat size. Thus, prenatal diagnosis of CMD should be based on a combination of factors, including maternal pregnancy history, clinical findings, and cautious interpretation of maternal and fetal DNA analysis.

Database: Medline
43. Myotonic dystrophy in pregnancy: a report of two cases within one family.

**Author(s):** Risseeuw, J J; Oudshoorn, J H; van der Straaten, P J; Kuypers, J C

**Source:** European journal of obstetrics, gynecology, and reproductive biology; Jun 1997; vol. 73 (no. 2); p. 145-148

**Publication Date:** Jun 1997

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 9228495

**Abstract:** Myotonic dystrophy, also called the Curschmann-Steinert syndrome, is an autosomal dominant inherited neuromuscular disorder characterized by progressive muscular dystrophy, muscle weakness and myotonia, which can affect both mother and child. Complications may arise during pregnancy, delivery, including anaesthetic problems, and in the neonatal period. During pregnancy hydramnion can be a first sign of the disease leading to premature labor and also muscle weakness and myotonia can aggravate complicating the course of delivery. The affected neonate may display severe hypotonia, facial diplegia and respiratory distress. The clinical diagnosis can be confirmed by direct DNA analysis in serum and in chorionvillus biopsy material. In this case report two sisters with myotonic dystrophy are described, their pregnancies, deliveries and the outcome of their affected babies.

**Database:** Medline

44. Myotonic dystrophy and pregnancy. A report of two cases and a review of the literature.

**Author(s):** Dufour, P; Berard, J; Vinatier, D; Savary, J B; Dubreucq, S; Monnier, J C; Puech, F

**Source:** European journal of obstetrics, gynecology, and reproductive biology; Apr 1997; vol. 72 (no. 2); p. 159-164

**Publication Date:** Apr 1997

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 9134395

**Abstract:** Myotonic dystrophy is a rare disease (1/8000), that is rarely associated with pregnancy, due to the fact that parents carrying the disease often encounter hypogonadism. Myotonic dystrophy is a neuro-endocrinian 'heredo-degenerative' dystrophy, with dominant autosomic transmission. Its association with pregnancy can lead to several problems. The myotony is often aggravated which leads to obstetrical complications turning into fetal loss, premature term delivery, hydrops, in-utero death, difficulties in expulsion, haemorrhage during delivery and/or anaesthetic accidents. The following signs during the pregnancy can diagnose fetal damage: presence of a hydrops, rare active fetal movements, and low fetal cardiac rhythm. They signify serious fetal damage leading to a diagnosis of myotonic dystrophy. Personal and family antecedents as well as an important hypotony and respiratory distress discovered in the new born are equally evocative elements. In congenital cases (6-30% of the time) the prognosis of the child is pessimistic. For all of the above elements, transmission is of maternal origin. The diagnosis of the congenital form is difficult because the disease is often unknown by the mother. The appearance of molecular tools permits a diagnosis to be formed much more rapidly in a new-born suspected to carry the illness of neonatal Steinert. Two observations illustrate this pathology. The occurrence of congenital myotonic dystrophy in a new-born allows us to diagnose the disease within the mother.

**Database:** Medline
45. Myotonic dystrophy in pregnancy.

Author(s): Vendola, N; Matarazzo, C; Bennici, S

Source: International journal of gynaecology and obstetrics: the official organ of the International Federation of Gynaecology and Obstetrics; Sep 1995; vol. 50 (no. 3); p. 297-298

Publication Date: Sep 1995
Publication Type(s): Letter Case Reports
PubMedID: 8543117
Database: Medline

46. Anaesthesia for caesarean section in a patient with myotonic dystrophy receiving warfarin therapy.

Author(s): Campbell, A M; Thompson, N

Source: Canadian journal of anaesthesia = Journal canadien d’anesthesie; May 1995; vol. 42 (no. 5); p. 409-414

Publication Date: May 1995
Publication Type(s): Case Reports Journal Article
PubMedID: 7614649
Available at Canadian journal of anaesthesia = Journal canadien d’anesthesie - from SpringerLink

Abstract:A 31-yr-old parturient with myotonic dystrophy and asthma presented for elective Caesarean section. The patient was receiving warfarin having had two previous episodes of thromboembolism. Anticoagulation was subsequently provided by heparin in the weeks prior to delivery. The combination of the patient’s medical conditions and the continuing need for anticoagulation presented a considerable anaesthetic problem in planning anaesthesia and analgesia for both elective and emergency delivery. Heparin was discontinued on the day prior to surgery and restarted immediately after surgery. During surgery flowtron anti-embolitic boots were used. Warfarin therapy was recommenced on the seventh postoperative day. Anaesthesia for Caesarean section was provided using a combined spinal epidural technique using a separate needle, separate interspace method. Postoperative pain was relieved by using a continuous epidural infusion, transcutaneous nerve stimulation and diclofenac. No new neurological problems arose despite the use of epidural analgesia in the presence of heparin anticoagulation. This method of providing anaesthesia and postoperative analgesia without the use of opioids in an anticoagulated, asthmatic, myotonic parturient has not been described elsewhere.

Database: Medline
47. Congenital myotonic dystrophy: molecular diagnosis and clinical study.

**Author(s):** Hojo, K; Yamagata, H; Moji, H; Fujita, T; Miki, T; Fujimura, M; Kidoguchi, K

**Source:** American journal of perinatology; May 1995; vol. 12 (no. 3); p. 195-200

**Publication Date:** May 1995

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 7612095

**Abstract:** Recently, an unstable DNA fragment specific to myotonic dystrophy (MyD) was discovered. In affected individuals, a DNA fragment is found that is larger than in normal siblings. Our objectives were to show whether the results of DNA analysis agree with the disease severity and prognosis in congenital myotonic dystrophy (CMyD) by DNA analysis. We investigated three pregnancies (two studied retrospectively) in three families. We genotyped the family members with the Southern blots and the polymerase chain reaction (PCR) analysis. In one case a prenatal diagnosis was carried out using chorionic villus sampling. This report also presents the three cases of affected mothers and CMyD babies with their growth courses. We clarify four main problems in CMyD, namely, respiratory distress, delayed motor development, feeding difficulty, and delayed mental development. The allele size in the range of 10 to 13 kb tended to be present as the adult form of MyD, and 14 to 15 kb as the CMyD. The three CMyD cases whose alleles size in the range of 14 to 15 kb showed various forms of disease and prognosis. We reached the following conclusions: the disease severity and prognosis in babies with CMyD did not correlate with the result of DNA analysis. The DNA analysis is a useful test for prenatal diagnosis. However, it is impossible to predict the disease severity and prognosis in babies with CMyD.

**Database:** Medline

48. Outcome of pregnancy in women with myotonic dystrophy and analysis of CTG gene expansion.

**Author(s):** Erikson, A; Forsberg, H; Drugge, U; Holmgren, G

**Source:** Acta paediatrica (Oslo, Norway : 1992); Apr 1995; vol. 84 (no. 4); p. 416-418

**Publication Date:** Apr 1995

**Publication Type(s):** Research Support, Non-u.s. Gov't Journal Article

**PubMedID:** 7795352

**Abstract:** Pregnancy outcome was investigated in 32 women with clinically obvious myotonic dystrophy. The results indicated that there are two groups of women, those whose children have the adult type of myotonic dystrophy and those whose children have the congenital type. The overall perinatal mortality was 14%. Polyhydramnios was an obvious sign of the congenital type. No subclinical gene carrier was found among the children. We conclude that prenatal diagnosis should be offered to women with myotonic dystrophy, particularly to those who have previously given birth to a child with the congenital type.

**Database:** Medline
49. Anesthetic management for cesarean section in patients with maternal myotonic dystrophy
Author(s): Takano Y.; Okada K.; Murata K.; Mizukami S.; Sato I.
Source: Japanese Journal of Anesthesiology; 1994; vol. 43 (no. 9); p. 1348-1351
Publication Date: 1994
Publication Type(s): Article
PubMedID: 7967031
Abstract: Myotonic dystrophy involves not only voluntary muscles of extremities, pharyngeal muscles and respiratory muscle but also smooth muscle in the gastrointestinal tract. This muscle involvement can cause difficulty in excreting sputa, delayed emptying time of stomach and regurgitation of gastric content, all of which can lead to disastrous complications of anesthetic management. Pregnancy is rare because of ovarian atrophy. We report two cases of cesarean section for patients with myotonic dystrophy. In one case, the newborn baby had dyspnea due to congenital myotonic dystrophy, and in another case, patient experienced postoperative pneumonia. Our cases and other reports suggest that spinal or epidural anesthesia is safely applied for a cesarean section of a patient with myotonic dystrophy.
Database: EMBASE

50. Congenital myotonic dystrophy; a report on thirteen cases and a review of the literature
Author(s): Hageman A.T.M.; Gabreels F.J.M.; Liem K.D.; Renkawek K.; Boon J.M.
Source: Journal of the Neurological Sciences; 1993; vol. 115 (no. 1); p. 95-101
Publication Date: 1993
Publication Type(s): Article
PubMedID: 8166775
Abstract: The congenital variant of myotonic dystrophy (CMD) is a severe disease with a high mortality. CMD is only seen in the offspring of mothers who themselves have myotonic dystrophy (MD). We present 13 patients with clinical symptoms of CMD and neuropathological findings of five of them. The most characteristic symptoms during pregnancy are reduced fetal movements and polyhydramnios. In the neonatal period generalized hypotonia, facial weakness, hyporeflexia, feeding and respiratory difficulties are present. Most of the children have a characteristic tented upper lip. The symptoms greatly diminish after a few weeks. All the children who survive the neonatal period are psychomotor retarded. On pathological examination no specific features were found in muscle tissue or in the brain. The pathogenesis and the cause of the maternal inheritance of CMD is not clear. A review of the literature is provided.
Database: EMBASE

51. Acute cord prolapse in an obstetric patient with myotonia dystrophica.
Author(s): Walpole, A R; Ross, A W
Source: Anaesthesia and intensive care; Nov 1992; vol. 20 (no. 4); p. 526-528
Publication Date: Nov 1992
Publication Type(s): Case Reports Journal Article
PubMedID: 1463191
Database: Medline
52. Genetic risks for children of women with myotonic dystrophy.
Author(s): Goodship, J; Gibson, D E; Burn, J; Honeyman, J; Cubey, R B; Schofield, I
Source: American journal of human genetics; Jun 1992; vol. 50 (no. 6); p. 1340-1342
Publication Date: Jun 1992
Publication Type(s): Letter Case Reports Comment
PubMedID: 1530708
Available at American journal of human genetics - from PubMed
Database: Medline

53. Dystrophia myotonica--emergency caesarean section with spinal anaesthesia.
Author(s): Stevens, J D; Wauchob, T D
Source: European journal of anaesthesiology; Jul 1991; vol. 8 (no. 4); p. 305-308
Publication Date: Jul 1991
Publication Type(s): Case Reports Journal Article
PubMedID: 1874230
Database: Medline

54. Placenta accreta and myotonic dystrophy. Two case reports.
Author(s): Freeman, R M
Source: British journal of obstetrics and gynaecology; Jun 1991; vol. 98 (no. 6); p. 594-595
Publication Date: Jun 1991
Publication Type(s): Case Reports Journal Article
PubMedID: 1873252
Database: Medline
55. Genetic risks for children of women with myotonic dystrophy.

Author(s): Koch, M C; Grimm, T; Harley, H G; Harper, P S

Source: American journal of human genetics; Jun 1991; vol. 48 (no. 6); p. 1084-1091

Publication Date: Jun 1991

Publication Type(s): Research Support, Non-u.s. Gov't Journal Article

PubMedID: 2035529

Abstract: In genetic counseling, the recommended risk estimate that any heterozygous woman with myotonic dystrophy (DM) will have a congenitally affected child is 3%-9%. However, after already having had such an offspring, a DM mother's risk increases to 20%-37%. The risks of 10% and 41%, respectively, calculated in this study are similar to the estimates in the literature. However, our data on clinical status of the mothers demonstrate that only women with multisystem effects of the disorder at the time of pregnancy and delivery are likely to have congenitally affected offspring. No heterozygous woman with polychromatic lens changes but no other clinically detectable multisystem involvement had a congenitally affected child. In addition, our data suggest that the chance of having a more severely affected child increases with greater severity of maternal disease. The findings of this study are relevant for genetic counseling, as the risk of having a congenitally affected child for women with classical manifestations of the disease is shown to be higher than predicted by the overall risk estimate for any heterozygous woman. We consider it appropriate to give these classically affected women risk figures which approach the recurrence risk given to mothers with congenitally affected children. However, the risk of having a congenitally affected child for heterozygous women with no multisystem involvement appears to be minimal. Our findings support the earlier proposed hypothesis of maternal metabolites acting on a heterozygous offspring. Neither genomic imprinting nor mitochondrial inheritance is able to explain the correlation between the clinical status of heterozygous mothers and that of their children.

Database: Medline

56. Anesthetic management of a parturient with myotonia dystrophica: a case report.

Author(s): Camann, W R; Johnson, M D

Source: Regional anesthesia; 1990; vol. 15 (no. 1); p. 41-43

Publication Date: 1990

Publication Type(s): Case Reports Journal Article

PubMedID: 2275912

Abstract: We report the case of a 22-year-old parturient with myotonia dystrophica. She underwent two separate intraabdominal surgical procedures in one day, both under lumbar epidural anaesthesia. Management was directed toward prevention of shivering, a known trigger of myotonic crises. Measures used included warm ambient atmosphere, warmed IV fluids, warming blankets and administration of epidural sufentanil, an opioid recently ascribed as an inhibitor of shivering in parturients.

Database: Medline
57. Severe congestive heart failure and cardiomyopathy as a complication of myotonic dystrophy in pregnancy.

**Author(s):** Fall, L H; Young, W W; Power, J A; Faulkner, C S; Hettleman, B D; Robb, J F

**Source:** Obstetrics and gynecology; Sep 1990; vol. 76 (no. 3); p. 481-485

**Publication Date:** Sep 1990

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 2381631

**Abstract:** The pregnancy of a patient with myotonic dystrophy and heart failure due to cardiac involvement is described. Endomyocardial biopsy was performed at 32 weeks' gestation with echocardiographic guidance to establish the diagnosis. Severe congestive heart failure, refractory to conventional therapy, was encountered. Continuous arteriovenous hemofiltration was used to relieve pulmonary edema before cesarean delivery.

**Database:** Medline

---

58. Obstetric anaesthesia in dystrophia myotonica.

**Author(s):** Blumgart, C H; Hughes, D G; Redfern, N

**Source:** Anaesthesia; Jan 1990; vol. 45 (no. 1); p. 26-29

**Publication Date:** Jan 1990

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 1969250

**Available at Anaesthesia - from Unpaywall**

**Abstract:** Two patients with dystrophia myotonica presented for urgent Caesarean section. Their per- and postoperative courses illustrate the anaesthetic problems posed by this disease. Respiratory difficulties are compounded by pregnancy and there is increased susceptibility to uterine haemorrhage. Choice of anaesthetic agent is discussed. Both had general anaesthetics; muscle relaxation was achieved with vecuronium.

**Database:** Medline

---

59. Obstetric complications as the first sign of myotonic dystrophy.

**Author(s):** Fossen, D; Gjerstad, L

**Source:** Acta obstetricia et gynecologica Scandinavica; 1986; vol. 65 (no. 6); p. 667-668

**Publication Date:** 1986

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 3799165

**Abstract:** A patient with undiagnosed myotonic dystrophy presented in pregnancy with a history of severe obstetric and neonatal complications in her two previous pregnancies. Problems arising after cesarean section led to the diagnosis of myotonic dystrophy. The features of this condition and its implications for pregnancy are pointed out. Complications of pregnancy and parturition may be the first symptoms of myotonic dystrophy.

**Database:** Medline
**Strategy 595287**

<table>
<thead>
<tr>
<th>#</th>
<th>Database</th>
<th>Search term</th>
<th>Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Medline</td>
<td>exp &quot;MYOTONIC DYSTROPHY&quot;/</td>
<td>4932</td>
</tr>
<tr>
<td>2</td>
<td>Medline</td>
<td>(&quot;Dystrophia Myotonica&quot; ADJ2 1).ti,ab</td>
<td>14</td>
</tr>
<tr>
<td>3</td>
<td>Medline</td>
<td>(&quot;Myotonic dystrophy&quot; ADJ2 1).ti,ab</td>
<td>1263</td>
</tr>
<tr>
<td>4</td>
<td>Medline</td>
<td>(1 OR 2 OR 3)</td>
<td>5271</td>
</tr>
<tr>
<td>5</td>
<td>Medline</td>
<td>(pregnan*).ti,ab</td>
<td>457713</td>
</tr>
<tr>
<td>6</td>
<td>Medline</td>
<td>exp PREGNANCY/</td>
<td>849381</td>
</tr>
<tr>
<td>7</td>
<td>Medline</td>
<td>(5 OR 6)</td>
<td>948556</td>
</tr>
<tr>
<td>8</td>
<td>Medline</td>
<td>(4 AND 7)</td>
<td>248</td>
</tr>
<tr>
<td>9</td>
<td>Medline</td>
<td>(&quot;Steinert* disease&quot;).ti,ab</td>
<td>75</td>
</tr>
<tr>
<td>10</td>
<td>Medline</td>
<td>(7 AND 9)</td>
<td>6</td>
</tr>
<tr>
<td>11</td>
<td>Medline</td>
<td>(&quot;congenital Myotonic dystrophy&quot;).ti</td>
<td>170</td>
</tr>
<tr>
<td>12</td>
<td>EMBASE</td>
<td>(&quot;Dystrophia Myotonica&quot; ADJ2 1).ti,ab</td>
<td>22</td>
</tr>
<tr>
<td>13</td>
<td>EMBASE</td>
<td>(&quot;Myotonic dystrophy&quot; ADJ2 1).ti,ab</td>
<td>1897</td>
</tr>
<tr>
<td>14</td>
<td>EMBASE</td>
<td>exp &quot;MYOTONIC DYSTROPHY 7784 TYPE 1&quot;/</td>
<td>7784</td>
</tr>
<tr>
<td>15</td>
<td>EMBASE</td>
<td>(12 OR 13 OR 14)</td>
<td>7880</td>
</tr>
<tr>
<td>16</td>
<td>EMBASE</td>
<td>(pregnan*).ti,ab</td>
<td>583318</td>
</tr>
<tr>
<td>17</td>
<td>EMBASE</td>
<td>exp PREGNANCY/</td>
<td>623991</td>
</tr>
<tr>
<td>18</td>
<td>EMBASE</td>
<td>(16 OR 17)</td>
<td>835292</td>
</tr>
<tr>
<td></td>
<td>Database</td>
<td>Query</td>
<td>Count</td>
</tr>
<tr>
<td>---</td>
<td>-----------</td>
<td>------------------------------------------------------------</td>
<td>-------</td>
</tr>
<tr>
<td>19</td>
<td>EMBASE</td>
<td>(15 AND 18)</td>
<td>273</td>
</tr>
<tr>
<td>20</td>
<td>EMBASE</td>
<td>(&quot;congenital Myotonic dystrophy&quot;).ti</td>
<td>216</td>
</tr>
</tbody>
</table>