IN BRIEF

NEURODEGENERATIVE DISEASE

New insights into CMT2A

Zhou et al. *J. Clin. Invest.* **129**, 1756–1771 (2019)

Charcot-Marie-Tooth disease type 2A (CMT2A) is a neurodegenerative disease caused by mutations in the gene encoding mitofusin-2 (MFN2), a GTPase involved in mitochondrial dynamics. Although MFN2 is ubiquitously expressed, CMT2A preferentially affects the nervous system. According to a new study in mice, the low expression of MFN1 in the nervous system explains the tissue specificity of the disease.

The investigators generated transgenic mice that overexpress a mutated form of the human MFN2 protein specifically in neurons. *Thy1.2–MFN2*^{R94Q} mice showed common clinical features of CMT2A, including sensorimotor deficits and vision loss, axonal degeneration as well as cytoplasmic and axonal mitochondrial accumulation. Crossing *Thy1.2–MFN2*^{R94Q} mice with mice that overexpress human MFN1 in the nervous system (*PrP-MFN1*) rescued the CMT2A phenotype. Augmenting MFN1 in the nervous system could therefore be a viable strategy to treat CMT2A. *ALB*

https://doi.org/10.1038/s41684-019-0297-7

GENOMICS

A new model for Stargardt disease?

Mäkeläinen et al. *PLoS Genet.* **15**, e1007873 (2019)

Approximately 1 in 8,000–10,000 people are affected by Stargardt disease (STGD), an autosomal recessive retinal degenerative disease leading to visual impairment. STGD is caused by mutations in the *ABCA4* gene, which encodes a membrane transporter protein expressed by photoreceptors. No treatment exists and only mouse models are available to study the disease. For over a decade, researchers have been trying to identify a canine model because unlike mice, dogs have a macula, the part of the retina primarily affected in patients with STGD.

Recently, whole-genome sequencing performed on a family quartet of dogs—two offspring showing clinical signs similar to human STGD and their unaffected parents—led to the identification of a loss-of-function mutation in the *ABCA4* gene, which could be used to develop a large animal model for human STGD. *ALB*

https://doi.org/10.1038/s41684-019-0298-6

IMAGING

3D imaging of the sheep uterus

Wu et al. *Sci. Transl. Med.* **11**, eaau1428 (2019)

Preterm babies are at high risk of neonatal mortality and long-term neurological morbidity; understanding the changes in human uterine electrophysiology during preterm and term labor could inform strategies to prevent preterm labor. Techniques currently used to monitor uterine contractions are either invasive or can only measure limited uterine areas. A study describes the development of a new technique, electromyometrial imaging (EMMI) that combines body-surface electrical recording with body-uterus geometry acquired with MRI to create electrical maps of the entire 3D uterus. EMMI safely, noninvasively and accurately imaged electrical activity of the uterus during contractions in sheep. In the future, EMMI might become a resource to measure uterine contractility in humans. *ALB*

https://doi.org/10.1038/s41684-019-0299-5

GENETICS

Repertoires of circRNAs in mammals

Ji et al. *Cell Rep.* **26**, 3444–3460 (2019)

Emerging evidence suggests that circular RNAs (circRNAs), a newly discovered class of noncoding RNAs, regulate many biological and pathological processes such as development, atherosclerosis and cancer. Advances in deep sequencing have identified numerous novel circRNAs and spurred interest in these molecules. However most databases are not comprehensive because they were established from only one cell-type, tissue or species, and fail to identify functionally important candidates. A new large-scale study of circRNAs repertoires from multiple tissues from human, macaque and mouse identified thousands of evolutionary conserved circRNAs and systematically elucidated their diversity in various tissues. These datasets, which are available online (http://circatlas.biols.ac.cn/), allowed the investigators to infer circRNA functions on a global scale and to prioritize promising functional candidates. *ALB*

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