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LES SÉMINAIRES DE L'INMG

Actinopathies : From Mutations to Treatment

Par

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Salle des Conférences
Médiathèque Paul Zech
Faculté de Médecine Lyon Est
8, Avenue Rockefeller
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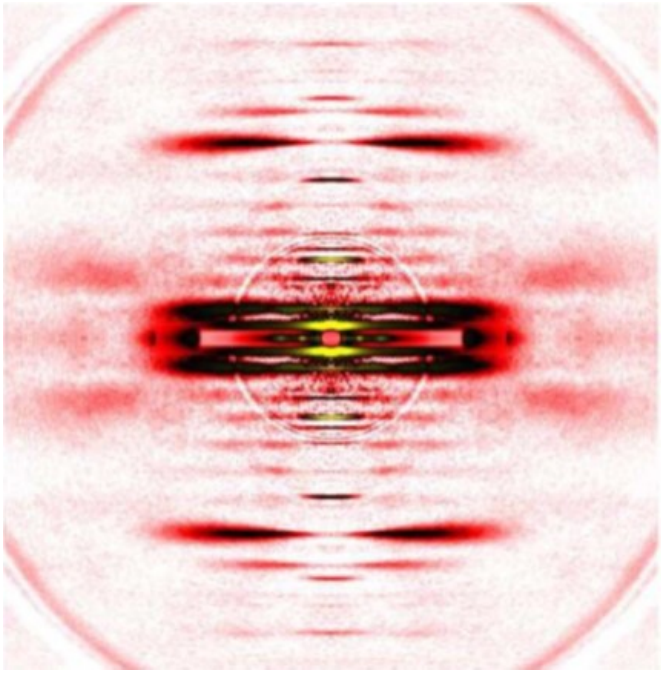
Abstract:

Actinopathies are genetically and clinically heterogeneous disorders mainly characterized by generalized muscle weakness. The understanding of this group of disorders has advanced in recent years through the identification of the causative mutations in the gene encoding one of the major proteins of the basic contractile unit of skeletal muscle, i.e., actin. In the present seminar, I will present (i) how these gene mutations lead to generalized muscle weakness and (ii) the advances regarding potential therapies.



Typical x-ray diffraction pattern from contracting skeletal muscle fibres.

Publications:

Chan C, Fan J, Messer AE, Marston SB, Iwamoto H, Ochala J. (2016) Myopathy-inducing mutation H40Y in ACTA1 hampers actin filament structure and function. Biochim Biophys Acta. 1862: 1453-1458.

Lindqvist J, Levy Y, Pati-Alam A, Hardeman EC, Gregorevic P, Ochala J. (2016) Modulating myosin restores muscle function in a mouse model of nemaline myopathy. Ann Neurol. In press.

Ochala J, Sun YB. (2016) Novel myosin-based therapies for congenital cardiac and skeletal myopathies. J Med Genet. In press

Gokhin DS, Ochala J, Domenighetti AA, Fowler VM. (2015) Tropomodulin 1 directly controls thin filament length in both wild-type and tropomodulin 4-deficient skeletal muscle. Development. 142: 4351-4362.