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Congenital Myotonic Muscular Dystrophy: The Diagnostic Role Of Nerve Conduction Study And Needle Electromyography

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Abstract

Background: Congenital myotonic muscular dystrophy (Congenital dystrophia myotonica) is a very rare genetic disorder that was probably first recognized by Vanier in 1960. Vanier emphasized that the manifestations of the congenital form differ from the adult form. The condition is characterized by marked hypotonia and mental retardation. The disorder has not been reported in Iraq. The aim of this paper is to describe the possible diagnostic challenges and difficulties.

Keywords: Congenital myotonic muscular dystrophy; Brain CT abnormalities; Diagnostic electromyography

Patients and Methods

A girl who was first referred at the age of four years because of marked hypotonia, growth and mental retardation with delayed speech. The case was studied and the diagnostic work-up is described.

Results

Brain CT-scan performed at about the age of two years showed mild atrophic changes in the form of ventricular dilatation and evidence of subarachnoid hemorrhage in the left parietal and frontal lobes. Diagnostic changes on electromyography didn't appear until the age of four years and allowed a confident diagnosis.

Conclusion

This case illustrated the possible delay in diagnosis and diagnostic difficulties associated with later appearance of diagnostic changes on electromyography.

Introduction

Congenital myotonic muscular dystrophy (Congenital dystrophia myotonica) is a very rare genetic disorder that was probably first recognized by Vanier in 1960. Vanier emphasized that the manifestations of the congenital form differ from the adult form. The condition is characterized by marked hypotonia and mental retardation [1-11]. The disorder has not been reported in Iraq. The aim of this paper is to describe the possible diagnostic challenges and difficulties.

Patients and Methods

A girl who was first referred at the age of four years because of marked hypotonia, growth and mental retardation with delayed speech. The case was studied and the diagnostic work-up is described.

Results

At the age of four years, the girl was markedly hypotonic and was showing no spontaneous movements. She was unable to sit without support *[Figure-1]* and she was not saying any word, she was not reacting normally to the environment and the parents were not sure she recognizing them, but they were sure that she did not understand anything said to her. She had feeding difficulties since early infancy and her weight was less than 9 kilograms.

Figure 1: At the age of four years, the girl was markedly hypotonic and was showing no spontaneous movements. She was unable to sit without support

Brain CT-scan performed during October, 2016 at about the age of two years showed mild atrophic changes in the form of ventricular dilatation and evidence of subarachnoid

hemorrhage in the left parietal and frontal lobes.Needle electromyography (EMG) study was performed at about the age of one year on right deltoid, biceps, and the right thoracic paraspinal muscles in the upper limbs, and vastus lateralalis, and left tibialis anterior in the lower limbs. The study showed evidence of active myopathic changes with spontaneous activity involving the limbs and paraspinal muscles. Nerve conduction study and needle electromyography study were performed on the 25th of December 2017, at about the age of three years. Nerve conduction study was performed by surface and needle electrode on right median nerve, right ulnar nerve, and right and left common peroneal nerves. The nerve conduction study *(Table-1)* showed reduced amplitude of the compound action potentials with normal sensory parameters, distal motor latencies, motor conduction velocities, and F-wave latencies. Needle electromyography study was performed on right deltoid, biceps, brachioradialis, vastus medialis, right and left tibialis anterior, EDB muscles, and gastocenemius. Needle electromyography study *(Table-1)* showed markedly increased resistance to needle insertion, and spontaneous activity grade 1-2 in the form of fibrillation, positive sharp waves, and CRD with no myotonic discharges,

The average duration of 20 motor units:
Right deltoid= 4.5 msec (n=8.6 msec).
Right biceps = 5.1 msec (n=8.4 msec).
Right vastus medialis = 4.1 msec (n=8.6 msec).
Right and left tibialis anterior = 6.1, 6, 1 msec (n= 10.1 msec).
Polyphasia of short duration and low amplitude was observed in 50-60%.
Early full recruitment pattern of low amplitude.

These electro-diagnostic nerve and muscle studies performed on the 25th of December, 2017 suggested moderate to severe chronic diffuse dystrophic myopathic changes mostly caused by congenital dystrophy. The proximal lower limbs muscles were more significantly involved. Normal motor and sensory nerve studies supported the clinical decision of excluding polyneuropathy and anterior horn cell disease from the differential diagnosis.

Nerve	Sensory			Motor			
	Latency msec/cm	Amplitude µV	SNCV m/sec	Muscle	DML msec/cm	MNCV m/sec	F-wave Latency m/sec
Right Median	2.5	22	56.6	APB	3.6	56.5	20.3
Right Ulnar	2.3	18	57	ADM	3.1	56.9	21.2
Left common peroneal				Tibialis anterior	3.7	44.9	
				EDB	4.6		42.2
Right common peroneal				Tibialis anterior	3.6		
				EDB	4.5	44	41.7
L sural	2.2	15.3	44.3				

Table 1: Findings of nerve conduction study and needle electromyography (EMG) study were performed on the 25th of December 2017

At the age of four years, nerve conduction study and needle electromyography (EMG) study were performed again. Nerve conduction study showed bilateral low amplitude common peroneal and posterior tibial motor responses with normal sural sensory response, and normal median and ulnar sensory and motor nerve conduction studies. Electromyography detected myotonic changes. It also showed spontaneous activity in the form of fibrillation and positive sharp waves. Increased percentage of polyphasic short duration, low amplitude motor units. The findings were consistent with moderate to severe myopathic changes affecting all limbs mostly caused by congenital myotonic muscular dystrophy.

Discussion

Myotonic dystrophy or dystrophia myotonica was originally regarded a disease of adults and adolescents commonly presented with characteristic diagnostic features including myotonia and progressive muscle degeneration affecting facial, jaw, neck, and distal limb muscles. Congenital myotonic muscular dystrophy (Congenital dystrophia myotonica) is a very rare genetic disorder that was probably first recognized by Vanier in 1960. Vanier emphasized that the manifestations of the congenital form differ from the adult form. The condition is characterized by marked hypotonia and mental retardation *[1-11]*. During the 1960s and 1970s, sever authors including (Parker et al., [2-6,11] emphasized that hypotonia, rather than myotonia is the principal feature of congenital myotonic muscular dystrophy *[1-11]*. The frequent occurrence of mental retardation has also been recognized *(Parker, et al., [2,4,5])*. Although, congenital progressive muscular dystrophy of the Fukuyama type which is observed mostly in Japan is also associated with marked hypotonia and mental retardation *[12,13]*, the non progressive nature of the disorder of this Iraqi patient, helped in excluding this condition. In addition to CT-scan evidence of mild brain atrophy, this patient had evidence of intra-cranial hemorrhage which is a well-recognized cause of early death in this condition *[14]*. In this case, diagnostic delay occurred before referral can be attributed to the late appearance of diagnostic changes on electromyography.

Conclusion

This case illustrated the possible delay in diagnosis and diagnostic difficulties associated with later appearance of diagnostic changes on electromyography.

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