Neurosciences Quiz

Submitted by: Hussein A. Algahtani, *MD FRCP(C),* ***Wail N. Yar,*** *MBBS.*
From the Department of Clinical Affairs (Algahtani), King Saud Bin Abdulaziz University for Health Sciences, National Guard Health Affairs, and the Department of Medicine (Yar), College of Medicine, King Abdulaziz University, Jeddah, Kingdom of Saudi Arabia.
Address correspondence to: Dr. Hussein Algahtani, Department of Clinical Affairs, King Saud Bin Abdulaziz University for Health Sciences, National Guard Health Affairs, PO Box 8202, Jeddah 21482, Kingdom of Saudi Arabia. Tel/Fax. +966 (2) 6240000 Ext. 26232. E-mail: halgahtani@hotmail.com

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E-mail: sdouglas@rmh.med.sa

A middle aged man with respiratory failure and weakness

Clinical Presentation

A 55-year-old man presented with 5 days history of shortness of breath, cough, and fever. When he arrived at the emergency department, he was hypoxic with respiratory distress and oxygen saturation of 85%. He was resuscitated and eventually intubated and mechanically ventilated. On questioning, his family reported a long-standing history of difficulty climbing stairs, rising from a chair, lifting his children, brushing his teeth, or combing his hair. They had also noticed change in his voice and occasional chocking attacks when he drinks water. He was not diabetic or hypertensive, but 10 years earlier he developed cataracts. His family history was positive for a similar weakness in his father and 2 of his children. Neurological examination revealed normal mentation. Examination of facial muscles are shown in **Figure 1.** A specific test was carried out (**Figure 2**).

Figure 1 - Examination of the facial muscles (patient in the intensive care unit).

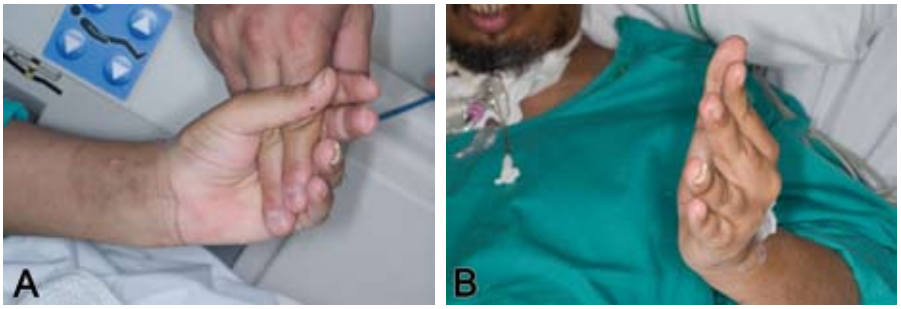

Figure 2 - Patient **A)** gripping examiner's fingers and **B)** immediately after releasing the fingers.

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Questions:

1. What is the diagnosis?

2. What is the sign shown in **Figure 2?** How can you confirm the diagnosis?

3. What is "genetic anticipation"?

4. What are the systemic manifestations of this disease?

Answers & Discussion

1. **Figure 1** shows classical myotonic dystrophy with facial weakness and nasogastric and tracheostomy tubes. When sleeping, the mouth was kept wide open (**A**). The patient had difficulty showing his teeth (**B**), and closing his eyes (**C**). The history was that of acute respiratory failure with long-standing progressive, non-fluctuating weakness. The respiratory failure was due to respiratory and bulbar muscles weakness (precipitated by pneumonia). The pattern of inheritance is autosomal dominant.
2. The clinical sign is grip myotonia. It is a feature of muscle fiber dysfunction characterized by an involuntary muscle tension that is caused by a lowered electrical threshold and action potentials, which repetitively fire because of a hyperexcitability of the muscle fiber membrane.[1] On EMG examination, myotonic muscles exhibit myotonic runs that are action potentials characterized by a modulation of frequency and amplitude.
3. Genetic anticipation is a phenomenon of increasing disease severity and decreasing age of onset in successive generations.[2]
4. In addition to neuromuscular manifestations of myotonic dystrophy, patients may suffer from many other systemic features (summarized in **Table 1**).

Table 1 - Systemic features of myotonic dystrophy.

Organ/System	Manifestations
Ocular	Cataract, low intraocular pressure, retinal degeneration, sluggish pupillary reactions, corneal exposure keratopathy
Cardiac	Arrhythmias, mitral valve prolapse, cardiomyopathy[3]
Respiratory	Excessive daytime sleepiness
Gastrointestinal	Abdominal pain, dysphagia, emesis, aspiration, diarrhea (or constipation), megacolon, laxity of the anal sphincter
Liver/gallbladder	Elevated γ-glutamyl transaminase, gallstones
Renal	Abnormal handling of calcium
Peripheral nerve	Polyneuropathy
Cutaneous	Balding, pilomatricoma (hair follicle tumor)
Endocrine	Insulin resistance and diabetes mellitus, testicular atrophy, abnormal growth hormone release, reduce adrenal secretion of androgenic hormones, parathyroid adenoma, hypothyroidism or hyperthyroidism
Skeletal	Cranial hyperostosis, talipes, small pituitary fossa
Immune	Reduced serum concentration of immunoglobulins
Behavioral	Changes in personality, affect, and motivation
Others	Increased risk of complications of general anesthesia, such as hypotension, aspiration, and respiratory depression.[4]

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4. Longman C. Myotonic dystrophy. ***J R Coll Physicians Edinb*** 2006; 36: 51-55.