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Myotonic Dystrophy: A Rare Autosomal Dominant Disorder

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Abstract

Myotonic Muscular Dystrophy is inherited form of an autosomal disease which may include cataract, low I.Q. , and heart conduction problems. In men their may be early balding and an inability to have children and gastric tract problems are common. It is a form of muscular dystrophy that affects muscles and many other organs in the body. Myotonia means an inability to relax muscles at will, which makes it difficult to relax the fingers after a firm hand grip. Muscular Dystrophy means progressive muscle degeneration leading to weakness and shrinkage of muscle tissues. It is caused by CTG triplet repeat expansion in non coding region of DMPK gene on chromosome 19q13.3, encoding myotonin. Myotonin is required for inter cellular conduction.

Keywords: Myotonic Dystrophy; Myotonia; Autosomal Dominant Disorder; Dmpk Gene; Myotonin.

Introduction

Clinical presentation of myotonic dystrophy is extremely variable, even in families. It can vary from severe respiratory insufficiency in infancy to cataract alone in adulthood. Molecular DNA analysis and electromyogram (EMG) is routinely available for myotonic dystrophy, including pre-natal diagnosis.

Clinical Presentation

A 51 year old male patient was brought to our E.D. at 2.00 pm with C/O slurring of speech, distension of abdomen and mild breathing difficulty since 2 days with B/L Upper and Lower Limb weakness since 9-10 months with B/L drooping of eyelids and diminision of vision since 4-5 years. No h/o fever, vomiting and change in bowel habits.

Primary Survey:

Airway Assesment: Patent

Breathing Assesment:

Respiratory rate – 16 CYCLES / MIN

Laboured breathing present

SPO2 at room air – 92%

SPO2 with oxygen–100% @ 3L/MIN O2 VIA nasal prongs.

Peripheral Pulsations

all peripheral pulsations present

Temperature : 98.2 F

Cardiac Monitor: Shows ST Depression.

Pupils: B/L Cataract Noted.

Secondary Survey:

Sample History

Physical Signs and Symptoms:- slurring of speech with laboured breathing with abdominal distension with B/L upper and lower limb weakness.

No Drug Allergy Known

Medications- not taken any treatment in the past

Past History: No H/O Dm, HTN, COPD, Weight

Loss in the Past.

Investigations and Management in E.D.:

12 Lead ECG Done Shows Sinus Rhythm @63 B/ Min with Minimal St Elevation in I and AVL with Deep T inversion in Antero-Lateral Leads.

2D ECHO : NO LV RWMA , EF = 60%

Troponin I – NEGATIVE

SOB Profile: CPKMB – 3.0 ng/ml

MYO — 220 ng/ml

TNI — < .05 ng/ml

BNP – 63.1 pg/ml

DDIM – 102 ng/ml

Nerve Conduction Study: Normal nerve conduction study in B/L upper and lower limbs.

NCCT Head: shows normal study.

EMG Study: Shows myopathic pattern/ muscular atrophy – Using Concentric Needle EMG Done In APB, FDI, ADM, Biceps, Tibialis Anterior, Vastus Lateralis, And L5 Paraspinous Muscles.

Reduced Mup'S and Incomplete IP'S are Recorded With Sign of Muscular Atrophy.

Management in E.D.: Patient was managed conservatively with NIV support and other supportive medications and supportive care as advised by Neuro physcian and Cardiologist.

Discussion

What is Already Known on this Topic

Myotonic dystrophy type 1 is the most common adult onset of muscular dystrophy, presenting as a multi systemic disorder with extremely variable clinical manifestation, from asymptomic adults to severely affected neonates.

Commonly Seen Complications

Myopathy, Lens opacities, heart conduction defects, gastrointestinal dysfunction, obstructive sleep apnea and daytime hyperinsomnlence, higher incidences of miscarriage in pregnancy are commonly seen.

Life Expectancy

Mean age of Death is 60 years.

Mortality is most commonly due to pneumonia and cardiac dysarrhythmias.

How this Might Change the Clinical Practice

High level of clinical suspicion by ER Physcian is needed for diagnosis.

Bed side general history, past history and drug history must be taken.

Prompt intervention with NIV to assist labored breathing is needed to reduce the work of breathing.

Genetic counseling is recommended to discuss the implications including the psychosocial and offspring risk reduction.

All survivors should undergo Annual Check-up for ECG, Urine Dipstick for Glucose and Ophthalmologist.

Conclusion and Take Home Message

Myotonic Dystrophy is the most common heritable autosomal neuromuscular disorder.

As a ER Physcian we should keep in mind regarding the typical presentation of such patients including the physical signs like early frontal balding, Ptosis, Lens opacities, inability to frown, clench teeth, smile and limb weakness. We should elicit the signs of Myotonia by asking the patient to rapidly relaxing the clenched fist or by tapping thenar eminence and last but not least look for the ability to swallow and the pattern of breathing and gait of the patient will give us a good clue for early diagnosis and prompt treatment in highly suspected cases.

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