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A Case Series on Central Core Disease in Pregnancy

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Abstract

Congenital myopathies are a group of minimally progressive or non-progressive neuromuscular conditions which is present from birth. A classical type of congenital myopathy is called central core disease. This condition is often confused with muscular dystrophy. Central core disease can be associated with comorbidities which affect pregnancy and its management. In this case series, we describe two cases, who are siblings affected by the same condition but at varied levels and their management during pregnancy. We also would like to illustrate a management plan for congenital myopathy during pregnancy, for a good maternal and fetal outcome.

Keywords

Congenital Myopathy, Central Core Disease, Pregnancy, Obstetric Management, Malignant Hyperthermia

1. Introduction

This is a group of slowly progressive or non-progressive disorders which are present from birth and cause muscle weakness [1]. The term "congenital" means something which is present since birth and "myopathy" means muscle weakness. Contradicting the previously made statement, clinical variants can exist who present as late onset forms. The most common type of congenital myopathy is central core disease. The point prevalence of this condition in the North of England, has been estimated as 0.4 (0.2 - 0.4) in 100,000 for central core disease and 0.2 (0 - 0.4) in 100,000 for Nemaline myopathy [2]. As the other subtypes are very rare, we could not get data regarding their prevalence in England. It is predominantly inherited as autosomal dominant, although recessive and sporadic forms can exist [3]. There can be a variable level of penetrance, which means that

the severity of each symptom can vary between individuals, although they have the same condition [3].

In our case series, we report two sisters who had "central core" type of congenital myopathy and one was wheelchair bound, while the other was able to walk independently without the use of any aids. However, she could only walk up to limited distances. Congenital myopathy occurs due to missense mutation in the ryanodine receptor gene on chromosome 19 [4]. There are about forty different types of congenital myopathy, the most common among them are:

- Central core disease: It is the most common type of myopathy associated with muscle weakness and developmental problems. "Central core" is an area of central clearing in the muscle with the loss of myofibrils, mitochondria and glycogen. This subtype has a significant association with malignant hyperthermia.
- Multiminicore disease: This condition has many associated subtypes and often causes severe muscle weakness in the arms, legs and scoliosis.
- Centronuclear myopathies: This subtype has associated muscle weakness in the face, arms, legs, and eye muscles, and could have associated breathing problems.
- Congenital fibre type disproportion myopathy: Here small fibres are found on muscle tissue during a biopsy and are associated with muscle weakness in the face, neck, arms, legs and trunk.
- Nemaline myopathy: Nemaline myopathy is a common type of congenital myopathy and causes muscle weakness in the face, neck, arms, and legs, and could sometimes have associated scoliosis. It may also cause breathing and feeding problems.
- Myotubular myopathy: This rare condition, presents only in males and could cause muscle weakness, floppiness and breathing problems.

There are several other myopathies with associated muscle weaknesses, but the description of all the subtypes is beyond the scope of this article. There could also be mixed myopathies.

Patients usually present with difficulty rising, climbing stairs, lifting weights, limitation to the distance to which they can walk. In severe cases, they could be wheelchair bound. During pregnancy fetus affected by congenital myopathy causes mothers to present with recurrent reduced fetal movements. In infancy children usually present with breathing difficulty, feeding difficulty and developmental delay. As they grow up, the commonly associated conditions are osteopenia, gastrointestinal malabsorption, skeletal deformities like kyphoscoliosis, which could in turn cause reduced lung function. Those with Nemaline myopathy typically have "Myopathic faces" [5] with marked facial weakness and facial dysmorphism. Sometimes it can be so severe that these patients are unable to close their eyes, mouth and have dysarthria. Congenital myopathies are usually confirmed by muscle biopsy with histopathology (electron microscopy/enzyme histochemical technique) or genetic mutation analysis. In this case series, we de-

scribe two cases with a similar diagnosis but with varied presentations and management during pregnancy.

2. Case Report

This report consists of siblings with central core disease. Their diagnosis and management in pregnancy has been reported after informed consent.

Case 1: This is a report of 31-year-old Caucasian women, who presented in her first pregnancy at 11 weeks of gestation. She was known to have a diagnosis of congenital myopathy. She had a strong family history of congenital myopathy and members were affected with variable penetrance. She was wheelchair bound and had a central core disease with weakness of central muscles, predominantly proximal muscles. The weakness was more in lower limbs compared to the upper limbs. She had an associated vitamin B12 deficiency. She suffered from osteopenia and had surgeries on the hip and knee at 9 and 11 years respectively.

Her first encounter with the maternity team was at 11 weeks of gestation and she did not have any prenatal checks. Her partner did not carry the genes for congenital myopathy. She was explained about the autosomal dominant nature of transmission and was offered genetic testing for the fetus in the first trimester to screen for congenital myopathy. However, she declined screening. She had an anatomy scan at 20 weeks and was seen by a consultant with special interest in maternal medicine and a specialist midwife throughout pregnancy. She had regular monthly growth scans from 28 weeks. She presented with recurrent reduced fetal movements during pregnancy, which is one of the signs of fetal congenital myopathy. She had a neurological opinion in pregnancy and a plan was made, which suggested that she had no contraindications for a vaginal delivery. She went into preterm labour at 33 weeks and had a category 1 caesarean section in view of breech presentation and abnormal CTG (Cardio toco graph). At the time of delivery, she had a general anaesthesia. A vapour free general anaesthetic with total intravenous anaesthesia was used. The perioperative and postoperative period was uneventful. She had no features of malignant hyperthermia and had a successful surgery. She delivered a baby which had hypotonia, slow respiration, weak cry, and needed respiratory support at birth. The baby also had feeding problems and developmental delay later. The baby was subsequently diagnosed with congenital myopathy.

Case 2: The second case is a sibling of the women described in the first case. She is a 32-year-old women with previous two miscarriages and first presented to the community midwife in the antenatal period at 6 weeks of gestation, with a background diagnosis of congenital myopathy. She had muscle weakness and could not walk long distances or climb stairs but could walk independently, without the need for any kind of aids. She also had vitamin B12 deficiency along with severe kyphoscoliosis. She had a surgery on the spine in the past. There was no compromise in lung function due to kyphoscoliosis. In the first trimester she

was also counselled regarding the nature of transmission and was offered genetic testing to screen the fetus for congenital myopathy, but she also declined fetal screening. She had an anatomy scan at 20 weeks and was seen by the consultant with regular growth scans from 28 weeks onwards. Like her sister, she presented with recurrent reduced fetal movements throughout pregnancy. Neurology and anaesthetic input were requested during pregnancy. The obstetric team was made aware of the condition and a paediatric alert was initiated with an intention that the baby might need respiratory support at birth. The patient had a didelphic uterus and had an elective caesarean at 38weeks in view of breech presentation under spinal anaesthesia. The patient was flagged by the obstetric team and referred to anaesthetic team during the antenatal period. During her preoperative visit, cardiology and respiratory team reviewed her and laid down a clear management plan. The plan made by the multidisciplinary team was followed on the day of the surgery. The patient had an elective caesarean section under spinal anaesthetic. A vapour free Anaesthetic machine was in place with a backup plan of TIVA (Total intravenous anaesthesia) using Propofol/Remifentanil. Intensive care team was alerted; however, the procedure was uneventful. The patient was satisfied with the analgesia she had received. At birth the baby had meconium aspiration, needed CPAP (continuous positive airway pressure) ventilation for 2 days to support respiration and was later discharged. The baby also had feeding difficulties at birth. The baby is now under follow up to monitor developmental milestones and will eventually have a screening test for congenital myopathy.

3. Discussion

Mothers with central core disease can have muscle weakness at varied levels. It is usually associated with kyphoscoliosis, osteopenia and vitamin deficiency. The condition can be associated with life threatening malignant hyperthermia [3] [6]. This is an autosomal dominant condition and neonates affected by this condition can have hypotonia, muscle weakness, weak cry and are likely to need respiratory support at birth. They usually have feeding difficulties and developmental delay.

This discussion aims to describe the various types of congenital myopathy, mode of inheritance, general onset, associated symptoms, appearance, involvement of the various organ systems and intends to compare it with central core disease, which is the diagnosis of both patients in our case series.

The primary problem with congenital myopathy is the missense mutation in the ryanodine receptor gene on chromosome 19, which causes muscle weakness due to sparse mitochondria. Various pathophysiological concepts have emerged to justify the defects in congenital myopathy. Some of the proposed defects include:

1) Sarcolemmal and intracellular membrane remodelling; excitation-contraction coupling; 2) Mitochondrial distribution and function; 3) Myofibrillar force generation; 4) Atrophy; 5) Autophagy [7].

The most common inheritance for this defect is autosomal dominant pattern, although recessive and sporadic forms exist. In both of our patients, the inheritance was autosomal dominant pattern. Their family tree is as described in **Figure 1**.

3.1. Onset of the Disease

Congenital myopathies are a group of muscular disorders, which can be present at birth, begin in infancy or childhood. They either progress very slowly or do not progress at all. It is worth noting that late onset forms and clinical variants exist. Nemaline myopathy is notorious and sometimes can have a late onset. Although, this disease is minimally progressive, rarely the progression may be rapid, and children may die early [1]. This is often noted in Nemaline myopathy and Myotubular myopathy. In our case series both siblings were diagnosed to have central core disease in childhood, which was confirmed by a muscle biopsy. Both declined chorionic biopsy and genetic testing during pregnancy. The baby which was born to the first patient has been diagnosed to have a heterozygous gene mutation confirming congenital myopathy. Baby born to patient 2 has not been tested yet.

3.2. Symptoms

The symptoms and signs of congenital myopathy largely depend on the type of myopathy. During pregnancy, the most common symptom suggestive of an affected infant in-utero is recurrent reduced fetal movements and the most common sign of an infant affected at birth is floppy infant, most often needing respiratory support at birth. As the infant grows older, there could be feeding problems and developmental delay. Individuals show symptoms based on the penetrance. There are clinical findings which are specifically observed in certain

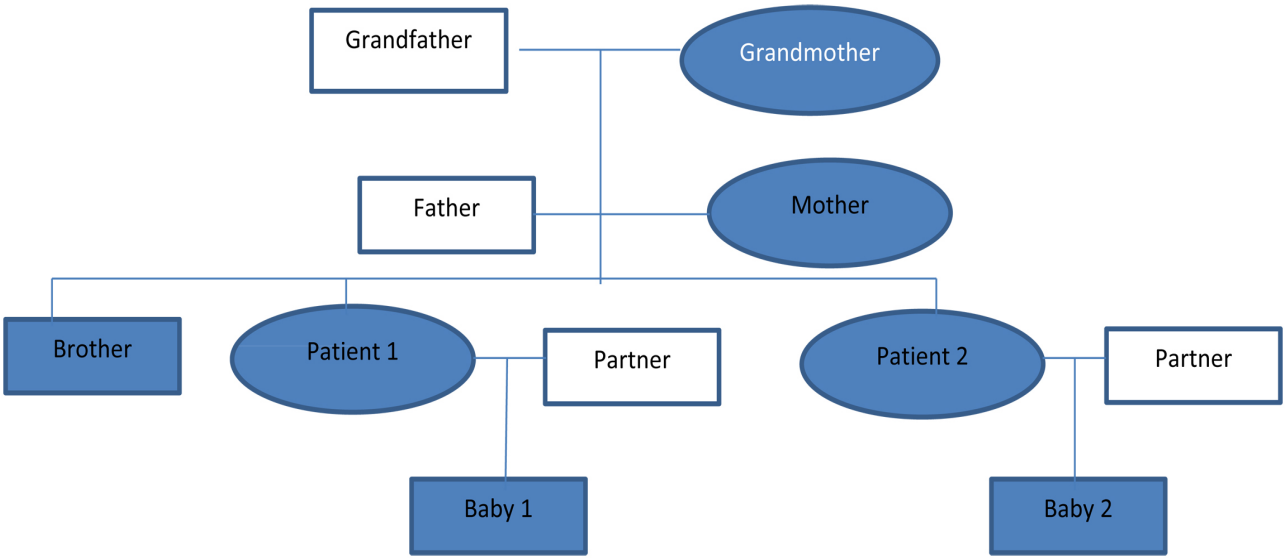

Figure 1. Family tree.

types of congenital myopathy, they are as follows:

- Eyelid ptosis and extraocular muscle weakness is usually seen in multiminicore disease, nemaline myopathy [8] and centronuclear myopathy [9].
- Congenital myasthenic syndromes and respiratory insufficiency is seen in central core and multiminicore diseases [10].
- Cardiac involvement [11] is usually associated with multicore disease and nemaline myopathy.
- Dysphagia is seen with spheroid and cytoplasmic body myopathy.
- Mental retardation in fingerprint myopathy [12] and congenital fibre type disproportion.
- Malignant hyperthermia in central core disease.
- Rigid spine is associated with central core, centronuclear, multiminicore and nemaline myopathy. Scoliosis may be an early and consistent sign.

In our case series both the patients had central core disease. Patient 1 had severe muscle weakness diagnosed in teens and needed pulling on the bannister when climbing stairs. Now she needs to sit while coming downstairs and uses wheelchair for longer distances. She has associated osteopenia and kyphoscoliosis and needed surgery on her hip and knee at 9 and 11 years of age. Her child was floppy at birth, needed respiratory and feeding support at birth and has been diagnosed with central core disease. Patient 2 can walk short distances and can manage daily activities. However, she cannot climb stairs and had severe osteopenia with kyphoscoliosis that needed spinal surgery. Her baby had meconium aspiration at birth and needed positive pressure ventilation for 2 days. The baby later has developed some feeding difficulties. There has been no genetic diagnosis on the baby yet.

3.3. Appearance

Skeletal deformities and contractures of the face and body could cause a "myopathic face" with high arched palate, facial dysmorphism and dysarthria. They are usually unable to close their eyes or mouth completely. This is typical of nemaline myopathy. In our case series both patients and babies did not have any of these features.

3.4. Congenital Myopathy and Lung Function

Congenital myopathy can cause difficulty breathing at birth due to diaphragmatic weakness. In our case series, baby born to patient 1 needed resuscitation at birth due to breathing difficulties. The lung function can be affected in those with severe kyphoscoliosis. The other lung function abnormalities could include restrictive lung disease, respiratory muscle weakness, chest wall abnormalities, chronic aspiration, and infections leading to abnormal ventilation [13]. Hence, a lung function test needs to be done in those with severe kyphoscoliosis and before planning pregnancy. In our case series, although both patients had kyphoscoliosis, their lung function tests were normal.

3.5. Congenital Myopathy and Cardiac Problems

Congenital myopathies can be associated with cardiac insufficiency due to problems with impulse generation or conduction defects causing arrhythmia, focal or diffuse myocardial thickening, dilation of the cardiac cavities, relaxation abnormality, hypertrophic, dilated, restrictive cardiomyopathy, apical form of hypertrophic cardiomyopathy causing sudden cardiac arrest or heart failure with systolic or diastolic dysfunction, Takotsubo phenomenon, secondary valve insufficiency, intra-cardiac thrombus formation [14]. Those with congenital myopathy must have a cardiac evaluation soon after their diagnosis is established and must be followed up by a cardiologist. In our case series, both patients did not have cardiac problems.

3.6. Congenital Myopathy and Obstetric Problems

As discussed earlier, women with affected babies usually present with recurrent reduced fetal movements. There is no direct evidence to prove that congenital myopathy can be associated with obstetric problems, but some studies have shown that patients may experience worsening of motor function and pulmonary function during pregnancy [15] [16] [17]. In a review article described by Sabine Rudnik-Schoenborn *et al*., they have found no significant relationship between congenital myopathy and miscarriages, preterm births, prolonged labour or instrumental deliveries, abnormal placentation or postpartum haemorrhage [13]. However, the rate of caesarean section was high in this group. Both patients in our case series had no problems during pregnancy but patient 1 had a preterm labour with breech presentation. Patient 2 also had fetus with breech presentation, and both needed a caesarean section.

3.7. Congenital Myopathy and Anaesthesia

The anaesthetic management of a parturient with Congenital myopathy is challenging. Along with the physiological changes in pregnancy there are added anaesthetic concerns with regards to Malignant Hyperthermia susceptibility (MHS), Anaesthesia Induced Rhabdomyolysis (AIR) and associated multisystem involvement [18]. The commonly associated systemic manifestations and anaesthetic implications of the different types of congenital myopathies have been summarised in **Table 1**.

In an elective case one should plan and prepare after liaising with the specialists. A clear documentation of classification and extent of disease progression must be made. It is crucial wherever possible to establish a genetic diagnosis due to various overlaps with clinical phenotypes however anaesthesia should not be delayed in an emergency situation [19].

Ryanodine receptor 1 (RYR1) mutation can be associated with MHS, respiratory insufficiency, rigid spine, scoliosis, bulbar weakness, hypotonia, facial dysmorphism and eye involvemnt [20]. A person at risk of malignant hyperthermia (MH) should be subjected to further testing (IVCT-invitro contracture test) as

Table 1. Anaesthetic implications and systemic manifestations associated with congenital myopathy.

Systemic manifestations	Commonly associated congenital myopathy	Preoperative Tests	Anaesthetic implications
Respiratory Insufficiency	Multi/Minicore disease Myotubular myopathy	Pulmonary function test	Sensitive to opioids, Benzodiazepines Avoid Suxamethonium Non depolarising muscle relaxants and magnesium can cause prolonged block
Corpulmonale Cardiomyopathies, MVP	Multicore disease Nemaline myopathy	ECG, 2D ECHO	Exaggerated cardiac decompensation with anaesthetic induction
Scoliosis, osteopenia, previous corrective surgeries, Spine deformities	Central Core disease Multi/Minicore disease Evans Syndrome King-Denborough disease	USG of spine Preoperative baseline Creatine Kinase, Serum Potassium	Careful positioning, moving and handling Technical difficulties in performing central neuraxial block Risk of Rhabdomyolysis
Facial dysmorphism elongated face, high arched palate, dolichocephaly	Nemaline myopathy		Difficult intubation can be anticipated
Bulbar symptoms-difficulty swallowing, coughing	Nemaline myopathy		Risk of aspiration
Life threatening Emergency MH AIR	**MHS** Central core disease Evans Syndrome King-Denborough Disease **AIR** All Congenital myopathies	Genetic diagnosis	Avoid triggers—Succinylcholine, Inhalational anaesthetics Prepare Anaesthetic machine Keep Dantrolene available for reversal

per the European Malignant Hyperthermia Group (EMHG) published guideline [21].

A positive family history and a history of corrective surgeries with anaesthetics along with patient logbook can provide invaluable information. Otherwise, in a case of undiagnosed myopathy, involvement of a specialist centre early in the pregnancy can help aid diagnosis and preparation. MHS and AIR is common with congenital myopathy and can be fatal. A previous uneventful anaesthetic exposure cannot be used to rule out risk of predisposition to MHS and AIR [22] [23].

Regional anaesthesia (Spinal, Epidural, peripheral nerve block) should be chosen whenever possible with prior documentation of the neurological function [24]. Pre-existing respiratory insufficiency can be exacerbated with high spinal anaesthesia. Technical difficulties can be anticipated due to pre-existing spine deformities. Bilateral Transversus Abdominis plane block (TAP) can help reduce

opioid requirements [25]. Perioperative management of Malignant Hyperthermia in suspected or susceptible patients should be as per the guidelines published by the EMHG. The recommendation is to provide anaesthesia free of the triggering agents *i.e.*, volatile anaesthetics and succinylcholine by preparing the anaesthetic workstation. Activated Charcoal Filters can be cost efficient in decontamination of anaesthetic machine from volatile agents [21].

General anaesthesia with endotracheal intubation with total intravenous anaesthesia (TIVA) aided with or without nondepolarising neuromuscular blockade would be ideal [26]. Standard monitoring with high degree of suspicion can help in early and prompt management of life-threatening emergencies such as MH, AIR. Rocuronium with rapid sequence intubation has the advantage of complete reversal with sugammadex which can prevent residual respiratory paralysis [27]. Prolonged neuromuscular blockade is anticipated. Hence, titration with neuromuscular monitoring is ideal. Succinylcholine can cause dangerous hyperkalaemia.

Pain relief should be optimised with short acting opioids and non-opioid medication. Standard post operative monitoring should be in place focussing on early detection of MH, AIR, and cardiopulmonary compromise. In case of life-threatening Malignant hyperthermia, management should be commenced as per the available guidelines [28].

Table 2. Care plan for congenital myopathy in preconception period.

Preconception
Cardiac evaluation
Evaluation of lung function
Evaluation of osteopenia, skeletal deformities and other nutritional deficiencies
Preconceptionally folic acid and supplementation of deficient vitamins and nutrients
Genetic counselling explaining the mode of inheritance and estimating the risk to the offspring. This discussion should cover talks about preimplantation genetic diagnosis, gamete donation, adoption and post conceptional screening techniques.

Table 3. Care plan for congenital myopathy during pregnancy.

Pregnancy
Offer screening with chorionic villous sampling/amniotic fluid testing
As the mobility may be restricted, a venous thromboembolism risk assessment must take place in the first trimester and a plan for thromboprophylaxis must be made.
The patient should be followed up in a consultant led unit with specialist midwife, occupational health therapist and social worker.
Second trimester anomaly scan
Anaesthetic evaluation, assess risk for malignant hyperthermia, kyphoscoliosis and to plan for pain relief and anaesthetic medications that can be used during labour and delivery.
Aim for vaginal delivery, however positioning during labour must be discussed in a multidisciplinary meeting based on individual situation, considering risk factors like osteopenia and previous surgeries.

Table 4. Care plan for congenital myopathy in postpartum period.

Postpartum
Active management of third stage of labour
Reassessment of venous thromboembolism scoring and thromboprophylaxis.
Neonatal assessment and plan for follow up
If the baby is affected, the mother will need additional help with feeding, mental health support, early acquaintance with suitable nursery and toddler groups.
Home visits by family doctor, paediatrician, specialist nurse and social worker may be arranged.

3.8. Ideal Obstetric Management

An ideal obstetric management plan must begin in the preconception period and the patient must receive appropriate antenatal care and postpartum care by a multidisciplinary team. A care plan has been summarised in Tables 2-4.

4. Conclusion

Although there is no direct effect of this condition on pregnancy except possible reduced fetal movements, a multi-disciplinary team which involves a specialist nurse, Neurologist, Cardiologist, Physician, Orthopaedician, Obstetrician, Anaesthetist, Paediatrician and social care is needed for the management of such cases.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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