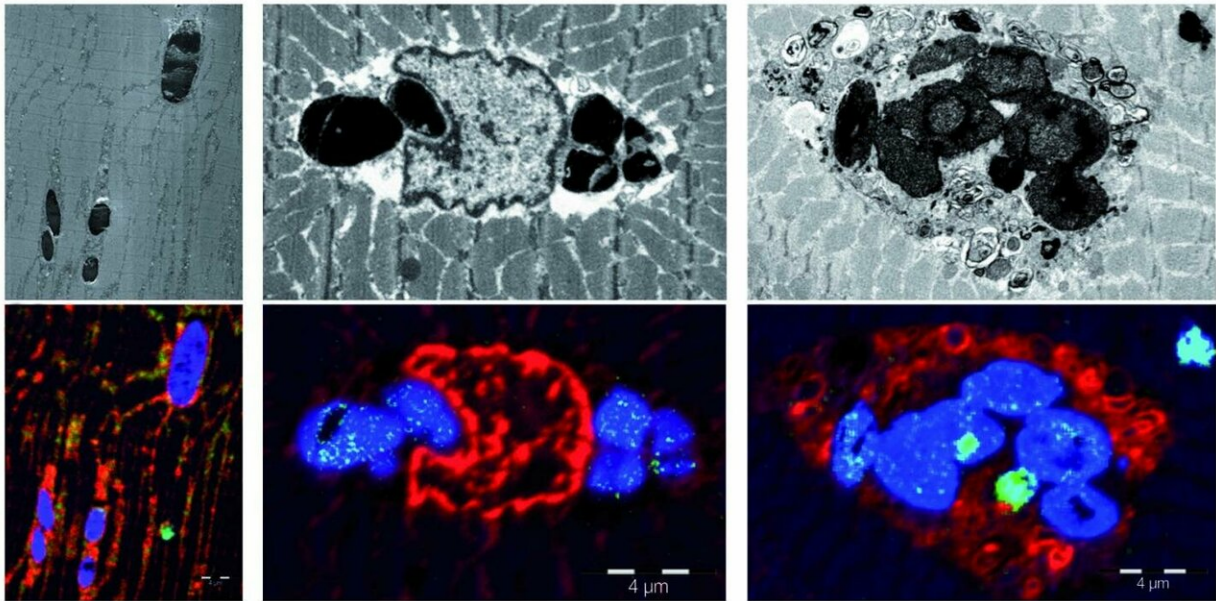


# New muscular disease myoglobinopathy described

March 27 2019

---



Credit: Bellvitge Biomedical Research Institute (IDIBELL)

Researchers at the Bellvitge Biomedical Research Institute (IDIBELL) led by Dr. Montse Olivé have described in *Nature Communications* a new muscular disease caused by a mutation in the myoglobin gene. The study was led by Prof. Nigel Laing and researchers from the Karolinska Institute (Stockholm, Sweden).

Myoglobin, the protein that gives muscles their red colour, has as its

main function the transportation and intracellular storage of oxygen, acting as an oxygen reservoir when there are low levels (hypoxia) or a total lack thereof (anoxia). It also acts as scavenger of free radicals and other [reactive oxygen species](#), avoiding cell damage due to oxidative stress.

"This is the first time that a [disease](#) caused by a mutation in the myoglobin gene has been identified," says Montse Olivé, doctor at the Bellvitge University Hospital, responsible for the muscular disease area and researcher of the Human Molecular Genetics group of the Genes, Disease and Therapy at IDIBELL. "We have identified the same mutation in several members of 6 unrelated European families, all of whom had the same symptoms and showed very characteristic lesions—called sarcoplasmic bodies—in [muscle biopsies](#)."

Myoglobinopathy, the name that has been proposed for the new disease, manifests between the fourth and fifth decade of life. It causes a progressive weakness of the axial and limb muscles, and in more advanced stages, affects the respiratory musculature and the heart.

The infrared microscopy of the MIRAS beamline at the synchrotron, which is able to characterize the [chemical composition](#) and structure of proteins, allowed the researchers to demonstrate the presence of oxidized lipids in the damaged [muscle](#) cells. "We have observed that sarcoplasmic bodies correspond to oxidized lipids and misfolded proteins," says Nuria Benseny, a postdoctoral researcher at the ALBA synchrotron.

Finally, researchers from the universities of Modena and Vienna identified an alteration in the biochemical and [thermodynamic properties](#) of the mutated myoglobin, including an alteration in the oxygen-binding capacity that contributes to the formation of the lipid and protein aggregates that are found in the muscles of the patients. This discovery

has allowed the diagnosis for some patients after many years, and will allow researchers to focus on finding a cure for this disease.

**More information:** Montse Olivé et al. Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions, *Nature Communications* (2019). DOI: 10.1038/s41467-019-09111-2 , [www.nature.com/articles/s41467-019-09111-2](https://www.nature.com/articles/s41467-019-09111-2)

Provided by IDIBELL-Bellvitge Biomedical Research Institute

Citation: New muscular disease myoglobinopathy described (2019, March 27) retrieved 20 April 2024 from <https://medicalxpress.com/news/2019-03-muscular-disease-myoglobinopathy.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.