Enrico Baruffini

Date of birth: 27/11/1980

Nationality: Italian

Gender: Male

CONTACT

Viale delle Scienze 11/A,
43124 Parma, Italy **(Work)**

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ABOUT ME

Scopus ID: 14630046700
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RESEARCH ID: C-1057-2015

WORK EXPERIENCE

30/11/2022 – CURRENT Parma, Italy

Associate Professor in Genetics University of Parma, Department of Chemistry, Life Sciences and Environmental Sustainability SCVSA

- Lecturer in Genetics courses and lab trainer for the BS degree course in Biology and for the MS degree course in Biomolecular, Genomic and Cellular Sciences at the SCVSA Department
- Experimental scientific research
- Local coordinator of national public and private projects

30/11/2019 – 29/11/2022 Parma, Italy

Fixed-term tenure track Researcher in Genetics Universi ty of Parma, Department SCVSA

- Lecturer in Genetics courses and lab trainer for the BS degree course in Biology at the SCVSA Department
- Experimental scientific research
- Local coordinator of national public and private projects

20/12/2015 – 29/11/2019 Parma, Italy

Fixed-term Researcher in Genetics University of Parma, Department SCVSA

- Lecturer in Genetics courses and lab trainer for the BS degree course in Biology at the SCVSA Department
- Experimental scientific research
- Local coordinator of national public and private projects

09/2015 – 12/2015 Parma, Italy

Adjunct Professor in Genetics University of Parma, Department of Life Sciences

- Lecturer in Genetics courses and lab trainer for the BS degree course in Biology at the SCVSA Department

15/03/2014 – 19/12/2015 Parma, Italy

Research Fellow "FIRB" in Genetics University of Parma, Department of Life Sciences

- Experimental scientific research
- Local coordinator of national public and private projects

31/01/2012 – 30/01/2014 Parma, Italy

Research Fellow in Genetics University of Parma, Department of Genetics, Biology of Microorganisms, Anthropology, Evolution

- Experimental scientific research
- Lab trainer for the BS degree course in Biotechnology at the Faculty of Sciences

27/10/2010 – 26/10/2011 Parma, Italy

Research Fellow in Genetics University of Parma, Department of Genetics, Biology of Microorganisms, Anthropology, Evolution

- Experimental scientific research

27/10/2008 – 26/10/2010 Parma, Italy

Post-doc research fellow University of Parma, Department of Genetics, Biology of Microorganisms, Anthropology, Evolution

- Experimental scientific research
- Lab trainer for the BS degree course in Biotechnology at the Faculty of Sciences

EDUCATION AND TRAINING

31/12/2004 – 30/12/2007 Parma, Italy

Ph.D. in Biotechnology University of Parma, Department of Genetics, Anthropology, Evolution

- Ricerca scientifica sperimentale sul lievito come sistema modello di patologie mitocondriali

Address Viale delle Scienze 11/A, Parma, Italy | **Field of study** Biotecnologie | **Final grade** Assolutamente soddisfacente | **Thesis** Studio nel sistema modello Saccharomyces cerevisiae di mutazioni patologiche nel gene POLG codificante la DNA polimerasi mitocondriale", docenti guida Iliana Ferrero Fortunati e Tiziana Lodi La tesi è riportata al link: https://www.repository.unipr.it/handle/1889/751

31/12/2005 – 30/12/2006 Louvain-la-Neuve, Belgium

Guest Ph.D. student Catholyc University of Louvain, Department FYSA

- Ricerca scientifica sperimentale sul lievito come sistema modello di patologie mitocondriali, sotto la supervisione della Prof.ssa Francoise Foury

Address Louvain-la-Neuve, Belgium

14/10/2002 – 15/11/2004 Parma, Italy

MS in Industrial Biotechnology University of Parma, Faculty of Sciences

- Studio di materie attinenti alla classe delle lauree specialistiche in Biotecnologie Industriali

Address Parma, Italy | **Final grade** 110/110 e lode | **Thesis** - Studio di materie attinenti alla classe delle lauree specialistiche in Biotecnologie Industriali

09/10/1999 – 20/07/2002 Parma, Italy

BS in Biotechnology University of Parma, Faculty of Sciences

- Studio di materie attinenti alla classe delle lauree in Biotecnologie

Address Parma, Italy | **Final grade** 110/110 e lode

LANGUAGE SKILLS

MOTHER TONGUE(S): Italian

Other language(s):

English

Listening	Reading	Spoken production	Spoken interaction	Writing
B2	C1	C1	B2	C1

French

Listening	Reading	Spoken production	Spoken interaction	Writing
A1	A2	A1	A1	A1

DIGITAL SKILLS

Use of Windows and Android | Use of suites such as Office and Acrobat Reader | Use of softwares for the visualization and molecular graphics (SPDBViewer, Rasmol) | Use of the most important softwares of Bioinformatics | Outlook | Teams | Skype | Zoom

ADDITIONAL INFORMATION

Communication and interpersonal skills

Communication skills Good communication skills acquired thanks to participation in various conferences and the carrying out of lectures for the students

Organisational skills

Organizations skills Excellent ability to work both independently and in teams acquired thanks to 16 years of research in Italian and foreign university research laboratories

Professional skills Good ability to coordinate research groups thanks to the funding of five peer-reviewed projects, of which I am responsible for the Unit or PI

Acquired techniques:
Molecular and biochemical methodologies: gene cloning and sequence analysis, PCR, colony PCR, mutagenic PCR, RT-PCR, qPCR, DNA extraction from E. coli, nucleic acid and protein extraction from yeast, DNA and RNA manipulation, transformation of yeast and E. coli, gene destruction in yeast, extraction of mitochondria and mitochondrial proteins, Southern blot, Northern blot, SDS-PAGE, Western blot
Yeast physiology and genetics: crossing, respiratory cytochrome analysis, measurement of respiratory activity, cytoduction, analysis of phenotypes associated with mitochondrial DNA loss, complex II, III and IV assays, ATPase gel assay, mitochondrial protein synthesis in vivo

Microscopy: use of the fluorescence microscope, processing of images obtained by expression of GFP and mitochondrial probes.

Ulteriori Informazioni

Abilitazione Scientifica Nazionale

1. Qualified as Full Professor in the sector 05/I1-Genetics, SSD BIO/18-Genetics in the 2018-2020 session (duration of the qualification: 01/06/2020-01/06/2030)
2. Qualified as Associate Professor in the sector 05/I1-Genetics, SSD BIO/18-Genetics in the 2016-2018 session (duration of the qualification: 26/07/2018-26/07/2028).
3. Qualified as Associate Professor in the sector 05/F1- Applied Biology, SSD BIO/13-Applied Biology in the 2016 session (duration of the qualification: 4/04/2017-4/04/2027).
4. Qualified as Associate Professor in the sector 05/I1: Genetics and microbiology, SSD BIO/18-Genetics and BIO/19-Microbiology in the 2013 session (duration of the qualification: 1/12/2014-1/12/2024) .

Responsibility and participation to funded projects

16/11/2019-15/5/2021: "FIL Incentivante di Ateneo 2019" at University of Parma, project “Identification and characterization of drugs targeting POLG disorders by using yeast models”, **P.I., 12,300 euros**

1/11/2019-30/10/2023: Italian Telethon Research Grant, project no. GGP19287 “Pre-clinical identification of drugs targeting POLG disorders by using a Zebrafish/Yeast trans-species approach (ZIPPY)”, P.I. Francesco Argenton (University of Padova, Italy), **Unit Coordinator, 66,000 euros**

17/10/2018-16/03/2023: Ministry of Health finalized research 2016 project no. GR-2016-02361449 “Italian Project on Hereditary Optic Neuropathies (IPHON): from genetic basis to therapy”, P.I. Leonardo Caporali (IRCCS Istituto delle Scienze Neurologiche di Bologna, Italy), **Unit Coordinator, 89,350 euros**

17/4/2017-16/4/2018: "FIL Incentivante di Ateneo 2016" at University of Parma, project no. FIL2016_10007492 “Identification of the molecular target of clofilium tosylate, a drug rescuing mitochondrial defects due to pathological mutations in the mitochondrial DNA polymerase”, **P.I., 6,564 euros**

10/3/2014-10/3/2017: Ministry of University and Research Futuro in Ricerca 2013 call, project no RBFR13IWDS “From yeast to humans: role of OPA1 isoforms and pathogenic mutations in neurodegeneration characterized by mitochondrial genome instability”, P.I. Claudia Zanna (University of Bologna, Italy) **Unit Coordinator, 158,082 euros**

2016-2019: Italian Telethon Research Grant, project no. GGP1504"MitMed consortium: from the identification and characterization of nuclear genes responsible for human mitochondrial disorders towards potential therapeutic approaches in experimental models", P.I. Daniele Ghezzi (Istituto Besta, Milan, Italy), participant

2012-2015: Italian Telethon Research Grant, project no. GGP11011 "MitMed: a multicenter consortium for the identification and characterization of nuclear genes responsible for human mitochondrial disorders", PI Massimo Zeviani (Istituto Besta. Milan, Italy), participant

Teaching activity

Activities carried out at the University of Parma

1. **from the academic year (ay) 2022/2023: Lecturer in the Human Molecular Genetics course in the MS degree course in Biomolecular, Genomic and Cellular Sciences, SSD BIO/18-Genetics, 6 ECTS per year, corresponding to 48 hours of frontal lessons.**
2. ay 2021/2022 and ay 2022/2023: Lecturer for the course of Human Molecular Genetics and Genetics of Model Organisms at the MS degree course in Genomic, Molecular and Industrial Biotechnology, SSD BIO/ 18-Genetics, 1 ECTS per year, corresponding to 8 hours of frontal lessons.
3. **from the ay 2020/2021: Lecturer of Genetics in the BS degree course in Biology, SSD BIO/18-Genetics, 9 ECTS per year, corresponding to 64 hours of frontal lessons and 12 hours of classroom exercises.**
4. **ay 2015-2016-ay 2019-2020: Lecturer of the Human Genetics course in the BS degree course in Biology, SSD BIO/18-Genetics, 6 ECTS per year, equal to 40 hours of frontal lessons and 12 hours of classroom exercises.**
5. from ay 2015-2016: Lab co-trainer of the integrated course of Biological Techniques Laboratory in the BS degree course in Biology, about 32 hours per year
6. ay 2015-2016: Co-lecturer of the course of Genetics and Integrated Laboratory of Biotechnology I in the BS degree course of Biotechnology,16 hours
7. 1/11/2018-15/01/2022, tutor of the Ph.D. student Andrea Degiorgi, Ph.D. in Biotechnology and Biosciences, XXXIV cycle
8. 1/11/2021-31/20/2024, tutor of the Ph.D. student Alexandru Ionut Gilea, Ph.D. in Biotechnology and Biosciences, XXXVII cycle
9. 2010-2015 Subject Expert in the BIO/18-Genetics sector
10. Supervisor and co-supervisor of more than 50 research theses of undergraduate students in Biology and Biotechnology and graduated students in Industrial Biotechnology or Molecular Biology. Participation in more than 20 Graduation Commissions.

Activities connected to didactic

2020-current: Secretary of the BS degree course in Biology, University of Parma
2020-current: Member of the Biology Didactic Commitee
2017-2019 Member of the students-teachers joint commission for the Department SCVSA
2016-current Member of the PhD Board in "Biotechnologies and Biosciences", University of Parma

2023: Member of the Commission for the Final Examination of Dr. Cristina Calderan in "Biomedical Sciences" at the University of Padova, Italy
2021: Member of the I Commission for the Final Examination of the XXXIII Ph.D. cycle in "Biotechnologies and Biosciences" at the University of Parma, Italy
2018: Organization of the "Next Generation Sequencing" course for the Ph.D. degree in Biotechnology and Biosciences, 1 ECTS for attending Ph.D. students
2016: Member of the Ph.D. Commission for the Final Examination of Dr. Sharma Vasundhara in "Biomolecular Sciences" at the University of Trento, Italy
2013. Lecturer of the presentation "Yeast as a model of mitochondrial pathologies: why, when and how" of the Workshop "Updates in neurogenetics" at the Besta Institute, Milan, Italy

Research activity

I carried out research regarding the creation of a model system based on the yeast Saccharomyces cerevisiae for the study of pathological mutations in the POLG gene encoding the catalytic subunit of the mitochondrial DNA polymerase, or Polg, whose mutations are associated with a wide range of mitochondrial pathologies. I dealt with five main aspects related to mutations in the MIP1 yeast gene coding for Polg (I) Validation of poithological human mutations, i.e. evaluation, by studying the corresponding mutations in MIP1, of the effects of the mutations, in relationship to the OXPHOS functionality and to the mutability of mitochondrial DNA. (ii) Chemical rescue of the phenotype induced by mutations in MIP1, i.e. identification of molecules, in particular antioxidants, able to reduce the effects on mitochondrial DNA instability induced by mutations. (iii) Genetic rescue of the pathological phenotype by overexpression of a gene, RNR1, encoding the R1 subunit of ribonucleotide reductase or by deletion of the SML1 gene, encoding a ribonucleotide reductase inhibitor. (iv) Study of the interactions between pathological mutations in the MIP1 gene in cis and in trans and (v) Understanding of the molecular mechanisms associated with the pathogenicity of mutations in MIP1 by measuring mitochondrial protein levels and polymerase activity in vitro of the mutated forms of Mip1.
Since April 2008, in the research field of yeast as a model system for the study of the molecular basis of human mitochondrial pathologies and for the identification of potential therapeutic strategies, I have been dealing with the following lines of research in the yeast system: study of effects of deletions and overexpression by multicopy plasmids of genes involved in repair on mitochondrial DNA stability; effects of nucleoside analogs used in anti-HIV therapy on mutations in MIP1 equivalent to human SNPs in POLG; study of the effects of a thiosemicarbazonic compound with anticancer action on the entire collection of yeast deletion mutants, in order to identify the pathways that are inhibited by the treatment and consequently the potential cellular targets of the compound and the mechanisms of action; search for molecules capable of reducing the phenotypic effects of mutations in MIP1; validation of human mutations involved in the pathogenesis of mitochondrial diseases in the yeast model system; effect of

mutations in the MES1 gene, encoding the enzyme methionyl-tRNA synthetase, equivalent to human pathological mutations; construction and validation of a chimeric model for the study of mitochondrial pathologies associated with mutations in the human OPA1 gene.
I recently used the models previously constructed for the research of molecules with potential therapeutic action through drug repurposing techniques.

Activities connected to the research

2018-current: Elected member and secretary of the Research Committee 05 (Biology) of the University of Parma

2017-2021: Member of the Research Commission of the Department SCVSA of the University of Parma

2021: Tutor of the research fellowship of Dr. Camilla Ceccatelli Berti "Analysis in yeast of mutations equivalent to potentially pathological human mutations associated to optic neuropathies found through whole exome sequencing" funded on the project "Italian Project on Hereditary Optic Neuropathies (IPHON): from genetic basis to therapy" (12 months)

2021: Tutor of the Research fellowship of Dr. Giulia di Punzio "Screening in yeast of FDA-approved drugs libraries for the research of compounds with a potential therapeutic action aginst mitochodnrail diseases" funded on the project "Italian Project on Hereditary Optic Neuropathies (IPHON): from genetic basis to therapy" (12 months)

2020-2021: Tutor of the Research fellowship of Dr. Alexandru Ionut Gilea "Pre-clinical identification of drugs targeting POLG disorders by using a yeast model" funded on the project "Pre-clinical identification of drugs targeting POLG disorders by using a Zebrafish/Yeast trans-species approach (ZIPPY)" (15,5 months)

2020: Tutor of the Research fellowship of Dr. Marina Bove "Identification and characterization of drugs targeting POLG disorders by using yeast models" funded on the project "Identification and characterization of drugs targeting POLG disorders by using yeast models" (4,5 months)

Bibliometric hindexes

1/03/2023:
Publication on peer-reviewed journals with IF: 50
h-index: 20 (Scopus)
Total citations: 1809 (Scopus)
Citations per publication: 36,2
Citations per year: 110

Editing and reviewing activity

2017- Associate Editor for the journal "PLOS ONE"
2016- Associate Editor for the journal "Frontiers in Pediatrics"
2015- Associate Editor for the journal "Frontiers in Genetics"
2014-2015 Guest Associate Editor for the journal "Frontiers in Genetics", "Research Topic" entitled "Impact of nuclear genetic variants on mitochondrial pathophysiology"
2019- Evaluator of candidates applying for the position of Review Editor for Frontiers in Genetics

Reviewer for the journals Proceedings of the National Academy of Sciences, India Section B: Biological Sciences, PLoS One, Microbial Cell Factories, African Journal of Biotechnology, FEMS Yeast Research, Mitochondrion, FEBS Letters, Frontiers in Genetics, Clinical Genetics, AIMS Microbiology, BBA General Subjects, PLoS Genetics, Oxidative Medicine and Cellular Longevity, Scientific Reports, Cytogenetic and Genome Research, Journal of Translational Genetics and Genomics, Genes, Yeast, Food Control, Nucleic Acid Research, Molecular Genetics and Metabolism Reports

Registration in the Reprise database for the evaluation of Italian scientific projects
Evaluation activity of three PRIN 2020 projects
Evaluation activity of a project for research funded by the University of Florence, Italy
Evaluation activity of two projects through the Cineca consortium, Italy

Third mission: public engagement

Participation in the Researchers' Night 2012, 2013, 2014 and 2019
Participation in the BS degree course in Biology Open Day in 2016, 2019, 2020 and 2021
Participation in the "Banca Nazionale del lavoro" day dedicated to Telethon in 2019
Participation in the Social Dinner held with Telethon volunteers from Parma with the relative presentation of the research project in 2020
QuBiTv interview in 2021: "interview with the researcher of the Department of Chemical Sciences, Life and Environmental Sustainability Enrico Baruffini (Research) - interview with Enrico Baruffini (Young researchers in Parma section, edited by the Informagiovani of the Municipality of Parma)"

Collaborators

Ongoing

1. Francesco Argenton and Natascia Tiso, Department of Biology, University of Padova, Padova, Italy
2. Leonardo Caporali and Valerio Carelli, IRRCS Istituto Scienze Neurologiche, Hospital Bellaria, Bologna, Italy
3. Claudia Zanna, Department of Pharmacy and Biotechnology, University of Bologna, Bologna, Italy

4. Rosalba Carrozzo and Enrico Bertini, Department of Neurosciences and Neurorehabilitation, Pediatric Hospital Bambino Gesù, Rome, Italy
5. Daniele Ghezzi, Department of Medical-Surgical and Transplant Pathophysiology, University "La Statale" of Milan, Milan, Italy
6. Claudia Castiglioni, Departamento de Neurología Pediátrica, Clínica Las Condes, Santiago, Chile
7. Massimo Zeviani, Department of Neurosciences, University di Padova, Padova, Italy
8. Robert Kopajtich, Helmholtz Zentrum München, Institut für Neurogenomik, Neuherberg, Germany

Past

1. Luis Brieba, Laboratorio Nacional de Genómica para la Biodiversidad, Irapuato, Guanajuato, Mexico
2. Gerarda Cappuccio and Nicola Brunetti-Pierri, TIGEM, Pozzuoli (NA), Italy
3. Ophry Pines, Department of Microbiology and Molecular Genetics, Hebrew University, Jerusalem, Israel
4. Duccio Cavalieri, Department of Biology, University of Florence, Sesto Fiorentino (FI), Italy
5. Reeval Segel, Department of Pediatrics, Shaare Zedek Medical Center, Hebrew University-Hadassah School of Medicine, Jerusalem, Israel
6. Tim M. Strom, Institute of Human Genetics,Technische Universitat Munchen, Munich, Germany
7. Robert W. Taylor, Wellcome Trust Centre for Mitochondrial Research, Institute for Ageing and Health, The Medical School, Newcastle University, Newcastle upon Tyne, UK
8. Patrick F. Chinnery, Institute of Human Genetics, Newcastle University, Newcastle upon Tyne, UK
9. Agnès Delahodde, Institut de Biologie Intégrative de la Cellule (I2BC), Universitè Paris-sud, Paris, France

Partecipation to congresses and schools

2022: Congress CureARS (Virtual: New York, USA)
2021: Congress AGI 2021 (Virtual: Rome, Italy)
2021: Mitochondrial Medicine (Virtual: Hinxton, Cambridge, UK)
2020: Mitochondrial Medicine (Virtual: Hinxton, Cambridge, UK)
2019: Convention Telethon 2019 (Riva del Garda, TN, Italy)
2018: Mitochondrial Medicine (Hinxton, Cambridge, UK)
2016: Molecular biology of mitochondrial gene expression. EMBO lecture (Bro, Sweden)
2016: Mitochondrial Medicine: Developing New Treatments For Mitochondrial Disease. (Hinxton, Cambridge, UK)
2015: 27° International conference on yeast genetics and molecular biology (Levico, TN, Italy)
2013: Mitochondrial Disease: Translating biology into new treatments (Hinxton, Cambridge, UK)
2012: National Congress FISV 2012 (Rome, Italy)
2011: XIX annual congress SIMA (Parma, Italy)
2011: Joint meeting: AGI-SIBV-SIGA (Assisi, PG, Italy)
2010: Yeast, an evergreen model (Rome, Italy)
2009: FEBS Advanced Lecture Course 2009: Mitochondria in Life, Death and Disease (Aussois, France)
2008: X Congress FISV (Riva del Garda, TN, Italy)
2007: Model organisms to study DNA repair and genome stability (Gargnano, BS, Italy)
2007: IX congress FISV (Riva del Garda, TN, Italy)
2007: ZYMI 2007 – Meeting of the italian yeast group (Florence, Italy)
2007: FEBS Advanced Lecture Series – Mitochondria in life, death and disease (Aussois, France)
2005: VII congress FISV (Riva del Garda, TN, Italy)
2005: Bioinformatics school (Turin, Italy)
2005: Meeting of the Italian researchers on yeast (Cortona, AR, Italy)

Honors and awards

Special mention for the PhD thesis awarded by the Italian Genetic Association in 2009 for having addressed "subjects of general interest concerning genetics, molecular biology and medicine".

Membership in Scientific Societies

2010-present Member of the Italian Genetic Association (AGI)
2009-present Member of the Italian Association of General Microbiology and Microbial Biotechnology (SIMGBM)
2011-2016 Member of the Italian Society of Environmental Mutagenesis (SIMA)

Publications

Publication on peer-reviewed Journals

I am author of 50 scientific publications in peer-reviewed international journals with impact factor (attachment 1)

Proceedings

I am author of 45 conference proceedings, including 2 journal conference proceedings (attachment 2)

Patents 2019

Delahodde, Agnès, NUGROHO PITAYU, Laras, Baruffini, Enrico, Lodi, Tiziana, Rötig, Agnès, Procaccio, Vincent (2019) COMPOUNDS FOR THE TREATMENT OF MITOCHONDRIAL DISEASES. (EP3226850B1, US10639287B2)

Parma, 01/03/2023

Attachment 1: publication list

All the publications are present in the following databases:

- Pubmed
- Scopus
- WOS

50) Magistrati M., Gilea A.I., Ceccatelli Berti C., Baruffini E., Dallabona C. (2023) Modopathies Caused by Mutations in Genes Encoding for Mitochondrial RNA Modifying Enzymes: Molecular Mechanisms and Yeast Disease Models. *Int J Mol Sci.* 24:2178. doi: 10.3390/ijms24032178. ISSN: 1422-0067 (**Corresponding author**)

49) Gilea A.I., Ceccatelli Berti C., Magistrati M., di Punzio G., Goffrini P., **Baruffini E.**, Dallabona C. (2021) *Saccharomyces cerevisiae* as a Tool for Studying Mutations in Nuclear Genes Involved in Diseases Caused by Mitochondrial DNA Instability. *Genes (Basel)*. **12**:1866. doi: 10.3390/genes12121866. ISSN: 2073-4425 (**Corresponding author**)

48) di Punzio G., Gilberti M., **Baruffini E.**, Lodi T., Donnini C., Dallabona C. (2021) A Yeast-Based Repurposing Approach for the Treatment of Mitochondrial DNA Depletion Syndromes Led to the Identification of Molecules Able to Modulate the dNTP Pool. *Int. J. Mol. Sci.* **22**:12223. doi: 10.3390/ijms222212223. ISSN: 1422-0067

47) Cappuccio G., Ceccatelli Berti C., **Baruffini E.**, Sullivan J, Shashi V., Jewett T, Stamper T., Maitz S., Canonico F., Revah-Politi A., Kupchik G.S., Anyane-Yeboa K., Aggarwal V., Benneche A., Bratland E., Berland S., D'Arco F., Alves C.A., Vanderver A., Longo D., Bertini E., Torella A., Nigro V.; D'Amico A., van der Knaap M.S., Goffrini P., Brunetti-Pierri N. (2021) Bi-allelic *KARS1* pathogenic variants affecting functions of cytosolic and mitochondrial isoforms are associated with a progressive and multisystem disease. *Hum. Mutat.* **42**:745-761. doi: 10.1002/humu.24210. ISSN: 1098-1004 (**Co-first author**)

46) Figuccia S., Degiorgi A., Ceccatelli Berti C., **Baruffini E.**, Dallabona C., Goffrini P. (2021) Mitochondrial Aminoacyl-tRNA Synthetase and Disease: The Yeast Contribution for Functional Analysis of Novel Variants. *Int. J. Mol. Sci.* **22**:4524. doi: 10.3390/ijms22094524. ISSN: 1422-0067

45) Hytönen M.K., Sarviaho R., Jackson C.B., Syrjä P., Jokinen T., Matiasek K., Rosati M., Dallabona C., **Baruffini E.**, Quintero I., Arumilli M., Monteuuis G., Donner J., Anttila M., Suomalainen A., Bindoff LA., Lohi H. (2021) In-frame deletion in canine *PITRM1* is associated with a severe early-onset epilepsy, mitochondrial dysfunction and neurodegeneration. *Hum. Genet.* **140:**1593-1609. doi: 10.1007/s00439-021-02279-y. ISSN: 0340-6717

44) Ceccatelli Berti C., di Punzio G., Dallabona C., **Baruffini E.**, Goffrini P., Lodi T., Donnini C. (2021) The Power of Yeast in Modelling Human Nuclear Mutations Associated with Mitochondrial Diseases. *Genes (Basel)*. **12**:300. doi: 10.3390/genes12020300. ISSN: 2073-4425

43) Marchi L., Degola F., **Baruffini E.**, Restivo F.M. (2021) How to easily detect plant NADH-glutamate dehydrogenase (GDH) activity? A simple and reliable *in planta* procedure suitable for tissues, extracts and heterologous microbial systems. *Plant. Sci.* **304**:110714. doi: 10.1016/j.plantsci.2020.110714. ISSN: 0168-9452

42) Facchinello N., Laquatra C., Locatello L., Beffagna G., Brañas Casas R., Fornetto C., Dinarello A., Martorano L., Vettori A., Risato G., Celeghin R., Meneghetti G., Santoro M.M., Delahodde A., Vanzi F., Rasola A., Dalla Valle L., Rasotto M.B., Lodi T., **Baruffini E.**, Argenton F., Tiso N. (2021) Efficient clofilium tosylate-mediated rescue of POLG-related disease phenotypes in zebrafish. *Cell. Death Dis.* **12**:100. doi: 10.1038/s41419-020-03359-z. ISSN 2041-4889 (**Co-corresponding author**)

41) Aleo S.J., Del Dotto V., Fogazza M., Maresca A., Lodi T., Goffrini P., Ghelli A., Rugolo M., Carelli V., **Baruffini E.**, Zanna C. (2021) Drug repositioning as a therapeutic strategy for neurodegenerations associated with *OPA1* mutations. *Hum. Mol. Genet.* **29**:3631-3645. doi: 10.1093/hmg/ddaa244. ISSN: 0964-6906 (**Co-senior author**)

40) Benincá C., Zanette V., Brischigliaro M., Johnson M., Reyes A., Valle D.A.D., J Robinson A., Degiorgi A., Yeates A., Telles B.A., Prudent J., **Baruffini E.**, S. F. Santos M.L., R. de Souza R.L., Fernandez-Vizarra .E, Whitworth A.J., Zeviani M. (2021) Mutation in the MICOS subunit gene *APOO* (MIC26) associated with an X-linked recessive mitochondrial myopathy, lactic acidosis, cognitive impairment and autistic features. *J. Med. Genet.* **58**:155-167. doi: 10.1136/jmedgenet-2020-106861. ISSN: 0022-2593

39) **Baruffini E.**, Ruotolo R., Bisceglie F., Montalbano S., Ottonello S., Pelosi G., Buschini A., Lodi T. (2020) Mechanistic insights on the mode of action of an antiproliferative thiosemicarbazone-nickel

complex revealed by an integrated chemogenomic profiling study. *Sci. Rep.* **10**:10524. doi: 10.1038/s41598-020-67439-y. ISSN: 2045-2322

38) Hoyos-Gonzalez N., Trasviña-Arenas C.H., Degiorgi A., Castro-Lara A.Y., Peralta-Castro A., Jimenez-Sandoval P., Diaz-Quezada C., Lodi T., **Baruffini E.**, Brieba LG. (2020) Modeling of pathogenic variants of mitochondrial DNA polymerase: insight into the replication defects and implication for human disease. *Biochim. Biophys. Acta Gen. Subj*. 1864:129608. doi: 10.1016/j.bbagen.2020.129608. ISSN:0304-4165 (**Co-corresponding author**)

37) Trasviña-Arenas C.H., Hoyos-Gonzalez N., Castro-Lara A.I., Rodriguez-Hernandez A., Sanchez-Sandoval M.E., Jimenez-Sandoval P., Ayala-García V.M., Díaz-Quezada C., Lodi T., **Baruffini E.**, Brieba L.G. (2019) Amino and carboxy-terminal extensions of yeast mitochondrial DNA polymerase assemble both the polymerization and exonuclease active sites. *Mitochondrion*, **49**:166-177. doi: 10.1016/j.mito.2019.08.005. ISSN: 1567-7249

36) Chin H., Goh D.L., Wang F.S, Hong Tay S.K., Heng C.K., Donnini C., **Baruffini E.**, Pines O. (2019) A combination of two novel *VARS2* variants causes a mitochondrial disorder associated with failure to thrive and pulmonary hypertension. *J. Mol. Med. (Berl).* **97**:1557-1566. doi: 10.1007/s00109-019-01834-5. ISSN: 0946-2716 (**Co-corresponding author**)

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Attachment 2: proceedings list

45) Gilea, Alexandru Ionut; Branas Casas, Raquel; Argenton, Francesco; Tiso, Natascia; Lodi, Tiziana; **Baruffini, Enrico** (2021) Identification of drugs for the treatment of *POLG*-related diseases by means of a high throughput drug repurposing approach performed in *Saccharomyces cerevisiae* - Mitochondrial Medicine 2021, Hinxton, Cambridgeshire, UK (telematico), 30/11-2/12/2021. (Poster)

44) Di Punzio, Giulia; Gilberti, Micol; Magistrati, Martina; Leotta, Antonella; Gilea, Alexandru Ionut; **Baruffini, Enrico**; Lodi, Tiziana; Donnini, Claudia; Dallabona, Cristina. (2021) A yeast-based repurposing approach revealed modulation of dNTP pool as a therapeutic target to treat mitochondrial DNA depletion syndromes in: ICY15 (International Congress on Yeasts) meets 30 ICYGMB (International Conference on Yeast Genetics and Molecular Biology) (Conferenza Virtuale), p. 417, 23-27/08/2021 (Poster)

43) Ceccatelli Berti, Camilla; Cappuccio, Gerarda; **Baruffini, Enrico**; Figuccia, Sonia; Brunetti-Pierri, Nicola; Goffrini, Paola. Modeling in yeast of *KARS* pathogenic variants associated with a progressive and multi-systemic disease: impact on cytosolic and mitochondrial isoforms (2020) -Mitochondrial Medicine 2020, Hinxton, Cambridgeshire, UK (telematico), 30/11-2/12/2020.

42) **Baruffini, Enrico**; Pitayu-Nugroho, Laras; Facchinello, Nicola; Giroux, Xavier; Beffagna, Giorgia; Degiorgi, Andrea; Donnini, Claudia; Argenton, Francesco; Delahodde, Agnès; Tiso, Natascia; Lodi, Tiziana (2019) Yeast, worm, patient's fibroblasts and zebrafish as models for drug repositioning of molecules targeting POLG-related diseases – Mitochondrial Medicine 2019, Wellcome Genome Campus, Hinxton, Cambridge nei giorni 11-13/12/2019.

41) **Baruffini, Enrico**; Lodi, Tiziana; Donnini, Claudia; Delahodde, Agnes; Beffagna, Giorgia; Facchinello, Nicola; Tiso, Natascia; Argenton, Francesco. (2019) Pre-clinical identification of drugs targeting polg disorders by using a zebrafish/yeast trans-species approach (ZIPPY) - XX Telethon Scientific Convention, Riva del Garda (TN), P. 24, 28-30/10/2019. (**Selected oral presentation**)

40) G. di Punzio, C. Ceccatelli Berti, F. Pelosi, M. Di Gregorio, C. Dallabona, C. Zanna, **E. Baruffini**, P. Goffrini, T. Lodi, C. Donnini (2018). *Saccharomyces cerevisiae* as a system to discover beneficial molecules for mitochondrial diseases. In: XV FISV CONGRESS Sapienza University of Rome, Italy September 18-21, 2018. p. 168, Roma, 18-21/9/2018 (Poster)

39) G. di Punzio, C. Ceccatelli Berti, F. Pelosi, M. Di Gregorio, C. Dallabona, C. Zanna, **E. Baruffini**, P. Goffrini, T. Lodi, C. Donnini (2018). *Saccharomyces cerevisiae* as a system to discover beneficial molecules for mitochondrial diseases. In: 8° Convegno Nazionale sulle Malattie Mitocondriali - Mitocon. Roma, 25-27/5/2018 (Poster)

38) C. Ceccatelli Berti, G. di Punzio, F. Pelosi, C. Dallabona, **E. Baruffini**, P. Goffrini, T. Lodi, C. Donnini (2018). The power of yeast in modeling human mutations leading to mitochondrial disease. In: 8° Convegno Nazionale sulle Malattie Mitocondriali. Roma, 25-27/5/2018 (Poster)

37) **E. Baruffini**, L. Pitayu-Nugroho, X. Giroux, A. Delahodde, T. Lodi (2018). Yeast as a model for drug repositioning: analysis of molecules acting as potential therapeutics for POLG-related diseases. In: Mitochondrial Medicine. p. P4, Wellcome Genome Campus, Hinxton, Cambridge, UK, 9-11/5/2018 (Poster)

36) M. Gilberti, C. Ceccatelli Berti, G. di Punzio, A. Degiorgi, **E. Baruffini**, C. Dallabona (2017). *Saccharomyces cerevisiae* as a model for the identification of beneficial molecules for mitochondrial diseases. In: AGI - Associazione Genetica Italiana. Cortona (Arezzo, Italia), 7-9/9/2017 (Poster)

35) C. Ceccatelli Berti, M. Gilberti, A. Degiorgi, G. di Punzio, **E. Baruffini**, C. Dallabona (2017). The power of yeast in modeling human mutations leading to mitochondrial disease:the case of ANT1, YARS2, DNM1L and LYRM7. In: AGI - Associazione Genetica Italiana. Cortona (Arezzo,Italia), 7-9/9/2017 (Poster)

34) Marchi L., Polverini E., Degola F., **Baruffini E.**, Restivo F.M. (2016). Glutamate dehydrogenase isoenzyme 3 (GDH3) of *Arabidopsis thaliana* is less thermostable than GDH1 and GDH2 isoenzymes. In: FISV XIV Congress. -, Roma, 20-23/9/2016 (Poster)

33) Marchi L., Polverini E., Degola F., **Baruffini E.**, Restivo F.M. (2016). Glutamate dehydrogenase isoenzyme 3 (GDH3) of *Arabidopsis thaliana* is less thermostable than GDH1 and GDH2 isoenzymes. In: Plant Biology Europe EPSO/FESPB 2016 Congress. Praga, Repubblica Ceca, 26-30/6/2016

32) Dallabona C., **Baruffini E.**, Gilberti M., Goffrini P., Donnini C.. (2016). The power of yeast in modeling human mutations: mitochondrial aminoacyl tRNA synthetases and mitochondrial tRNA modifiers. In: Molecular biology of mitochondrial gene expression. p. 33, EMBO, Bro, Svezia, 23-26/5/2016 (Poster)

31) **Baruffini E.**, Pitayu L., Rodier C., Rötig A., Lodi T., Delahodde A.. (2016). Yeast as a model for drug discovery: identification of a molecule acting as potential therapeutics for POLG-related diseases. In: Molecular biology of mitochondrial gene expression. p. 28-29, EMBO, Bro, Svezia, 23-26/05/2016 (Poster)

30) Nasca A., Legati A., Carrara G., Lamantea E., Ardissone A., Moroni I., Nolli C., **Baruffini E.**, Goffrini P., Ghezzi D. (2016). Mitochondrial disorders caused by mutations impairing miotchondrial dynamics. In: Mitochondrial Medicine: Developing New Treatments For Mitochondrial Disease. p. P71, Wellcome Genome Campus, Hinxton, Cambridge, UK, 4-6/05/2016 (Poster)

29) Nolli C., Goffrini P., Soliani L., Lazzaretti M., Zanna C., Lodi T., **Baruffini E.** (2016). *S. cerevisiae* carrying a *MGM1*/OPA1 chimeric gene: a model for the study of dominant optic atrophy and for drug discovery. In: Mitochondrial Medicine: Developing New Treatments For Mitochondrial Disease. WElcome Genome Campus, Hinxton, Cambridge, UK, 4-6/05/2016. Wellcome Trust Sanger Insitute, Welcome Genome Campus, Hinxton, Cambridge, UK, 4-6/05/2016 (Poster)

28) **Baruffini E.**, Pitayu L., Rodier C., Rotig A., Lodi T., Delahodde A. (2016). Yeast as a model for drug discovery: Identification of a molecule acting as potential therapeutics for POLG-related diseases. In: Miotchondrial Medicine: Developing New Treatments For Mitochondrial Disease. p. P10, Elcome Genome Campus, Hinxton, Cambridge, UK, 4-6/05/2016 (Poster)

27) Rivero D., Bernà ., Stefanini I., **Baruffini E.**, Bergerat A., Csikàsz-Nagy A., De Filippo C., Cavalieri D. (2015). Hsp12p and *PAU* genes are involved in ecological interactions between natural yeast strains. In: 27th International Conference on Yeast Genetics and Molecular Biology. YEAST, vol. 32, p. S81, ISSN: 0749-503X, Levico Terme, Trento, Italia, 6-12/09/2015 (Poster)

26) Dallabona C., **Baruffini E.**, Gilberti M., Goffrini P., Donnini C. (2015). Mitochondrial Aminoacyl-tRNA Synthetase: The Power Of Yeast In Modeling Human Pathological Mutations. In: 27th International Conference on Yeast Genetics and Molecular Biology. YEAST, vol. 32, p. 85, ISSN: 0749-503X, Levico Terme (TN), 6-12/9/2015 (Poster)

25) C. Nolli, P. Goffrini, M. Lazzaretti, C. Zanna, R. Vitale, T. Lodi, **E. Baruffini**, Enrico (2015). Validation of a MGM1/OPA1 chimeric gene for functional analysis in yeast of mutations associated with dominant optic atrophy. In: 27th International Conference on Yeast Genetics and Molecular Biology. YEAST, vol. 32, p. S185, ISSN: 0749-503X, Levico Terme, Trento, Italia, 6-12/09/2015 (poster)

24) **E. Baruffini**, L. Pitayu, A. Rotig, T. Lodi, A. Delahodde (2015). Yeast model for drug discovery: Identification of molecules acting as potential therapeutics for POLG-related diseases. In: 27th International Conference on Yeast Genetics and Molecular Biology. YEAST, vol. 32, p. S173, ISSN: 0749-503X, Levico Terme, Trento, Italia, 6-12/09/2015 (Poster)

23) L. Melchionda, F. Invernizzi, A. Nasca, D. Diodato, E. Lamantea, S. Marchet, C. Lamperti, **E. Baruffini**, C. Dallabona, C. Donnini, P. Goffrini, T. Lodi, I. Ferrero, C. De Pittà, C. Da Re, S. Corrà, M. Zordan, R. Costa, M. Zeviani, D. Ghezzi (2015). Mitmed: a multicenter consortium for the identification and characterization of nuclear genes responsisble for human mitochondrial disorders. In: Fondazione Telethon: XVIII Scientific Convention. Riva del Garda (TN), 9-11/03/2015 (Poster)

22) C. Dallabona, D. Diodato, T.B. Haack, **E. Baruffini**, L. Melchionda, C. Mariotti, T.M. Strom, H. Prokisch, I. Ferrero, C. Lamperti, M. Zevianci, D. Ghezzi (2013). Yeast model for novel AARS2 mutations associated with progressive leukoencephalopathy and cerebellar ataxia. In: Mitochondrial Disease: Translating biology into new treatments. Cambridge, UK, 2-4/10/2013 (Poster)

21) **E. Baruffini**, C. Dallabona, F. Invernizzi, J.W. Yarham, L. Melchionda, C. Donnini, A. Burlina, T.M. Strom, T.B. Haack, H. Prokish, R.W. Taylor, M. Zeviani, I. Ferrero, D. Ghezzi (2013). MTO1 mutations cause respiratory chain

deficiency in humans and yeast. In: Mitochondrial Disease: Translating biology into new treatments. Cambridge, UK, 2-4/10/2013 (Poster)

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19) **E. Baruffini**, A. Buschini, R. Ruotolo, S. Ottonello, G. Pelosi, F. Bisceglie, T. Lodi (2013). A chemical genomic approach to characterize the antiproliferative activity of metal based drugsdriven by phenotypic screening in yeast as a model organism. In: 6th Central Europe Conference. Trieste, 10-13/09/2013 (Poster)

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