Obstetrical Care in Myotonic Dystrophy Type 1 (Steinert Disease)

Original title: Schwangerschaftsbetreuung bei Myotoner Muskeldystrophie Curschmann Steinert

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Background: The Myotonic Dystrophy Curschmann Steinert (MDCS), also known as Steinert’s disease is the commonest type of myotonic muscular diseases. It is characterized by an inhibition of relaxation after rapid, intentional contractions. The incidence in Germany is about 5 in 100 000 citizens. It is inherited as autosomal-dominant trait which shows an unstable amplification of a CTG-trinucleotide on chromosome 19. The severity depends on the age of onset and is closely related to its amplification. 5–35 CTG triplets are normal whereas > 45 CTG repeats are pathological. There are three forms of Steinert’s disease:

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   - adult / classic form: it counts 200–1000 CTG repeats setting in mainly between the age of 15–30. The symptoms are general muscular pain and weakness beginning with distal muscle groups. The typical signs are facial myopathy, atrophy of distal muscle groups (especially hypothenar), myotonia after repetitive intentional contractions. General manifestations affect various systems including cardiovascular (arrhythmia / cardiac arrest), endocrine (diabetes), respiratory (recurring lower respiratory infections due to hypoventilation), gastrointestinal (constipation due to impaired peristalsis), vision (cataract, ptosis).
   - mild / senile form: 45–200 CTG repeats beginning in old age. All signs and symptoms are reduced.
   - congenital / neonatal form: This is the most severe form counting > 1000 CTG triplets. Affected neonates present as “floppy infants” with facial paralysis, respiratory insufficiency and difficulty in sucking and swallowing. The mortality is 50%. A retrospective case report of a neonate with MDCS in 2000 describes polyhydramnios, reduced fetal movement followed by intrapartum asphyxia and 37 days on a ventilator due to respiratory insufficiency. The authors pointed out the essence of a careful case and family history to recognise rare diseases. Steinert’s disease is the Myotonic Dystrophy type 1. Type 2 is known as PROMM (Proximale Myotonic Myopathy). Differential diagnosis include myasthenia gravis, Prader Willi or Wernig Hoffmann Syndrome.

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Case Report: The striking general appearance of a 23 year old primigravida in the 25th week of pregnancy prompted a detailed history which led to a strong suspicion of Steinert’s disease. An obvious diagnosis to those familiar with the disease. Prior to this the patient had followed all recommended antenatal care examinations, but the MDCS had remained undetected. She had experienced muscle pain throughout her teens. This had abated when she became pregnant. Unclenching of her fist was delayed to > 2 seconds – typical for Steinert’s disease. The father and grandfather had suffered a progressive muscular illness. The grandfather had died of cardiac arrest. The patient was referred to the perinatal centre for observation and diagnostics, especially to evaluate obstetrics related risks. Apart from a mild pulmonary restriction there were no additional clinical findings of note. Genetic screening indicated MDCS with a CTG repeat of 700. In the 39th week of pregnancy a baby boy was delivered by c-section without intrapartal complications. When discharged home the mother was breastfeeding successfully. The boy had a normal genotype.

Discussion: The MDCS protein kinase phosphorylates various proteins. A mutation leads to dysfunctional sodium channels. As a consequence repetitive membrane depolarisations and contractions of myotonic muscular fibres occur. Any protein kinase dysfunction leads to a general clinical picture beyond the muscular complaints. Confirmation by genetic testing: The first step analyses the length of fragments on chromosome 19q13.2 to 19q13.3 separating normal from (pre)pathological genomes. When above the cut off range of 35 trinucleotide repeats the DNA is analysed by southern blot hybridisation for quantification. Due to anticipation especially when passed on by the mother the chances for presentation of a neonatal form are 90% compared to 29% for any other form. MDCS pregnant women have a higher risk for pre-eclampsia, pre-term contractions or labour. The fetus might present with intrauterine death. Perinatal risks are prolonged labour, asphyxia or adjustment disorders. It is recommended to avoid general anaesthesia because muscle relaxant drugs might cause severe laryngeal spasm with impossibility for intubation. There is an association with malignant hyperthermia.

Conclusion: Always be aware of rare cases. Diseases with genetic disorder require multidisciplinary team work. The possibility of diagnostics prior to implantation is very likely to play a central role in future.