

GABBA Programme

2018 – NEURODEGENERATIVE DISEASES

April 4-6, i3S

Coordination – Saraiva, MJ (IBMC)

Faculty – Outeiro T (Gottingen)

Gomes Pereira M (Paris)

Saraiva MJ (i3S)

April 4

9.30- 11.00 a.m. Outeiro, T. Biology of alpha-synuclein in health and disease

11.30 -13.00 a.m. Outeiro,T. Spreading of synuclein pathology: is it a prion?

2.30 – 4.00 p.m. Gomes Pereira M. Unstable DNA repeats and human disease

4.30 – 6.00 p.m. Gomes Pereira M. From expanded DNA repeats to cell dysfunction: main trends in toxic RNA diseases

April 5

11.00 – 1 p.m. Outeiro, T. Journal Club in PD

3-5-p.m - Gomes-Pereira, M. Journal Club on MD

April 6

10-1 p.m. Saraiva MJ. Unfolding Transthyretin-related diseases

4-6 p.m. Saraiva MJ. Journal Club on TTR.

List of papers for classes:

Review references on MD:

1. Zhao et al. *The Repeat Expansion Diseases: The dark side of DNA repair (2006)*. **DNA repair**, 32: 96-105.
2. Nelson, Orr and Warren. *The Unstable Repeats—Three Evolving Faces of Neurological Disease (2013)*. **Neuron**, 77: 825-843
3. Zhang and Ashizawa. *RNA toxicity and foci formation in microsatellite expansion diseases (2017)*. **Current Opinion in Genetics and Development**, 44: 17-29

Review references on PD:

1. Lázaro et al., 2017. Cellular models as tools for the study of the role of alpha-synuclein in Parkinson's disease. **Exp. Neurology**.
2. Dehay et al., (2015). Targeting α -synuclein for treatment of Parkinson's disease: mechanistic and therapeutic considerations. **Lancet Neurol** 14: 855-66.

Review references TTR amyloidoses

Nuvolone M & Merlini G. 2017. Emerging therapeutic targets currently under investigation for the treatment of systemic amyloidosis. **Expert Opinion on Therapeutic Targets**, 21:12, 1095-1110

List of papers for seminars

On MD:

1. Wang et al., 2017. Reduced cytoplasmic MBNL1 is an early event in a brain-specific mouse model of myotonic dystrophy. *Human Molecular Genetics* Vol. 26, No. 12 2247–2257.
2. Zou, T. 2017. RAN Translation Regulated by Muscleblind Proteins in Myotonic Dystrophy Type 2. *Neuron* 95, 1292–1305

On PD:

1. Ordonez, D., 2018. α -synuclein Induces Mitochondrial Dysfunction through Spectrin and the Actin Cytoskeleton. *Neuron* 97, 108–124.
2. Abdelmotilib H et al., 2017- α -Synuclein fibril-induced inclusion spread in rats and mice correlates with dopaminergic Neurodegeneration. *Neurobiology of Disease* 105: 84–98

On TTR Amyloidoses:

1. Gonçalves, NP, et al., 2016. Overexpression of Protocadherin-10 in Transthyretin-Related Familial Amyloidotic Polyneuropathy. *The American Journal of Pathology* 186:1913-1924.
2. Gonçalves NP, et al., 2016. Tissue remodeling after interference RNA mediated knockdown of transthyretin in a Familial Amyloidotic Polyneuropathy mouse model. *Neurobiology of Aging*. 47: 91-101.
3. Panayiotou E., 2017. C1q ablation exacerbates amyloid deposition: a study in a transgenic mouse model of ATTRV30M amyloid neuropathy. *Plos One*, i13;12(4):e0175767s.