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Missed Diagnosis

Dystrophia myotonica and pregnancy – an instructive case

H.T. Chung,[1] A.Y.C. Tam,[2] V. Wong,[2] D.F.H. Li,[3] J.T.C. Ma,[1] C.Y. Huang,[1] Y.L. Yu[1] and E. Woo[1]

Departments of [1]Medicine, [2]Paediatrics and [3]Obstetrics, Queen Mary Hospital, University of Hong Kong, Hong Kong.

Summary: Two cases of dystrophia myotonica, a mother and her newborn child, are reported. The diagnosis in the mother was only made after seven years and two eventful deliveries. The obstetrical and anaesthetic complications, as well as the neonatal form of dystrophia myotonica, are reviewed.

Introduction

Dystrophia myotonica is an uncommon disease with a published prevalence of 4.5 to 5.5 per 100,000.[1] However, this might be an underestimate because mild disease is usually undiagnosed.[1] The age of onset is usually between 20 to 50 years but a neonatal form of the disease is well recognized albeit rare.[2] The disease can present in many different ways and, as illustrated in our case, the diagnosis can be obscured for a long period of time unless a high index of suspicion is maintained.

Case report

A 38 year old housewife presented in January 1979 with her third pregnancy complicated by hydramnios. Her two previous pregnancies had been relatively uneventful, terminating in a normal spontaneous delivery in the first and a lower segment Caesarian section, because of placenta praevia, in the second. This third pregnancy was allowed to proceed to term and she went into spontaneous labour. However, Caesarean section was required because impending rupture of the uterus was suspected. Immediately after intubation and during induction of anaesthesia, she developed rapid atrial fibrillation and shock. With supportive measures a baby boy was delivered. The baby died on day 5 from respiratory distress. Subsequent examination of the mother revealed only paroxysmal atrial fibrillation. The electrocardiogram (ECG) showed a sinus rhythm with a PR interval of 0.20 seconds. The creatine phosphokinase was elevated at 2650 μmol/min.1 (normal 5–165 and the lactate dehydrogenase and aspartate aminotransferase were slightly above normal but there were no serial changes of ECG or cardiac enzyme suggesting acute myocardial infarction. Mild mitral valve prolapse was documented in the echocardiogram. Holter cardiac monitoring showed paroxysmal atrial fibrillation, ventricular ectopics and ventricular tachycardias. She was initially treated with digoxin, verapamil and lignocaine and finally stabilized with oral verapamil.

In 1985, she conceived again and this pregnancy was complicated by diabetes mellitus which required insulin treatment. There was hydramnios and intrauterine growth retardation. A baby girl was delivered at 37 weeks' gestation by Caesarian section in December 1985. She weighed 2 kg and was severely asphyxiated, requiring resuscitation and ventilation. Examination revealed a severely hypotonic infant with a tented mouth, bilateral ptosis, micrognathia and bilateral talipes equinovarus. The ribs were thin and slender on X-ray. Heart failure due to persistent ductus arteriosus developed on day 5. This further compromised her respiration and she was dependent on the ventilator. Motor nerve conduction velocities of the right peroneal and posterior tibial nerves were normal. Electromyography of the deltoid, tibialis anterior, and quadriceps muscles showed typical myopathic changes but no myotonia. The diagnosis of neonatal dystrophia myotonica was made. The mother was re-examined and admitted to dysphagia in the past three months. She had a high forehead, a tented mouth and a myopathic face (Figure 1). There was wasting and weakness of the sternomastoids and myotonia was demonstrated in her left hand and tongue. The baby remained grossly hypotonic until 3

Correspondence: Y.L. Yu, M.D., M.R.C.P.
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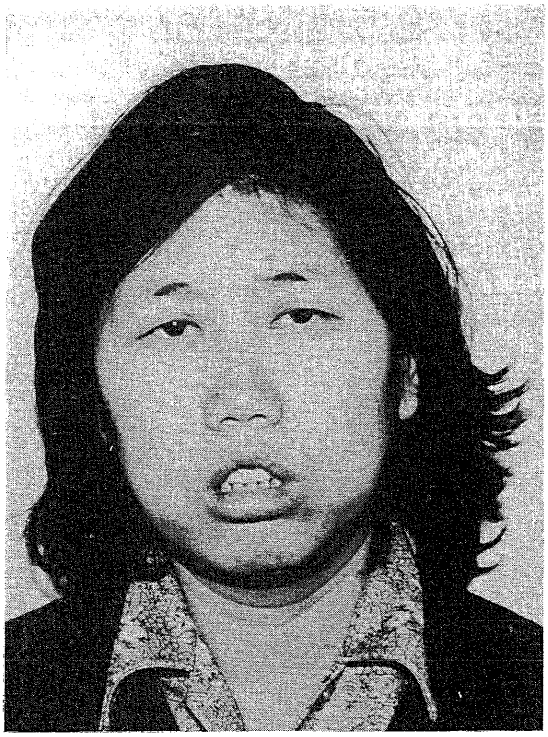

Figure 1 Picture of the mother taken after the birth of her fourth child showing a high forehead, tented mouth and myopathic face.

months later when limb movements were observed. She subsequently developed bronchopulmonary dysplasia and pneumothorax, and succumbed at the age of 7 months.

In retrospect, the third child of the mother probably died from neonatal dystrophia myotonica. A younger sister of the mother had a baby girl who died in the neonatal period but she declined examination. Otherwise, no other members of the family were affected.

Discussion

Neonatal dystrophia myotonica is usually preceded by hydramnios and decreased fetal movement *in utero*. The neonate suffers from severe hypotonia at birth with facial diplegia. However, myotonia does not happen in the neonatal period. These babies have a characteristic tented-shaped mouth. Joint deformities such as talipes equinovarus, arthrogryposis, kyphoscoliosis and lordosis are common and there is increased incidence of respiratory distress, aspiration pneumonia, impaired sucking and swallowing, delayed motor development and mental retardation. Death is commonly caused by respiratory failure, atelectasis, cerebral haemorrhage and prematurity. With supportive care, the affected child will improve after the neonatal period, but will develop symptoms and signs of the adult onset disease at an earlier age and with greater severity.[2,3]

Harper & Dyken[4] found that the mother was the affected parent in the majority of neonatal patients. Ascertainment bias was considered unlikely as both parents were examined. The difference in fertility between affected males and females could only explain part of the discrepancy since their own investigation concluded that the former were slightly less fertile than the latter. The author then proposed, without supporting evidence, two further explanations, namely different alleles responsible for the neonatal and adult form of disease, and an as yet unidentified maternal factor related to the neonatal disease. In a detailed analysis of the pedigrees of the neonatal and adult-onset probands, Glanz & Fraser[5] found that the incidence of neonatal disease was much higher in the offspring of affected mothers of the neonatal probands, when compared with offspring of the affected female relatives of these mothers. These data would argue against the former and favour the latter explanation

The diagnosis of dystrophia myotonica was not made in our patient until her second presentation. In retrospect, there were several clues at the initial presentation, namely hydramnios, the anaesthetic complication, cardiac arrhythmia and the neonatal death of her third child due to respiratory failure. Pregnancy in patients with dystrophia myotonica may exacerbate the disease resulting in increase in myotonia and muscle weakness. There are also various obstetrical complications associated with dystrophia myotonica. In the antepartum period, spontaneous abortion, threatened abortion, hydramnios and premature onset of labour are more common than in the normal population. During labour, prolonged first stage, poor voluntary effort in the second stage and failure of the uterus to contract during the third stage are well recognized complications. The postpartum period may be complicated by excessive haemorrhage, and loss of consciousness. Deaths due to haemorrhage and effects of anaesthetics and analgesics have been reported.[6]

General anaesthesia administered for Caesarean section or other operations can be hazardous in patients with dystrophia myotonica.[7] As illustrated in our patient, various forms of cardiac arrhythmias are more likely to occur during the intra- and post-operative period because anaesthesia and surgical stimulation may aggravate any preexisting conduction block by increasing vagal tone or by causing transient hypoxia.[7] Other manifestations include prolonged PR interval, widened QRS complex, non-specific ST and

T wave changes, various degrees of heart block, transient atrial flutter or fibrillation, mitral valve prolapse, cardiac enlargement and heart failure.[8,9] The major respiratory problems for anaesthesia are impaired vital capacity, aspiration, atelectasis and prolonged mechanical ventilation in the postoperative period. These patients are particularly sensitive to the respiratory depressive action of narcotics, barbiturates and benzodiazepines. Succinylcholine may cause persistent contraction of respiratory and pharyngeal muscles and therefore difficulties in ventilation and intubation, whereas using neostigmine to reverse the action of competitive muscle relaxants such as curare may precipitate myotonia.[7] In a review of 71 cases of dystrophia myotonica, Kaufman[10] found that of 25 cases with complete records of anaesthesia, 15 were uneventful, five had respiratory complications and four died. Only one patient had troublesome myotonia during the operation. Diagnosis of the disease before surgery will certainly reduce the complications by better preparation as well as identification of high risk cases with Holter monitoring and lung function tests.[7]

While specific treatment for dystrophia myotonica is not available, early diagnosis will enable prompt treatment of cardiac arrhythmias, prevention of anaesthetic risks, genetic counselling and prenatal diagnosis.[11] Our patient enjoyed a healthy life for seven years after the initial presentation. Her disease manifested again during her last pregnancy with diabetes mellitus but the diagnosis was made only after the birth of a floppy baby. It has been suggested that we should shake hands with mothers with floppy babies,[12] but would it also be a good idea to look for myotonia in patients with hydramnios, anaesthetic accident, unexplained cardiac arrhythmias and diabetes mellitus.

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