Journal of Neurology & Neurophysiology

Vavrinkova B, et al., J Neurol Neurophysiol 2015, 6:2
DOI: 10.4172/2155-9562.1000279

Review Article Open Access

Pregnancy and Delivery in Leyden-Möbius Muscular Dystrophy

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Received date: February 10, 2015; **Accepted date:** March 11, 2015; **Published date:** March 16, 2015

Abstract

This case report subscribes a rare case of pregnancy and delivery in a patient with progressive Leyden-Möbius muscular dystrophy. Due to muscle weakness, diaphragm weakness, atrophy of individual muscle groups, spine deformities and often dislocation of thoracic organs, these patients cannot avoid caesarean section to end pregnancy, followed by prolonged intubation and controlled ventilation. During pregnancy, the growing uterus elevates the diaphragm and impairs breathing.The authors have not found any published case of pregnancy in Leyden-Möbius muscular dystrophy in local or international literature. Gravidity in advanced muscular dystrophy is rare and associated with high risk.

Keywords: Leiden-Moebius muscular dystrophy; Pregnancy; Muscle weakness; Atrophy

Introduction

Leyden-Möbius muscular dystrophy is an autosomal recessive hereditary disease of unknown aetiology; it is an inborn disorder of protein metabolism primarily affecting the proximal muscle groups leading to progressive muscular dystrophy. Later it spreads to the muscles of the pelvic floor and lower extremities. The estimated incidence is 1:200,000. The disease has specific electromyographic finding and typically, serum levels of creatine kinase and lactate dehydrogenase are increased. ECG changes may develop in the disease; however, they are not typical. Stretch reflexes are not elicitable, intellect is normal. The disease symptoms develop in the 2nd decennium – weakness of arm and gluteal muscles, back muscles and decreased function of diaphragm. This leads to problems with gait and standing up, decrease of breathing volume and spine statics disruption. In further progression, weakness and atrophy of shank muscles, peroneal muscles, equinovarous position of lower extremities, Achilles tendon contractures, weakness of mimic muscles, neck muscles, pectoral muscles and atrophy of brachial biceps muscles develop.

Causal therapy is not known. Only physiotherapy and orthopaedic management of contractures are available.

Disease prognosis is uncertain, it depends on the disease progression rate [1-4].

A female patient, initials K.J., second gravidity, first delivery, was transferred to the department of obstetrics and gynaecology in the 30th week of gravidity with the following diagnosis: imminent premature delivery, progressive Leyden-Möbius muscular dystrophy, bronchial asthma, severe thoracic spine scoliosis, thyroidopathy and history of vocal cords paresis and myocarditis.

Pregnancy course before admission to the department of obstetrics and gynaecology

In the 9th week of gravidity, she was hospitalised due to bleeding and amniocentesis was performed in the 17th week. The foetus karyotype was without chromosomal aberrations – 46/XY. From the 27th gestation week, she was hospitalised due to aches in the right hypochondrium and signs of imminent premature delivery.

The consulting neurologist stated progression of the underlying disease and recommended ending the pregnancy via caesarean section in case of the condition worsening. Due to the higher availability of specialised consultations and the availability of a well-equipped ICU after eventual surgery, the patient was transferred to the perinatology centre in the 30th week of gravidity.

Finding at admission (3 July)

A female patient of asthenic stature, with clear atrophies of muscle groups in both upper and lower extremities, chest and spine with deformed severe scoliosis. Significantly limited muscle performance. Fully mobile, dyspnoea signs even in minimal strain. Height , weight .

Uterus size corresponding to the 30th gravidity week, slightly irritable. Vaginal examination: cervix length , sacral position, closed, foetus head over the entrance, arches not expanded. Ultrasonography finding without pathology, foetus biometry corresponding to the 30th gestation week.

Laboratory examinations: blood gas tests according to Astrup: pH 7.422, pCO2 35.6 mmHg, pO2 89.6 mmHg, SpO2 97%. Blood count: Hgb l0.l g/dl • Hct 29.4% • RBC 3.28.10/l • WBC 4.9.l0/l, creatinine, urea, ALT, AST, minerals and total proteins normal.

Hospitalisation course

At the beginning of the hospitalisation, uterine contractions decreased after infusion therapy, the patient felt well. There was no clear progression of the underlying disease, pregnancy finding calm. On 23 August, pain in the neck developed with further progression.

Otorhinolaryngology specialist diagnosed epiglottitis and recommended ampicillin 4x1 g. The condition temporarily stabilised. On 8 August, aphonia developed. Otorhinolaryngologist recommended cephoxitin 4x1 g. The general condition worsened, breathing problems developed, dyspnoea at rest and general muscle weakness. Based on the previous neurologic consultation, a caesarean section was scheduled to end the gravidity due to the progression of the underlying disease. The surgery would be performed as soon as there were no intubations contraindications according to the recommendation of the otorhinolaryngology specialist.

On 9 August, the caesarean section was performed without technical complications; 5,000 units of heparin were administered subcutaneously intraoperatively. During skin suture, slight diffuse oozing from the upper subcutaneous layers; wound compression. Further care of the neonate of male gender (2,140 g / 46 cm, good condition – Apgar score 9-10-10) provided by a paediatrist. Blood loss intraoperatively was about 600 ml. The female patient was transferred to the ICU, controlled ventilation was provided due to the underlying disease as scheduled.

On day 0, she tolerated the intubation tube. In the afternoon, the heart rate increased to 120/min with awakening; at 23:00, it was 134/min. Slight bleeding from the laparotomy wound even with compression bandage. Blood pressure stable, 117-130/71-77 mmHg.

On day 1, the patient was conscious, suture with subcutaneous haematoma in the left pole. HR was 156/min.

BP 118/77. Acute blood count and coagulation tests (WBC 23.7 • Hgb 73 • Hct 21.9 • Plt 207 • INR 1.0 • APTT 57.3s; afterwards 32.3s • thrombin time 89.8s, afterwards 11.8s • fibrinogen 3.9 g/l, afterwards 3.0 • ethanol negative • antithrombin III 79%, afterwards 89 • D-dimer 1000-2000. 3x300 ml of RBC mass administered and 1 unit of frozen human plasma. In the afternoon, she was extubated, breathing spontaneously, heart rate decreased to 115-120/min. Blood count at 18:00: WBC 24.2 • RBC 3.02 • Hgb 89 • Hct 28.2 • Plt 148. Blood gases tests according to Astrup normal. Therapy: cephoxitin 4xl g i.v., heparin 5,000 units s.c. every 12 hours, infusion of glucose 10%, Ringer solution, saline – in total 2,500 ml.

On day 2 from 04:00, there was a gradual increase of heart rate oscillating around 136/min.

Further acute coagulation and blood count tests were performed: WBC 25.8 • Hgb 92 • Hct 28.1 • Plt 139 • INR 1.7 • APTT 109.2 • Thrombin time more than l00 • Fibrinogen 5.2 • Ethanol negative • D-dimers more than 2,000 • Total proteins 45 g/l. Consulted haematologist – recommended continuation of heparin in continual infusion 5,000 units/24 hours, fibrinogen 3g, K vitamin 2 ampules i.v., further dose of RBC mass (600 ml) and 2 units of frozen human plasma. At 11:00 the heart rate increased to 162/min. In control blood count tests there was haemoglobin decrease despite the administered RBC mass to 83, Hct 25.3. Abdominal ultrasonography was performed at the bed: free fluid, probably haemoperitoneum. Surgical revision of the abdominal cavity was scheduled after stabilisation of coagulation parameters. 1,000 units of antithrombin III, 2 ampules, vitamin K, infusion of saline with 60 ml of 7.5% KCl (2.8 mmol/l) due to a critically low potassium level were administered. At 13:00, the surgical revision started. During the revision, deposits of 200-300 ml of liquid blood were found in the subcutaneous and subfascial layer. In the abdominal cavity, there was free blood with peritoneal exudate, about 1,000 ml. The uterus suture was strong, with diffuse bleeding. Blood did not coagulate. Supravaginal amputation of the uterus was performed and left-side adnexectomy was added due to bleeding from the left ovarium that did not stop even after further injections. Coagula started to form at the end of the procedure. The abdominal cavity was drained with a strong silicone drain.

After surgical revision, the condition of the patient was stable. Drain discharge was minimal, sanguinolent fluids, HR 120/min, BP 110/75.

On day 3, controlled ventilation was still performed. Abdomen and suture were calm, coagulation parameter reached normal levels. Blood count: Hgb 103 • Hct 30.2 • total protein level 41. Due to the severity of the condition and assumed necessity of long-term controlled ventilation, the patient was transferred to the intensive medicine department.

At the intensive medicine department, the patient was stable for the first 2 days. Extubation attempts were unsuccessful. From day 3 the condition worsened. Severe septic condition developed with diarrhoea, fevers up to 39°C, developing bronchopneumonia. Clostridium difficile was found in the stool. Patient was suffering from tachycardia up to 186/min and cardiovascular failure was imminent. Vancomycine 4x1 g was administered in microinfusion with digoxin, hydrocortisone, dobutamine and fluconazole infusion therapy. Septic condition retreated after 3 days, circulation became stable, heart rate decreased to 90/min. The patient was still under artificial ventilation, conscious, she was able to communicate.

On 22 August, it was necessary to perform tracheostomy due to repeated unsuccessful extubation attempts. The patient suffered from strong depression. She tried to extract the intubation tube twice in suicidal attempts. Only after confrontation with photos of her child, the psychic condition of the patient stabilised. She cooperated and underwent physiotherapy. On 28 August, artificial pulmonary ventilation was stopped and on 3 September the tracheostomy tube could be extracted. On 9 September, the patient was transferred back to the department of obstetrics and gynaecology in good condition. Here, she underwent intensive breathing physiotherapy and learnt to care for the child. She was dismissed home on 2 October, i.e. after 91 days of hospitalisation.

Currently, the condition of the patient is stable. She is able to continue in her normal life and to care for the child.

Discussion and conclusion

The authors have not found any published case of pregnancy in Leyden-Möbius muscular dystrophy in local literature. Few papers have been published on this disorder in pregnancy in intrnational literature, mainly in the form case reports [5]. Gravidity in advanced muscular dystrophy is rare and associated with high risk.

Due to muscle weakness, diaphragm weakness, atrophy of individual muscle groups, spine deformities and often dislocation of thoracic organs, these patients cannot avoid caesarean section to end pregnancy, followed by prolonged intubation and controlled ventilation. During pregnancy, the growing uterus elevates the diaphragm and impairs breathing. Therefore, it will always be necessary to end the pregnancy prematurely in such patients. Rudnik-Schöneborn et al evaluated 27 patients with different myopathies retrospectively. Operative deliveries were more frequent and most women experienced worsening of weakness in pregnancy and required assistance after delivery [6].

The causes leading to severe complications of this extent that could incur the death of the mother are not known. The caesarean section was not technically difficult; it was performed without complication and in accordance with the specific guidelines and procedures. Literature mentions no causality of the underlying disease and coagulation disorder or higher sensitivity to the administered heparin. The radical procedure (supravaginal uterus amputation) during surgical revision was chosen due to the risk of further surgical intervention if coagulopathy were not manageable – a condition lethal for the patient. Positive psychic motivation of the patient was also significant; it has largely contributed to the turnaround and successful treatment of the critical medical condition.

References

1. Appel SH (1978) The muscular dystrophies. Clinical update on two major types. Postgrad Med 64: 93-102.
2. Brooks MH (1979) Muscular dystrophies and congenital myopathies. Neurol.neourosurg 36: 2-7.
3. Girdany B, Danowski TS (1956) Muscular dystrophy. II. Radiologic findings in relation to severity of disease. AMA J Dis Child 91: 339-345.
4. Golg AP (1988) Diagnosis and management of muscle disease. New York, SP Medical and Scientific Books
5. Ayoubi JM, Meddoun M, Jouk PS, Favier M, Pons JC (2000) Vaginal delivery in a woman with limb-girdle muscular dystrophy. A case report. J Reprod Med 45: 498-500.
6. Rudnik-Schöneborn S, Glauner B, Röhrig D, Zerres K (1997) Obstetric aspects in women with facioscapulohumeral muscular dystrophy, limb-girdle muscular dystrophy, and congenital myopathies. Arch Neurol 54: 888-894.