

**BIOGRAPHICAL SKETCH**

Provide the following information for the Senior/key personnel and other significant contributors.  
Follow this format for each person. **DO NOT EXCEED FIVE PAGES.**

NAME: **Valeria Di Leo**

eRA COMMONS USER NAME (credential, e.g., agency login):

POSITION TITLE: **Post Doctoral Research**

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable. Add/delete rows as necessary.)*

INSTITUTION AND LOCATION	DEGREE <i>(if applicable)</i>	Start Date MM/YYYY	Completion Date MM/YYYY	FIELD OF STUDY
University of Bari (Italy)	Undergrad	09/2011	07/2014	Bachelor of Science in Medical and Pharmaceutical Biotechnology
University of Trieste (Italy)	MSc	09/2015	03/2017	Master of Science in Functional Genomics
Newcastle University (UK)	PhD	10/2018	04/2022	Doctor of Philosophy in Translational and Clinical Research
Newcastle University (UK)	PostDoc	05/2022	ongoing	Neuromuscular disorders and mitochondrial dysfunction

**A. Personal Statement**

I am a motivated young researcher with six years of experience in the research field of neuromuscular disorders. I have secured independent funding throughout my career, including scholarships to support my studies, travel grants to attend international conferences and research grants to invest in my research interests. This year, I have secured nearly £175,000. I am a regular organizer of public and patient engagement events with Lily Foundation and the Wellcome Centre for Mitochondrial Research. In September 2023, I will run half a marathon (21k) fundraising money and awareness for Muscular Dystrophy UK.

Education and award

- **MDUK Research Grant** for 2 years (around £150,000 in total)
- **British Neuropathological Society** travel bursary to attend EUROMIT 2023 (£500)
- Nomination as “**Inspiring women**” for the International Women’s Day Event in Newcastle University
- **Wellcome Trust Translational Partnership Award** (£7000 in total) & **EMBO Scientific Exchange Grant** (€7,550 in total) to collaborate and visit the Institute of Bioengineering for Catalonia
- Presentation prize granted by **World Muscle Society** (Elsevier WMS Membership Award)
- **World Muscle Society Fellowship award** to attend WMS Congress 2022 (£1000 in total)
- **Guarantors of Brain** travel grant to attend WMS Congress 2022 (£750 in total)
- First prize for poster presentation awarded by **Wellcome Trust Translational Partnership** (£150 in total)
- Travel grant from **Newcastle University** to attend UMDF, Mitochondrial Medicine 2022 (£1,800 in total)
- **Doctor of Philosophy** in Translational and Clinical Research awarded within the Wellcome Centre for Mitochondrial Research, Newcastle University, UK (£60,000 in total)
- **Erasmus+ Traineeship scholarship** awarded from University of Trieste for 12 months (€10,000 in total)
- **Master of Science scholarship** awarded from University of Trieste (€10,000 in total)
- **Bachelor of Science scholarship** awarded from University of Bari (€5,000 in total)

**B. Positions, Scientific Appointments and Honors**

- 2023 – Visiting researcher at the Institute for Bioengineering of Catalonia, project supervised by Dr Fernandez-Costa

- 2022 - Postdoc Project: Professor Gorman, Dr Vincent, Wellcome Centre for Mitochondrial Research, Newcastle University, UK
- 2021 - Seminar co-leader/leader for undergraduate students' sessions
- 2021 - Undergraduate student demonstrator, invigilator, and examiner
- 2021 - ongoing - Reviewer in Experimental Physiology
- 2020 - Undergraduate student demonstrator, invigilator, and examiner
- 2018 – Doctor of Philosophy Project: Dr Vincent, Dr Russell, Dr Tuppen, Prof Gorman, Wellcome Centre for Mitochondrial Research, Newcastle University, UK
- 2017 - Traineeship and master project: Professor Straub laboratory, John Walton Muscular Dystrophy Research Centre, Newcastle University, UK

### C. Contributions to Science

During my MSc project, I investigated the co-segregation of *SRPK3* and *TTN* variants in congenital myopathy patients (manuscript accepted by Nature Genetics).

During my PhD project, I looked at the effects of resistance exercise training on mitochondrial dysfunction in mitochondrial myopathy and myotonic dystrophy type 1 (DM1) patients (manuscript under revision and in preparation). Thanks to the multi-level analysis performed, I acquired competence with many techniques, of which the most relevant involve immunofluorescence, imaging mass cytometry and RNA analysis (Menger et al. 2022).

During my current position as Research Associate, I am characterizing some novel therapeutic targets identified during my PhD project to provide insight for the cure of mitochondrial myopathy patients. To do this, in March 2023 I joined the Institute of Bioengineering of Catalonia (Spain) as a visiting researcher where, supervised by Dr Fernandez-Costa, I generated *in vitro* 3D models of mitochondrial myopathy of human skeletal muscles. Moreover, my research in the field of DM1 demonstrated that 12-week resistance exercise training induces ameliorations in oxidative phosphorylation deficiency in DM1 patients.

In the next few years as Research Associate, I will expand the investigation of mitochondrial signaling pathways in DM1 skeletal muscle biopsies to underpin the crosstalk between mitochondrial activity and DM1 pathogenesis. This will be supported by 2-years MDUK Research Grant and by the collaboration established with Prof Duchesne at University of Québec in Chicoutimi (Canada).

#### Manuscripts published:

Menger KE, Chapman J, Díaz-Maldonado H, Khazeem MM, Deen D, Erdinc D, Casement JW, Di Leo V, Pyle A, Rodríguez-Luis A, Cowell IG, Falkenberg M, Austin CA, Nicholls TJ. Two type I topoisomerases maintain DNA topology in human mitochondria. *Nucleic Acids Res.* 2022 Oct 10:gkac857. doi: 10.1093/nar/gkac857. Epub ahead of print. PMID: 36215039.

Töpf A, Zaharieva IT, Di Leo V, ..., Francesco Muntoni, Volker Straub. “Digenic inheritance involving a muscle specific protein kinase and the giant titin protein causes a skeletal muscle myopathy”. Paper accepted in *Nature Genetics*.

#### Manuscripts under revision:

Di Leo V, Lawless C, Roussel M-P, Gomes T.B, Gorman G.S, Russell O.M, Tuppen H.L.A, Duchesne E & Vincent A.E. “Resistance training rescues mitochondrial dysfunction in skeletal muscle of patients with myotonic dystrophy type 1”. Paper under second revision with *Journal of Neuromuscular Diseases*.

#### Manuscripts in preparation:

Di Leo V et al. “Resistance exercise training induces molecular changes in mitochondrial myopathy patients”. Manuscript in preparation.

### D. Scholastic Performance

YEAR	COURSE TITLE	GRADE
2022	Doctor of Philosophy	awarded
2018	Master of Science in Functional Genomics	110/110 cum laude
2015	Bachelor of Science in Medical and Pharmaceutical Biotechnology	100/110
2011	High School Classical Lyceum	98/100