

CASE 2000 - 10

SUBMITTED BY:

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CLINICAL HISTORY:

The patient is 34 years of age and the mother of a normal 9 month old boy. She has a history of familial neuromuscular disorder affecting her mother (now bed-ridden), her sister and a niece; two nephews are unaffected.

As a toddler she walked on her toes, had an Achilles tendon release at the age of 9, at the age of 12 had eye surgery for strabismus and she never walked or run well. Beginning in her teens she developed slowly progressive muscle weakness involving lower extremities, eventually she complained of weakness in the shoulder girdle.

CPK determination is 460 and the sedimentation rate is slightly elevated.

The patient had a muscle biopsy at the age of 11 which was inconclusive.

Muscle biopsy from the left deltoid was performed.

MATERIAL SUBMITTED:

Any one of:

- a) cryosection stained with H&E
- b) cryosection stained with Gomori-trichrome
- c) semithin sections

POINT FOR DISCUSSION:

Diagnosis