Becker muscular dystrophy: an unusual presentation

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Abstract

A 15 year old boy who presented with passing painless dark urine was found to have myoglobinuria. His creatine phosphokinase was raised, and a muscle biopsy specimen showed non-specific dystrophic changes. Subsequent DNA analysis led to the diagnosis of Becker muscular dystrophy. Myoglobinuria may be a presenting symptom of Becker muscular dystrophy.

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Becker muscular dystrophy is an X linked condition characterised by progressive muscular weakness with a varied spectrum of severity.¹ Diagnosis is based upon clinical features, creatine phosphokinase levels, findings on muscle biopsy, and DNA analysis. Creatine phosphokinase and myoglobin are raised in muscular disorders,² but frank myoglobinuria is a rare presentation. We report a case where myoglobinuria was the initial presentation leading to the diagnosis of Becker muscular dystrophy.

Case report

A 15 year old boy presented with a history of the painless passing of dark urine once or twice a year for three years. These episodes lasted approximately 24 hours and were not preceded by strenuous exercise, illness, drug or alcohol abuse. There were no other problems on systematic inquiry.

He was born after a full term normal delivery with no history of birth complications. Development milestones were age appropriate. At the age of 5 years he had surgical elongation of bilateral Achilles tendons for a toe walking gait. He had no further medical or physical problems until the first episode of passing dark urine. He played rugby and football regularly and only occasionally complained of calf pains. His maternal uncle was suspected of having Becker muscular dystrophy based upon clinical history, though this had never been confirmed by any investigations.

On examination he was of average height and weight and physically well developed with a slight prominence of the calf muscles. He had depressed knee and ankle reflexes but the power was not diminished. General examination was otherwise unremarkable.

Biochemical investigations conducted during the first outpatient appointment showed a

raised aspartate aminotransferase level of 606 IU/l (normal 4-30) and a lactate dehydrogenase level of 2886 IU/l (normal 225-550). Full blood count and urea and electrolyte concentrations were normal. Lactate dehydrogenase isoenzymes showed muscle origin and creatine phosphokinase was initially 1280 IU/l (reference <195 IU/l) and when repeated two months later was 11 650 IU/l. After 24 hours' bed rest, creatine phosphokinase was 1915 IU/l before and increased to 5870 IU/l after exercise. Neither discoloration nor myoglobin were present in the urine.

A few months later, after an episode of passing dark coloured urine, this was found to be positive for myoglobin. Creatine phosphokinase was raised to 100 000 IU/l, aspartate aminotransferase was 1963 IU/l, and lactate dehydrogenase 7715 IU/l. A muscle biopsy specimen taken on two occasions demonstrated non-specific dystrophic changes. DNA analysis confirmed the diagnosis of Becker muscular dystrophy.

Discussion

In Becker muscular dystrophy symmetrical muscles of the pelvic girdle and thighs are prominently involved, calf muscle hypertrophy being an almost universal feature. The first symptoms may be noted between 1 and 45 years of age but in the majority they are seen between 5 and 15 years.³ In our case the patient had normal developmental milestones and was able to carry out strenuous exercise. Although he had bilateral Achilles tendon repair at the age of 5 years, early development of contractures is not a consistent feature in Becker muscular dystrophy.¹ Apart from mild calf muscle hypertrophy he had no other features consistent with Becker muscular dystrophy.

In Becker muscular dystrophy, creatine phosphokinase levels may be increased early in the disease; thereafter values fall with age. Values may be raised 20 to 200 times the normal limit.³ In our case the creatine phosphokinase was shown to be raised at random and further so after exercise.

The myoglobin content in blood and urine is increased in a number of muscle disorders and is a fairly sensitive indicator of muscle necrosis.⁴ Only on one occasion when the creatine phosphokinase was raised to 500 times the normal range was myoglobinuria confirmed. In this case the myoglobinuria

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associated with marked increase in lactate dehydrogenase isoenzymes was a result of rhabdomyolysis. There was no deterioration in renal function of the patient at any time.

To the best of our knowledge, presentation of Becker muscular dystrophy with myoglobinuria has not been documented in the past.

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- 3 Gardner-Medwin D. Clinical features and classification of the muscular dystrophies. *Br Med Bull* 1980; **36**: 109–15.
- 4 Turnbull DM, Bindoff LA. Biochemical investigation muscle disease. *Ann Clin Biochem* 1989; **26**: 472–6.