The diagnostic value of EM in skeletal muscle biopsy: A service evaluation study at Leeds Teaching Hospitals - NHS Trust

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Background

- Skeletal muscle diseases include a wide range of disorders that are often challenging to diagnose due to overlapping clinical and histological features.

- Electron microscopy (EM) can provide valuable ultrastructural detail when other investigations are inconclusive.

- This service evaluation aimed to assess the diagnostic value of EM in skeletal muscle biopsies by determining how often it contributed additional diagnostic information in routine practice.

Methods

- A total of 84 reports were reviewed from the Leeds Teaching Hospital Trust database between Jan 2018 – Dec 2025.
- Cases were evaluated by two reviewers with oversight from a consultant histopathologist.
- Data were summarised descriptively in Microsoft Excel, and the diagnostic yield with 95% confidence intervals was determined using the Wilson method.
- EM findings were compared with light microscopy results and categorised as either:
  - Confirm / exclude specific diagnosis.
  - No change to histological diagnosis.

Results

- EM contributed additional diagnostic value in 67 of the 84 reviewed cases (80%).

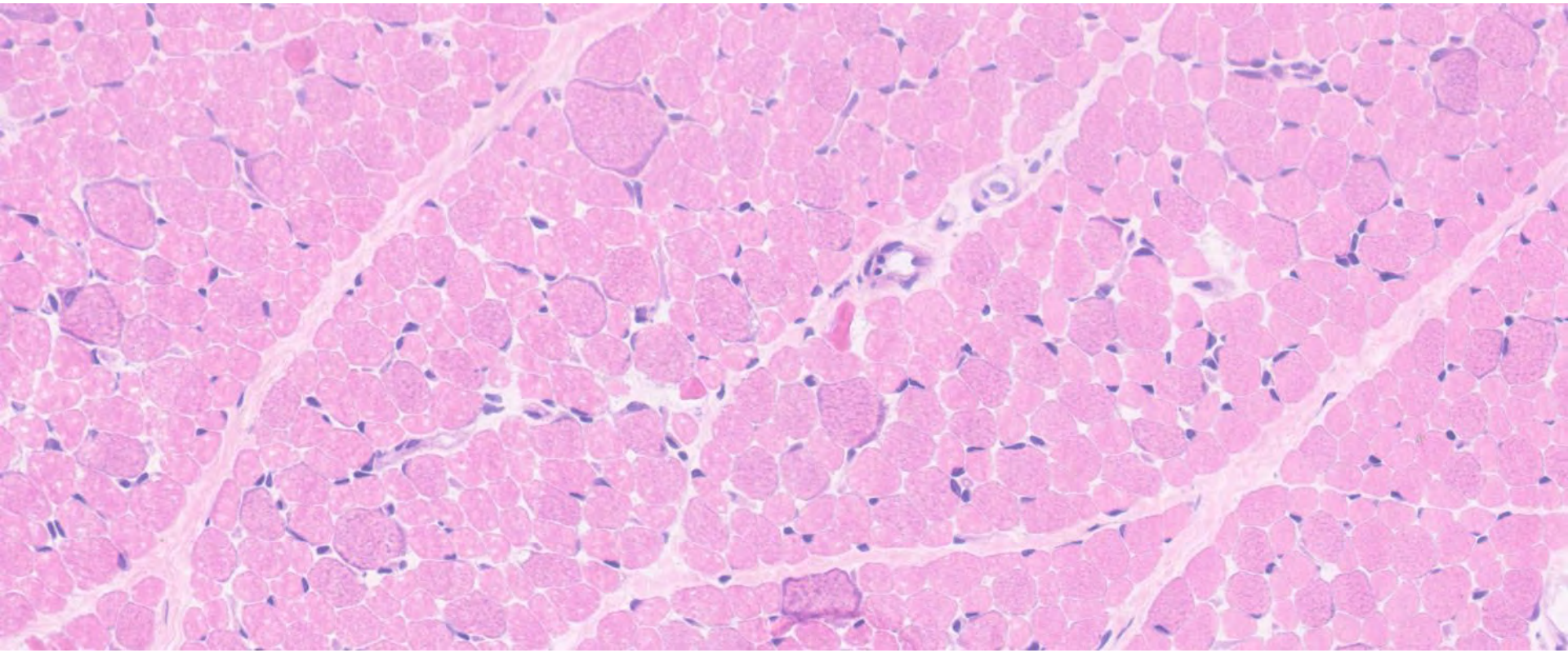
- The greatest yield was observed in:
  - Mitochondrial myopathies (95%).
  - Inflammatory myopathies (73 %).

- Biopsies showing minimal or no structural abnormalities demonstrated a lower yield (57%).

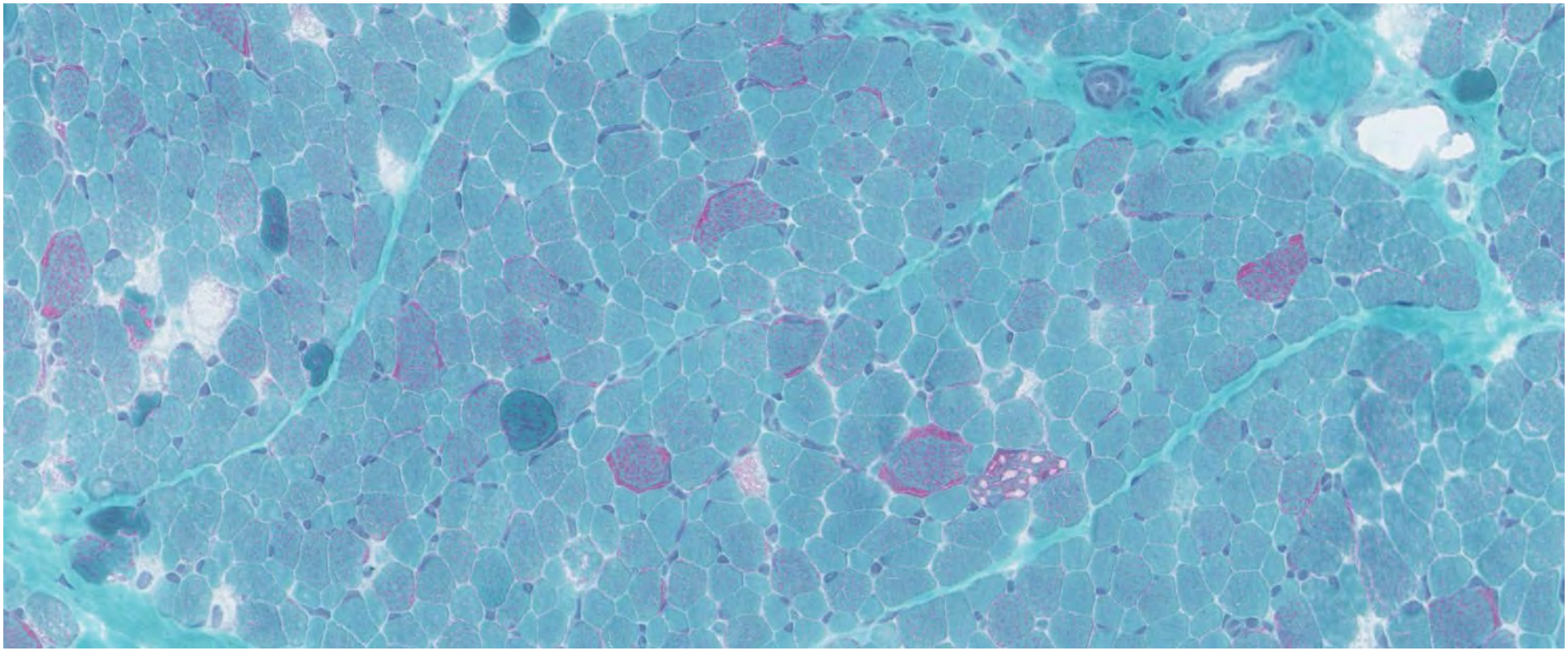
Patient 1

- 2 y f baby
- 1st year hypotonia, gross ascites, dysmorphic features, multiple systems involved.
- High lactate
- Nodular liver
- MRI brain: normal
- ? Mitochondrial disorder / storage disorder

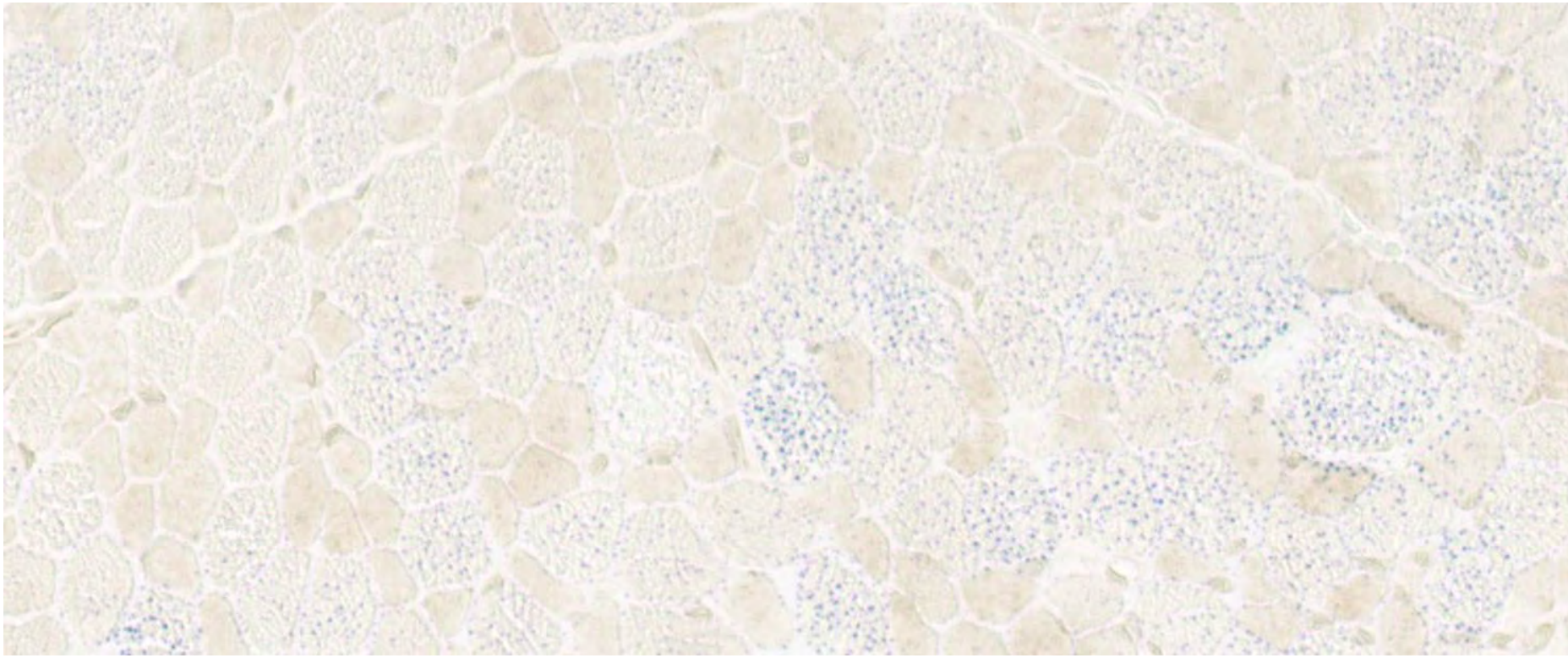
Histology: H&E stain

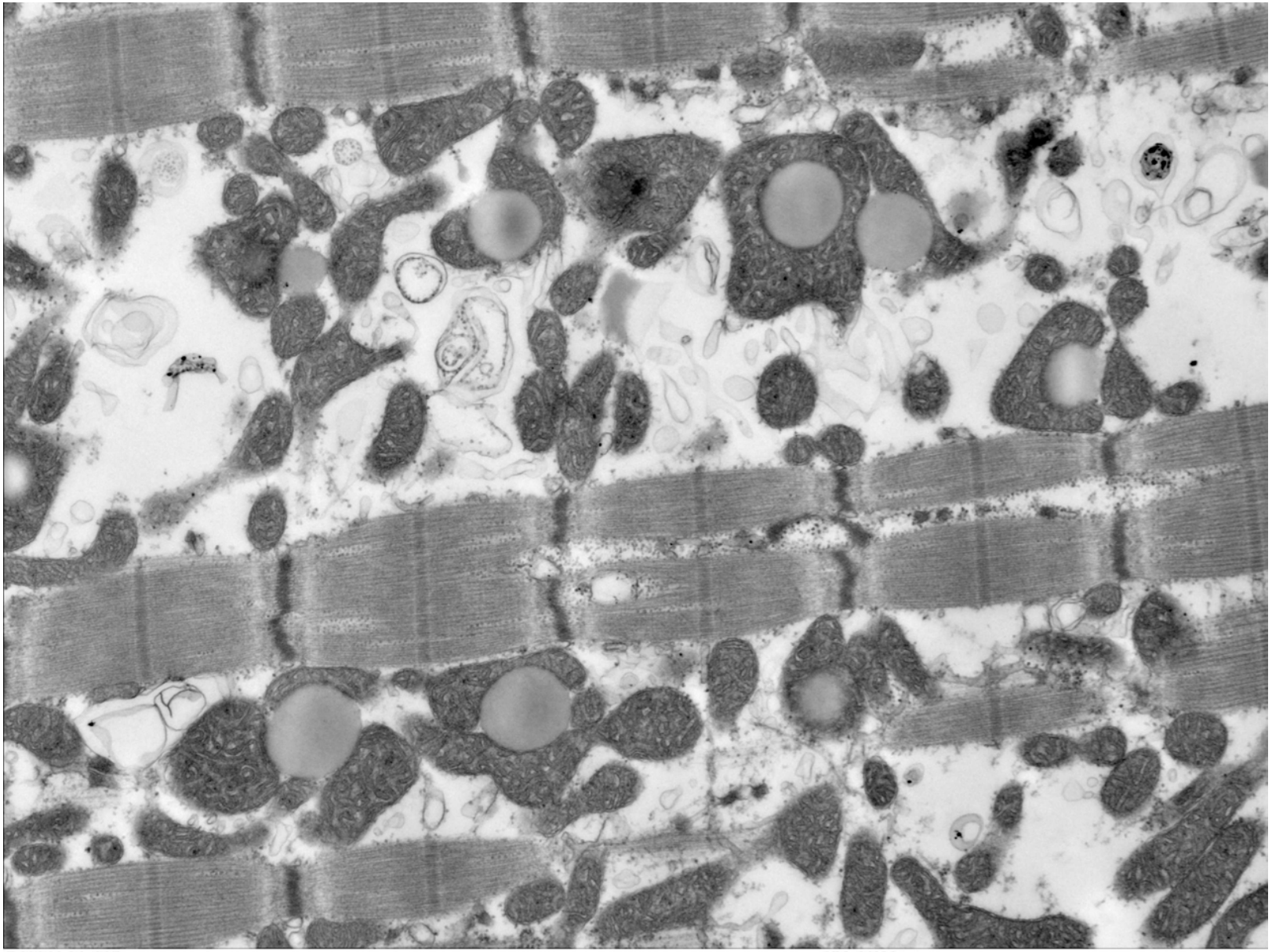


Histology: Gomori stain



Histology SDH-COX stain

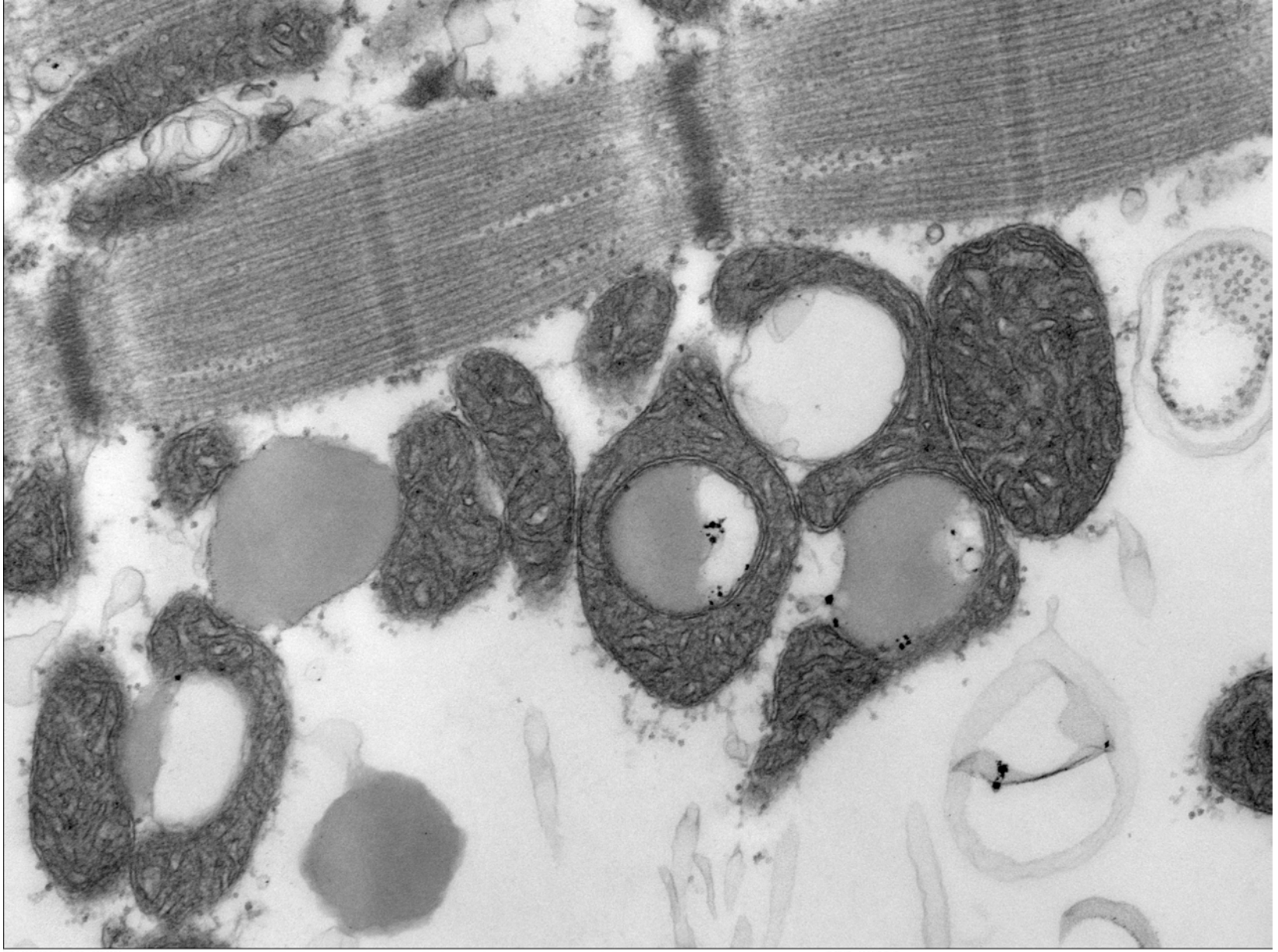




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R23341 24
LH24-35583

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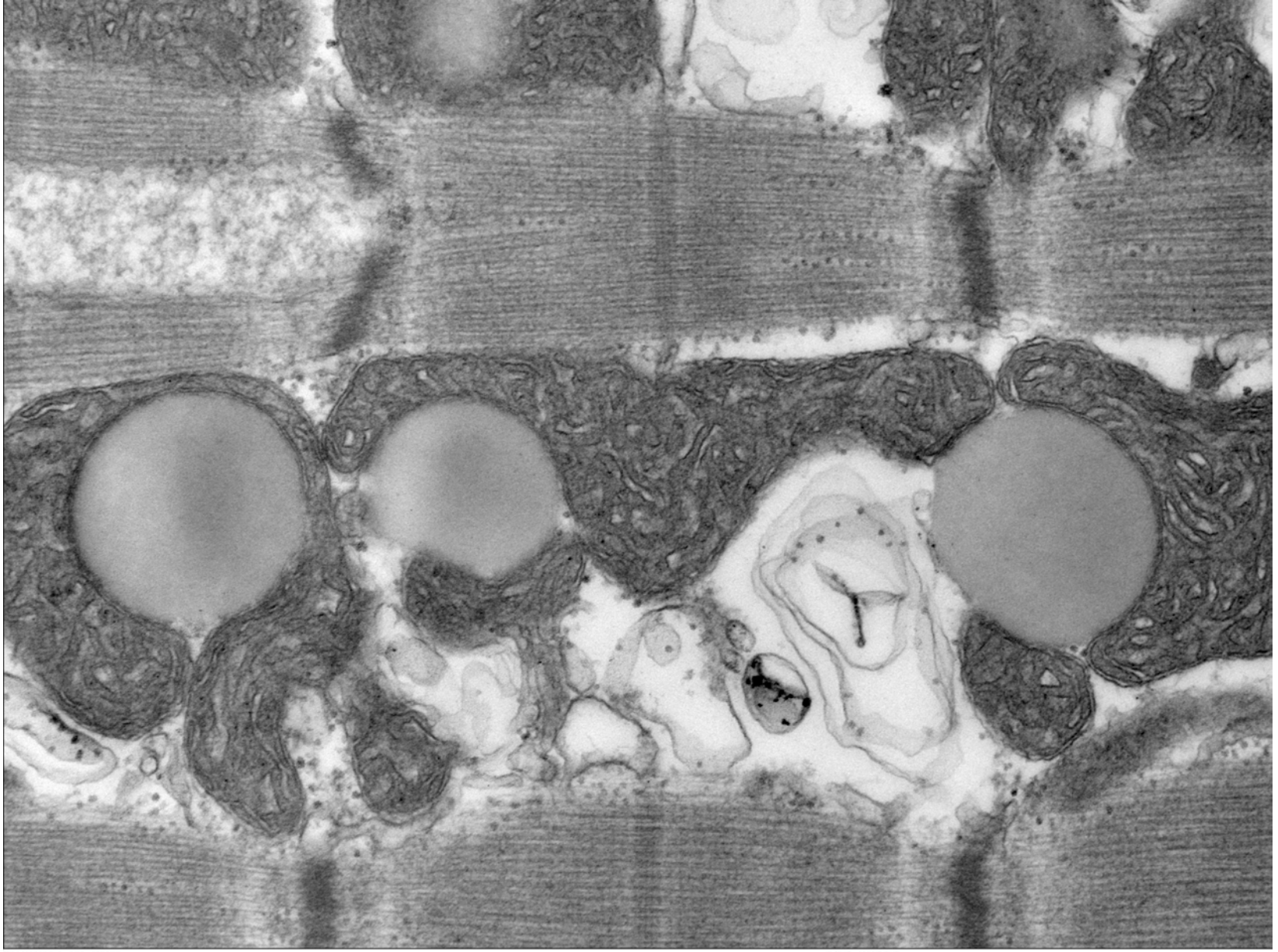
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Leicester Royal Infirmary



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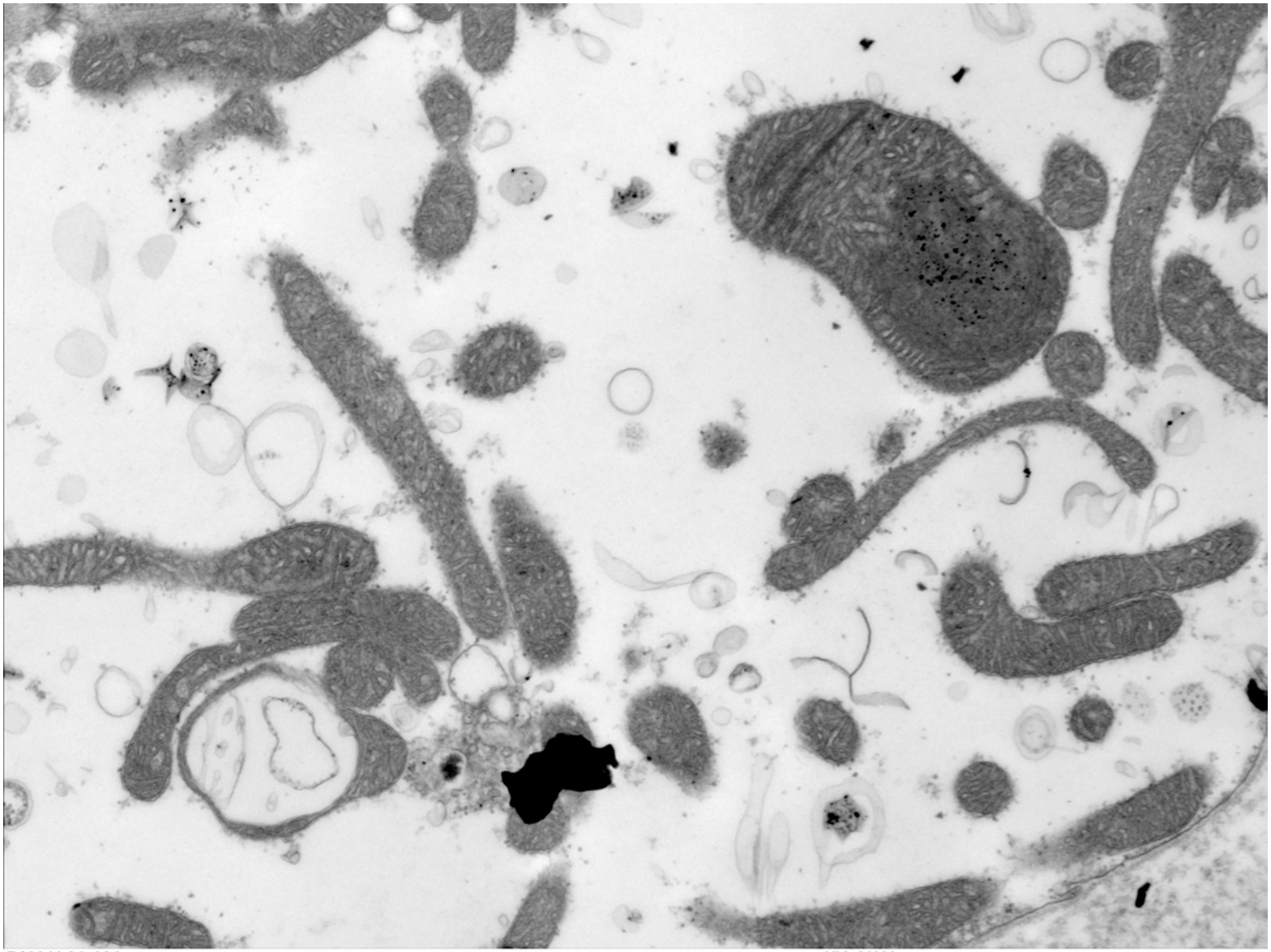
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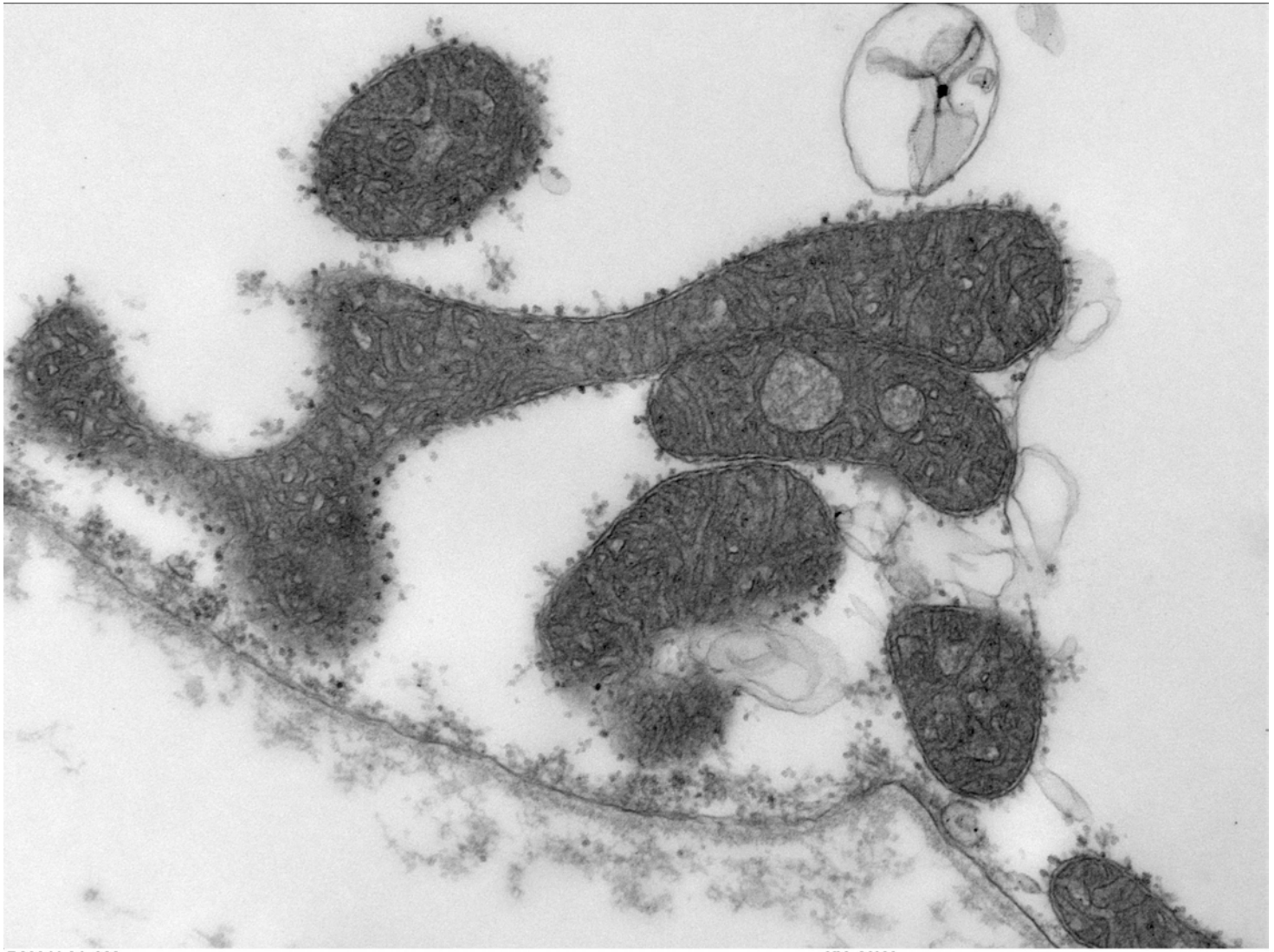
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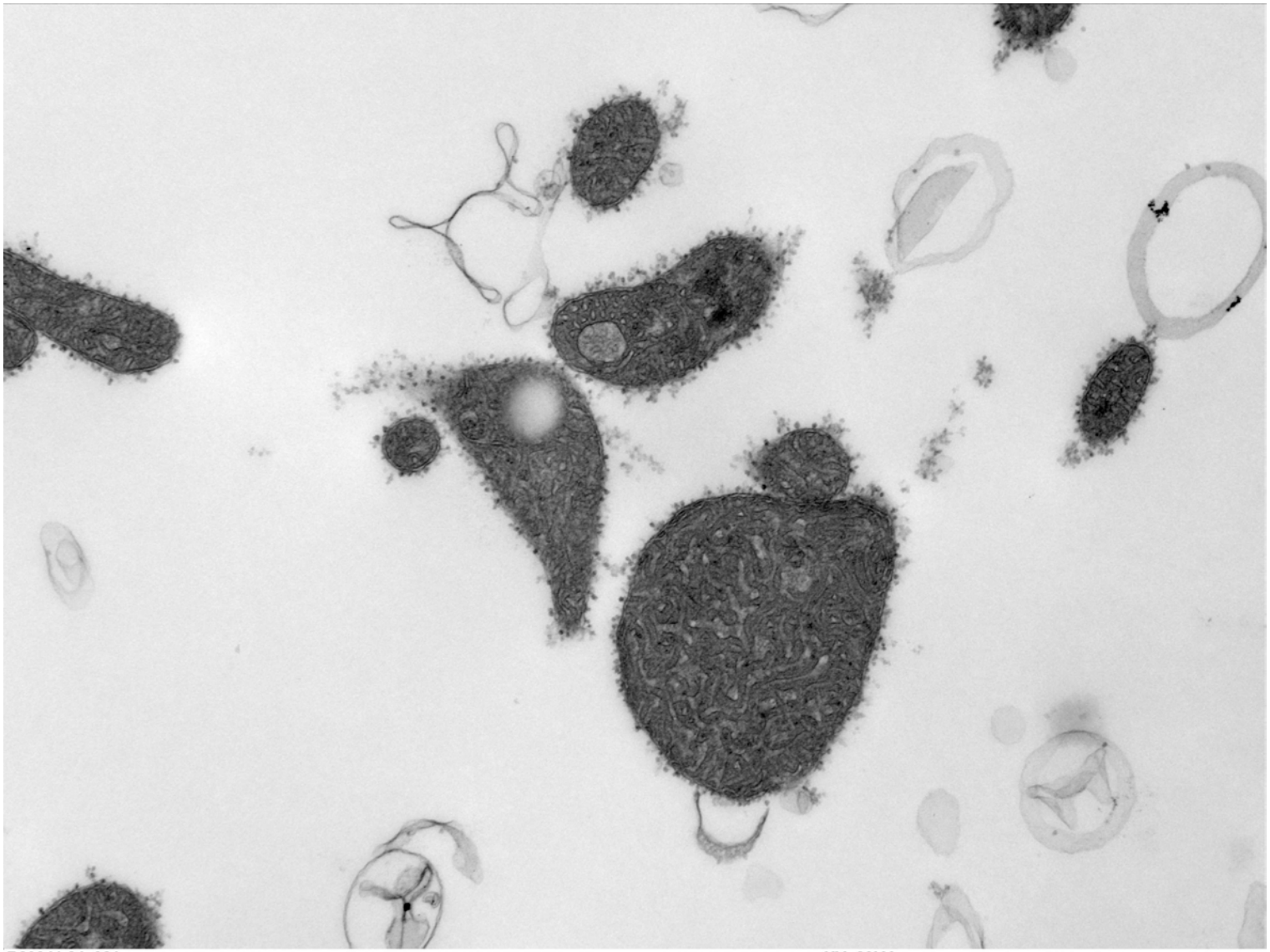
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HV=80kV
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Leicester Royal Infirmary



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R23341 24
LH24-35583

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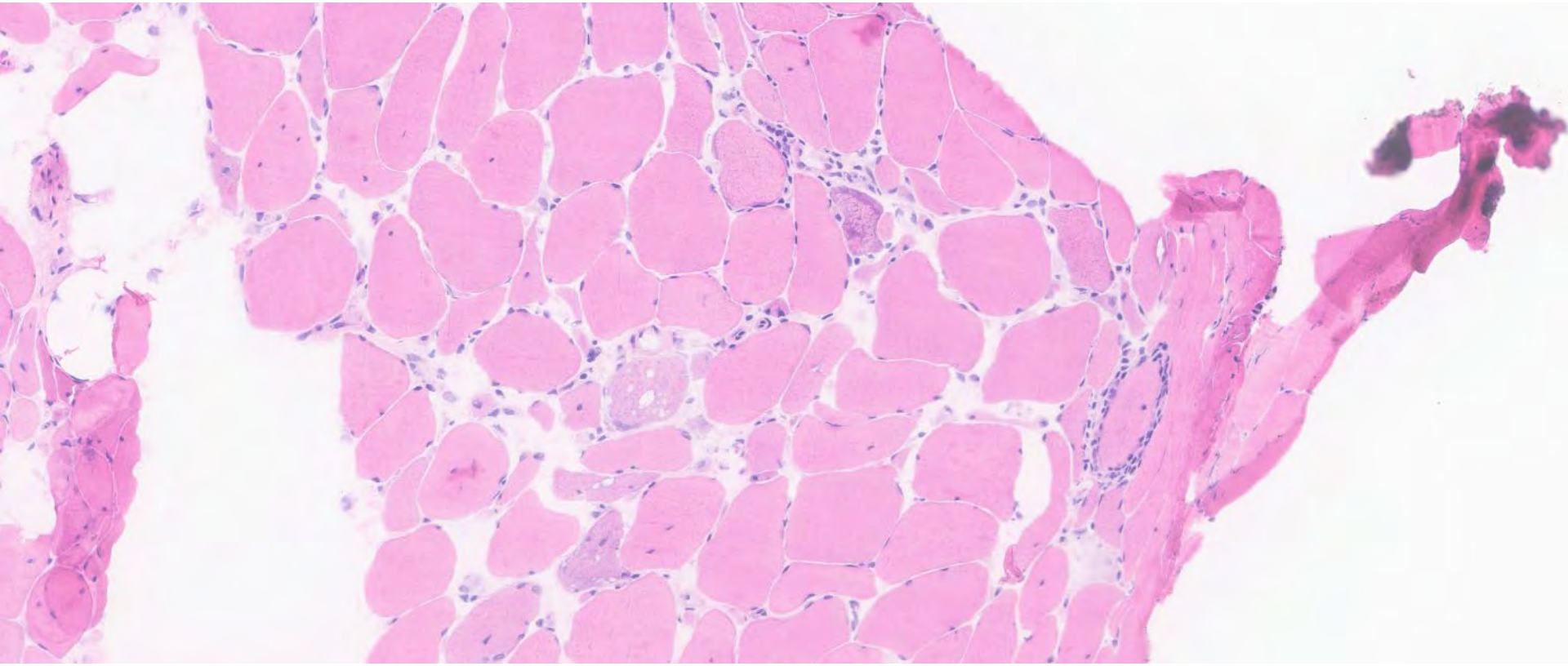
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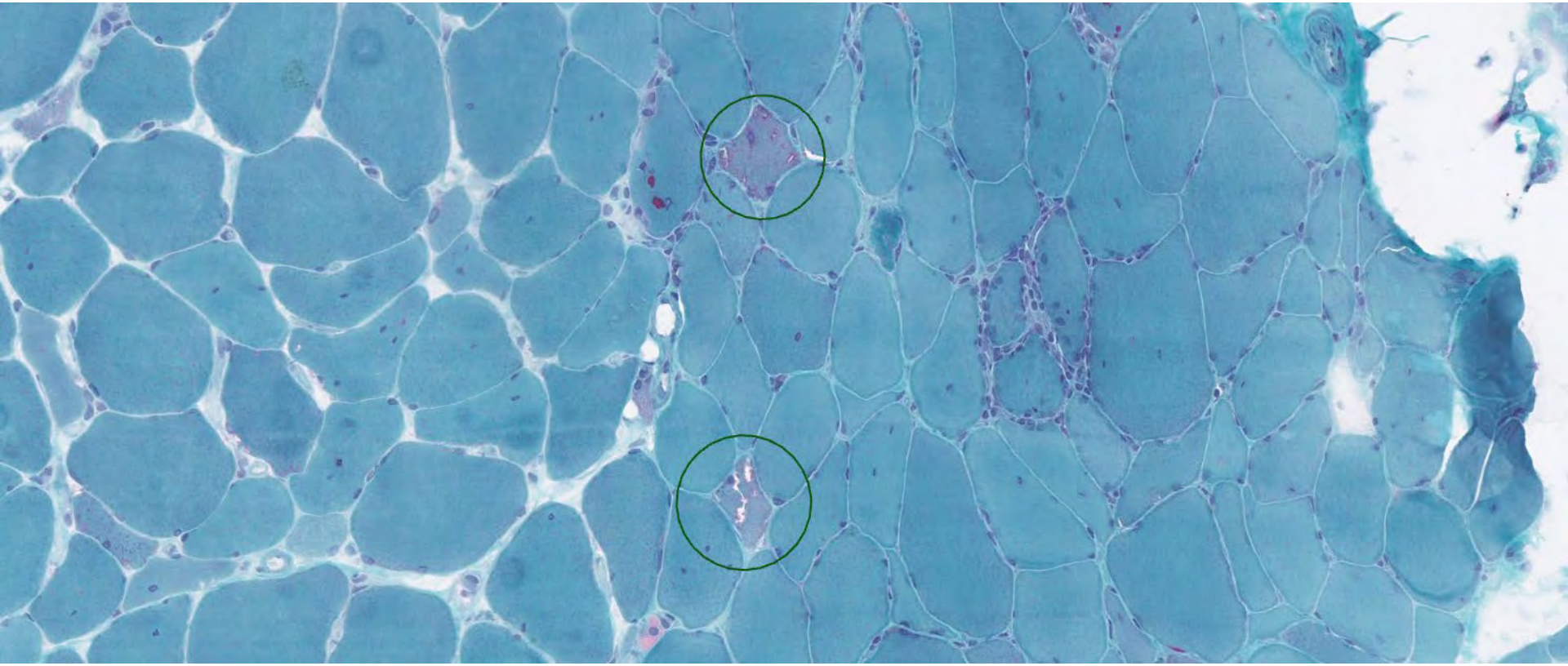
Final diagnosis

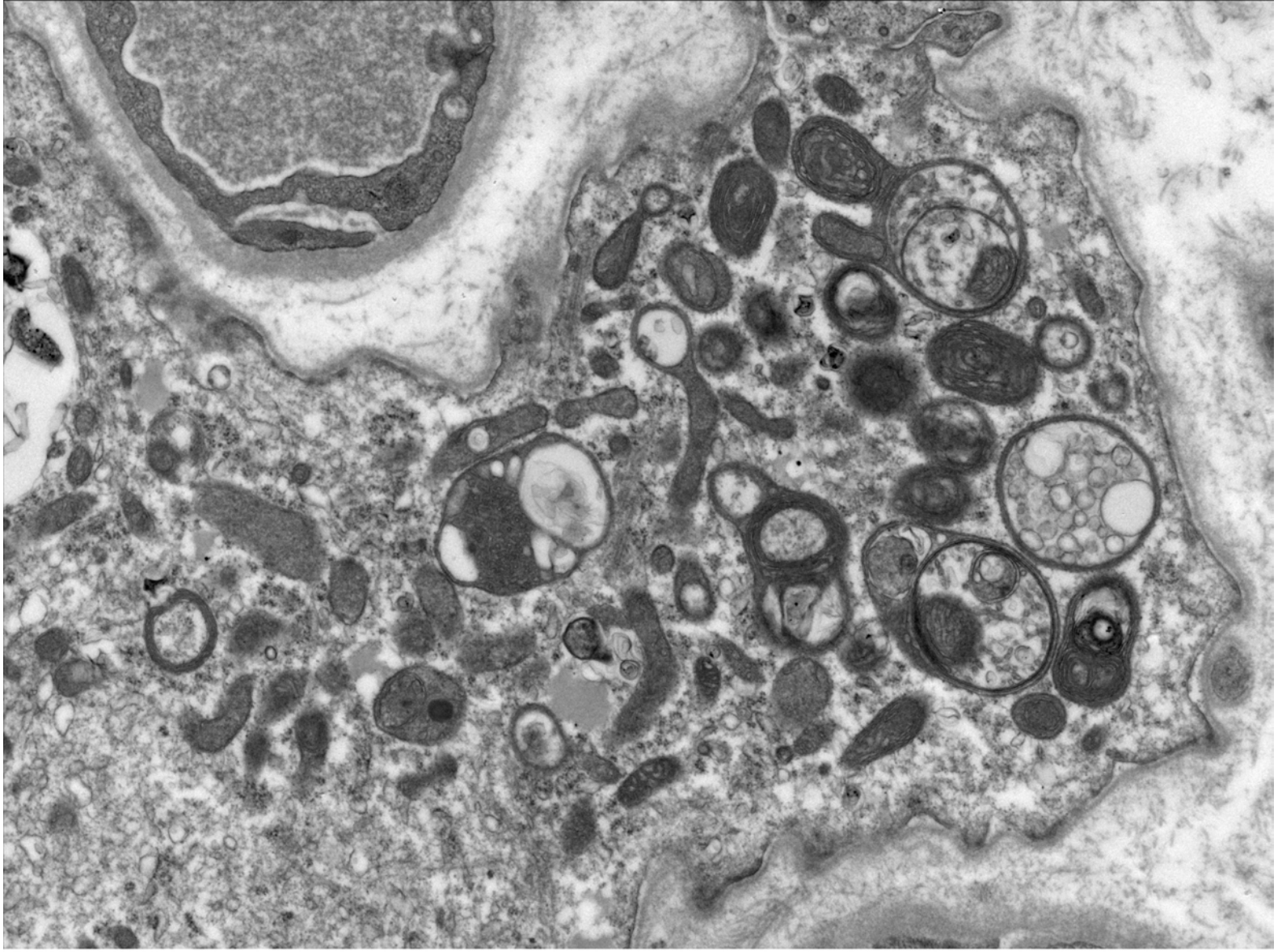
- Mitochondrial myopathy on muscle biopsy with EM.
- Genetic tests: Mitochondrial TRMU-related genetic disorder

Patient 2

- 55 y m
- Weakness in hand grip and proximal legs
- NCS/EMG: myopathic features in long finger flexure and knee extensor with spontaneous active necrosis in muscle
- ? Inclusion body myositis



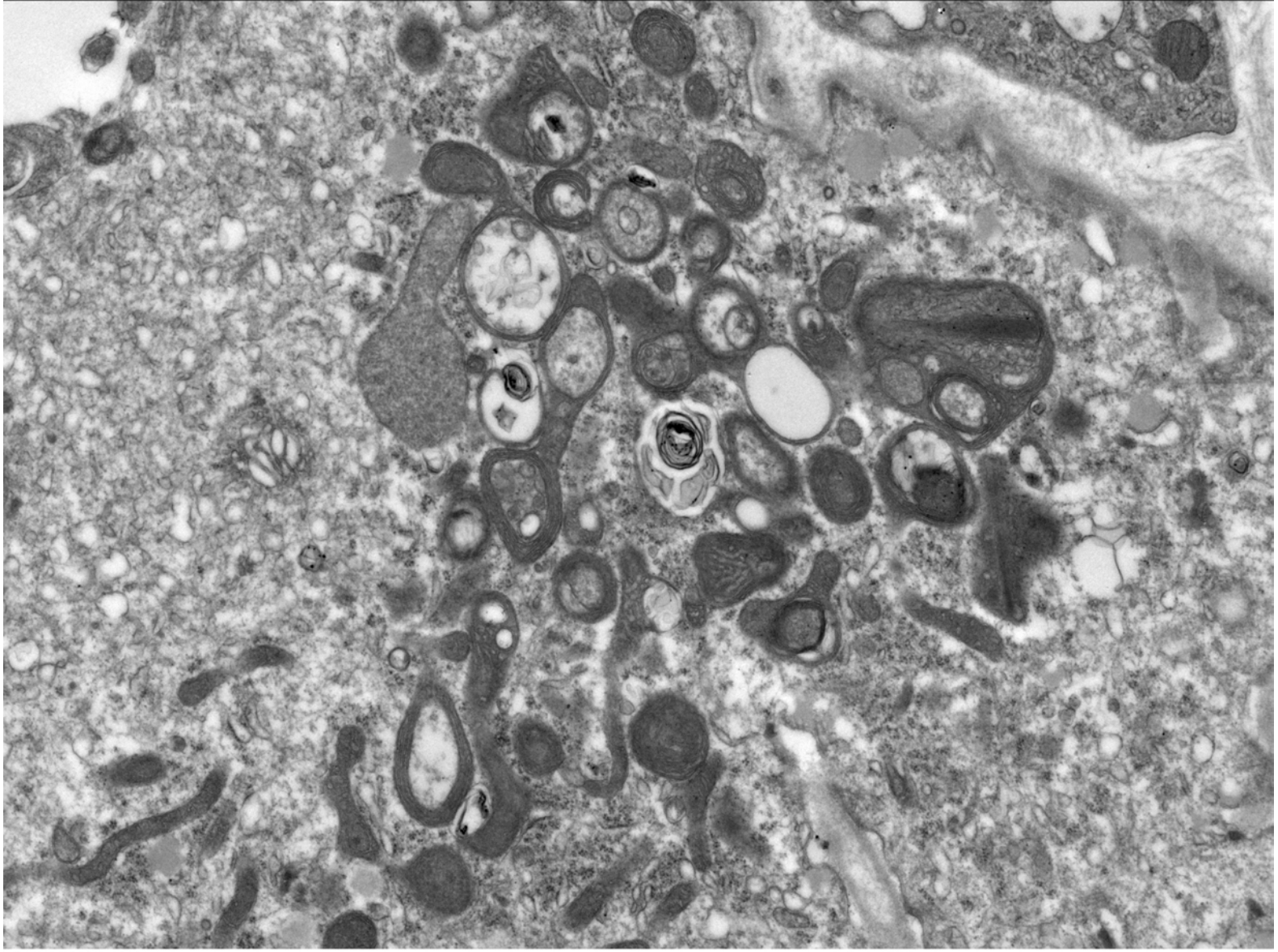




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Muscle biopsy LH24-38426
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Direct Mag: 3000 x
AMT Camera System

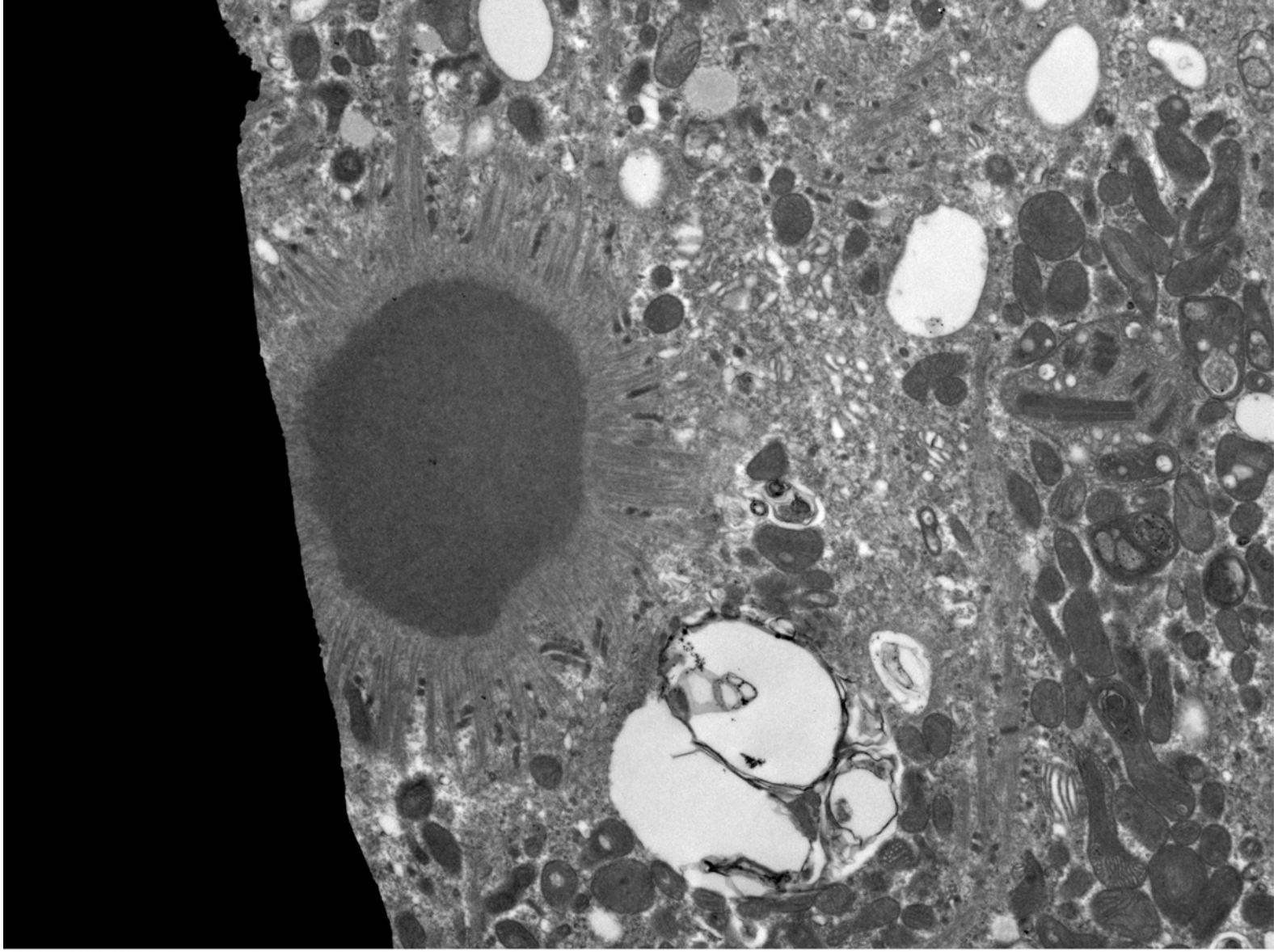
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R24596 24_013.jpg
Muscle biopsy LH24-38426
R24596 24

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AMT Camera System

Microscopist: Natalie Wilson Scope 2
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R24596 24_014.jpg
Muscle biopsy LH24-38426
R24596 24

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AMT Camera System

Microscopist: Natalie Wilson Scope 2
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Gamma: 0.95, No Sharpening, Normal Contrast

Diagnosis

- Inclusion Body myositis

Conclusion

- EM is essential diagnostic tool in skeletal muscle disease.
- EM best use as targeted rather than routine use.

Thanks to:

- Electron Microscopy Facility, Core Biotechnology Services, The University of Leicester.

- Medical school, The University of Leeds.

- Histopathology department at Leeds Teaching Hospitals – NHS Trust.

Declaration:

- Researchers declare No conflict of interest.

- This service evolution aimed to improve diagnostic service and teach medical students.