

Pregnancy and delivery in Leyden-Möbius muscular dystrophy

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Abstract

Leyden-Möbius muscular dystrophy is an autosomal recessive hereditary disease of unknown aetiology; it is a congenital disorder of protein metabolism primarily affecting proximal muscle groups leading to progressive muscular dystrophy. It later spreads to the muscles of the pelvic floor and lower extremities. The estimated incidence is 1:200,000. This paper describe a case of pregnancy and delivery in woman with progressive Leyden-Moebius muscular dystrophy. Cesarean section was performed due to progression of the underlying disease. First postoperative day DIC occure and surgical revision of abdominal cavity was performed. Although the uterine suture was strong, diffuse bleeding was present. Blood was not coagulating. Supravaginal amputation of the uterus was performed including left-sided adnexectomy due to bleeding from the left ovarium. Due to the severity of the condition and assumed necessity of long-term controlled ventilation, the patient was transferred to the intensive medicine department. She was dismissed home after 91 days of hospitalisation. Gravity in advanced muscular dystrophy is rare and associated with a 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 the caesarean section to end their pregnancy, followed by prolonged intubation and controlled ventilation. During pregnancy, the growing uterus elevates the diaphragm and impairs breathing. Therefore, pregnancies in such patients will probably always have to be ended prematurely.

INTRODUCTION

Leyden-Möbius muscular dystrophy is an autosomal recessive hereditary disease of unknown aetiology; it is a congenital disorder of protein metabolism primarily affecting proximal muscle groups leading to progressive muscular dystrophy. It later spreads to the muscles of the pelvic floor and lower extremities. The estimated incidence is 1:200 000. The disease has specific elec-

tromyographic findings, and serum levels of creatine kinase and lactate dehydrogenase are typically elevated. ECG alterations may develop, although they are not typical. Stretch reflexes are not elicitable; the intellect is normal. The disease symptoms develop in the 2nd decennium and are manifested by weakness of arm and gluteal muscles, back muscles, and by a decreased function of the diaphragm. This causes problems with gait and standing up, a decrease of the respiratory volume,

and spine statics disruption. Further progression is characterised by weakness and atrophy of shank muscles, peroneal muscles, by the equinovarus position of lower extremities, Achilles tendon contractures, weakness of mimic muscles, neck muscles, pectoral muscles, and an atrophy of brachial biceps muscles.

Causal therapy is not known. The care is limited to only physiotherapy and orthopaedic management of the contractures.

Prognosis of the disease is uncertain and depends on the disease progression rate (Appel 1978; Brooks 1979; Girdany & Danowski 1956; Golg 1988).

THE CASE

A female patient, 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, the patient was hospitalised due to bleeding, and amniocentesis was performed in the 17th week. The foetus karyotype showed no chromosomal aberrations – 46/XY. From the 27th gestation week, the patient 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 the condition worsens. Due to higher availability of specialised consultations and 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 (July 3)

A female patient of asthenic stature, with clear atrophies of muscle groups in both upper and lower extremities and in the chest, and a spine with deformed severe scoliosis. Significantly limited muscle performance. Fully mobile; dyspnoea signs present even in minimal strain. Height 153 cm, weight 46 kg.

Uterus size corresponding to the 30th gravidity week, slightly irritable. Vaginal examination: cervix length 2 cm, in the sacral position, closed, foetus head over the entrance, arches not expanded. Ultrasonography finding without pathology; foetus biometry corresponding to the 30th gestation week.

Laboratory assessments: Blood gas tests according to Astrup: pH 7.422, pCO₂ 35.6 mmHg, pO₂ 89.6 mmHg, SpO₂ 97%. Blood count: Hgb 10.1 g/dl, Hct 29.4%, RBC 3.28×10¹²/l, WBC 4.9×10⁹/l; creatinine, urea, ALT, AST, minerals and total proteins normal.

Hospitalisation course

In the first days of hospitalisation, uterine contractions subsided after infusion therapy and the patient was generally feeling better. There was no clear progression of the underlying disease, and the pregnancy finding was calm, which allowed releasing the patient temporarily home. On August 23, sore throat developed with further progression. An otorhinolaryngology specialist diagnosed epiglottitis and recommended Ampicillin 4×1 g. The condition temporarily stabilised. On August 08, aphonia developed. The otorhinolaryngologist recommended Mefoxin 4×1 g. The general condition was altered, and the respiratory problems, dyspnoea at rest and general muscle weakness were progressing. 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 August 09, the caesarean section was performed without any technical complications; 5 000 units of Heparin were administered subcutaneously intraoperatively. During skin suture, slight diffuse oozing emerged from the upper subcutaneous layers; wound compression. Further care of the male neonate (2 140 g / 46 cm, good condition – Apgar score 9-10-10) was provided by a paediatrist. Intraoperative blood loss was about 600 ml. The female patient was transferred to the ICU where controlled ventilation was provided due to the underlying disease as scheduled.

On day 0, the patient tolerated the intubation tube. In the afternoon, the heart rate increased to 120/min with awakening, reaching 134/min at 23:00. Slight bleeding from the laparotomy wound occurred 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 57.3 s; afterwards 32.3 s, thrombin time 89.8 s, afterwards 11.8 s, fibrinogen 3.9 g/l, afterwards 3.0, ethanol negative, antithrombin III 79%, afterwards 89, D-dimer 1 000–2 000. 3×300 ml of RBC mass and 1 unit of frozen human plasma were administered. In the afternoon the patient 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 were normal. Therapy: Mefoxin 4×1 g i.v., Heparin 5,000 units s.c. every 12 hours, infusion of glucose 10%, Ringer solution, saline – 2 500 ml in total.

On day 2 from 04:00, the heart rate was gradually increasing again, 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 100,

Fibrinogen 5.2, Ethanol negative, D-dimers more than 2000, total proteins 45 g/l. The consulted haematologist recommended to continue Heparin in a continual infusion 5000 units/24 hours, fibrinogen 3g, K vitamin 2 ampules i.v., and a further dose of RBC mass (600 ml) and 2 units of frozen human plasma. At 11:00 the heart rate increased to 162/min. Control blood count tests revealed a haemoglobin decrease to 83 despite the administered RBC mass, Hct 25.3. Abdominal ultrasonography was performed at the bedside: free fluid, probably haemoperitoneum. Surgical revision of the abdominal cavity was scheduled after stabilisation of coagulation parameters. 1000 units of antithrombin III and 2 ampules of vitamin K were administered, and saline was infused with 60 ml of 7.5% KCl (2.8 mmol/l) due to a critically low potassium level. 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 layers. In the abdominal cavity, there was free blood with peritoneal exudate, about 1000 ml. Although the uterine suture was strong, diffuse bleeding was present. Blood was not coagulating. Supravaginal amputation of the uterus was performed including left-sided adnexectomy due to bleeding from the left ovary that did not stop even after further injections. Coagula started to form towards the end of the procedure. The abdominal cavity was drained with a strong silicone drain.

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

On day 3, controlled ventilation still continued. The abdomen and the suture were calm; coagulation parameter reached their 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. The patient was suffering from tachycardia up to 186/min and cardiovascular failure was imminent. Vancocin 4×1 g was administered in a microinfusion with Digoxin, Hydrocortisone, Dobutamine and Diflucan infusion therapy. The septic condition retreated after 3 days, the circulation stabilised, and the heart rate decreased to 90/min. The patient was still under artificial ventilation, conscious, and able to communicate.

On August 22, tracheostomy had to be approached due to repeated unsuccessful extubation attempts. The patient suffered from strong depression. She tried to extract the intubation tube twice in suicidal attempts. Her psychic condition only stabilised after confrontation with photos of her child. She cooperated and

underwent physiotherapy. On August 28, artificial pulmonary ventilation was stopped and on September 03, the tracheostomy tube could be extracted. On September 09, the patient was transferred back to the department of obstetrics and gynaecology in a good condition. Here, she underwent intensive breathing physiotherapy and was learning to care for the child. She was dismissed home on October 02, i.e. after 91 days of hospitalisation.

Currently, the condition of the patient is stable. She is able to continue 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 national or international literature. Gravidity in advanced muscular dystrophy is rare and associated with a 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 the caesarean section to end their pregnancy, followed by prolonged intubation and controlled ventilation. During pregnancy, the growing uterus elevates the diaphragm and impairs breathing. Therefore, pregnancies in such patients will probably always have to be ended prematurely.

The causes leading to these severe complications that could result in 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 yet another 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.

Author's conflict of interest disclosure

The authors stated that there are no conflicts of interest regarding the publication of this article.

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