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Case Report

# Suspicion of Duchenne Muscular Dystrophy in A 7-Year-Old Madurese Boy: A Rare Case Report

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## ABSTRACT

**Introduction:** Duchenne Muscular Dystrophy (DMD) is an X-linked disorder presenting in early childhood and characterized by proximal muscle weakness and calf hypertrophy in affected boys associated with a functional deficiency of dystrophin. The incidence of these cases reached 1 in 3500-6000 male births. Research at RSUPN Dr Cipto Mangunkusumo (RSCM) showed that DMD is the second most frequent neuromuscular disorder in children regardless there is no prevalence data available yet in Indonesia. It is quite complicated to diagnose DMD due to the limited facilities of genetic testing and its high cost.

**Case Presentation:** A 7-year-old boy came with the main complaint of weakness in both legs progressively two years before admission. He had to hold onto the floor and then both knees when started to stand. There was a history of motor and speech delay but family history was denied. Vital signs and nutritional status were within normal. Laboratory examination revealed significantly elevated levels of CK, ALT, and AST. The EMG result supported the presence of myopathy. Due to a lack of facilities, genetic testing or muscle biopsy could not be done. He received prednisone once a day with a 14 mg dose and was scheduled to attend regular physiotherapy.

**Conclusion:** In a limited facilities area, the clinician should have a suspicion of DMD not only by anamnesis and clinical features, but also by considering the elevated levels of CK and myopathies on EMG.

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## INTRODUCTION

Duchenne Muscular Dystrophy (DMD) is the most common muscular dystrophy in early childhood characterized by proximal muscle weakness and calf hypertrophy in affected boys and is associated with a functional deficiency of dystrophin [1]. The incidence of these cases reached 1 in 3500-6000 male births [2]. But until now, there is no data about pediatric cases in Indonesia while RSUPN Dr Cipto Mangunkusumo's (RSCM) research showed that DMD is the most second frequent neuromuscular disorder in children [3]. It is quite complicated to diagnose DMD due to the limited facilities of genetic testing and its high-cost [4].

The accurate and early diagnosis of DMD plays an important role in effective patient management. Management of DMD requires multidisciplinary approaches and treatment with glucocorticoids has been the only effective drug [1]. In areas with limited facilities, the clinician should have a suspicion of DMD not only by anamnesis and clinical features, but also by considering the elevated levels of creatine kinase and myopathies on EMG.

This case report describes suspicion of Duchenne Muscular Dystrophy in a 7-year-old Madurese boy. Establishing a diagnosis requires genetic testing which is not available. This case report aimed to provide an understanding of suspicion of DMD by laboratory and electromyography examination.

## CASE PRESENTATION

A 7-year-old Madurese boy was admitted to Paediatric Polyclinic RSUD Mohammad Noer, Pamekasan with the main complaint of weakness in both legs. This complaint appeared progressively two years before admission. The parents reported repeated falls,

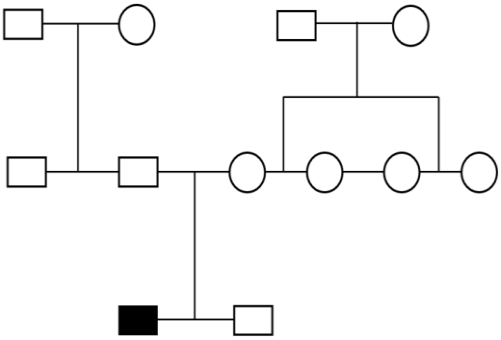


**Fig.1**. Patient's Pedigree

difficulty standing up for a while, climbing stairs, and doing other sports activities. He had to hold onto the floor and then both knees when started to stand. The parents also reported a history of motor delay; head up at 4 months, rolling up at 6 months, sitting at 8 months, standing at 16 months, and walking at 24 months. There was no family history of having the same experience. He was the first child out of 2 children. His younger brother did not have any complaints like him (Fig.1).

Physical examination on admission disclosed an alert boy with blood pressure 100/70 mmHg, pulse 90x/min, respiratory of rate 20x/min, temp 36.5 C, SpO2 98%, weight 18.5 kg, and height 111 cm. There were no skin abnormalities, pale conjunctiva, cyanosis, jaundice, or dyspnea. The heart and lungs were normal, and the liver and spleen were not palpable. The extremities were warm and had no edema. There was pseudohypertrophy on both calves with firm rubber palpation (Fig.2). Physiological reflexes BPR/TPR +2/+2, KPR +1/+1. Pathological reflexes: Babinski -/-, Chaddock -/-, Clonus -/-. The motor strength of the superior extremity was 5555/5555 while the inferior extremity was 4433/3344.



**Fig.2.** Swelling Calves Inpatient

Sensory was normal. The Gower sign was positive (Fig.3).

Laboratory examination revealed significantly elevated levels of creatine-kinase 5089 U/l (normal values: < 190 U/l), alanine aminotransferase 209 U/l (normal values: < 31 U/l), and aspartate aminotransferase 172 U/l (normal values: < 35 U/l). Chest X-Ray was normal. Electromyography revealed muscle abnormality without any nerve involvement supporting the presence of myopathy. Due to a lack of facilities, genetic testing or muscle biopsy could not be done.

The patient was diagnosed with suspicion of DMD. He received steroid therapy prednisone once a day with a 14 mg dose and was scheduled to attend regular physiotherapy. In the first month, there was a significant improvement. He was referred to the Department of Child Neurology at RSUD Dr Soetomo Surabaya for further diagnostic tests and management.

## DISCUSSION

The suspicion of Duchenne Muscular Dystrophy is based on anamnesis, clinical features, laboratory findings, and electromyography results. Male sex, family history, progressive proximal muscle weakness, calf hypertrophy, delayed walking, difficulty climbing or descending stairs, difficulty rising from the floor, difficulty running or walking, frequent falls, and Gower Sign were typical characteristics of DMD. Deficits of cognitive, autism spectrum disorder, speech delay, and gross motor delay also can be found though not always present. Laboratory findings on DMD were elevated CK levels, ALT, and AST. EMG may reveal myopathy (Fig.4) [5].

In our case, the clinical manifestation was a progressive proximal muscle weakness that started when he was 5 years old, frequently repeated falls, difficulty standing up for a while, or climbing the stair and doing other sports activities. The parents also reported gross motor delay and speech delay. Affected boys experience delays in being able to walk compared to normal children where 50% of them can only walk after the age of 18 months. He also had calves swelling and Gower Sign. Gower Sign is a condition when children use their arms to lift themselves from a seated position on the ground [Fig.5]. In our case, the patient used his arms to push himself erect by walking his hands up his thighs. It is also common for DMD to have enlarged calves because fat and connective tissue replace the build-up of scar tissue in the muscle [6–8].

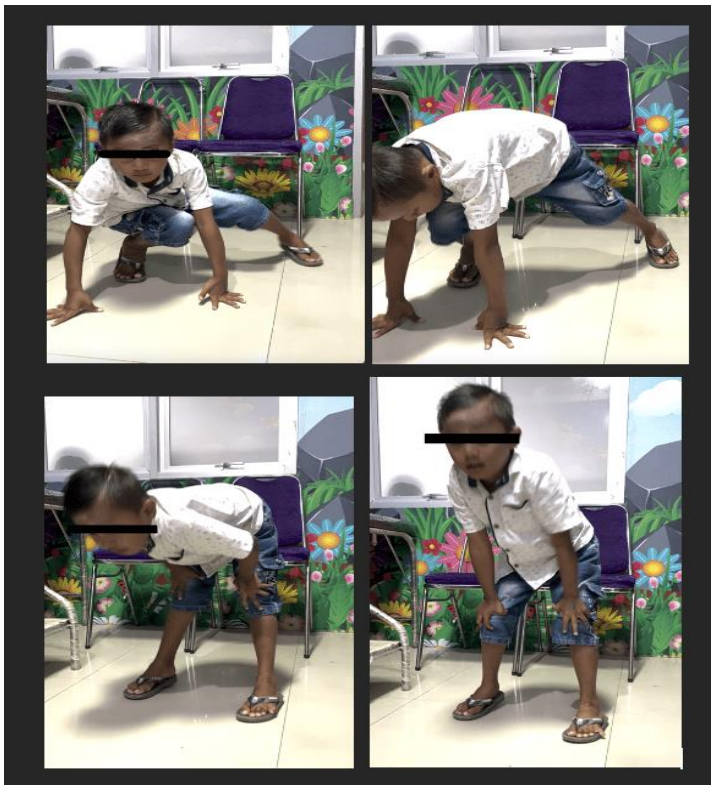


**Fig.3.** Gowers Sign Inpatient

There was no family history of DMD in this patient. It can occur in 30% of patients with DMD because of a spontaneous mutation (*de novo*) when the mother is a carrier without a family history before. The younger brother of this patient should be observed for the chance of having DMD later [6].

The incidence of these cases reached 1 in 3500-6000 male births each year [1,2]. Although there is no data about pediatric cases in Indonesia, retrospective research in Rumah Sakit Dr Cipto Mangunkusumo showed that DMD is the second most frequent neuromuscular disorder in children [3]. The manifestation of DMD is a progressive loss of muscle strength presenting with delayed motor milestones with or without intellectual disability. Mutations in the DMD gene encoding dystrophin, which localizes to the cytoplasmic face of the sarcolemma of the skeletal muscle, forming one component of a large dystrophin-associated glycoprotein complex is the cause of DMD. Approximately 0.6% of the DMD genes are exons, while the rest are large introns. The large size of the DMD gene makes it susceptible to mutations, with one-third of all mutations arising *de novo.* The precise mechanism of dystrophin deficiency-induced muscle fibre degeneration remains unclear. The absence of dystrophin at the plasma membrane leads to the delocalization of dystrophin-associated proteins from the membrane, disruption of the cytoskeleton with the resultant membrane instability, increased susceptibility to mechanical stress, and irreversible degeneration of muscle tissue. Unlike DMD, Becker Muscular Dystrophy (BMD) is a milder dystrophinopathy phenotype resulting from partial dystrophin mutations. Females are usually asymptomatic but some female carriers show a milder form of disease (Fig.6). As progressivity, DMD may cause respiratory, cardiac, and orthopedic complications [1,2,9].

The laboratory findings on DMD revealed elevated levels of creatine-kinase CK), alanine aminotransferase (ALT), aspartate aminotransferase (AST), aldolase, and lactate dehydrogenase (LDH). Creatine kinase is an enzyme that is abundant in muscle and leaks into the bloodstream upon muscle damage. This enzyme is a sensitive indicator of skeletal and cardiac muscle injury and reliable means of determining if high transaminase levels are associated with underlying muscle disease. Elevated levels of CK on DMD at least 10-20 times (often 50-200 times) should raise the suspicion of DMD. Patients with DMD have peak levels of CK at two or three years old, and then levels decline with increasing age due to the progressive loss of dystrophic muscle fibres replaced by fatty cells and fibrous tissue. Our patient's laboratory findings were suitable with DMD [4,9].

Based on the diagnostic steps of DMD in Fig.4, this patient is suspected of dystrophinopathy and should do a genetic test or muscle biopsy for diagnostic confirmation. Due to limited facilities and its high cost, those tests could

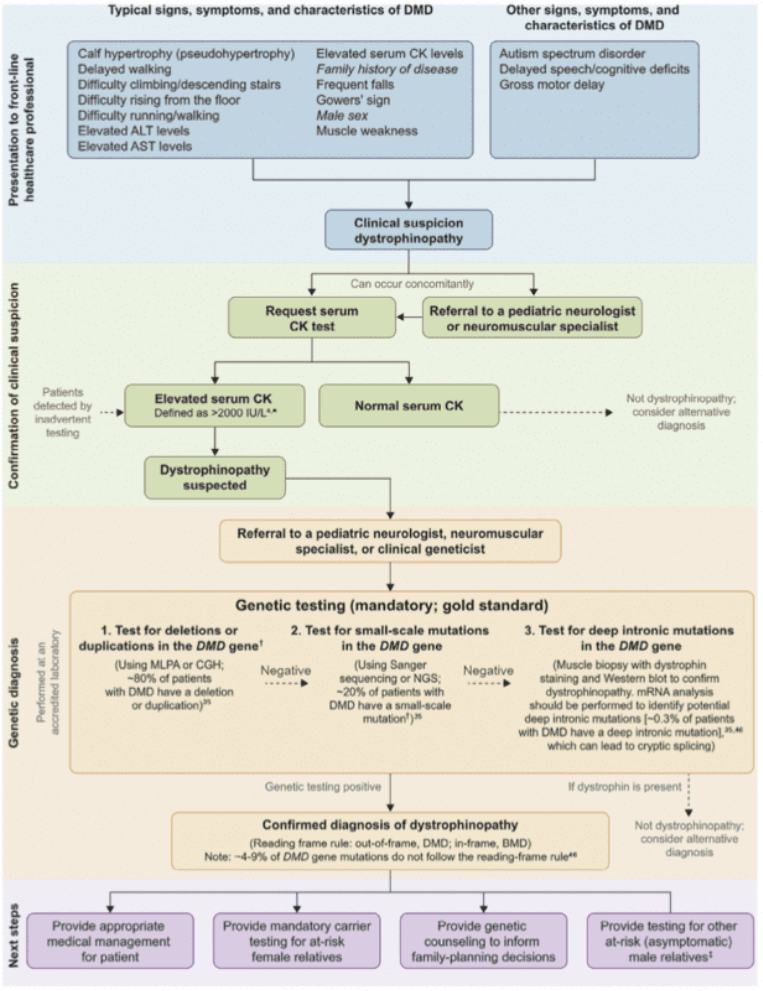



**Fig.4.** Diagnostic Steps for Reaching Complete Diagnosis of DMD

not be done. Meanwhile, electromyography (EMG) and nerve conduction studies are rarely required in the diagnosis of DMD. These investigations can help clinicians distinguish between a condition that begins in the muscle and nerve disorders that mimic muscular dystrophy and make sure if there is any nerve damage. EMG findings on DMD are myopathic with short duration, low amplitude polyphasic motor unit potentials, particularly in proximal muscle. EMG and nerve conduction studies on this patient showed muscle abnormalities without any nerve involvement that support the presence of myopathy [9].

It is essential to rule out dystrophinopathies when considering DMD diagnosis. Common differential diagnoses for DMD are Becker Muscular Dystrophy (BMD), Limb-Girdle Muscular Dystrophy, Emery-Dreifuss Disease, and others. Besides it is milder than DMD, Becker Muscular Dystrophy has slower progression than DMD. The levels of CK are not as high as those of DMD. Limb-Girdle Muscular Dystrophy as a clinical manifestation during the second decade with normal cognitive function generally, rarely a calf pseudohypertrophy, and levels of transaminase are usually not elevated. In DMD, CK levels are elevated >2000 U/l because of active muscle fibre necrosis and injury so it is a very good screening test for DMD [8,10].

Patients in DMD required a multidisciplinary approach for optimum management. The complications of DMD can lead to fatal morbidity and mortality. Giving additional steroids to boys with DMD who are still walking can improve muscle strength and prolong walking ability for 2-5 years, thus improving the quality of life. The steroid also can reduce the incidence of scoliosis, upper extremity, and cardiorespiratory dysfunction. International guidelines for DMD treatment recommend Prednisone and/or Prednisolone (0.75mg/kg/day) and Deflazacort (0.9 mg/kg/day). Since DMD patients have lifelong steroid usage generally, an optimal follow-up is necessary for monitoring its side effects (behavioral disturbance, weight



**Fig.5**. Gowers Sign

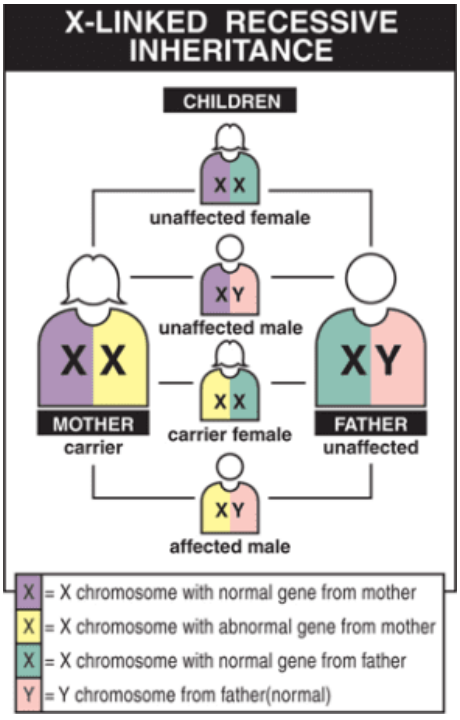



**Fig.6**. X-linked Recessive Inheritance on DMD

gain, gastric irritation, growth-bone health, delayed puberty, and adrenal suppression). Several parameters should be monitored routinely for those who got steroid medication including blood pressure, heart rate, oxygen saturation levels, height, weight, Cushingoid features, and ophthalmology evaluation. The patient of our case received steroid therapy equivalent to prednisone 0.75mg/kg/day, a monthly follow-up plan for physiotherapy and a regular assessment of progressive muscle and cardiac/respiratory damage [1,11–13].

Physiotherapy on DMD is important for the prevention of contractures and the maintenance of muscle function. Almost 90% of sufferers tend to develop scoliosis later. Monitoring for this should be initiated before the loss of the ability to walk including prophylaxis with physiotherapy and appropriate seating to prevent pelvic asymmetry and provide postural support. The patient in our case did not show scoliosis but we suggest he undergo routine physiotherapy monthly [6].

DMD patients will get live dependant on a wheelchair. Death results from respiratory failure, pulmonary infection, or cardiomyopathy can be major causes of morbidity and mortality. Generally, patients survive up to the second decade, but life expectancy is increasing with advances in cardiac and respiratory care.

We were unable to determine the prognosis of our patient because it is still to be observed. Periodic reevaluation is mandatory in this situation [6,14].

## CONCLUSION

A 7-year-old Madurese boy was diagnosed with suspicion of DMD. Clinical manifestations are a progressive weakness in both legs, calves pseudohypertrophy, and Gowers Sign. Elevated levels of CK and myopathies on EMG were found. In this case, the patient received daily steroids, regular physiotherapy, and education about the disease also referral planning to a tertiary hospital. Evaluation at one month revealed significant improvement.

## ACKNOWLEDGMENT

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## CONFLICT OF INTEREST

The authors declare there is no conflict of interest.

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