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Characteristic of Duchenne Muscular Dystrophy

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Abstract

Background: Duchenne Muscular Dystrophy (DMD) is the most common X-linked disorder muscular dystrophy in children. It has incidence 1:3500 infants. It occurs most commonly in boys between 3-5 years old and caused by mutation in the gene for the protein dystrophin which important to maintain the muscle cell membrane. Its characterized by symmetrical proximal muscle weakness and calf hypertrophy. This can result in trouble of standing up and walking. One of the clinical presentation of DMD is positive Gower's sign. There's usually delay in motor development and followed by death from cardiac or respiratory complications. The diagnostic test for DMD are Creatine Kinase, genetic testing, and muscle biopsy. Although we still aim to provide cure for this disorder, treatment such as corticosteroid therapy have provided improvements in function, quality of life, and life expectancy. Aim is to identify clinical features of signs, symptoms and characteristic of DMD.

Method: Observational descriptive study of patient's medical records. A 10 year old boy patient presented with weakness of both lower limb. His parents gave medical history of repeated falls, and has family history that one of his maternal uncle died of the same illness at young age. The general physical examination, the child presented with difficulty in standing, walking, getting up from sitting position, proximal weakness, mild hamstring muscle and achilles tendon contracture, calf hypertrophy and positive Gower's sign. With the thoracolumbal x-ray showed that he had mild scoliosis. For the treatment, patient got Prednisolone daily and physical rehabilitation and showed improvement of clinical sign.

Conclusion: DMD is inherited musculoskeletal disorder that affects boys with age range between 3-5 year. The simple clinical diagnostic of DMD is Gower's sign and calf hypertrophy. The average life expectancy of DMD is 26 yo, some cases may live into their 30s.

Keyword: DMD, X-linked disorder, calf hypertrophy, Gower's sign.

Biography

Sri Hastuti was a lecturer and neurologist in Aceh, Indonesia. She has completed her MD from Universitas Syiah Kuala, Aceh in 2002 and Neurology Specialist from Universitas Indonesia in 2010. In June 2017 to June 2018, I become a Neurology Pediatric fellow at University Malaya Medical Centre, Kuala Lumpur. After dedicating herself in remote area for 3 years and observing many neuropediatric field, she had motivation to continue research in the subject, particularly in neuropediatric cases. In the middle of September 2019, she has got consultant title for Neurology Pediatric at The National Congress of Indonesian Neurological Association in Surabaya, Indonesia.

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