Picture quiz

Neuromuscular disorders

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(Compiled by A Arasalingam[1] and Saman B Gunatilake[2])

Q1

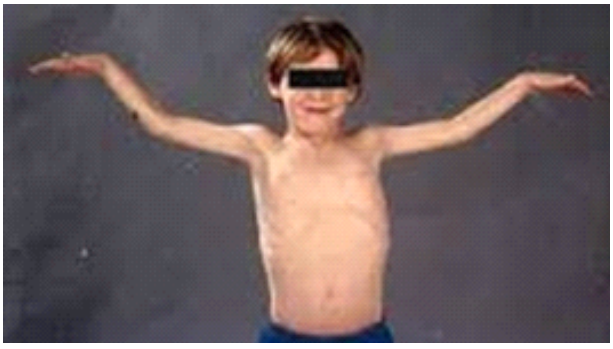
A 45-year-old male presented with progressive exercise induced muscle cramps, myalgia and stiffness over the last 20 years. There was no significant family history. On examination there was generalised muscle hypertrophy especially of the calf, but no weakness. Tapping or brief application of pressure to the upper arm produced short lasting muscle contractions. He had elevated serum CK levels. Western Blot pattern is shown:

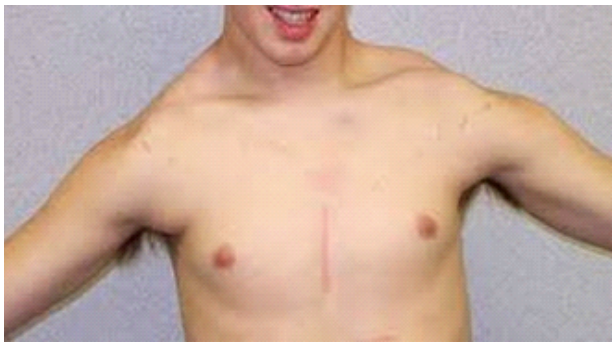
What is the diagnosis?

Q2

A 24-year-old male presented with a rigid spine and neck, contractors at the elbow and shortening of the Achilles tendon, scoliosis and increased lumbar lordosis. The muscles of the lower arms and legs and the scapular muscles were atrophied. There was mild weakness of the triceps and iliopsoas muscles. He had generalised areflexia. Cardiac investigations revealed a first degree AV block. Serum CK activity was elevated 6 times the upper limit of normal. He had noticed that he was unable to extend his arm fully and that his Achilles tendon was taut from the age of five. He had an elder brother with the same presentation.

What is the diagnosis?





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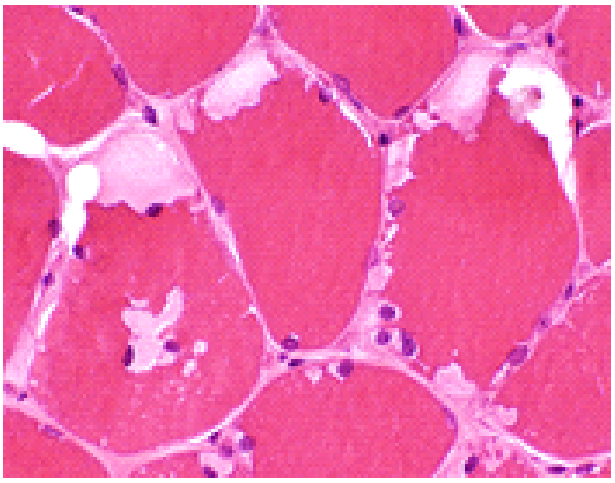
Q3

A 40-year-old female presented with cramps and swelling of the hands for more than 4 years. She had never had episode of passing dark urine nor had she noticed muscle weakness. Her past medical history was unremarkable except for a goiter. Her family history was negative for neuromuscular disorders. Her neurological examination was normal.

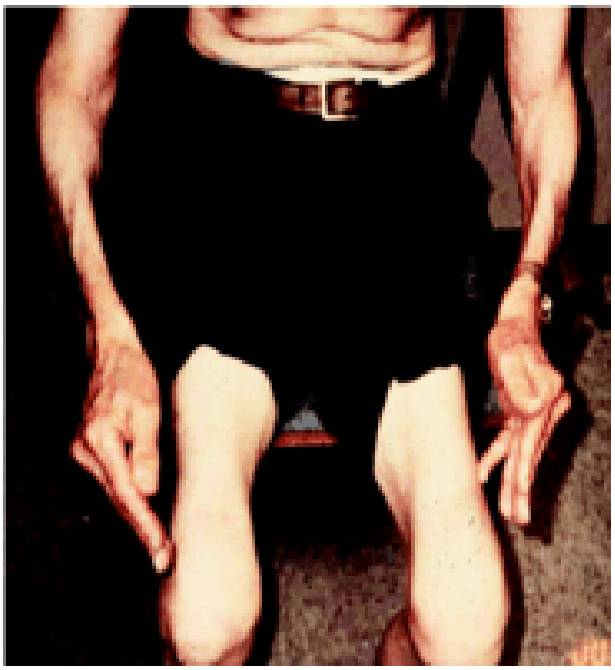
Initially she had a markedly elevated serum CK activity of > 13000 IU/L. Repeat measurements showed activities of 3389 IU/L and 1550 IU/L respectively. The ischaemic forearm test showed only a minimal increase in serum lactate but ammonia showed a considerable rise from 14 to 489 micromol/l. A skeletal muscle biopsy was done. Histology shown.



What is the diagnosis?

Q4

A 66-year-old man presented with progressive muscle weakness for more than 6 years. Had been treated for polymyositis with steroids without any benefit. Patient complained of difficulty going up and down stairs and turning door knobs and handles. Serum CK was 360 IU/L (normal <200).

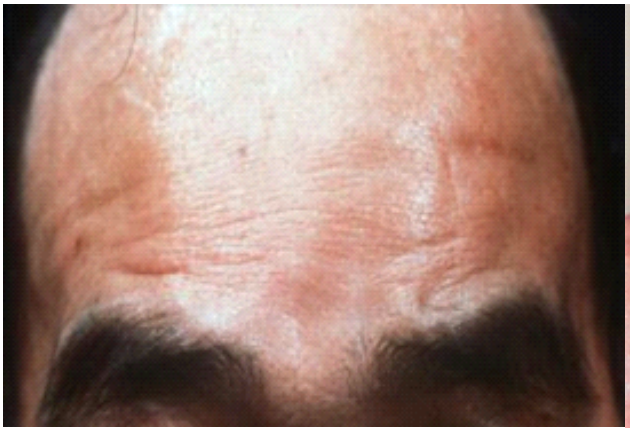
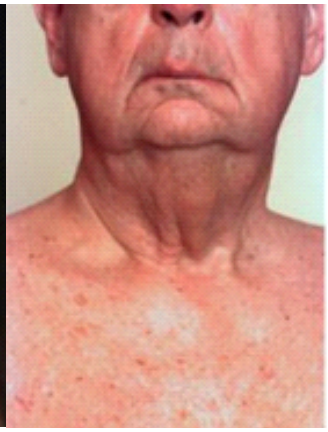
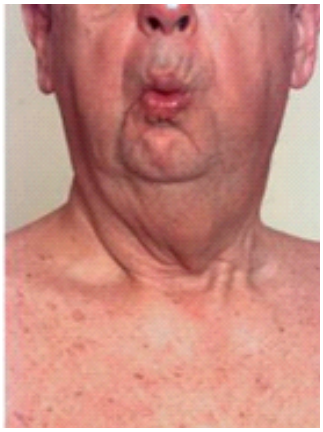
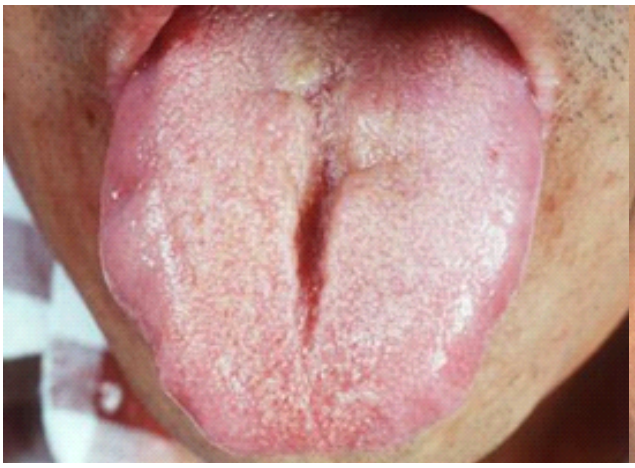
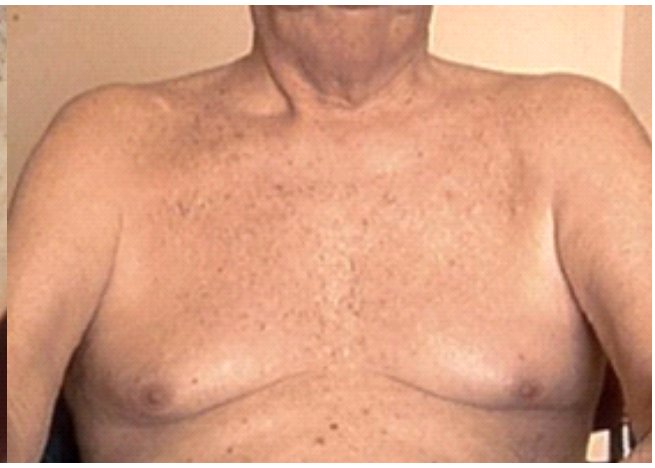


What is the diagnosis?

What is seen in a muscle biopsy?

Q5

What is the diagnosis?

(Answers on page 42-43)