## PERSONAL INFORMATION

# Enrico Baruffini

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Sex Male | Date of birth 28/11/1980 | Nationality Italian

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Scopus ID: 14630046700

ResearcherID: C-1057-2015

## WORK EXPERIENCE

2018-2021

### Unit Leader

Italian “Ricerca Finalizzata 2016” Project GR-2016-02361449: “ Italian Project on Hereditary Optic Neropathies (IPHON): from genetic basis to therapy”

University of Parma

Unit Leader of a project funded by the Italian Ministry of Health for 450,000 € (89500 € to our Unit)
Business or sector University

1/3/2016-today

### Member of the Doctorate School of Biotechnologies and Life Sciences

Department of Chemistry, Life Sciences and Environmental Sustainability

University of Parma

Unit Leader of a project funded by the Italian Ministry of Health for 450,000 € (89500 € to our Unit)
Business or sector University

21/12/2015-today

### Researcher (Italian RTD-A) and Assistant Professor

Department of Life Sciences, then Department of Chemistry, Life Sciences and Environmental Sustainability
University of Parma
Experimental Research and Coordination of a research group
Business or sector University

05/10/2015-20/12/2015

### Adjunct Professor

Teaching Course: Human Genetics at BSc Degree in Biology
University of Parma
Semester 42-hour course of Human Genetics
Business or sector University

16/03/2014-today

### Unit Leader

Italian “Futuro in Ricerca 2013” Project RBFR13IWDS: “From yeast to human: how OPA1 isoforms and pathogenic mutations cause neurodegenerations characterized by mtDNA instability”
University of Parma
Unit Leader of a project funded by the Italian Ministry of Education, University and Research for ~450,000 € (158,000 € to our Unit)
Business or sector University

8/10/2008-15/03/2014

### Post-doc Fellow

Department of Genetics, Biology of Microorganisms, Anthropology, Evolution, then Department of Life Sciences
University of Parma
Experimental Research
Business or sector University

## EDUCATION AND TRAINING

01/01/2005-31/12/2007

### PhD Degree

Department of Genetics, Biology of Microorganisms, Anthropology, Evolution
University of Parma

- Experimental Research on yeast as a model system of mitochondrial diseases
- Thesis supervisors: Ileana Ferrero and Tiziana Lodi
- Academic Qualification: PhD in Biotechnologies
- Evaluation: “Absolutely Satisfactory”

01/01/2006-31/12/2006

### PhD Course

Department FYSA
Catholic University of Louvain, Louvain-la-Neuve, Belgium

- Experimental Research on yeast as a model system of mitochondrial diseases caused by POLG mutations
- Thesis local supervisors: Françoise Foury

15/10/2002-16/11/2004

### Master Degree

University of Parma

- Study of subjects related to Industrial and Molecular Biotechnologies
- Thesis supervisor: Tiziana Lodi
- Academic Qualification: MSc in Industrial Biotechnologies
- Evaluation: 110/110 cum laude

10/10/1999-21/07/2004

## Bachelor Degree

University of Parma

- Study of subjects related to Biotechnologies
- Thesis supervisor: Giorgio Dieci
- Academic Qualification: BSc in Biotechnologies
- Evaluation: 110/110 cum laude

## PERSONAL SKILLS

Mother tongue(s): Italian

Other language(s)

| | UNDERSTANDING | | SPEAKING | | WRITING |
|---|---|---|---|---|---|
| | Listening | Reading | Spoken interaction | Spoken production | |
| English | C1 | C1 | C1 | C1 | C1 |

Other language(s)

| | UNDERSTANDING | | SPEAKING | | WRITING |
|---|---|---|---|---|---|
| | Listening | Reading | Spoken interaction | Spoken production | |
| French | A1 | A2 | A1 | A1 | A1 |

Digital competence

| SELF-ASSESSMENT | | | | |
|---|---|---|---|---|
| Information processing | Communication | Content creation | Safety | Problem solving |
| Independent User | Proficient user | Basic User | Basic User | Independent user |

Good command of Office, software for image processing (Adobe Photoshop), molecular graphic software (SPDBViewer, Rasmol), bioinformatics software.

Driving licence: B

Technical skills: Molecular and biochemical methods: gene cloning and sequence analysis, PCR, colony PCR, mutagenic PCR, RT-PCR, qPCR, extraction of DNA from *E. coli*, extraction of nucleic acids and proteins from yeast, manipulation of DNA and RNA, transformation of yeast and *E. coli*, gene disruption in yeast, extraction of mitochondria and mitochondrial proteins, Southern blot, Northern blot, SDS-PAGE, Western blot, in vivo protein synthesis assay.
Physiology and genetics of yeast: sporulation, analysis of respiratory cytochromes, measurement of respiratory activity, analysis of the phenotypes associated with loss of mitochondrial DNA, assays of complexes II, III and IV, in gel ATPase assay.
Microscopy: use of a fluorescence microscope.

Publications: See Attachment 1

Patents: See Attachment 2

Projects: See Attachment 3

Conferences and Seminars: Co-author of 31 contributions in conference or journal proceedings

Honours and awards: I received the Italian “Abilitazione Scientifica Nazionale” as Associate Professor in the SSD BIO/13: Applied Biology in 2016 (from 4/4/2017 to 4/4/2023).

I received the Italian “Abilitazione Scientifica Nazionale” as Associate Professor in the competitive sector 05/I1: Genetics and Microbiology in 2013 (from 1/12/2014 to 1/12/2020).

I received a special mention for the PhD thesis granted by the Italian Genetics Association in 2009, having dealt with "matters of general interest related to genetics, molecular biology and medicine."

Memberships: 2016-today Member of the Doctorate School of Biotechnologies at the University of Parma, Parma, Italy
2010-today Member of AGI (Italian Genetics Association)
2011-2016 Member of SIMA (Italian Environment Mutagenesis Society)
2009-today "Cultore della Materia" in the Italian Sector BIO-18/Genetics
2009-today Member of SIMGBM (Italian Society of General Microbiology and Microbial Biotechnologies)

Research Activity: During my career, I dedicated myself to the study of genetics, molecular biology and physiology of yeast, with interest on the defects of mitochondrial metabolism due to mutations in genes which are the cause of mitochondrial diseases in humans.
During my PhD course and my post-doc fellowship, both held at the University of Parma (Italy) under the supervision of Tiziana Lodi and Ileana Ferrero, I studied, with the help of molecular approaches, several aspects linked to replication of mitochondrial DNA (mtDNA) in yeast. My main interest concerned the creation of a yeast model system for the analysis of pathological mutations in the POLG gene, encoding the catalytic subunit of the mitochondrial DNA polymerase, whose mutations are associated to a wide range of mitochondrial disorders. Yeast was chosen because mtDNA replication apparatus is conserved between yeast and man and yeast can be easily manipulated thanks to the well-known techniques of molecular genetics and genetic engineering used in this organism.
This yeast model allowed me to study four main aspects linked to mutations in the yeast gene encoding for Polg, called MIP1:
1. validation of potentially pathological human mutations, i.e. evaluation, through study of the equivalent mutations in MIP1, of the effects of these mutations related to OXPHOS function and to extended and point mutability of mitochondrial DNA;
2. chemical rescue of the phenotype induced by MIP1 mutations, i.e. identification of molecules, in particular antioxidants, capable to reduce the effects on mitochondrial DNA instability induced by mutations;
3. genetic rescue of the pathological phenotype by means of overexpression of RNR1 gene, encoding the ribonucleotide reductase subunit R1;
4. understanding the molecular mechanisms underlying the pathology. This goal has been reached during the one-year stage at the FYSA Department in Louvain-la-Neuve (Belgium), under the supervision of Francoise Foury. There I studied the interactions between pathological MIP1 mutations located in cis or in trans and the molecular mechanisms of MIP1 mutations by means of measurement of mitochondrial protein levels and of in vitro polymerase activity of mutant variants of Mip1p.
In the last years, thanks to national and international collaborations, I studied mutations affecting genes involved in the modification/maturation of mitochondrial tRNAs, such as MTO1, encoding the mitochondrial tRNA uridine 5-carboxymethylaminomethylase, human ELAC2/yeast TRZ1, encoding the mitochondrial RNase Z, hTRIT1/yMOD5, encoding the A37 tRNA adenylate isopentenyltransferase, in the aminoacylation of mitochondrial tRNAs, such as hAARS2/yALA1 and hVARS2/yVAS1, encoding the mitochondrial alanyl-tRNA synthetase and valyl-tRNA synthetase, respectively, and hDNM1L,yDNM1, encoding a dynamin like protein involved in mitochondrial fission.
In 2013 Tiziana Lodi and me, together with our collaborators Agnes Delahodde, Laras Pitayu and Agnes Rotig from University of Paris-Sud (France), discovered, by the meaning of a high-throughput screening of FDA-approved drugs on yeast mip1 mutants, a molecule able to reduced the frequency of deletions in mtDNA for which a France patent has been filed and an EU patent is pending.
Since 2013, I’m working, as unit leader, on the study of pathological mutations in OPA1 in a proper yeast model constructed in our lab where human OPA1, unable by itself to complement the deletion in the yeast orthologous gene MGM1, was fused with a region of MGM1, leading to the construction of a functional chimera.

Editing and Reviewing

2017- Today Associate Editor for the Journal “PLOS ONE”

2016-Today Associate Editor for the Journal “Frontiers in Genetics”

2014-2016 Guest Associate Editor for the Journal “Frontiers in Genetics” and proposer of a “Research Topic” entitled “Impact of nuclear genetic variants on mitochondrial pathophysiology”

2012-today Peer Review for the following journals:; PLOS One; Microbial Cell Factories;; FEMS Yeast Research; FEBS Letters, AIMS Microbiology, Mitochondrion, Clinical Genetics, Frontiers in Genetics

Teaching activity

Starting from the academic year 2015-2016, I am the lecturer of the course of “Human Genetics” for the Bachelor Degree Course of Biology at the University of Parma, for a total of 134 hours.

from the academic year 2015-2018, I am one of the teaching assistant at the Course of “Tecniche di Labporatorio Biologico) for the Bachelor Degree Course of Biology at the University of Parma

I am supervisor or co-supervisor of more than 35 BSc or Msc theses in Biotechnologies, Biology, Industrial Biotechnologies and Molecular Biology at The University of Parma, for a total of 24 hours

In 2004-2005, 2007-2008 and 2012-2013 I have been a teaching assistant for the course of Genetics for the BSc Biotechnologies Degree at the University of Parma, for a total of 60 hours.

In 2013-2014 I have been a teaching assistant at the course of Microorganisms and Plant Genetics for the MSc in Molecular Biology, University of Parma, for a total of 10 hours.

## Attachment 1: list of publications

32) Langer Y., Aran A., Gulsuner S., Abu Libdeh B., Renbaum P., Brunetti D., Teixeira PF, Walsh T., Zeligson S., Ruotolo R., Beeri R., Dweikat I., Shahrour M., Weinberg-Shukron A., Zahdeh F., **Baruffini E.**, Glaser E., King M.C., Levy-Lahad E., Zeviani M., Segel R. (2018) Mitochondrial PITRM1 peptidase loss-of-function in childhood cerebellar atrophy. J Med Genet. doi:10.1136/jmedgenet-2018-105330. [Epub ahead of print] PubMed PMID: 29764912.

31) Dallabona C., **Baruffini E.**, Goffrini P., Lodi T. (2017) Dominance of yeast *aac2*$^{R96H}$ and *aac2*$^{R252G}$ mutations, equivalent to pathological mutations in ant1, is due to gain of function. Biochem Biophys Res Commun. 493:909-913. doi:10.1016/j.bbrc.2017.09.122.

30) Del Dotto V., Mishra P., Vidoni S., Fogazza M., Maresca A., Caporali L., McCaffery J.M., Cappelletti M., **Baruffini E.**, Lenaers G., Chan D., Rugolo M., Carelli V., Zanna C. (2017) OPA1 Isoforms in the Hierarchical Organization of Mitochondrial Functions. Cell Rep. 19:2557-2571. doi: 10.1016/j.celrep.2017.05.073.

29) Nasca A., Legati A., **Baruffini E.**, Nolli C., Moroni I., Ardissone A., Goffrini P., Ghezzi D. (2016) Biallelic Mutations in DNM1L are Associated with a Slowly Progressive Infantile Encephalopathy. Hum Mutat. 37: 898-903. doi:10.1002/humu.23033.

28) Brunetti D., Torsvik J., Dallabona C., Teixeira P., Sztromwasser P., Fernandez-Vizarra E., Cerutti R., Reyes A., Preziuso C., D'Amati G., **Baruffini E.**, Goffrini P., Viscomi C., Ferrero I, Boman H., Telstad W., Johansson S., Glaser E., Knappskog P.M., Zeviani M., Bindoff L.A., In press. Defective PITRM1 mitochondrial peptidase is associated with Aβ amyloidotic neurodegeneration. EMBO Mol Med. doi: 10.15252/emmm.201505894.

27) Pitayu L., **Baruffini E.**, Rodier C., Rötig A., Lodi T., Delahodde A. In press. Combined use of *Saccharomyces cerevisiae, Caenorhabditis elegans* and patient fibroblasts leads to the identification of clofilium tosylate as a potential therapeutic chemical against POLG-related diseases. Hum Mol Genet.

26) Nolli C., Goffrini P, Lazzaretti M, Zanna C, Vitale R, Lodi T, **Baruffini E.** (2015) Validation of a MGM1/OPA1 chimeric gene for functional analysis in yeast of mutations associated with dominant optic atrophy. Mitochondrion. 25:38-48. doi: 10.1016/j.mito.2015.10.002. **(Corresponding author)**

25) Rivero D., Berná L., Stefanini I., **Baruffini E.**, Bergerat A., Csikász-Nagy A., De Filippo C., Cavalieri D. (2015) Hsp12p and *PAU* genes are involved in ecological interactions between natural yeast strains. *Environ. Microbiol.* **17**, 3069-3081. doi: 10.1111/1462-2920.12950. ISSN: 1462-2912

24) Hadchouel A., Wieland T., Griese M., **Baruffini E.**, Lorenz-Depiereux B., Enaud L., Graf E., Dubus J.C., Halioui-Louhaichi S., Coulomb A., Delacourt C., Eckstein G., Zarbock R., Schwarzmayr T., Cartault F., Meitinger T., Lodi T., de Blic J., Strom T.M. (2015) Biallelic Mutations of Methionyl-tRNA Synthetase Cause a Specific Type of Pulmonary

Alveolar Proteinosis Prevalent on Réunion Island. *Am. J. Hum. Genet.* **96**, 826-831. doi: 10.1016/j.ajhg.2015.03.010. ISSN: 0002-9297 **(Co-fist author)**

23) Lodi T., Dallabona C., Nolli C., Goffrini P., Donnini C., **Baruffini E.** (2015) DNA polymerase γ and disease: what we have learned from yeast. *Front. Genet.* **17**, 6:106. doi: 10.3389/fgene.2015.00106. ISSN: 1664-8021 **(Corresponding author)**

22) **Baruffini E.**, Ferrari J., Dallabona C., Donnini C., Lodi T.. (2015) Polymorphisms in DNA polymerase γ affect the mtDNA stability and the NRTI-induced mitochondrial toxicity in *Saccharomyces cerevisiae*. *Mitochondrion* **20**, 52-63. doi: 10.1016/j.mito.2014.11.003. ISSN: 1567-7249 **(Corresponding author)**

21) Marchi L., Polverini E., Degola F., **Baruffini E.**, Restivo F.M. (2014) Glutamate dehydrogenase isoenzyme 3 (GDH3) of Arabidopsis thaliana is less thermostable than GDH1 and GDH2 isoenzymes. *Plant. Physiol. Biochem.* **83**, 225-231. doi: 10.1016/j.plaphy.2014.08.003 ISSN:0981-9428

20) Yarham J.W., Lamichhane T.N., Pyle A., Mattijssen S., **Baruffini E.**, Bruni F., Donnini C., Vassilev A., He L., Blakely E.L., Griffin H., Santibanez-Koref M., Bindoff L.A., Ferrero I., Chinnery P.F., McFarland R., Maraia R.J., Taylor R.W. (2014) Defective i6A37 modification of mitochondrial and cytosolic tRNAs results from pathogenic mutations in *TRIT1* and its substrate tRNA. *PLoS Genet.* **10**: e1004424 doi: 10.1371/journal.pgen.1004424. ISSN: 1553-7390

19) Diodato D., Melchionda L., Haack T.B., Dallabona C., **Baruffini E.**, Donnini C., Granata T., Ragona F., Balestri P., Margollicci M., Lamantea E., Nasca A., Powell C.A., Minczuk M., Strom T.M., Meitinger T., Prokisch H., Lamperti C., Zeviani M., Ghezzi D. (2014) *VARS2* and *TARS2* mutations in patients with mitochondrial encephalomyopathies. *Hum. Mutat.* **35**, 983-989. doi: 10.1002/humu.22590. pp.983-989. ISSN: 1059-7794

18) Dallabona C., Diodato D., Kevelam S.H., Haack T.B., Wong L.J., Salomons G.S., **Baruffini E.**, Melchionda L., Mariotti C., Strom T.M., Meitinger T., Prokisch H., Chapman K., Colley A., Rocha H., Ounap K., Schiffmann R., Salsano E., Savoiardo M., Hamilton E.M., Abbink T.E., Wolf N.I., Ferrero I., Lamperti C., Zeviani M., Vanderver A., Ghezzi D., van der Knaap MS. (2014) Novel (ovario) leukodystrophy related to *AARS2* mutations. *Neurology* **82**, 2063-2071. doi: 10.1212/WNL.0000000000000497. ISSN: 0028-3878

17) **Baruffini E.**, Dallabona C., Invernizzi F., Yarham J.W., Melchionda L., Blakely E.L., Lamantea E., Donnini C., Santra S., Vijayaraghavan S., Roper H.P., Burlina A., Kopajtich R., Walther A., Strom T.M., Haack T.B., Prokisch H., Taylor R.W., Ferrero I., Zeviani M., Ghezzi D. (2013) *MTO1* Mutations are Associated with Hypertrophic Cardiomyopathy and Lactic Acidosis and Cause Respiratory Chain Deficiency in Humans and Yeast. *Hum Mutat.*, **34**, 1501-1509. doi: 10.1002/humu.22393. ISSN: 1059-7794

16) Haack T.B., Kopajtich R., Freisinger P., Wieland T., Rorbach J., Nicholls T.J., **Baruffini E.**, Walther A., Danhauser K., Zimmermann F.A., Husain R.A., Schum J., Mundy H., Ferrero I., Strom T.M., Meitinger T., Taylor

R.W., Minczuk M., Mayr J.A., Prokisch H. (2013) *ELAC2* Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. *Am J Hum Genet.*, **93**, 211-223. doi: 10.1016/j.ajhg.2013.06.006. ISSN: 0002-9297

15) Ghezzi D., **Baruffini E.**, Haack T.B., Invernizzi F., Melchionda L., Dallabona C., Strom T.M., Parini R., Burlina A.B., Meitinger T., Prokisch H., Ferrero I., Zeviani M. (2012) Mutations of the mitochondrial-tRNA modifier *MTO1* cause hypertrophic cardiomyopathy and lactic acidosis. *Am J Hum Genet.*, **90**, 1079-1087. doi: 10.1016/j.ajhg.2012.04.011. ISSN: 0002-9297 **(Co-fist author)**

14) **Baruffini E**., Serafini F., Ferrero I., Lodi T. (2012) Overexpression of DNA polymerase zeta reduces the mitochondrial mutability caused by pathological mutations in DNA polymerase gamma in yeast. PLoS One, 7 e34322. doi: 10.1371/journal.pone.0034322. ISSN: 1932-6203 (**Corresponding author**)

13) Serafini F., Bottacini F., Viappiani A., **Baruffini E.**, Turroni F., Foroni E., Lodi T., van Sinderen D., Ventura M. (2011) Insights into physiological and genetic mupirocin susceptibility in bifidobacteria. *Appl Environ Microbiol.* **77**, 3141-3146. 2011. doi: 10.1128/AEM.02540-10. ISSN: 0099-2240

12) **Baruffini E.**, Horvath R., Dallabona C., Czermin B., Lamantea E., Bindoff L., Invernizzi F., Ferrero I., Zeviani M., Lodi T. (2011) Predicting the contribution of novel POLG mutations to human disease through analysis in yeast model. *Mitochondrion.* **11**, 182-190. doi: 10.1016/j.mito.2010.09.007. ISSN: 1567-7249

11) Stewart J.D., Horvath R., **Baruffini E.**, Ferrero I., Bulst S., Watkins P.B., Fontana R.J., Day C.P., Chinnery P.F. (2010) Polymerase γ gene POLG determines the risk of sodium valproate-induced liver toxicity. *Hepatology.* **52**, 1791-1796. doi: 10.1002/hep.23891. ISSN: 0270-9139

10) **Baruffini E.**, Ferrero I., Foury F. (2010) In vivo analysis of mtDNA replication defects in yeast. *Methods.* **51**, 426-436. doi: 10.1016/j.ymeth.2010.02.023. ISSN: 1046-2023

9) **Baruffini E.**, Lodi T. (2010) Construction and validation of a yeast model system for studying in vivo the susceptibility to nucleoside analogues of DNA polymerase gamma allelic variants. *Mitochondrion.* **10**, 183-187. doi: 10.1016/j.mito.2009.10.002. ISSN:1567-7249 (**Corresponding author**)

8) Stricker S., Prüss H., Horvath R., **Baruffini E.**, Lodi T., Siebert E., Endres M., Zschenderlein R., Meisel A. (2009) A variable neurodegenerative phenotype with polymerase gamma mutation. *J Neurol Neurosurg Psychiatry.* **80**, 1181-1182. doi: 10.1136/jnnp.2008.166066. ISSN: 0022-3050

7) **Baruffini E.**, Serafini F., Lodi T. (2009) Construction and characterization of centromeric, episomal and GFP-containing vectors for *Saccharomyces cerevisiae* prototrophic strains. *J Biotechnol.* **143**, 247-254. doi: 10.1016/j.jbiotec.2009.08.007. ISSN: 0168-1656

6) Fukasawa T., Sakurai H., Nogi Y., **Baruffini E.** (2009) Galactose transporters discriminate steric anomers at the cell surface in yeast. *FEMS Yeast Res.* **9**, 723-731. doi: 10.1111/j.1567-1364.2009.00517.x. ISSN: 1567-1356

5) Spinazzola A., Invernizzi F., Carrara F., Lamantea E., Donati A., Dirocco M., Giordano I., Meznaric-Petrusa M., **Baruffini E.**, Ferrero I., Zeviani M. (2009) Clinical and molecular features of mitochondrial DNA depletion syndromes. *J Inherit Metab Dis.* **32**, 143-158. doi: 10.1007/s10545-008-1038-z. ISSN:0141-8955

4) **Baruffini E.**, Ferrero I., Foury F. (2007) Mitochondrial DNA defects in *Saccharomyces cerevisiae* caused by functional interactions between DNA polymerase gamma mutations associated with disease in human. *Biochim Biophys Acta. Molecular Basis of Disease* **1772**, 1225-1235. doi:10.1016/j.bbadis.2007.10.002. ISSN: 0925-4439

3) **Baruffini E.**, Lodi T., Dallabona C., Foury F. (2007) A single nucleotide polymorphism in the DNA polymerase gamma gene of *Saccharomyces cerevisiae* laboratory strains is responsible for increased mitochondrial DNA mutability. *Genetics.* **177**, doi: 10.1534/genetics.107.079293. ISSN: 0016-6731

2) **Baruffini E.**, Goffrini P., Donnini C., Lodi T. (2006) Galactose transport in *Kluyveromyces lactis*: major role of the glucose permease Hgt1. *FEMS Yeast Res.* **6**, 1235-1242. doi: 10.1111/j.1567-1364.2006.00107.x. ISSN: 1567-1356

1) **Baruffini E.**, Lodi T., Dallabona C., Puglisi A., Zeviani M., Ferrero I. (2006) Genetic and chemical rescue of the *Saccharomyces* cerevisiae phenotype induced by mitochondrial DNA polymerase mutations associated with progressive external ophthalmoplegia in humans. *Hum Mol Genet.* **15**, 2846-2855. doi: 10.1093/hmg/ddl219. ISSN: 0964-6906

## Attachment 2: patents

European and US patent number EP3226850 and US20170266136
Title: Compounds for the treatment of mitochondrial diseases
Inventors: Delahodde Agnès [Fr]; Pitayu Nugroho Laras [Fr]; Baruffini Enrico [It]; Lodi Tiziana [It]; Rötig Agnès [Fr]; Procaccio Vincent [Fr]
Applicants: Université Paris-Sud [Fr]; Centre Nat Rech Scient [Fr]; Univ D'angers [Fr]; Centre Hospitalier Univ D'angers [Fr]

French Patent pending number WO2016087771.
Title: COMPOUNDS FOR THE TREATMENT OF MITOCHONDRIAL DISEASES
Inventors: DELAHODDE AGNÈS [FR]; PITAYU LARAS Ajeng [FR]; BARUFFINI ENRICO [IT]; LODI TIZIANA [IT]; RÖTIG AGNÈS [FR]; PROCACCIO VINCENT [FR]
Applicants: UNIV PARIS SUD [FR]; CENTER NAT RECH Scient [FR]; UNIV ANGERS [FR];
CT HOSPITALIER UNIVERSITAIRE D ANGERS [FR]

## Attachment 3: funded projects

**Ongoing**

2018-2021 PI. Leonardo Caporali
Italian “Ricerca Finalizzata 2016” Project GR-2016-02361449: “ Italian Project on Hereditary Optic Neropathies (IPHON): from genetic basis to therapy”
Role: Unit Leader

01/11/2015-31/10/2018 PI: Ghezzi Daniele
Italian TELETHON Project GGP15041: "MitMed consortium: from the identification and characterization of nuclear genes responsible for human mitochondrial disorders towards potential therapeutic approaches in experimental models"
Role: Participant

**Completed**

03/14/2014-3/14/2017 PI: Zanna Claudia
FIRB (Italian Future in Research) Project RBFR13IWDS: "From yeast to human: how OPA1 isoforms and pathogenic mutations cause neurodegenerations characterized by mtDNA instability” at the Department of Life Sciences, University of Parma, Parma, Italy
Role: Unit Leader

12/15/2011-12/15/2014 PI: Zeviani Massimo
Italian TELETHON Project GGP11011: “MitMed: a multicenter consortium for the identification and characterization of nuclear genes responsible for human mitochondrial disorders”
Role: Participant

2007-2010 PI: Zeviani Massimo
Italian TELETHON Project GGP07019: “Identification and characterization of nuclear genes responsible for human mitochondrial disorders”
Role: Participant