

CURRICULUM VITAE



PERSONAL INFORMATION

Name **CLARICE PATRONO**
E-mail **clarice.patrono@enea.it**
clarypat@gmail.com
Nationality Italian
Date of birth 12 OCTOBER 1968

WORK EXPERIENCE

- Employer's name and locality
• Sector
• Position
• Main activities and responsibilities

December 2008 – today
ENEA, Casaccia Research Center (Rome, Italy), Division of Health Protection Technologies, Laboratory of Health and Environment
Research organization
Research Scientist
Research activities in the field of cell biology and radiobiology. Studies of *in vitro* radiation-induced biological effects, particularly concerning the evaluation of cytogenetic damage, cell viability and cell death mechanisms. Analysis of *in vitro* effects of hyperthermia as radiosensitiser.
Molecular biology studies on the role of polymorphisms of DNA repair genes in cancer susceptibility and individual radiosensitivity.
Biomonitoring and retrospective biological dosimetry studies. Evaluation of cytogenetic damage in circulating lymphocytes of patients undergoing radiotherapy or targeted radionuclide therapy.
- Participation in research projects

From April 2020: Participation in the Multi-Year Project **BioPhyMeTRE "Novel Biological! and Physical Methods for Triage in Radiological and Nuclear (RIN) Emergencies"** funded by NATO "Science for Peace and Security Programme"
2019: Participation in the Tender HOME Tor13 **"The preparation of a Biosecurity toolbox to strengthen European Biosecurity"** funded by the European Commission
2012-2015: Participation in the EU-EMRP Joint Research Project **BioQuaRT "Biologically weighted Quantities in radiotherapy"**
2012-2015: Participation in the experimental activities of the Coordination Action **RENEB "Realizing the European Network of Biodosimetry"** within the EU FP-7 (EURATOM Fission Programme)
2009-today: Participation in the national project **TOP-IMPLART "Intensity Modulated Proton Linear Accelerator for Radiotherapy"**, funded by Regione Lazio.
- Participation in Working Groups

From 2020: Participation in the "Biosecurity: monitoring and surveillance measures" Working Group of the **"Italian National Committee for Biosafety, Biotechnology and Sciences of Life" - CNBBSV.**
- Employer's name and locality
• Sector
• Position
• Main activities and responsibilities

January 2006 – December 2008
"Fondazione Santa Lucia" (Rome, Italy), Laboratory of Neurogenetics
Scientific Institute for Research, Hospitalisation and Health Care (IRCCS)
Research fellow in experimental neurology
Laboratory and research activities on the molecular genetics of hereditary spastic paraplegias and on the role of genetic polymorphisms in neurodegenerative diseases.
- Employer's name and locality
• Sector
• Position
• Main activities and responsibilities

April 2004 - December 2005
San Raffaele Pisana (Rome, Italy), Laboratory of Molecular Biology
Scientific Institute for Research, Hospitalisation and Health Care (IRCCS)
Research Fellow
Collaboration with the Institute of Neurobiology and Molecular Medicine of the Italian national research council (CNR): study of oxidative stress and apoptosis on primary murine cells and human and murine cell lines.

- Employer's name and locality
 - Sector
 - Position
- Main activities and responsibilities

1997 – 2003

Children's Hospital "Bambino Gesù" Rome, Italy

Scientific Institute for Research, Hospitalisation and Health Care (IRCCS), Laboratory of Molecular Medicine

Research grant

Participation in research projects on molecular diagnostics and genetics of mitochondrial and neurodegenerative diseases.

- Employer's name and locality
 - Sector
 - Position
- Main activities and responsibilities

2001

Instituto de Investigaciones Biomédicas "Alberto Sols" CSIC-UAM (Madrid, Spain), Department of Biochemistry

University

Research fellow

Research activities in the fields of biochemistry, cell and molecular biology within the project "Upgrade in cellular systems of mtDNA-related disorders".

- Employer's name and locality
 - Sector
 - Position
- Main activities and responsibilities

1996 – 1997

"BIOS" S.p.A. (Rome, Italy)

Centre of laboratory analysis and specialistic diagnostics

Consultant

Laboratory activities in the Molecular Biology Unit: molecular diagnostics of HCV, HBV, HIV, paternity testing.

- Employer's name and locality
 - Sector
 - Position
- Main activities and responsibilities

1990 – 1994

University of Rome "La Sapienza" (Rome, Italy), Department of Biochemical Sciences

University

Student, fellow intern

Research activities in biochemistry, in the fields of enzyme and protein purification, peptide purification and sequencing, enzyme kinetics and inhibition.

EDUCATION AND TRAINING

- Education/Training organization's name and locality
 - Qualification awarded

April-June 2020

ENEA – Online training course

Training course "Project Management e nuovi modelli di leadership".

- Education/Training organization's name and locality
 - Qualification awarded

February 2020

IVTECH - ENEA Casaccia Research Center (Rome, Italy)

IVTECH theoretical-practical course "12th Workshop on 3D advanced in-vitro Models"

- Education/Training organization's name and locality
 - Qualification awarded

June 2017

IRSN – Institut de Radioprotection et de Sûreté Nucléaire (Paris, France) - EURADOS

Training course "Uncertainty analysis for retrospective dosimetry and associated research"

- Education/Training organization's name and locality
 - Qualification awarded
 - PhD Thesis

2010

"University of Perugia" (Perugia, Italy), Department of Experimental Medicine and Biochemical Sciences

PhD in "Molecular Biology and Biotechnology"

"Molecular approaches for the genetic study of autosomal dominant forms of hereditary spastic paraplegia"

- Education/Training organization's name and locality
- Principal subjects / skills acquired
 - Qualification awarded

- Education/Training organization's name and locality
 - Qualification awarded
 - Master's Thesis

- Education/Training organization's name and locality
 - Qualification awarded

- Education/Training organization's name and locality
 - Qualification awarded
 - Thesis

PERSONAL SKILLS
MOTHER TONGUE

OTHER LANGUAGES

- Reading
- Writing
- Speaking

- Reading
- Writing
- Speaking

TECHNICAL SKILLS

2000

University of Rome "La Sapienza" (Rome, Italy)

Biological Sciences

Grant for the attendance of master classes abroad

1996

University of Rome "La Sapienza" (Rome, Italy)

Master Course in "Biotechnology"

"Design of an immunoenzymatic assay for the detection of gene mutations after *in vitro* DNA amplification: applications to cystic fibrosis and β -thalassemia"

1994

University of Rome "La Sapienza" (Rome, Italy)

Professional qualification as biologist

1992

University of Rome "La Sapienza" (Rome, Italy)

Degree in Biological Sciences (110/100 cum laude)

"Determination of the primary structure of the GABA-transaminase enzyme from pig liver"

ITALIAN

ENGLISH

VERY GOOD

VERY GOOD

GOOD

SPANISH

GOOD

GOOD

GOOD

Biochemistry: protein extraction and purification; chromatography, electrophoresis and spectrophotometry techniques; Western blot.

Molecular biology: molecular diagnosis of genetic diseases with PCR, screening of point mutations and DNA automatic sequencing. Genotyping, genetic association studies and linkage analysis. Cloning of PCR fragments and human cDNA in procariotic and eucariotic vectors. Transient and stable transfections. Southern and Northern blot.

Cell biology: maintenance and propagation of human and animal cell cultures. Study of apoptosis and oxidative stress in primary murine cells and in human and murine cell lines (determination of DNA Fragmentation, TUNEL assay, dosage of caspases). Cell viability and citotoxicity assays, clonogenic assay. Cell staining (Hoechst, Sybr gold, May-Grünwald and Giemsa) and immunofluorescence. Determination of chromosomal damage (chromosomal aberrations, micronuclei) induced by ionising and non-ionising radiation or genotoxic agents in human lymphocytes or cultured cells.

Radiobiology: cell culture exposure to ionising radiations (x-rays, gamma rays, protons, alpha-particles); cell irradiation with ion microbeam. Cytogenetic assays used in biological dosimetry (Dicentric Chromosome Assay and Cytokinesis-Block Micronucleus Assay); retrospective dose reconstruction for individuals accidentally exposed to ionising radiation.

PUBLICATIONS

- 1** Testa A, PATRONO C, Palma V, Kenzhina L, Mamyrbayeva A, Biyakhmetova D, Zhamaldinov F, Della Monaca S, Fattibene P, Quattrini MC, Maltar-Strmečki N, Erceg I, Vojnić Kortmiš M, Vidotto M, Bortolin E (2020). NATO Science for Peace and Security (SPS) project "BioPhyMeTRE": "Novel biological and physical methods for triage in radiological and nuclear (R/N) emergencies". *Il nuovo cimento C*, 43 (6). pp. 1-8. doi: 10.1393/ncc/i2020-20153-9
- 2** Picardi L, Ampollini A, Anello P, Balduzzi M, Bazzano G, Borgognoni F, Cisbani E, D'Andrea M, De Angelis C, De Angelis G, Della Monaca S, Esposito G, Ghio F, Giuliani F, Lucentini M, Marino C, Montereali RM, Nenzi P, Notaro C, PATRONO C, Placido C, Piccinini M, Ronsivalle C, Santavenere F, Spurio A, Strigari L, Surrenti V, Tabocchini A, Trinca E, Vadrucci M (2020). Status and development of the TOP-IMPLART Project. arXiv:2010.05608 (preprint).
- 3** Testa A, Palma V, PATRONO C (2019). Chapter 5: Dicentric Chromosome Assay (DCA) and Cytokinesis-Block Micronucleus (CBMN) Assay in the Field of Biological Dosimetry. Dhawan A & Bajpayee M Eds. *Genotoxicity Assessment. Methods and Protocols; Second Edition*. Humana Press. METHODS MOL BIOL. 2031:p.105-119. doi: 10.1007/978-1-4939-9646-9_5
- 4** Testa A, Palma V, PATRONO C (2019). A NOVEL BIOLOGICAL DOSIMETRY ASSAY AS A POTENTIAL TOOL FOR TRIAGE DOSE ASSESSMENT IN CASE OF LARGE-SCALE RADIOLOGICAL EMERGENCY. *RADIAT PROT DOSIMETRY*. 186(1):9-11. doi: 10.1093/rpd/ncz001
- 5** Testa A, Ballarini F, Giesen U, Gil OM, Carante MP, Tello J, Langner F, Rabus H, Palma V, Pinto M, PATRONO C (2018). Analysis of Radiation-Induced Chromosomal Aberrations on a Cell by-Cell Basis after Alpha-Particle Microbeam Irradiation: Experimental Data and Simulations. *RADIAT RES*. 189(6):597-604. doi: 10.1667/RR15005.1
- 6** Pietraforte D, Paulicelli E, PATRONO C, Gambardella L, Scorza G, Testa A, Fattibene P (2018). Protein oxidative damage and redox imbalance induced by ionising radiation in CHO cells. *FREE RADIC RES*. 52(4):465-479. doi:10.1080/10715762.2018.1446529
- 7** Gaddini L, Balduzzi M, Campa A, Esposito G, Malchiodi-Albedi F, PATRONO C, Matteucci A (2018). Exposing primary rat retina cell cultures to γ -rays: An in vitro model for evaluating radiation responses. *Exp Eye Res*;166:21-28. doi: 10.1016/j.exer.2017.09.009
- 8** Franconi R, Testa A, Giovanetti A, Lopresto V, Pinto R, Turchi R, Iliano E, Massa S, PATRONO C, Palma V, Rinaldi A, Bartoleschi C, Pardini MC, Demurtas O, Cappelli M, Marino C (2017). CBRN Emergencies: Integrated Approach to Develop Innovative Technology and Training of Operators. *BIOMEDICINE & PREVENTION*, vol. 4 (PART 2); 133. DOI:10.19252/000000085
- 9** Kulka U, Abend M, Ainsbury E, Badie C, Barquinero JF, Barrios L, Beinke C, Bortolin E, Cucu A, De Amicis A, Domínguez I, Fattibene P, Frøvig AM, Gregoire E, Guogyte K, Hadjidekova V, Jaworska A, Kriehuber R, Lindholm C, Lloyd D, Lumniczky K, Lyng F, Meschini R, Mörtl S, Della Monaca S, Monteiro Gil O, Montoro A, Moquet J, Moreno M, Oestreicher U, Palitti F, Pantelias G, PATRONO C, Piqueret-Stephan L, Port M, Prieto MJ, Quintens R, Ricoul M, Romm H, Roy L, Sáfrány G, Sabatier L, Sebastià N, Sommer S, Terzoudi G, Testa A, Thierens H, Turai I, Trompier F, Valente M, Vaz P, Voisin P, Vral A, Woda C, Zafiroopoulos D, Wojcik A (2017). RENEb - Running the European Network of biological dosimetry and physical retrospective dosimetry. *INT J RADIAT BIOL*, vol 93(1); p. 2-14. doi: 10.1080/09553002.2016.1230239
- 10** Brzozowska B, Ainsbury E, Baert A, Beaton-Green L, Barrios L, Barquinero JF, Bassinet C, Beinke C, Benedek A, Beukes P, Bortolin E, Buraczewska I, Burbidge C, De Amicis A, De Angelis C, Della Monaca S, Depuydt J, De Sanctis S, Dobos K, Domene MM, Domínguez I, Facco E, Fattibene P, Frenzel M, Monteiro Gil O, Gonon G, Gregoire E, Gruel G, Hadjidekova V, Hatzi VI, Hristova R, Jaworska A, Kis E, Kowalska M, Kulka U, Lista F, Lumniczky K, Martínez-López W, Meschini R, Moertl S, Moquet J, Noditi M, Oestreicher U, Orta Vázquez ML, Palma V, Pantelias G, Montoro Pastor A, PATRONO C, Piqueret-Stephan L, Quattrini MC, Regalbuto E, Ricoul M, Roch-Lefevre S, Roy L, Sabatier L, Sarchiapone L, Sebastià N, Sommer S, Sun M, Suto Y, Terzoudi G, Trompier F, Vral A, Wilkins R, Zafiroopoulos D, Wieser A, Woda C, Wojcik A (2017). RENEb accident simulation exercise. *INT J RADIAT BIOL*, vol. 93(1); p. 75-80. doi: 10.1080/09553002.2016.1206230
- 11** Ainsbury E, Badie C, Barnard S, Manning G, Moquet J, Abend M, Antunes AC, Barrios L, Bassinet C, Beinke C, Bortolin E, Bossin L, Bricknell C, Brzoska K, Buraczewska I, Castaño CH, Čemusová Z, Christiansson M, Cordero SM, Cosler G, Monaca SD, Desangles F, Discher M, Domínguez I, Doucha-Senf S, Eakins J, Fattibene P, Filippi S, Frenzel M, Georgieva D, Gregoire E, Guogyte K, Hadjidekova V, Hadjiiska L, Hristova R, Karakosta M, Kis E, Kriehuber R, Lee J, Lloyd D, Lumniczky K, Lyng F, Macaeva E, Majewski M, Vanda Martins S, McKeever SW, Meade A, Medipally D, Meschini R, M'kacher R, Gil

OM, Montero A, Moreno M, Noditi M, Oestreicher U, Oskamp D, Palitti F, Palma V, Pantelias G, Pateux J, PATRONO C, Pepe G, Port M, Prieto MJ, Quattrini MC, Quintens R, Ricoul M, Roy L, Sabatier L, Sebastià N, Sholom S, Sommer S, Staynova A, Strunz S, Terzoudi G, Testa A, Trompier F, Valente M, Hoey OV, Veronese I, Wojcik A, Woda C (2017). Integration of new biological and physical retrospective dosimetry methods into EU emergency response plans - joint RENEB and EURADOS inter-laboratory comparisons. *INT J RADIAT BIOL*, vol. 93(1); p.99-109. doi: 10.1080/09553002.2016.1206233

12 PATRONO C, Sterpone S, Testa A, Verna A, Palma V, Gentile P, Cozzi R (2015). Polymorphisms in X-Ray Repair Cross-Complementing Group 1 Gene: Haplotypes, Breast Cancer Risk and Individual Radiosensitivity. *OPEN MEDICINE JOURNAL*, vol 2; p. 25-30 ISSN: 1874-2203

13 PATRONO C, Monteiro Gil O, Giesen U, Langner F, Pinto M, Rabus H, Testa A (2015). 'BioQuaRT' project: design of a novel in situ protocol for the simultaneous visualisation of chromosomal aberrations and micronuclei after irradiation at microbeam facilities. *RADIATION PROTECTION DOSIMETRY*, vol 166; p. 197-9. ISSN 0144-8420, doi: 10.1093/rpd/ncv160

14 PATRONO C, Sterpone S, Testa A, Cozzi R (2014). Polymorphisms in base excision repair genes: Breast cancer risk and individual radiosensitivity. *WORLD JOURNAL OF CLINICAL ONCOLOGY*, vol 5; p. 874-82. ISSN 2218-4333, doi: 10.5306/wjco.v5.i5.874

15 Cornetta T, PATRONO C, Terrenato I, De Nigris F, Bentivoglio AR, Testa A, Palma V, Poggioli T, Padua L, Cozzi R (2013). Epidemiological, clinical, and molecular study of a cohort of Italian Parkinson disease patients: association with glutathione-S-transferase and DNA repair gene polymorphisms. *CELLULAR AND MOLECULAR NEUROBIOLOGY*, vol 33; p. 673-80. ISSN 0272-4340, doi: 10.1007/s10571-013-9933-8

16 Orlacchio A, Babalini C, Borreca A, PATRONO C., Massa R, Basaran S, Munhoz R P., Rogaeva E A., St George-Hyslop P H., Bernardi G, Kawarai T (2010). SPATACSIN mutations cause autosomal recessive juvenile amyotrophic lateral sclerosis. *BRAIN*, vol. 133; p. 591-598, ISSN: 0006-8950, doi: 10.1093/brain/awp325

17 Sterpone S, Mastellone V, Padua L, Novelli F, PATRONO C., Cornetta T, Giammarino D, Donato V, Testa A, Cozzi R (2010). Single-nucleotide polymorphisms in BER and HRR genes, XRCC1 haplotypes and breast cancer risk in Caucasian women. *JOURNAL OF CANCER RESEARCH AND CLINICAL ONCOLOGY*, vol. 136; p. 631-636, ISSN: 0171-5216, doi: 10.1007/s00432-010-0791-1

18 Pippucci T, Panza E, Pompili E, Donadio V, Borreca A, Babalini C, PATRONO C., Zuntini R, Kawarai T, Bernardi G, Liguori R, Romeo G, Montagna P, Orlacchio A, Seri M (2009). Autosomal recessive hereditary spastic paraplegia with thin corpus callosum: a novel mutation in the SPG11 gene and further evidence for genetic heterogeneity. *EUROPEAN JOURNAL OF NEUROLOGY*, vol. 16; p. 121-126, ISSN: 1351-5101, doi: 10.1111/j.1468-1331.2008.02367.x

19 Orlacchio A, PATRONO C., Gaudiello F, Rocchi C, Moschella V, Floris R, Bernardi G, Kawarai T (2008). Silver syndrome variant of hereditary spastic paraplegia - A locus to 4p and allelism with SPG4. *NEUROLOGY*, vol. 70; p. 1959-1966, ISSN: 0028-3878, doi: 10.1212/01.wnl.0000294330.27058.61

20 Orlacchio A, PATRONO C., Borreca A, Babalini C, Bernardi G, Kawarai T (2008). Spastic paraplegia in Romania: high prevalence of SPG4 mutations. *JOURNAL OF NEUROLOGY, NEUROSURGERY AND PSYCHIATRY*, vol. 79; p. 606-607, ISSN: 0022-3050, doi: 10.1136/jnnp.2007.128827

21 Orlacchio A, PATRONO C, Orlacchio A, St George-Hyslop PH, Bernardi G, Kawarai T (2006). Clinical and genetic studies on autosomal dominant hereditary spastic paraplegia. Online publication: www.siicsalud.com

22 Bornstein B, Mas JA, PATRONO C., Fernandez-Moreno MA, Gonzalez-Vioque E, Campos Y, Carrozzo R, Martin MA, del Hoyo P, Santorelli FM, Arenas J, Garesse R (2005). Comparative analysis of the pathogenic mechanisms associated with the G8363A and A8296G mutations in the mitochondrial tRNA(Lys) gene. *BIOCHEMICAL JOURNAL*, vol. 387; p. 773-778, ISSN: 0264-6021, doi: 10.1042/BJ20040949

23 Montagna G, Di Biase A, Cappa M, Melone MA, Piantadosi C, Colabianchi D, PATRONO C, Attori L, Cannelli N, Cotrufo R, Salvati S (2005). Identification of seven novel mutations in ABCD1 by a DHPLC-based assay in Italian patients with X-linked adrenoleukodystrophy Identification of seven novel mutations in ABCD1 by a DHPLC-based assay in Italian patients with X-linked adrenoleukodystrophy. *HUMAN MUTATION*. Vol. 25 (2), p. 222. doi: 10.1002/humu.9303

24 Patrono C, Scarano V, Cricchi F, Melone MA, Chiriaco M, Napolitano A, Malandrini A, De Michele G, Petrozzi L, Giraldi C, Santoro, L., Servidei, S., Casali, C., Filla, A., Santorelli, F.M (2005). Autosomal dominant hereditary spastic paraplegia: DHPLC-based mutation analysis of SPG4 reveals eleven novel mutations *HUMAN MUTATION*, Vol. 25 (5), p. 506. doi: 10.1002/humu.9340

- 25** Bertini E, PATRONO C, Dionisi-Vici C, Francalanci P, Fogli A, Eymard-Pierre E, Santorelli FM, Boespflug-Tanguy O (2004). Chapter 8: Experience on two recently discovered forms of undetermined leukodystrophy. Uziel G. & Taroni F. Eds. Hereditary Leukoencephalopathies and Demyelinating Neuropathies in Children. John Libbey Eurotext, Montrouge, France. Mariani Foundation Paediatric Neurology Series: 12; p. 79-86.
- 26** PATRONO C (2004). Megalencephalic-cystic leukodystrophy. Orphanet Encyclopedia. Patrono C. Megalencephalic-cystic leukodystrophy. Orphanet Encyclopedia. Online publication: www.orpha.net
- 27** Siciliano G, Pasquali L, Manca ML, Del Corona A, Tessa A, Patrono C, Prontera C, Zucchelli G, Santorelli FM (2003). The relationship between anaerobic lactate threshold and plasma catecholamines during incremental exercise in hereditary spastic paraplegia. *FUNCTIONAL NEUROLOGY*, vol. 18(2), p. 83-87, ISSN:0393-5264.
- 28** PATRONO C., Di Giacinto G, Eymard-Pierre E, Santorelli FM, Rodriguez D, De Stefano N, Federico A, Gatti R, Benigno V, Megarbane A, Tabarki B, Boespflug-Tanguy O, Bertini E (2003). Genetic heterogeneity of megalencephalic leukoencephalopathy and subcortical cysts. *NEUROLOGY*, vol. 61; p. 534-537, ISSN: 0028-3878, doi: 10.1212/01.WNL.0000076184.21183.CA
- 29** Tessa A, Casali C, Damiano M, Bruno C, Fortini D, PATRONO C, Cricchi F, Valoppi M, Nappi G, Amabile GA, Bertini E, Santorelli FM (2002). An additional family carrying a new atlanti mutation. *NEUROLOGY*, vol. 59; p. 2002-2005, ISSN: 0028-3878, doi: 10.1212/01.WNL.0000036902.21438.98
- 30** PATRONO C., Dionisi-Vici C, Giannotti A, Bembi B, Digilio MC, Rizzo C, Purificato C, Martini C, Pierini R, Santorelli FM (2002). Two novel mutations of the human Delta 7-sterol reductase (DHCR7) gene in children with Smith-Lemli-Opitz syndrome. *MOLECULAR AND CELLULAR PROBES*, vol. 16; p. 315-318, ISSN: 0890-8508, doi: 10.1006/mcpr.2002.0426
- 31** Valente EM, Brancati F, Caputo V, Bertini E, PATRONO C., Costanti D, Dallapiccola B (2002). Novel locus for autosomal dominant pure hereditary spastic paraplegia (SPG19) maps to chromosome 9q33-q34. *ANNALS OF NEUROLOGY*, vol. 51; p. 681-685, ISSN: 0364-5134, doi: 10.1002/ana.10204
- 32** PATRONO C., Casali C, Tessa A, Cricchi F, Fortini D, Carrozzo R, Siciliano G, Bertini E, Santorelli FM (2002). Missense and splice site mutations in SPG4 suggest loss-of-function in dominant spastic paraplegia. *JOURNAL OF NEUROLOGY*, vol. 249; p. 200-205, ISSN: 0340 5354, doi: 10.1007/PL00007865
- 33** Carrozzo R, Tessa A, Vazquez-Memije ME, Piemonte F, PATRONO C., Malandrini A, Dionisi Vici C, Vilarinho L, Villanova M, Schagger H, Federico A, Bertini E, Santorelli F (2001). The T9176G mtDNA mutation severely affects ATP production and results in Leigh syndrome. *NEUROLOGY*, vol. 56; p. 687-690, ISSN: 0028-3878, doi: 10.1212/WNL.56.5.687
- 34** Piemonte F, Casali C, Carrozzo R, Schagger H, PATRONO C., Tessa A, Tozzi G, Cricchi F, Di Capua M, Siciliano G, Amabile GA, Morocutti C, Bertini E, Santorelli FM (2001). Respiratory chain defects in hereditary spastic paraplegias. *NEUROMUSCULAR DISORDERS*, vol. 11; p. 565-569, ISSN: 0960-8966, doi: 10.1016/S0960-8966(01)00214-0
- 35** Tessa A, Carbone I, Matteoli MC, Bruno C, PATRONO C, Patera IP, De Luca F, Lorini R, Santorelli FM (2001). Identification of Novel WFS1 Mutations in Italian Children With Wolfram Syndrome. *HUMAN MUTATION*, 17(4), p. 348-349. doi: 10.1002/humu.32
- 36** Santorelli FM, PATRONO C., Fortini D, Tessa A, Comanducci G, Bertini E, Pierallini A, Amabile GA, Casali C (2000). Intrafamilial variability in hereditary spastic paraplegia associated with an SPG4 gene mutation. *NEUROLOGY*, vol. 55; p. 702-705, ISSN: 0028-3878, doi: 10.1212/WNL.55.5.702
- 37** Santoro L, Carrozzo R, Malandrini A, Piemonte F, PATRONO C., Villanova M, Tessa A, Palmeri S, Bertini E, Santorelli FM (2000). A novel SURF1 mutation results in Leigh syndrome with peripheral neuropathy caused by cytochrome c oxidase deficiency. *NEUROMUSCULAR DISORDERS*, vol. 10; p. 450-453, ISSN: 0960-8966, doi: 10.1016/S0960-8966(99)00122-4
- 38** Tessa A, PATRONO C., Santorelli FM, Giannotti A, Digilio MC, Pacifico C, Presuttari F, Tieri L (2000). Rapid detection of the 35delG mutation in the GJB2 gene in childhood deafness. *JOURNAL OF MEDICAL SCREENING*, vol. 7; p. 167, ISSN: 0969-1413, doi: 10.1136/jms.7.3.167
- 39** PATRONO C., Rizzo C, Tessa A, Giannotti A, Borrelli P, Carrozzo R, Piemonte F, Bertini E, Dionisi-Vici C, Santorelli FM (2000). Novel 7-DHCR mutation in a child with Smith-Lemli-Opitz syndrome. *AMERICAN JOURNAL OF MEDICAL GENETICS*, vol. 91; p. 138-140. ISSN:0148-7299, doi: 10.1002/(SICI)1096-8628(20000313)91:2<138::AID-AJMG12>3.0.CO;2-Q
- 40** Santorelli FM, Piemonte F, Carrozzo R, Tessa A, PATRONO C., Tozzi G, Bertini E (2000). OXPHOS and mtDNA alterations in a family with spastic paraparesis. *ACTA NEUROLOGICA SCANDINAVICA*, vol. 101; p. 255-258. ISSN: 0001-6314, doi: 10.1034/j.1600-0404.2000.101004255.x

41 Giannotti A, Tessa A, PATRONO C, De Florio L, Velardo M, Dionisi-Vici C, Bertini E, Santorelli FM (2000). A novel CBFA1 mutation (R190W) in an Italian family with cleidocranial dysplasia. HUMAN MUTATION, vol. 16, Issue 3, p. 277. doi: 10.1002/1098-1004(200009)16:3<277::AID-HUMU25>3.0.CO;2-V

Strigari L, Dini V, Ungania S, Testa A, Balduzzi M, Soriani A, Marconi R, Sciuto R, PATRONO C, Palma v, Tabocchini MA (2018). Biological effects evaluation in peripheral blood lymphocytes from patients undergoing radium-223 chloride (²²³RaCl₂) therapy. 2nd European Congress of Medical Physics 2008, Copenhagen, Denmark. PHYSICA MEDICA, Vol. 52 (s.1); p. 64-65, doi: 10.1016/j.ejmp.2018.06.240

Dini V, Testa A, PATRONO C, Balduzzi M, Palma V, Sciuto R, Soriani A, Strigari L, Marconi R, Tabocchini MA (2018). Biological effects evaluation in peripheral blood lymphocytes from patients undergoing radium-223 (²²³RaCl₂) therapy. Associazione Italiana di Fisica Medica, 10th National Congress 2018, Bari. PHYSICA MEDICA, Vol. 56(S.2); p. 115, doi: 10.1016/j.ejmp.2018.04.096

PATRONO C, Lopresto V, Balduzzi M, Leter G, Piccirillo P, Testa A, Palma V, Marino C (2018). U-251 human glioblastoma cell line model to study hyperthermia as radiosensitizer. 32nd Annual Meeting of the European Society for Hyperthermic Oncology 2018, Berlin. STRAHLENTHER ONKOL vol. 194, pp. 489-490, doi: 10.1007/s00066-018-1295-1

Orlacchio A, Babalini C, PATRONO C., Montieri P, Varlese M L, Borreca A, Moschella V, Mercuri N B, Bernardi G, Kawarai T (2009). ALS phenotypes with mutations in SPG11. 13th International Congress of Parkinson's Disease and Movement Disorders 2009, Paris, France. MOVEMENT DISORDERS, vol. 24(s.1); p. S151, ISSN: 0885-3185

Orlacchio A, Borreca A, PATRONO C., Babalini C, Mercuri N B, Bernardi G, Kawarai T (2008). Autosomal recessive juvenile ALS associated with spatacsin mutations. 12th International Congress of Parkinson's Disease and Movement Disorders 2008, MOVEMENT DISORDERS, vol. 23; p. S48, ISSN: 0885-3185

Orlacchio A, Babalini C, Varlese ML, PATRONO C, Borreca A, Esposito Z, Sancesario G, Orlacchio A, Kawarai T (2008). ASSOCIATION STUDY OF THE APOA1 GENE IN AN ITALIAN POPULATION WITH ALZHEIMER'S DISEASE. Alzheimer's Association International Conference on Alzheimer's Disease 2008, Chicago, IL. ALZHEIMER'S & DEMENTIA, vol. 4 (s. 4), p. T593, doi: 10.1016/j.jalz.2008.05.1815

Orlacchio A, Massini C, Tiribuzi R, Costanzi E, Makrypidi G, Mattoli F, Esposito Z, PATRONO C, Miele ML, Maiotti M, Sancesario G, Zampolini M, Orlacchio A, Martino S (2008). IDENTIFICATION OF BIOCHEMICAL MARKERS FOR THE DIAGNOSIS OF ALZHEIMER'S DISEASE IN PATIENTS AFFECTED BY TYPE II DIABETES. Alzheimer's Association International Conference on Alzheimer's Disease 2008, Chicago, IL. ALZHEIMER'S & DEMENTIA, vol. 4 (s. 4), p. T335, doi: 10.1016/j.jalz.2008.05.977

Orlacchio A, Miele M, PATRONO C, Borreca A, Varlese M, Babalini C, Dionisi L, Moschella V, Orlacchio A, Bernardi G, Kawarai T (2007). Association of APOE ε4 Allele with Vascular Dementia in Italian Population. Alzheimer's disease: new advanced. 10th International conference on and related disorders (Madrid, 16-20 July 2006). Medimond.

Orlacchio A, PATRONO C., Gaudiello F, Moschella V, Borreca A, Orlacchio A, Floris R, Bernardi G, Kawarai T (2007). Silver syndrome variant of hereditary spastic paraplegia: Identification of a novel locus. 11th International Congress of Parkinson's Disease and Movement Disorders 2007, Istanbul, Turkey. MOVEMENT DISORDERS, vol. 22(s.16); p. S2, ISSN: 0885-3185.

Orlacchio An, PATRONO C., Gaudiello F, Borreca A, Moschella V, Orlacchio AI, Floris R, Bernardi G, Kawarai T (2007). A new locus for the Silver syndrome variant of hereditary spastic paraplegia. 17th Meeting of the European Neurological Society 2007, Rhodes, Greece. JOURNAL OF NEUROLOGY, vol. 254 (s.3); p. 19-20, ISSN: 0340-5354, doi: 10.1007/s00415-007-3001-6

Orlacchio An, PATRONO C., Borreca A, Babalini C, Dionisi L, Moschella V, Orlacchio AI, Bernardi G, Kawarai T (2006). Clinical and genetic study of two Italian families with silver syndrome. 16th Meeting of the European Neurological Society 2006, Lausanne, Switzerland. JOURNAL OF NEUROLOGY, vol. 253 (s.2); p. 51-51, ISSN: 0340-5354, doi: 10.1007/s00415-006-2001-2

Orlacchio A, PATRONO C., Borreca A, Babalini C, Dionisi L, Moschella V, Orlacchio A, Bernardi G, Kawarai T (2006). Clinical and genetic findings of two Italian kindreds with Silver syndrome. 10th International Congress of Parkinson's Disease and Movement Disorders 2006, Kyoto, Japan. MOVEMENT DISORDERS, vol. 21(s15); p. S412, ISSN: 0885-3185.

Deodato F, Rizzo C, Waterham HR, Santorelli F, PATRONO C, Wanders RJA, Boenzi S, Giannotti A, Dionisi-Vici C (2002). Atypical Smith-Lemli-Opitz syndrome (SLOS) with normal circulating sterols and high residual enzyme activity. Society for the Study of Inborn Errors of Metabolism, 40th Annual Symposium 2002, Dublin. Journal of Inherited Metabolic Disease, vol. 25 (s.1); p. 164. doi: 10.1023/A:1016120125723

Grossi A, Boenzi S, PATRONO C, Rizzo C, Santorelli FM, Cotugno G, Dionisi-Vici C (2002). Overt adrenal insufficiency in a child with Smith-Lemli-Opitz syndrome (SLOS). Society for the Study of Inborn Errors of Metabolism, 40th Annual Symposium 2002, Dublin. *Journal of Inherited Metabolic Disease* vol. 25 (s.1): p. 164. doi: 10.1023/A:1016120125723

Fortini D, Casali C, Tessa A, Damiano M, PATRONO C., Cricchi F, Bernedetti L, Amabile GA, Bertini E, Santorelli FM (2002). A single-base insertion in SPG3A is associated with autosomal dominant hereditary spastic paraplegia. 52nd Annual Meeting of the American Society of Human Genetics, Baltimore, MD. *AMERICAN JOURNAL OF HUMAN GENETICS*, vol. 71; p. 516, ISSN: 0002-9297

Caputo V, Brancati F, Valente E, Bertini E, PATRONO C., Santorelli F, Salvi S, Pizzuti A, Dallapiccola B (2002). Linkage analysis in an Italian family affected by autosomal dominant pure hereditary spastic paraplegia. EUROPEAN HUMAN GENETICS CONFERENCE 2002, Strasbourg. *EUROPEAN JOURNAL OF HUMAN GENETICS*, vol. 10, s. 1; p. 265, ISSN: 1018-4813

PATRONO C., Cricchi F, Fortini D, Tessa A, Tozzi G, Piemonte F, Carrozzo R, Bertini E, Comanducci G, Casali C, Santorelli FM (2000). Hereditary spastic paraplegias in Italian families: Clinical and molecular investigations. 50th Annual Meeting of the American Society of Human Genetics, Philadelphia, PA, 2000. *AMERICAN JOURNAL OF HUMAN GENETICS*, vol. 67; p. 375. ISSN: 0002-9297

Carrozzo R, Tessa A, Piemonte F, Chichierchia G, PATRONO C., Capuano O, Dionisi-Vici C, Bertini E, Santorelli FM (2000). A novel mtDNA mutation (T9176G) hampers ATP production and results in Leigh syndrome. 50th Annual Meeting of the American Society of Human Genetics, Philadelphia, PA, 2000. *AMERICAN JOURNAL OF HUMAN GENETICS*, vol. 67; p. 280. ISSN: 0002-9297

Carrozzo R, Piemonte F, Casali C, Tozzi G, PATRONO C., Tessa A, Bertini E, Santorelli FM (1999). Respiratory-chain enzyme activities in muscle from HSP patients. 49th Annual Meeting of the American Society of Human Genetics, San Francisco, CA, 1999. *AMERICAN JOURNAL OF HUMAN GENETICS*, vol. 65; A232. ISSN: 0002-9297

Santorelli FM, Vici CD, Giannotti A, Bertini E, Rizzo C, PATRONO C., Tessa A, Piemonte F, Carrozzo R (1999). A novel 7-DHCR mutation in a Lebanese child with Smith-Lemli-Opitz syndrome. *ATHEROSCLEROSIS*, vol. 144, s1; p. 21-22, ISSN: 0021-9150, doi: 10.1016/S0021-9150(99)80079-3