

Management of Myotonia Congenita During Pregnancy and Labor

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Abstract

Myotonia congenita (MC) is a rare genetic disorder which affects skeletal muscles leading to delayed relaxation after voluntary contraction. Symptoms are aggravated during pregnancy requiring close monitoring antenatally. We describe the case of a patient in her third pregnancy affected with MC who had a successful delivery of a healthy baby via elective caesarean section. She had an uneventful pregnancy with no worsening of her symptoms and no abnormalities detected on routine scanning. This case is unique as it represents the challenges that obstetricians encounter in managing pregnant women with rare neurological disorders. Due to potential risks related to these conditions, the mainstay of care is multidisciplinary team management and planned labor.

Keywords: Myotonia congenita; Pregnancy; Labor; Anesthesia

Introduction

This is a rare muscle condition which is due to an autosomal dominant inheritance causing a mutation in the chloride channel of skeletal muscle. An autosomal recessive subtype of myotonia congenita (MC) called Becker disease has also been described. The prevalence of both types has been estimated as 6:100,000 worldwide.

Case Report

We describe the case of a 33-year-old patient of Scandinavian background who was diagnosed with MC at age of 12 when she presented with dysarthria. At the age of 15, her disease progressed further and at that point, she developed symptoms of myotonia. The patient described that her symptoms ameliorated in hot weather or after exercise. There was no family history.

She had two previous pregnancies both of which were delivered by elective caesarean section at 39 weeks. This was advised by her obstetrician due to her risk of mechanical obstruction in labor.

She was reviewed initially in the antenatal clinic at 15 weeks gestation of her third pregnancy. Neurological examination at that time was normal apart from hypertrophied proximal muscles of the upper limbs and some difficulty getting up from a sitting position. The patient was educated regarding the 50% risk of that the foetus could be affected with MC. She declined invasive testing as the results would not affect her decision to continue with her pregnancy.

The patient expressed her wishes to have a vaginal birth and the associated risks of vaginal delivery after having two previous caesarean sections were discussed. A consultant neurologist was involved in the discussion regarding the possibility of a vaginal delivery. Their advice was that due to limited information and no current guidance regarding preferred mode of delivery in pregnant women with MC, there are no absolute contraindications to vaginal delivery. An anesthetist reviewed the patient and suggested that she should not be a candidate for general anesthesia. After having an extensive discussion about possible complications from both her neurological condition and previous sections, it was decided by the patient that she would proceed with elective caesarean section at term.

She had an uneventful pregnancy with no worsening of her symptoms and no abnormalities detected on routine scanning. She was admitted electively at 39 weeks gestation for caesarean section under epidural anesthesia. She had an uncomplicated operation with minimum blood loss (300 mL). She gave birth to baby girl who cried immediately

Manuscript accepted for publication April 1, 2014

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doi: <http://dx.doi.org/10.14740/jcgo240w>

Discussion

Thomsen disease is an inherited disorder that affects the gene *CLCN* located on chromosome 7 (7q35) which encodes skeletal muscle chloride channels. The ClC-1 protein is composed of two subunits. Mutations responsible for the dominant type (Thomsen disease) are expected to affect both subunits, while mutation causing the recessive type (Becker disease) most likely affects the encoded subunit. It is important to know the functional characteristics of ClC-1 channel mutations, as this enables clinicians to offer better treatment for affected individuals [1]. There is a 50% possibility of the foetus being affected in Thomsen disease. Considering this, all patients are offered prenatal counselling and diagnostic options.

It typically presents in early childhood, and affects all muscles including face. It was first described in 1876 by the Danish physician Julius Thomsen as he was affected by the condition himself, along with several family members over many generations [2]. This condition causes muscle stiffness, delayed muscle relaxation after voluntary contractions and hypertrophy of the skeletal muscle. Severity of symptoms can vary greatly between individuals and throughout their lifetime.

Symptoms are aggravated during pregnancy requiring close monitoring antenatally. Previously asymptomatic patients may need medication during pregnancy depending on the severity of their symptoms. Due to this potential risk, the mainstay of care is multidisciplinary team management and planned labor [3].

There is limited evidence other than case studies and reports regarding the effects of MC on labor progression. However, some studies have suggested that there may be a risk of prolonged or obstructed labor. The current guidelines do not preclude vaginal delivery [3, 4].

Patients with MC are sensitive to anesthesia, so intra-

and post-operative complications are relatively common in this group especially with depolarizing muscle relaxants. General anesthesia is generally avoided in the condition with a preference for the use of local anesthesia and mild sedatives [3, 4]. A case report has described one patient with undiagnosed MC who had received suxamethonium which led to serious respiratory complications [5].

We describe the case of a patient, in her third pregnancy, affected with MC with the successful delivery of a healthy baby via elective caesarean section. We conclude that the mainstay to manage pregnancy and labor in women with rare neurological disorders is via early multidisciplinary involvement.

References

1. Desaphy JF, Gramegna G, Altamura C, Dinardo MM, Imbrici P, George AL, Jr., Modoni A, et al. Functional characterization of ClC-1 mutations from patients affected by recessive myotonia congenita presenting with different clinical phenotypes. *Exp Neurol*. 2013;248:530-540.
2. OMIM (Online Mendelian Inheritance in Man) www.ncbi.nlm.nih.gov/omim.
3. Gorthi S, Radbourne N, Drury C. Management of pregnancy with Thomsen's disease. *Rajagopalan European Journal of Obstetrics & Gynecology and Reproductive Biology*. 2013;170(1):293-294.
4. Abhijit B, Prathibha N, Olufela I. Pregnancy in women with myotonia congenita. *International Journal of Gynecology & Obstetrics*. 2009;106(1):62-63.
5. Farbu E, Softeland E, Bindoff LA. Anaesthetic complications associated with myotonia congenita: case study and comparison with other myotonic disorders. *Acta Anaesthesiol Scand*. 2003;47(5):630-634.