Acknowledgements
The nature of our mind leads us to seek the essence or the why of things

Descriptive anatomy is to physiology what geography is to history, and just as it is not enough to know the typography of a country to understand its history, so also it is not enough to know the anatomy of organs to understand their functions.

Claude Bernard (1813-1878)

More than six years has passed since I became a PhD student and moved to Amsterdam. Time has flown as it has been very busy and intense; filled with data collections, courses, reflections, preoccupations, writings, discussions and perseverance. In learning about sciences, I have learned about myself and have grown up. I have always had a passion for the human body and how the organs are working together. I really enjoy to discover and try to figure it out what is happening when the skeletal muscle is sick. How each molecule interacts with each other. The results of this work will contribute to the knowledge and understanding of Nemaline myopathy to finally find a cure one day.

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CURRICULUM VITAE

Barbara was born in 1985, in Les Lilas in Paris’ suburbs, France.

After finishing her Bachelor’s degree in Life Sciences at Pierre et Marie Curie University (UPMC, Paris VI, France), Barbara started a Master’s degree in Physiology and Integrative Biology, specializing in Physiology and Pathophysiology. During her first Masters internship, she took a particular interest in basic research by studying the genetic and cardiac pathophysiology of the Brugada syndrome under the supervision of Dr. N. Neyroud (INSERM UMRS 956, La Pitié-Salpêtrière hospital, Paris).

Her interest in research continued into her second Masters internship with clinical studies on dynamic ventilatory mechanics at rest and during exercise in patients with bronchial asthma, chronic obstructive pulmonary disease or pulmonary arterial hypertension under the supervision of Pr. T. Similowski and Dr. P. Laveneziana (ER 10, UPMC, La Pitié-Salpêtrière hospital, Paris).

In November 2012, Barbara moved to Amsterdam to start her PhD at the department of Physiology of the VUMC under the supervision of the Prof.dr. Coen Ottenheijm. The project was on muscle weakness in Nemaline myopathy patients, of which the results are presented in this thesis.
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