ELENA PERENTHALER

Nationality: Italian
Researcher unique identifier: [redacted]

WORK EXPERIENCE

PHD CANDIDATE
AUG 2017 – DEC 2021 (4 years 5 months)
Barakat laboratory - Clinical Genetics department
Erasmus University Medical Center - Rotterdam (NL)
Main activities:
• Discovery and molecular characterization of a new DEE syndrome (OMIM # 618744) caused by a start-loss mutation in the brain-expressed isoform of the *UGP2* gene
• Investigation of the chromatin architecture of the *UGP2* locus
• High-throughput identification of functional enhancers in neural stem cells
• Investigation of YY1 interactome in human embryonic and neural stem cells
• Investigation of the molecular mechanism by which YY1 regulates gene expression

ERASMUS + RESEARCH INTERN
JAN 2017 - JUL 2017 (6 months)
Gillingwater laboratory - Centre for integrative physiology & Euan MacDonald centre for motor neurone disease research
University of Edinburgh (UK)
Main activity:
• Investigation of the temporal and tissue-specific variability of SMN protein levels in mouse models of spinal muscular atrophy

RESEARCH ASSISTANT
FEB 2015 – AUG 2015 (6 months)
Viero laboratory of translational architectomics
Institute of biophysics - National research Council (IBF-CNR; Italy)
Main activity:
• Investigation of the role of the RNA-binding protein CELF3 in axonal outgrowth and translation

EDUCATION

PhD IN BIOMEDICAL SCIENCES
ERASMUS UNIVERSITY MEDICAL CENTER, NETHERLANDS;
AUG 2017 – DEC 2021
Date of award: 20-09-2022
Thesis title: "Neurodevelopmental disorders: from genes to regulatory elements"

M.SC IN CELLULAR AND MOLECULAR BIOTECHNOLOGY - NEUROBIOLOGY STUDY TRACK
UNIVERSITY OF TRENTO, ITALY;
OCT 2014 - OCT 2016
Date of award: 25-10-2016
Thesis title: "In vivo translatome analyses reveal translational defects in early symptomatic Spinal Muscular Atrophy"

24/09/2022

B.SC IN BIOMOLECULAR SCIENCES AND TECHNOLOGY
UNIVERSITY OF TRENTO, ITALY
SEP 2011 - SEP 2014
Date of award: 26-09-2014
Thesis title: "A possible new role for the RNA binding protein CELF3 in translation regulation and neurites outgrowth"

LANGUAGES

Italian: native speaker
English: highy proficient - IELTS 8.0 (CEFR C1)

PRACTICAL SKILLS

Various cell and molecular biology techniques including:
- Cell culture, differentiation, transfection and genome editing;
- Immunofluorescence and confocal microscopy;
- DNA, RNA and protein purification;
- Subcellular fractionation;
- Cloning;
- Chromatin immunoprecipitation (ChIP-qPCR; ChIP-seq);
- Chromatin conformation capture (T2C; Hi-ChIP followed by sequencing);
- PCR and qPCR;
- RNA sequencing;
- Polysome and ribosome profiling (POL-seq; RIBO-seq);
- Western Blotting;
- Immunoprecipitation.

Microsoft Office Word, Excel and PowerPoint
Adobe Illustrator, InDesign and basic Photoshop

PERSONAL SKILLS

I thrive in collaborative environments that allow both social interactions and scientific discussions to share ideas, opinions and advices. In my previous experiences I worked in teams of people trained in various disciplines ranging from medicine to biology and bio-informatics, strengthening my communication skills.

At the time I started my PhD I was the first member of a newly formed lab. Even though it wasn't easy at first, my determination allowed me to grow faster as an independent scientist. Furthermore, my organizing skills contributed to build the foundation of a succesful lab, as it is now, 5 years later. During this period I worked on multiple projects further developing my time-management skills to adequately advance on all the different tasks, and to achieve the set goals within the established deadlines.

I have a keen interest for graphic design, hence I pay particular attention to the visual aspect of presentations and posters, and to the design of figures for papers. I enjoy creating clear graphics delivering an immediate message, without overlooking details. Writing a PhD thesis in the Netherlands allowed me to further fine-tune this skill, as it gave me the opportunity to design not only the interior layout of the thesis book but also a cover representing the key concepts explored in my work.

24/09/2022

AWARDS AND FUNDINGS

- Second prize for best poster at the 2019 MGC PhD Student Workshop, Maastricht, Netherlands
- Travel grant from the cost action CA16118 for the conference "Brain Malformations: A Roadmap for Future Research" at the Weizmann Institute of Science, Rehovot, Israel (17-20/03/2019; 816.71€)
- Erasmus+ for traineeship after graduation (09/01/2017 – 08/07/2017; 2880€)

PUBLICATIONS

Yousefi S, Deng R*, Lanko K*, Medico Salsench E*, Nikoncuk A*, van der Linde HC, **Perenthaler E**, van Ham TJ, Mulugeta E#, Barakat TS#. (2021) Comprehensive multi-omics integration identifies differentially active enhancers during human brain development with clinical relevance. Genome Med Oct 19;13(1):162.

Sanderson LE*, Lanko K*, Alsagob M*, Almass R*, Al-Ahmadi N*, Najafi M, Al-Muhaizea MA+, Alzaidan H+, AlDhalaan H+, **Perenthaler E**, ..., Schmidts M#, Barakat TS#, van Ham TJ#, Kaya N#. (2021) Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain 144(3):769-780

Barish S, Barakat TS, Michel BC, Mashtalir N, Phillips JB, Valencia AM, Ugur B, Wegner J, Scott TM, Bostwick B, Undiagnosed Diseases Network, Murdock DR, Dai H, **Perenthaler E**, ..., Bellen HJ. (2020) BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. Am J Hum Genet 07, 096-2

Verheul TCJ*, van Hijfte L*, **Perenthaler E***, Barakat TS. (2020) The Why of YY1: Mechanisms of Transcriptional Regulation by Yin Yang. Front Cell Dev Biol 8: 59264

Lauria F*, Bernabò P*, Tebaldi T*, Groen EJN*, **Perenthaler E**, Maniscalco F, Rossi A, Donzel D, Clamer M, Marchioretto M, Omersa N, Orri J, Dalla Serra M, Anderluh G, Quattrone A, Inga A, Gillingwater TH# and Viero G#. (2020) SMN-primed ribosomes modulate the translation of transcripts related to spinal muscular atrophy. Nat Cell Biol 22: 239-25

Perenthaler E, Nikoncuk A*, Yousefi S*, Berdowski WM*, Alsagob M*, ..., Barakat TS. (2020) Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. Acta Neuropathol 39: 45-442

Perenthaler E*, Yousefi S*, Niggl E*, Barakat TS. (2019) Beyond the Exome: The Non-coding Genome and Enhancers in Neurodevelopmental Disorders and Malformations of Cortical Development. Front Cell Neurosci 3, 352

Clamer M, Tebaldi T, Lauria F, Bernabò P, Gómez-Biagi RF, Marchioretto M, Kandala DT, Minati L, **Perenthaler E**, Gubert D, Pasquardini L, Guella G, Groen EJN, Gillingwater TH, Quattrone A, and Viero G. (2018) Active Ribosome Profiling with RiboLace. Cell Rep 25: 097-08 e095

Groen EJN, **Perenthaler E**, Courtney NL, Jordan CY, Shorrock HK, van der Hoorn D, Huang Y, Murray LM, Viero G, Gillingwater TH. (2018) Temporal and tissue-specific variability of SMN protein levels in mouse models of spinal muscular atrophy. Hum Mol Genet 27: 285-2862.

Barakat TS*, Halbritter F*, Zhang M+, Rendeiro AF+, **Perenthaler E**, Bock C, Chambers I. (2018) Functional Dissection of the Enhancer Repertoire in Human Embryonic Stem Cells. Cell Stem Cell 23: 276-288 e278

Bernabò P*, Tebaldi T*, Groen EJN*, Lane FM*, **Perenthaler E**, Mattedi F, Newbery HJ, Zhou H, Zuccotti P, Potrich V, Shorrock HK, Muntoni F, Quattrone A, Gillingwater TH, Viero G. (2017) In Vivo Translatome Profiling in Spinal Muscular Atrophy Reveals a Role for SMN Protein in Ribosome Biology. Cell Rep 2: 953-965

* these authors contributed equally
+ these authors contributed equally
# these authors jointly supervised this work

24/09/2022

WORKSHOPS AND CONFERENCES

2017 - 27th MGC symposium (Rotterdam - NL)
2017 - Dutch neurodevelopmental disorders day (Rotterdam - NL)
2018 - MGC workshop (Texel - NL; short presentation)
2018 - UCSC Gene Browsing workshop
2018 - Gene transcription in health and disease symposium (Rotterdam - NL)
2019 - MGC workshop (Maastricht - NL; poster)
2019 - Brain malformations: a roadmap for future research conference (Rehovot - IL; poster)
2019 - 29th MGC symposium (Rotterdam - NL)
2019 - ACE SBM and SCORE day (Rotterdam - NL)
2019 - Mini symposium CRISPR-Cas9 (Rotterdam - NL)
2020 - ESHG (virtual; presentation)
2021 - MGC workshop (virtual; presentation)
2021 - Boost your research career with a personal grant (virtual)
2021 - Neuro-MIG conference (virtual; presentation)
2021 - Sophia research day (virtual; presentation)
2021 - Brain prize meeting (virtual)
2021 - ESHG (virtual; presentation)
2022 - 30th MGC symposium (Leiden - NL; presentation)

ORAL AND POSTER PRESENTATIONS

ORAL PRESENTATIONS

• Characterization of the non-coding regulatory DNA in human cortex development and disease
2018 - MGC workshop (Texel - NL)

• Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform specific start-loss mutations of essential genes can cause genetic diseases
2020 - ESHG (virtual)

• Characterization of the functional enhancer repertoire in human neural stem cells
2021 - MGC workshop (virtual)

• Characterization of the functional enhancers in human neural stem cells
2021 - Neuro-MIG conference (virtual)

• Characterization of the functional enhancers in human neural stem cells
2021 - Sophia research day (virtual)

• Characterization of the functional enhancers in human neural stem cells
2021 - ESHG (virtual)

• Dissecting the role of YY1 in determining enhancer activity and gene expression
2022 - 30th MGC symposium (Leiden - NL)

POSTERS

• Characterization of the regulatory DNA in human cerebral cortex development and disease
Perenthaler E, Yousefi S, Nikoncuk A, Barakat TS
2019 - MGC workshop (Maastricht - NL)

• Characterization of the non-coding regulatory DNA in human cerebral cortex development and disease
Perenthaler E, Yousefi S, Nikoncuk A, Barakat TS
Brain malformations: a roadmap for future research conference (Rehovot - IL)

24/09/2022