

## Publication

## "Get the Balance Right": Pathological Significance of Autophagy Perturbation in Neuromuscular Disorders

**JournalArticle (Originalarbeit in einer wissenschaftlichen Zeitschrift)****ID** 3660462**Author(s)** Castets, Perrine; Frank, Stephan; Sinnreich, Michael; Rüegg, Markus A.**Author(s) at UniBasel** [Rüegg, Markus A.](#) ; [Castets, Perrine](#) ; [Sinnreich, Michael](#) ;**Year** 2016**Title** "Get the Balance Right": Pathological Significance of Autophagy Perturbation in Neuromuscular Disorders**Journal** Journal of Neuromuscular Diseases**Volume** 3**Number** 2**Pages / Article-Number** 127-155

Recent research has revealed that autophagy, a major catabolic process in cells, is dysregulated in several neuromuscular diseases and contributes to the muscle wasting caused by non-muscle disorders (e.g. cancer cachexia) or during aging (i.e. sarcopenia). From there, the idea arose to interfere with autophagy or manipulate its regulatory signalling to help restore muscle homeostasis and attenuate disease progression. The major difficulty for the development of therapeutic strategies is to restore a balanced autophagic flux, due to the dynamic nature of autophagy. Thus, it is essential to better understand the mechanisms and identify the signalling pathways at play in the control of autophagy in skeletal muscle. A comprehensive analysis of the autophagic flux and of the causes of its dysregulation is required to assess the pathogenic role of autophagy in diseased muscle. Furthermore, it is essential that experiments distinguish between primary dysregulation of autophagy (prior to disease onset) and impairments as a consequence of the pathology. Of note, in most muscle disorders, autophagy perturbation is not caused by genetic modification of an autophagy-related protein, but rather through indirect alteration of regulatory signalling or lysosomal function. In this review, we will present the mechanisms involved in autophagy, and those ensuring its tight regulation in skeletal muscle. We will then discuss as to how autophagy dysregulation contributes to the pathogenesis of neuromuscular disorders and possible ways to interfere with this process to limit disease progression.

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