Mitochondrial medicine

RESEARCH ARTICLES

**TFE3 regulates whole-body energy metabolism in cooperation with TFEB**
Nunzia Pastore, Andrea Ballabio and colleagues

TFE3 plays a critical role in the metabolic response to environmental cues by regulating glucose homeostasis, lipid metabolism and mitochondrial dynamics.
EMBO Molecular Medicine Published online 10.03.2017
DOI: 10.15252/emmm.201607204

**CoQ deficiency causes disruption of mitochondrial sulfide oxidation, a new pathomechanism associated with this syndrome**
Marta Luna-Sánchez, Luis C López and colleagues

Disruption of the mitochondrial hydrogen sulfide oxidation pathway is identified as a new pathomechanism associated with primary CoQ deficiency. These findings may help explain the clinical heterogeneity of this syndrome.
EMBO Molecular Medicine Published online 17.11.2016
DOI: 10.15252/emmm.201606345

**Coenzyme Q deficiency causes impairment of the sulfide oxidation pathway**
Marcello Ziosi, Catarina M Quinzii and colleagues

Coenzyme Q (CoQ) is an electron acceptor for sulfide-quinone reductase (SQRI), the first enzyme of the hydrogen sulfide oxidation pathway. Lack of CoQ is here shown to cause impairment of hydrogen sulfide oxidation in vitro and in vivo.
EMBO Molecular Medicine Published online 17.11.2016
DOI: 10.15252/emmm.201606356

**Modified Atkins diet induces subacute selective ragged-red-fiber lysis in mitochondrial myopathy patients**
Sofia Ahola, Anu Suomalainen and colleagues

High-fat, low-carbohydrate modified Atkins diet (mAD) is a common weight-loss method, found to ameliorate mitochondrial myopathy in mice. In human patients, mAD induces muscle damage, especially of ragged-red fibers, the most affected by the disease.
EMBO Molecular Medicine Published online 19.09.2016
DOI: 10.15252/emmm.201606592

**Coenzyme A corrects pathological defects in human neurons of PANK2-associated neurodegeneration**
Daniel I Orellana, Sonia Levi and colleagues

Mutations in PANK2 cause PANK disease. This belongs to a group of disorders characterized by progressive neurodegeneration and excessive iron deposition in the brain. PANK2 enzyme catalyzes the first step in CoA synthesis. iPSC-derived neurons from PANK2 patients display abnormal phenotypes.
EMBO Molecular Medicine Published online 11.08.2016
DOI: 10.15252/emmm.201606391

**SLC25A46 is required for mitochondrial lipid homeostasis and cristae maintenance and is responsible for Leigh syndrome**
Alexandre Janer, Eric A Shoubridge and colleagues

Whole-exome sequencing in a Leigh syndrome patient identified mutations in SLC25A46, a degenerate member of the mitochondrial metabolite transport family, linking altered mitochondrial dynamics to early-onset neurodegenerative disease.
EMBO Molecular Medicine Published online 07.07.2016
DOI: 10.15252/emmm.201506159

**Reduction in mitochondrial iron alleviates cardiac damage during injury**
Hsiang-Chun Chang, Hossein Ardehali and colleagues

Modulation of mitochondrial iron is shown to be a viable therapeutic approach against ischemic heart disease and heart failure, highlighting the need to develop more targeted iron chelators.
EMBO Molecular Medicine Published online 19.02.2016
DOI: 10.15252/emmm.201505748

**Defective PITRM1 mitochondrial peptidase is associated with Aβ amyloidotic neurodegeneration**
Dario Brunetti, Laurence A Bindoff

A clinically peculiar neurodegenerative disorder in humans was indentified and shown to be caused by a pathogenic homozygous mutation in PITRM1, encoding an oligopeptidase of the mitochondrial inner compartment. The neuropathology of a PITRM1−/− mouse provides genetic evidence that Aβ is present within mitochondria, and demonstrates a link between impaired PITRM1 activity and Aβ amyloidotic neurodegeneration in mammals.
EMBO Molecular Medicine Published online 23.12.2015
DOI: 10.15252/emmm.201505894

REVIEWS

**Mitochondrial disease in adults: what’s old and what’s new?**
Patrick F Chinnery
EMBO Molecular Medicine Published online 26.11.2015
DOI: 10.15252/emmm.201505079

**Mitochondrial disorders in children: toward development of small-molecule treatment strategies**
Werner JH Koopman, Julien Beyrath, Cheuk-Wing Fung, Saskia Koene, Richard J Rodenburg, Peter HGM Willems, Jan AM Smeitink
EMBO Molecular Medicine Published online 07.03.2016
DOI: 10.15252/emmm.201506131

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