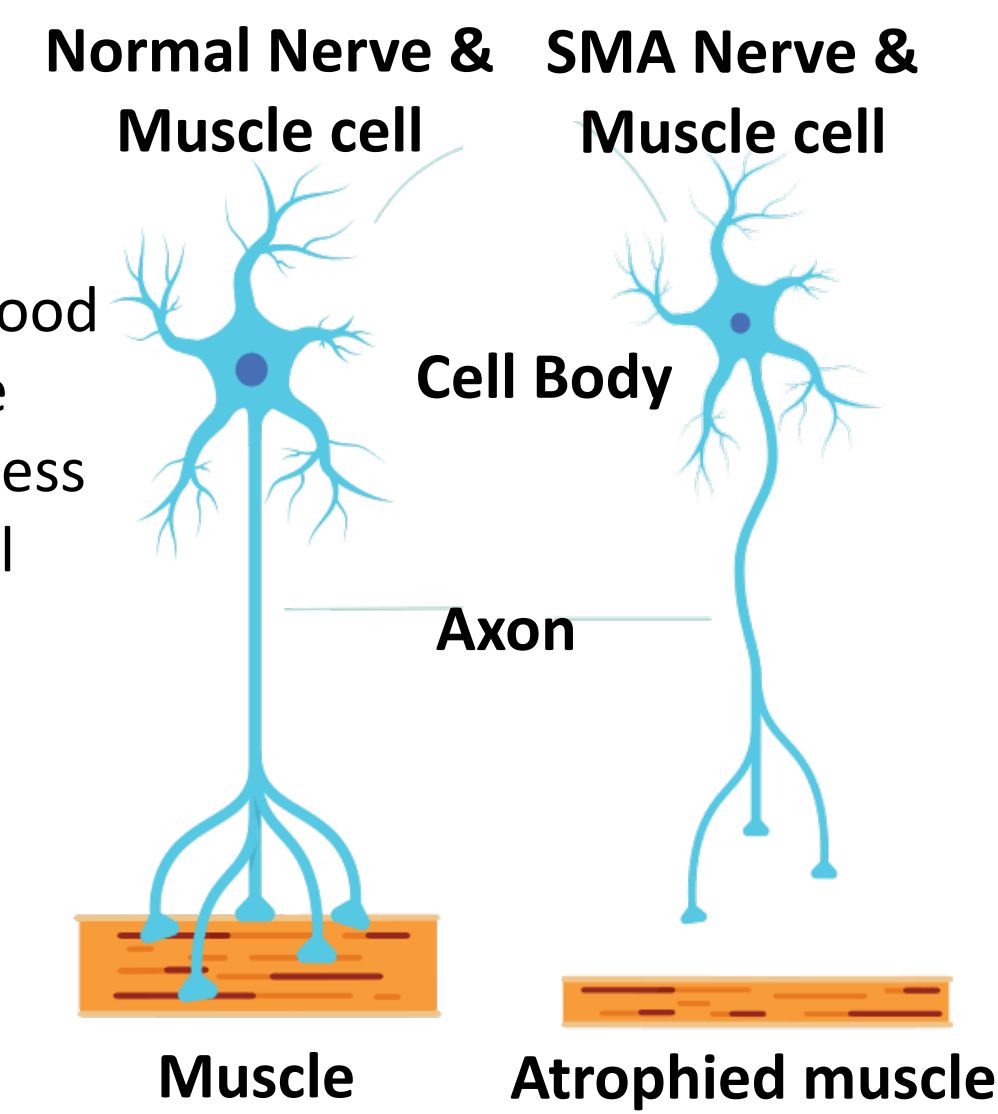
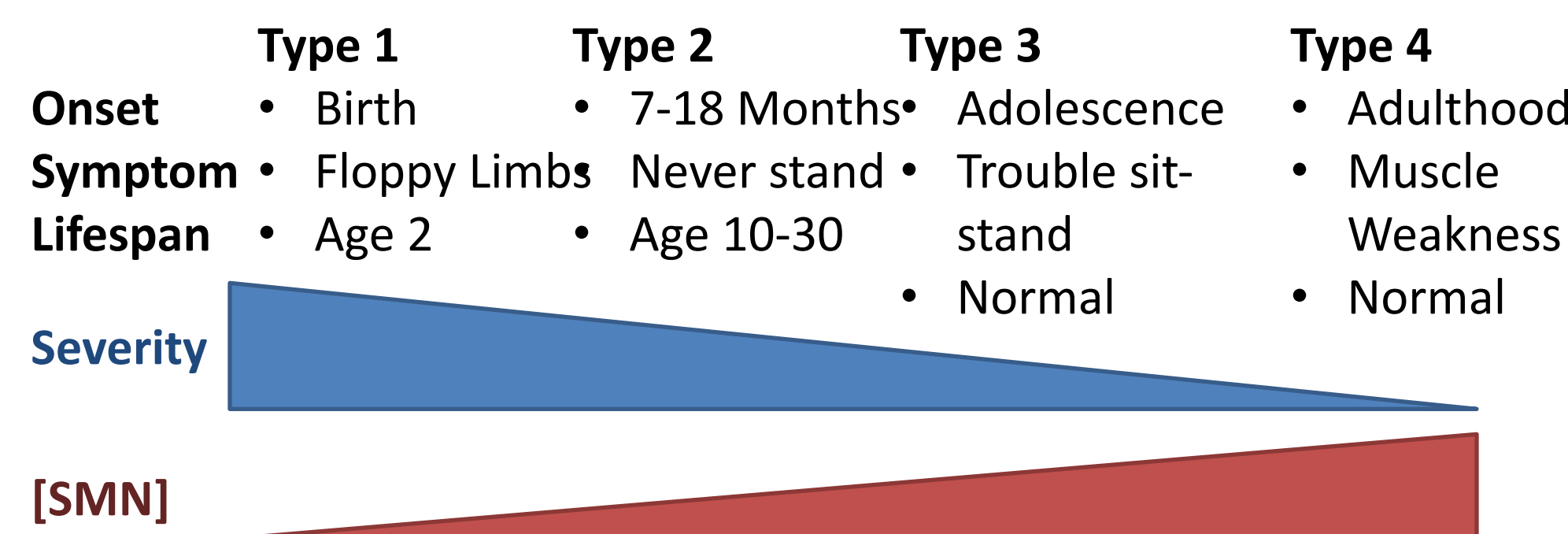
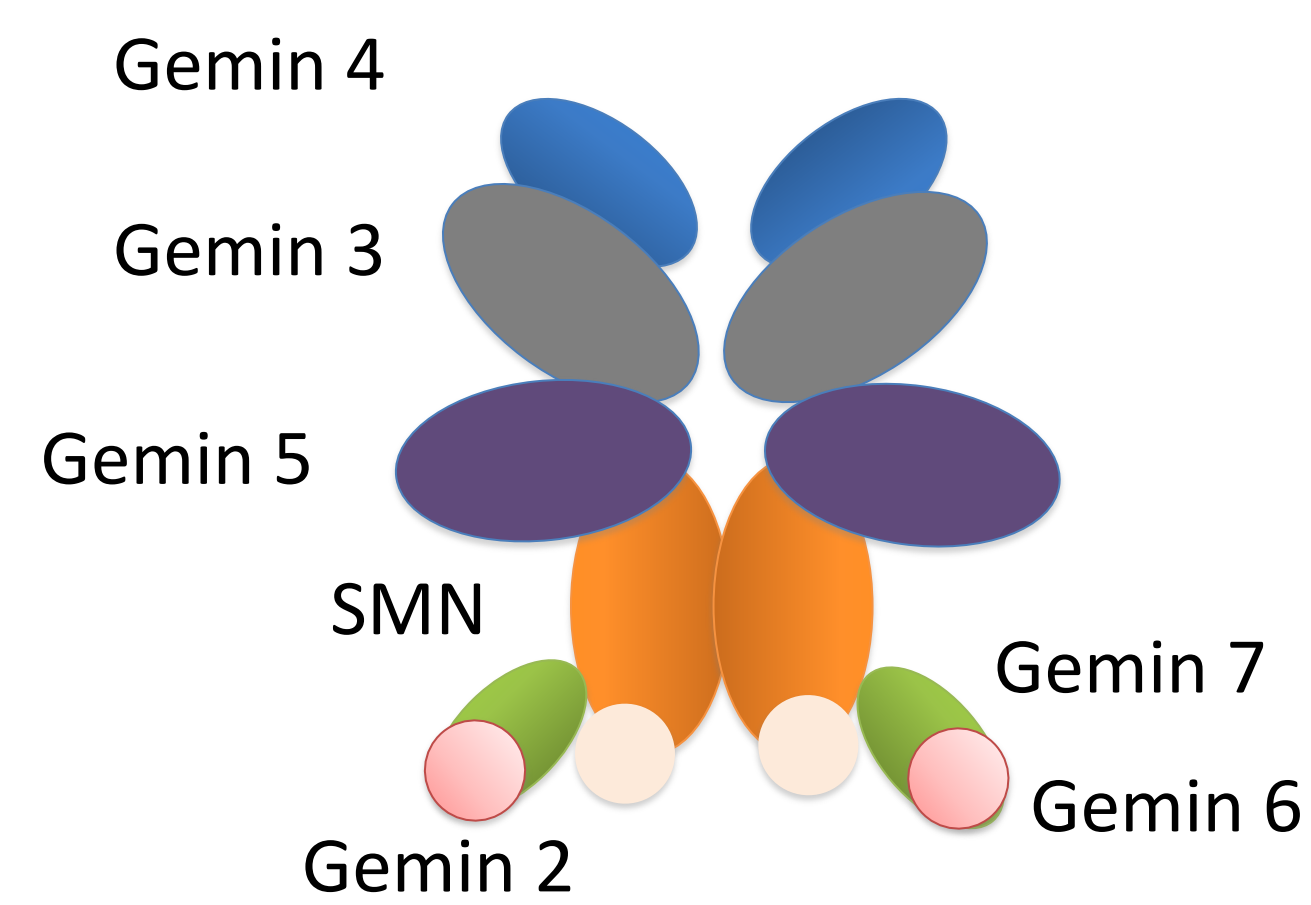


Introduction

Spinal Muscular Atrophy



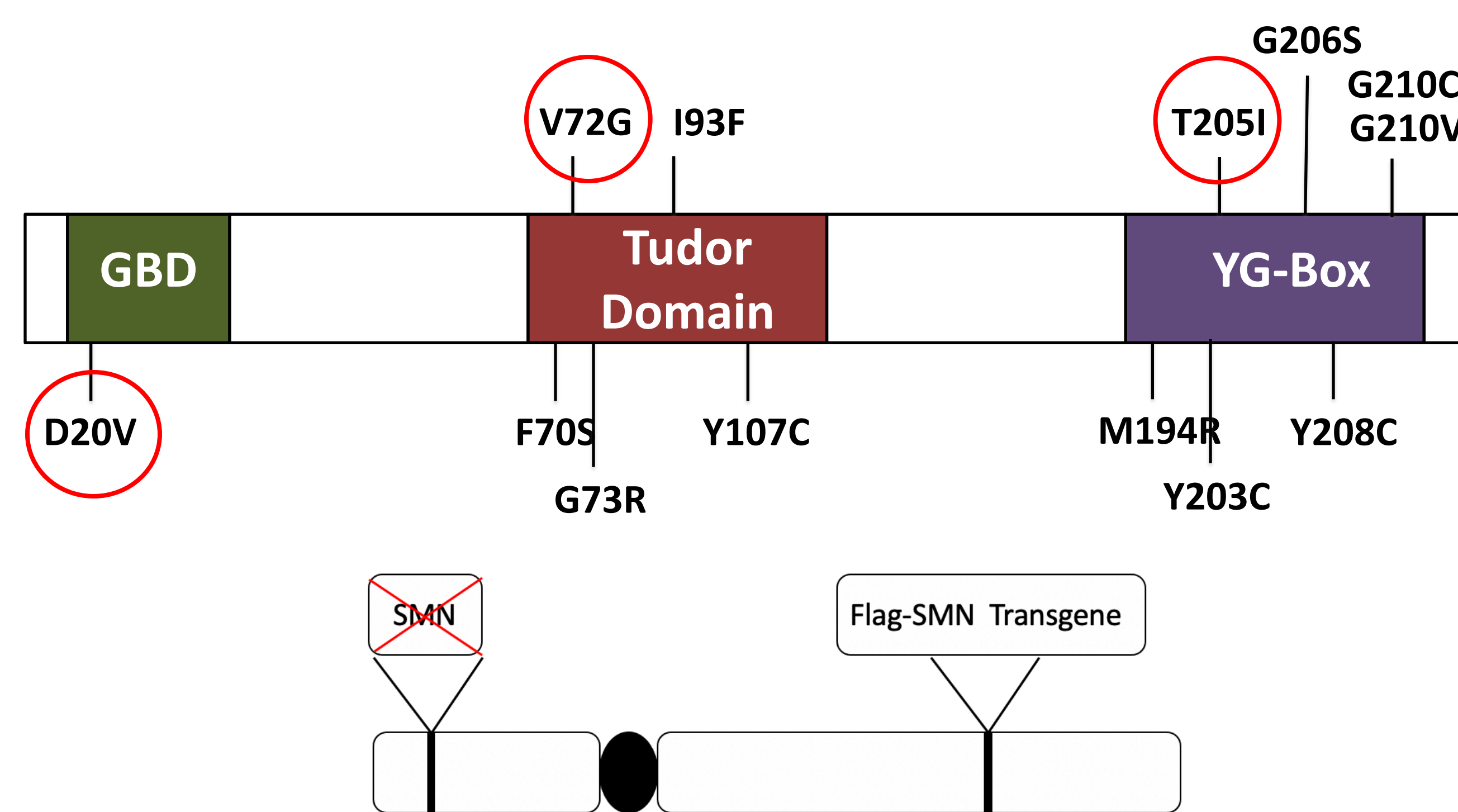
Survival Motor Neuron Complex¹



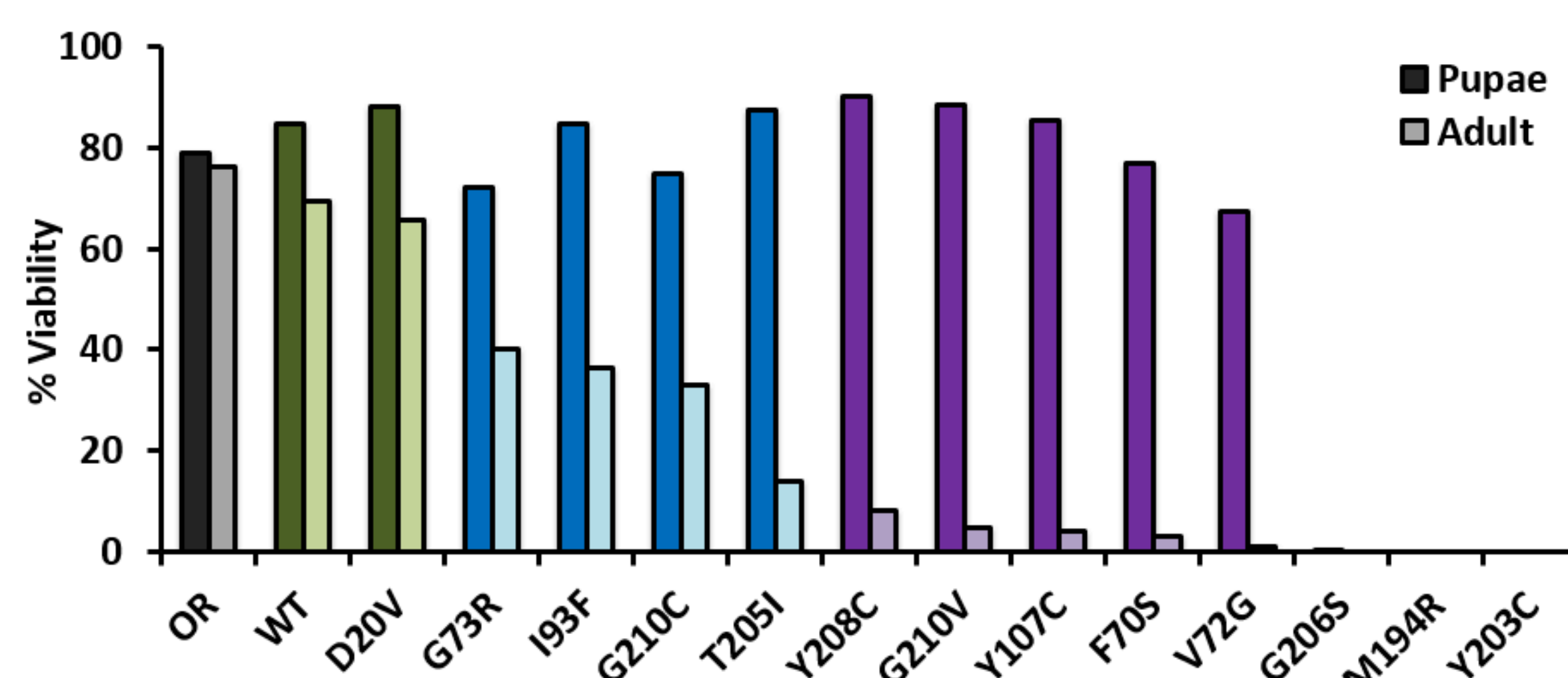
SMN Complex functions

- snRNP biogenesis.
- Protein Homeostasis
- Splicing
- Translation
- Immune Response
- Cell Trafficking

Human patient mutations in *Drosophila melanogaster*

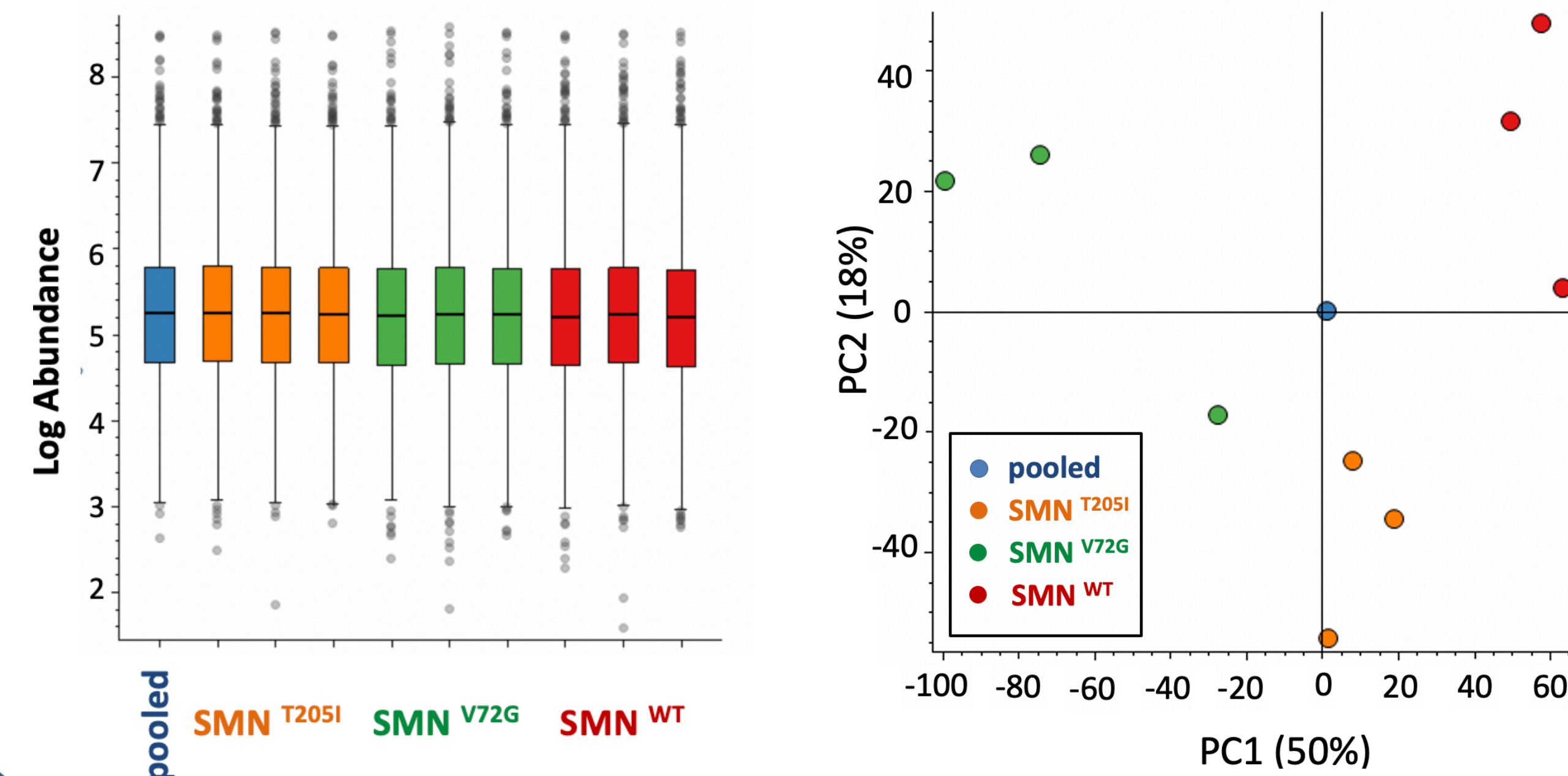


SMA mutations cause a spectrum of disease severities?



Methods

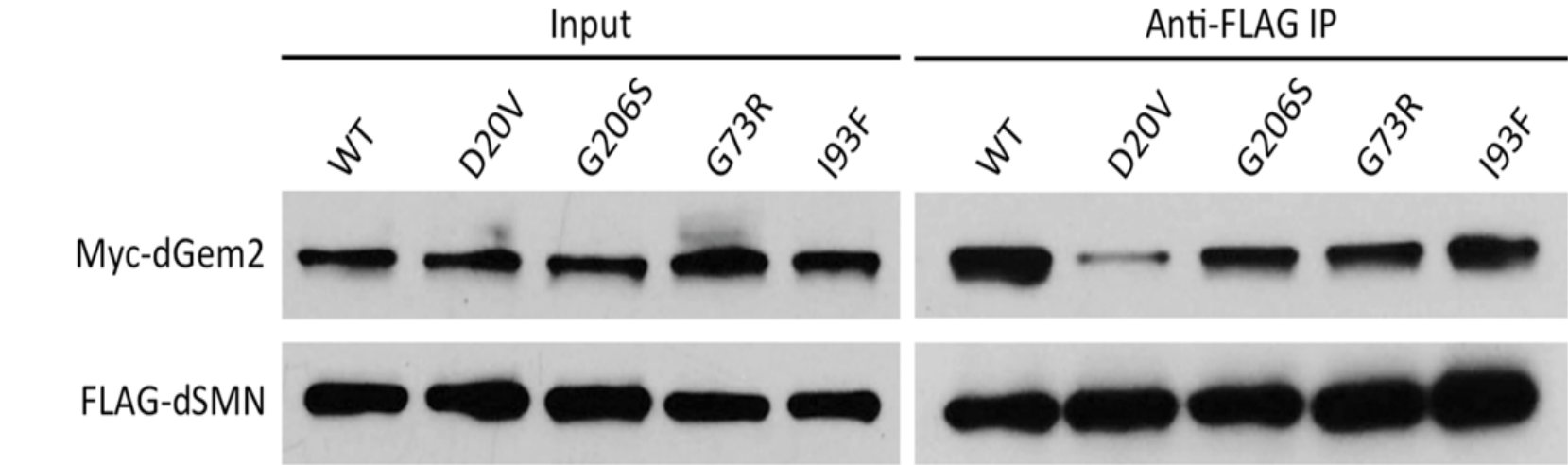
Tandem Mass Tagging – Mass Spectrometry



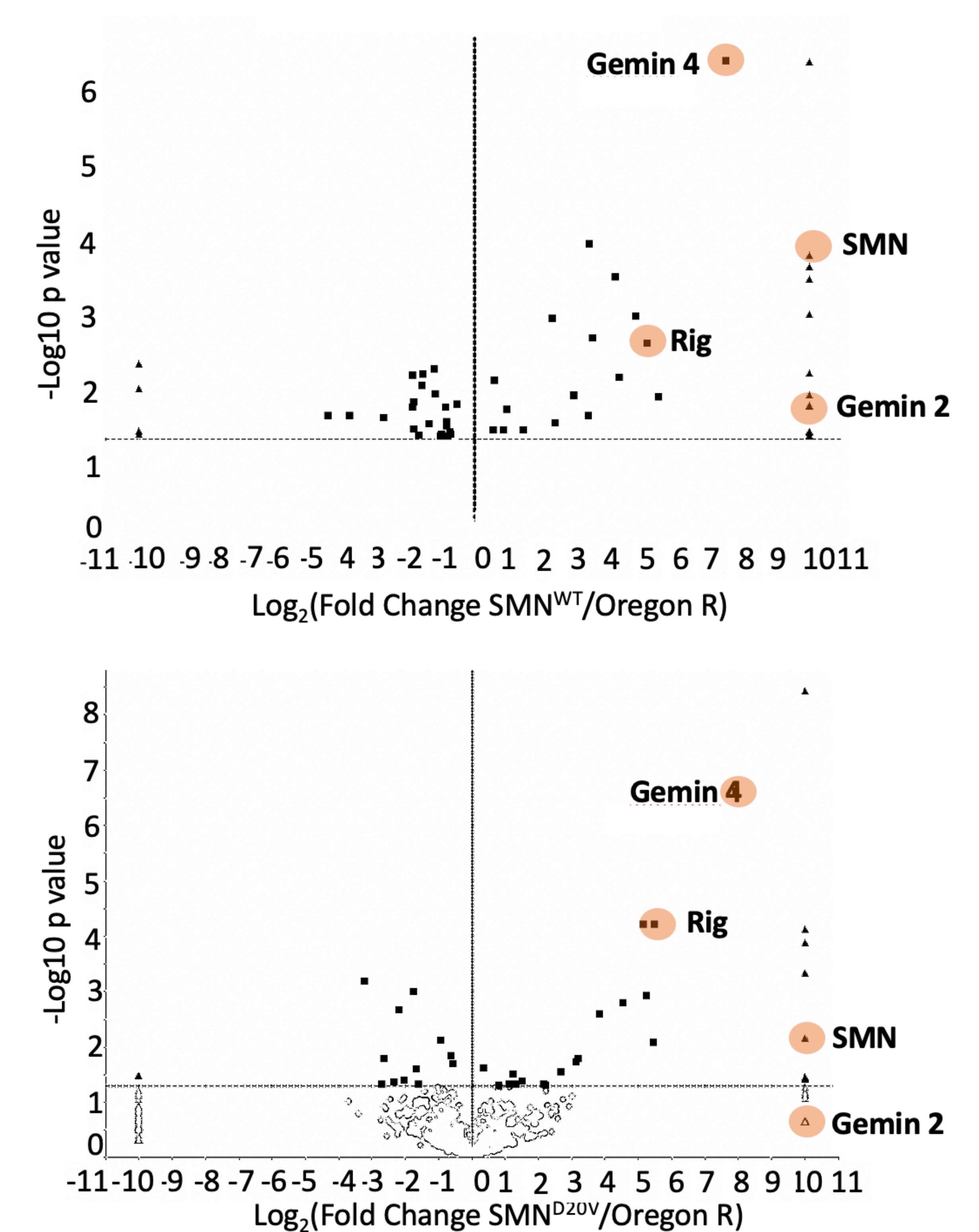
SMN Complex

Reduced Gemin2 Binding in SMN^{D20V}

In vitro³

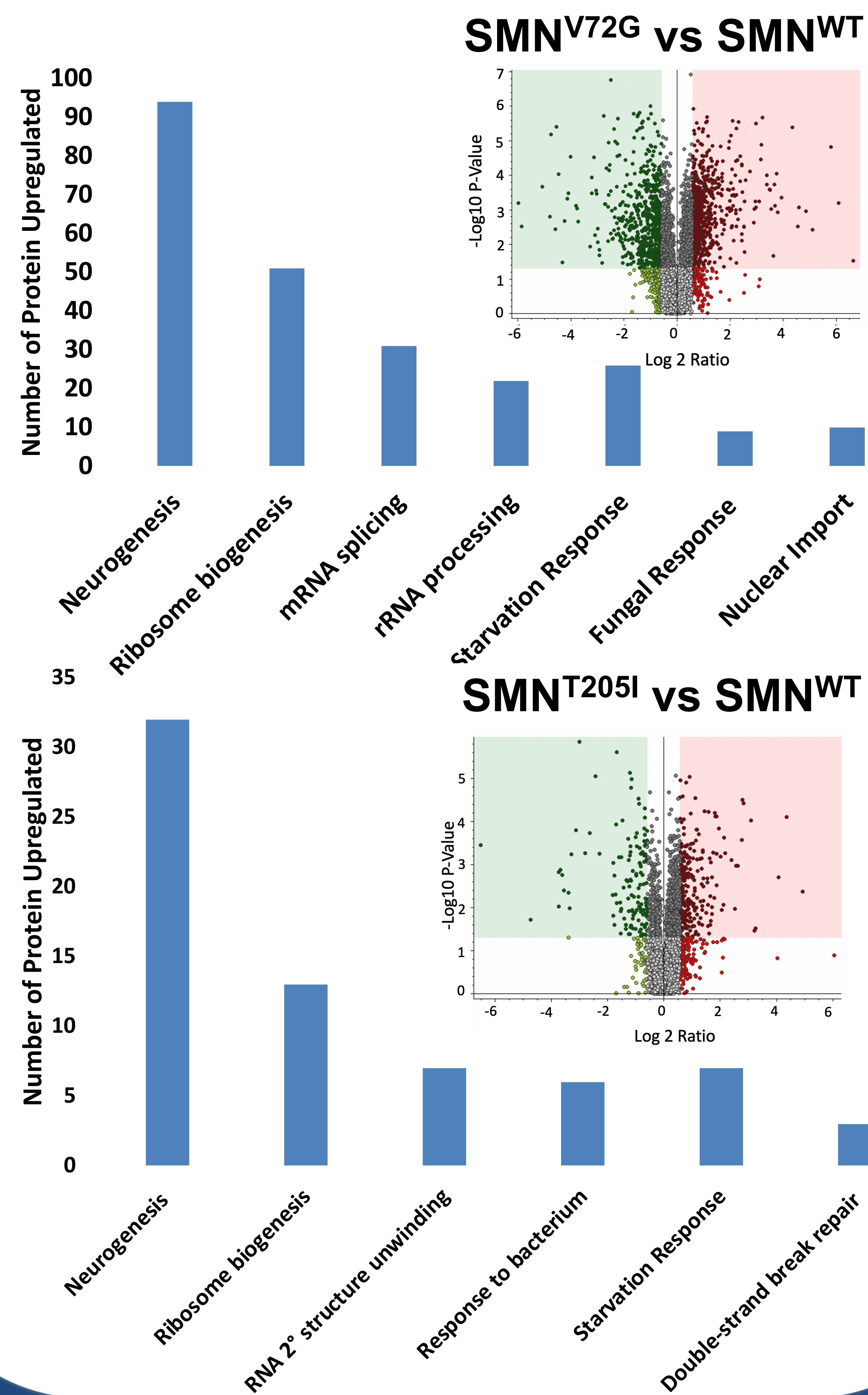


In vivo



Proteome

Functions of upregulated proteins in SMN mutants



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References

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- ³Praveen, K., Wen, Y., Gray, K.M., Noto, J.J., Patiolla, A.R., Van Duyne, G.D. Matera, A.G. *Plos Genetics*. 2014, 10(8), e1004489