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## **LES SÉMINAIRES DE L'INMG**

# *Spinal Muscular Atrophy: a multi-organ disease*

par

## **Rashmi KOTHARY**

*(Invité par Patrick LOMONTE)*

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<https://med.uottawa.ca/neuromuscular/>

**Jeudi 27 Septembre 2018  
11 heure**

**Amphi 3  
Faculté de Médecine Lyon Est  
3e étage  
8, Avenue Rockefeller  
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**Abstract :**

Spinal Muscular Atrophy was characterized in the late 1800s but it was almost 100 years later that the genetic cause was identified as a mutation in the human SMN1 gene. While humans do have two genes that code for SMN, SMN2 only produces 10% protein when compared to SMN1. Early work with mouse models was complicated by the fact that mice only have the one Smn gene which when mutated results in preimplantation lethality. This problem was solved by the development of a mouse model incorporating human SMN2 gene, which was for years the only viable mouse model. Now, there are over a hundred mouse models available that have served to inform our understanding of pathogenesis in SMA.

Classically, SMA is described as a motor neuron disease; however, SMN is expressed ubiquitously throughout the body. In fact, SMA is emerging to be a multi-organ disease with SMN depletion having impacts on many tissues in the body. Subcutaneous administration of available treatments may address these affected organs better than intrathecal administration. Also, systemic gene therapies that are currently under development may help repair function in these other organs.

Dr. Kothary will present on the work in his laboratory on the multi-organ nature of SMA. Defects in various tissues will be discussed.