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New variant of necklace fibres display peculiar lysosomal structures and mitophagy

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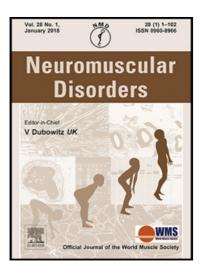


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Highlights

- We identified two families with the same mutation in the *DNM2* gene.
- They had slightly different clinical presentation but similar histological features.
- We describe a new variant of 'necklace fibres' by histology and 3D EM.
- Structural features are discussed in the context of the relevant literature.

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