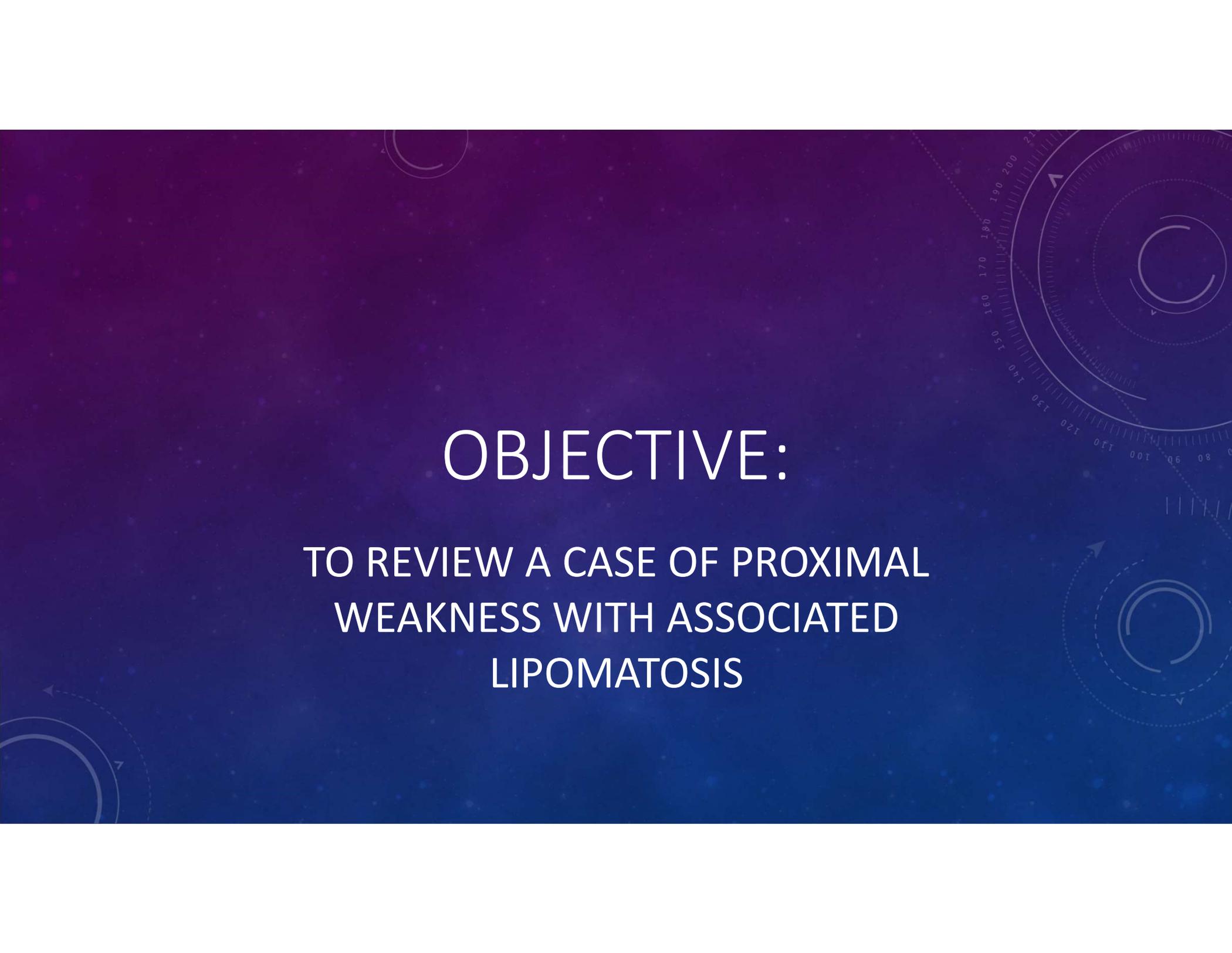
The background features a dark blue gradient with intricate white and light blue circular patterns. On the left side, there is a large circular scale with numerical markings from 140 to 260 in increments of 10. Several smaller circles and arcs are scattered across the field, some with arrows indicating direction. The overall aesthetic is technical and scientific.

A CASE OF LIPOMATOSIS AND PROXIMAL WEAKNESS

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FEBRUARY 21, 2018

The background features a dark blue gradient with a starry space pattern. On the right side, there are several technical diagrams, including a large circular gauge with numerical markings from 80 to 210 and a smaller circular diagram below it. Both diagrams have arrows indicating a clockwise direction. The text is centered in the middle of the slide.

OBJECTIVE:

TO REVIEW A CASE OF PROXIMAL
WEAKNESS WITH ASSOCIATED
LIPOMATOSIS

CASE PRESENTATION

52 y/o male

- 8 yr Hx of progressive proximal weakness
- No ocular or bulbar symptoms

PMHx:

Gout

Lipomas - 20 year history

- Symmetric Upper body (chest, back, and proximal UEs)

Family Hx : No history of weakness

Mother w/ multiple lipomas

CASE PRESENTATION

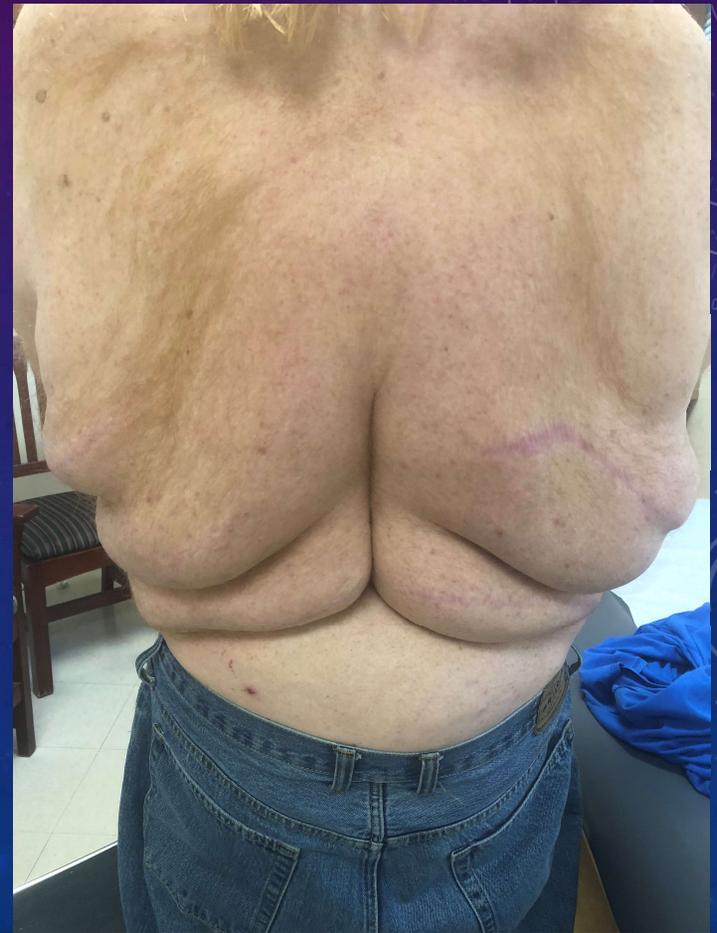
- On examination:

| Muscle | Right | Left |
|--------------------|-------|------|
| Neck Flexors | 4 | |
| Shoulder Abductors | 4+ | 4+ |
| Elbow Flexors | 5- | 5- |
| Hip Adductors | 4- | 4 |
| Hip Flexors | 4 | 4 |
| Hip Extensors | 4 | 4 |
| Knee Flexors | 4+ | 4+ |

- Sensory Examination: intact
- Gait: Waddling
- General examination: Multiple Symmetric Lipomas
 - Neck, Ant Chest, Back, Shoulders

- CK 543

CASE PRESENTATION



CASE PRESENTATION

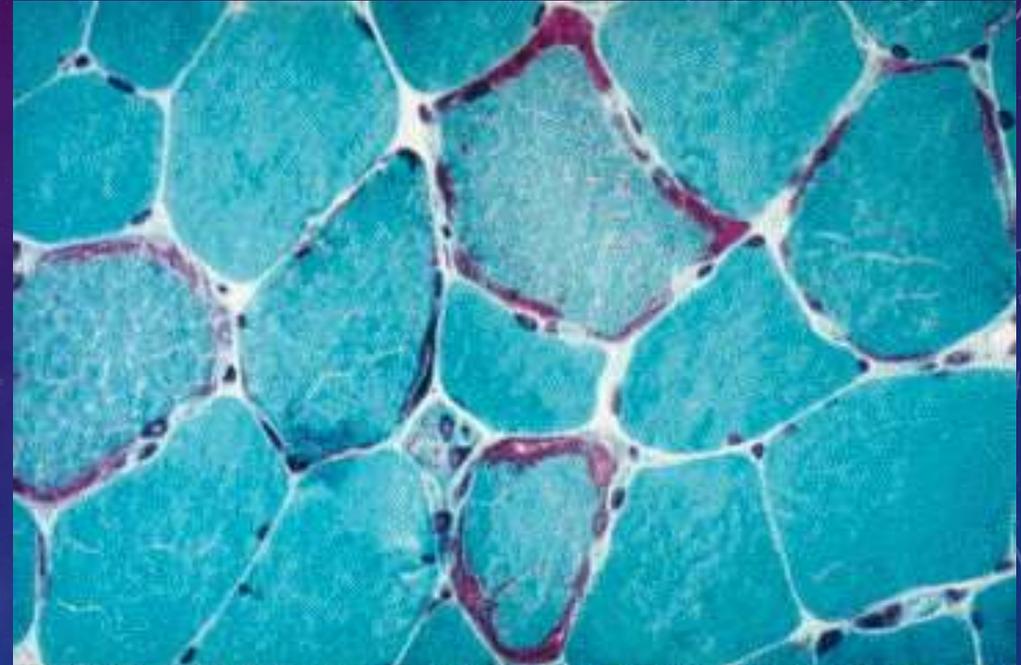
Nerve Conductions: Absent Sural Response B/L

Electrodiagnostic Study: Irritative Myopathy

| Muscle | Spont Act | | | MUAP | | | Recruitment |
|-----------|-----------|------|------|------|-----|----|-------------|
| | IA | Fib | PSW | Amp | Dur | PP | Pattern |
| R-side | | | | | | | |
| Tib Ant | 1+ | 1+ | 1+ | + | N | + | N |
| Gastroc | 1+ | None | None | N | N | N | N |
| Vast Lat | N | None | None | + | N | N | MLD Dcr |
| Iliopsoas | 2+ | 2+ | 1+ | +/- | +/- | + | Early |
| Glut Med | 1+ | 1+ | 1+ | - | - | N | Early |
| Biceps | 1+ | 1+ | 1+ | - | - | + | Early |
| Triceps | 1+ | N | None | N | N | + | Early |
| BR | N | N | None | - | - | N | Early |
| FDI | N | None | None | N | N | N | N |

CASE PRESENTATION

- Muscle Biopsy of R Quadriceps:
 - Ragged red fibers
 - + Fiber-type grouping
 - No inflammation
 - Impression: Suggestive of Mitochondrial Myopathy

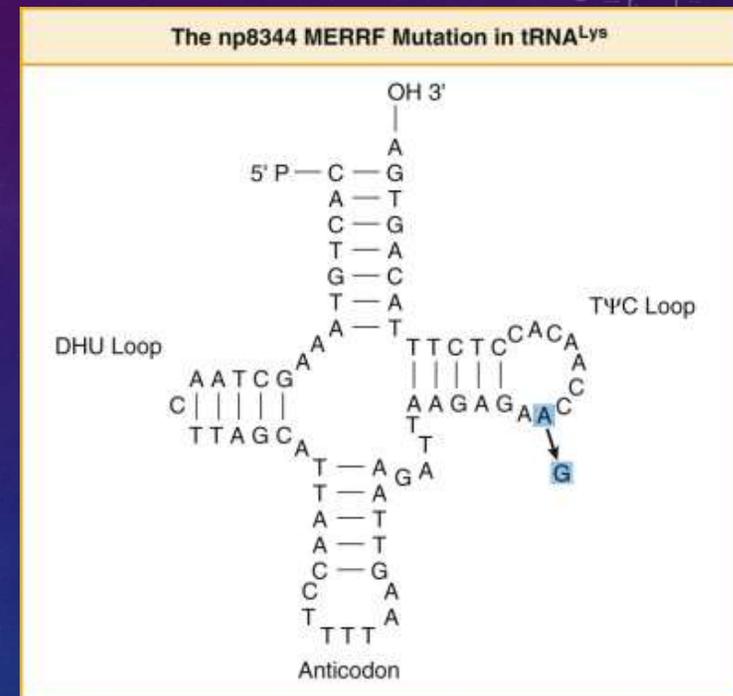


Biochimica et Biophysica Acta, 2004

WHAT IS THE DIAGNOSIS?
HOW TO PROCEED FROM HERE...?

GENETIC TESTING

- Genetic Testing on a serum sample:
 - **MT-TK mutation** detected
 - Specifically: **m.8344 A>G**
 - Heteroplasmy 83%



Adkison, L. *Integrated Review of Genetics*, 2012.

LITERATURE REVIEW

- m.8344 A>G associated w/ Myoclonic Epilepsy with Ragged Red Fibers (MERRF) Classically, MERRF described with:
 - Myoclonus
 - Seizures
 - Cerebellar Ataxia
 - Proximal Muscle Weakness
- Madelung's Disease, aka Multiple Symmetric Lipomatosis (MSL)
 - Adult-onset recurrent Lipomatosis
 - Affecting:
 - Nuchal
 - Upper thorax
 - Association w/ neuropathy
 - Rare Case reports described combination of: **MSL + Myopathy** ... including m.8344A>G mutation



Annals of Indian Academy of Neurology 2009.

LITERATURE REVIEW OF M.8344A>G MUTATION

| Study (Author, date) | Pt Population (n=) | Age at Sx Onset | Lipomatosis (%) | Myopathy (%) | Biopsy w/ RRFs (%) | Epilepsy (%) | Myoclonus (%) | Neuropathy (%) |
|----------------------|--------------------|-----------------|-----------------|--------------|--------------------|--------------|---------------|----------------|
| Altmann et al 2016 | 34 | 24.5 (±10.9) | 4/24 (17%) | 18 (53%) | 5 (14.7%) | 17/28 (61%) | 16/27 (59%) | 7/22 (32%) |
| Perera et al 2018 | 8 | 39.1 (±10.9) | 8 (100%) | 1 (12.5%) | N/A | 0 | N/A | 6/8 (75%) |
| Kazakos et al 2012 | 7 | 45.6 (±12.2) | 7 (100%) | 5 (71%) | 5/5 tested | 0 | 0 | 0 |
| Our Patient 2018 | 1 | 32 | + | + | + | Neg | Neg | + |

DISCUSSION

MERRF:

Myoclonus
Epilepsy
Ataxia

Our Pt:

m.8344A>G
Lipomatosis
Myopathy
Neuropathy

MSL:

Lipomatosis
w/o
Myopathy

CONCLUSION

- This is a rare case of m.8344 A>G mutation of the MT-TK gene presenting **with** MSL, myopathy, **and** neuropathy, but **without** epilepsy.
- Lingerin Questions:
 - *What is the link between muscle tissue and adipocytes?*
 - *Does the degree of heteroplasmy play a role?*

