

NAME Gasparre, Giuseppe (08 June 1979)		POSITION TITLE Assistant Professor in Medical Genetics	
eRA COMMONS USER NAME -			
EDUCATION/TRAINING (Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)			
INSTITUTION AND LOCATION	DEGREE (if applicable)	YEAR(s)	FIELD OF STUDY
Red Cross Nordic United World College, Norway	IB	1996-1998	Biology
University of Bologna, IT/University of Manitoba	MS	1998-2003	Pharm. Biotechnologies
University of Turin, Italy	PhD	2004-2008	Human Genetics
University of Bari, Italy	Post-Doc	2008-2010	Molecular Biology

A. Positions and Honors.

Positions and Employment

2003	Fellow at the Saint Boniface Research Center, Molecular Immunology, Winnipeg, Canada
2004	Employee at the European School of Genetic Medicine-European Genetics Foundation, Italy
2008	Post-doctoral fellow at University of Bologna, Unit of Medical Genetics, S.Orsola hospital, Italy
2009	Post-doctoral fellow at University of Bari, Biochemistry and Molecular Biology, Italy
2010	Associate researcher of Medical Genetics, S.Orsola hospital, University of Bologna, Italy

Other Experience and Professional Memberships

2004	Licensed from the <i>Collegio Superiore Alma Mater Studiorum</i> University of Bologna, IT
2009	Faculty at the EMBO lecture course in mitochondrial medicine, Bologna, IT
2010	Faculty at the 1 st course in Mitochondrial metabolism and Cancer, Bologna, IT
2010	College of Mentors member, Human Genetics PhD program, University of Turin, IT

Prizes

2008	Malpighi d'Oro prize for under-30 young investigators
2011	Salvatore Venuta Young Scientist Prize – MSO, Mediterranean School of Oncology

B. Peer-reviewed publications (in chronological order).

1. K.Stankov, A.Biondi, M.D'Aurelio, **G.Gasparre**, A.Falasca, G.Romeo, G.Lenaz. Mitochondrial Activities of a Cell Line Derived from Thyroid Hürthle Cell Tumors. **Thyroid**. 2006; 16(4):325-31. PMID: 16646677.
2. E.Bonora, A.M.Porcelli*, **G.Gasparre****et al.* (*co-first authors). Defective oxidative phosphorylation in thyroid oncocyctic carcinoma is associated with pathogenic mitochondrial DNA mutations affecting complexes I and III. **Cancer Res**. 2006; 66(12): 6087-96. PMID: 16778181.
3. **G.Gasparre**, A.M.Porcelli, E.Bonora *et al.* Disruptive mitochondrial DNA mutations in complex I subunits are markers of oncocyctic phenotype in thyroid tumors. **PNAS**. 2007; 104:9001-9006. PMID: 17517629.
4. **G.Gasparre**, E.Hervouet, E.de Laplanche *et al.* Clonal expansion of mutated mitochondrial DNA is associated with tumor formation and complex I deficiency in renal oncocyctomas. **Hum Mol Genet**. 2008; 17(7):986-95. PMID: 18156159.
5. A.M.Porcelli, A.Ghelli, L.Iommarini, E.Mariani, M.Hoque, C.Zanna, **G.Gasparre**, M.Rugolo. The antioxidant function of Bcl-2 preserves cytoskeletal stability of cells with defective respiratory complex I. **Mol Cell Life Sci**. 2008; 65(18):2943-51. PMID:18695940.
6. D.Lascaro, S.Castellana, **G.Gasparre**, G.Romeo, C.Saccone, M.Attimonelli. The RHNumtS compilation: features and bioinformatics approaches to locate and quantify Human NumtS. **BMC Genomics**. 2008; 9:267. PMID: 18522722.
7. **G.Gasparre**, L.Iommarini, A.M.Porcelli *et al.* An inherited mitochondrial DNA disruptive mutation shifts to homoplasmy in oncocyctic tumor cells. **Hum Mutat**. 2009; 30(3):391-6. PMCID: 19086058

8. **G.Gasparre**, E.Bonora, G.Tallini, G.Romeo. Molecular features of thyroid oncocytic tumors. *Mol Cell Endocrinol*. 2010; 321:67-76. PMID: 20184940.
9. A.M.Porcelli, A.Ghelli, C.Ceccarelli, M.Lang , G.Cenacchi, M.Capristo, L.F.Pennisi, I.Morra, E.Ciccarelli, A.Melcarne, A.Bartoletti-Stella, N.Salfi, G.Tallini, A.Martinuzzi, V.Carelli, M.Attimonelli, M.Rugolo, G.Romeo, **G.Gasparre**. The genetic and metabolic signature of oncocytic transformation implicates HIF1 α destabilization. *Hum Mol Genet*. 2010; 19:1019-32. PMID: 20028790.
10. A.Bartoletti-Stella, N.Salfi, C.Ceccarelli, M.Attimonelli, G.Romeo, **G.Gasparre**. Mitochondrial DNA mutations in oncocytic adnexal lacrimal glands of the conjunctiva. *Arch Opht*. 2011; 129:664-6. PMID: 21555623.
11. **G.Gasparre**, G.Romeo, M.Rugolo, A.M.Porcelli. Learning from oncocytic tumors: why choose inefficient mitochondria? *BBA Bioenergetics*. 2011; 1807:633-42 PMID: 20732299.
12. G.Elachouri, S.Vidoni, C.Zanna, A.Pattyn, H.Boukhaddaoui, K.Gaget, P.Yu-Wai-Man, **G.Gasparre**, E.Sarzi, C.Delettre, A.Olichon, D.Loiseau, P.Reynier, P.F.Chinnery, A.Rotig, V.Carelli, C.P.Hamel, M.Rugolo, G.Lenaers. OPA1 links human mitochondrial genome maintenance to mtDNA replication and distribution. *Genome Res*. 2011; 21:12-20. PMID: 20974897.
13. I.Kurelac, G.Romeo, G.Gasparre. Mitochondrial metabolism and cancer. *Mitochondrion*. 2011; 11:635-7 PMID: 21447406.
14. F.Guerra, I.Kurelac, A.Cormio, R.Zuntini, L.B.Amato, C.Ceccarelli, D.Santini, G.Cormio, F.Fracasso, L.Selvaggi, L.Resta, M.Attimonelli, M.N.Gadaleta, **G.Gasparre**. Placing mitochondrial DNA mutations within the progression model of type I endometrial carcinoma. *Hum Mol Genet*. 2011; 20:2394-405 PMID: 21470976.
15. F.C.Geyer, D.de Biase, M.B.K.Lambros, M.Ragazzi, M.A.Lopez-Garcia, R.Natrajan, A.Mackay, I.Kurelac, **G.Gasparre**, A.Ashworth, V.Eusebi, J.S.Reis-Filho, G.Tallini. Genomic profiling of mitochondrion-rich breast carcinoma: chromosomal changes may be relevant for mitochondria accumulation and tumour biology. *Breast Cancer Res Treat*. In press. PMID:21509527.
16. F.Guerra, I.Kurelac, P.Magini, D.Santini, C.Ceccarelli, **G.Gasparre**. Mitochondrial DNA genotyping reveals synchronous nature of simultaneously detected endometrial and ovarian cancers. *Gyn Oncology*. 2011;122(2):457-8. PMID: 21592547.
17. I.Kurelac, M.Lang, R.Zuntini, C.Calabrese, D.Simone, S.Vicario, M.Santamaria, M.Attimonelli, G.Romeo, **G.Gasparre**. Searching for a needle in the haystack: Comparing six methods to evaluate heteroplasmy in difficult sequence context. *Biotechnol Adv*. In press. PMID:21689740.
18. L.M.Pradella, R.Zuntini, P.Magini, C.Ceccarelli, I.Neri, S.Cerasoli, C.Graziano, **G.Gasparre**, D.Turchetti. Two distinct thyroid tumours in a Cowden Syndrome patient carrying both a 10q23 and a mitochondrial DNA germline deletion. *J Med Genet*. 2011; 48(11):779-82. PMID:2192610.
19. D.Simone, F.M.Calabrese, M.Lang, **G.Gasparre**, M.Attimonelli. The Reference Human Nuclear Mitochondrial Sequences Compilation Validated and Implemented on the UCSC Genome Browser. *BMC Genomics*. 2011;12(1):517. PMID:22013967.
20. **G.Gasparre**, I.Kurelac, M.Capristo, L.Iommarini, A.Ghelli,C.Ceccarelli, G.Nicoletti, P.Nanni, C.De Giovanni, K.Scotlandi, C.M.Betts, V.Carelli, P.L.Lollini, G.Romeo, M.Rugolo, A.M.Porcelli. A mutation threshold distinguishes the anti-tumorigenic effects of the mitochondrial gene MTND1, an oncojanus function. *Cancer Res*. 71(19):6220-6229. PMID: 21852384.
21. F.Rubino, R.Piredda, F.M.Calabrese, D.Simone, M.Lang, C.Calabrese, V.Petruzzella, M.Tommaseo-Ponzetta, **G.Gasparre**, M.Attimonelli. HmtDB, a genomic resource for Mitochondrion Based Human Variability studies. *Nucleic Acid Res*. In press. PMID: 22139932
22. M.Lang, M.Sazzini, F.M.Calabrese, D.Simone, A.Boattini, G.Romeo, D.Luiselli, M.Attimonelli, **G.Gasparre**. Polymorphic NumtS trace human population relationships. *Hum Genet*. In press. PMID: 22160368.
23. V.Petruzzella, R.Carrozzo, C.Calabrese, R.Dell'Aglio, R.Trentadue, R.Piredda, L.Artuso, T.Rizza, M.Bianchi, A.M.Porcelli, S.Guerriero, **G.Gasparre**, M.Attimonelli. Deep sequencing unearths NumtS under false heteroplasmic LHON-associated mtDNA variants. *Hum Mol Genet*. 2012. 21(17):3753-64. PMID: 22589247.
24. L.Iommarini, M.Calvaruso, I.Kurelac, **G.Gasparre***, A.M.Porcelli. Complex I impairment in mitochondrial diseases and cancer: parallel roads leading to different outcomes. *Int J Biochem Cell Biol*. 2013. 45(1):47-63. PMID: 22664328.
25. F.Guerra, A.M.Perrone, I.Kurelac, D.Santini, C.Ceccarelli, M.Cricca, C.Zamagni, P.De Iaco, **G.Gasparre**. Mitochondrial DNA Mutation in Serous Ovarian Cancer: Implications for Mitochondria-Coded Genes in Chemoresistance. *J Clin Oncol*. 2012. 30(36):e373-8

26. L.Artuso, S.Zoccolella, P.Favia, A.Amati, R.Capozzo, G.Logroscino, L.Serlenga, I.Simone, **G.Gasparre**, V.Petruzzella. Mitochondrial genome aberrations in skeletal muscle of patients with motor neuron disease. **ALS**. In press.
27. I.Kurelac, A.MacKay, M.Lambros, E.Di Cesare, G.Cenacchi, C.Ceccarelli, I.Morra, A.Melcarne, L.Morandi, F.M.Calabrese, M.Attimonelli, G.Tallini, J.S.Reis-Filho, **G.Gasparre**. Somatic mitochondrial DNA disruptive mutations are modifiers of tumorigenesis that correlate with low genomic instability in pituitary adenomas. **Hum Mol Genet**. 2013. 22(2):226-38
28. L.M.Pradella LM, M.Lang, I.Kurelac, E.Mariani, F.Guerra, R.Zuntini, G.Tallini, A.MacKay, J.S.Reis-Filho, M.Seri, D.Turchetti, **G.Gasparre**. Where Birt-Hogg-Dubé meets Cowden Syndrome: Mirrored genetic defects in two cases of syndromic oncocytic tumours. **Eur J Hum Genet**. In press.
29. A.Ghelli, A.Marchesini, C.Tropeano, M.A.Calvaruso, L.Iommarini, A.M.Porcelli, C.Zanna, V.De Nardo, A.Martinuzzi, F.Wibrand, J.Vissing, I.Kurelac, **G.Gasparre**, N.Selamoglu, F.Daldal, M.Rugolo. The Cytochrome B p.278Y>C mutation causative of a multisystem disorder enhances superoxide production and alters supramolecular interactions of respiratory chain complexes. **Hum Mol Genet**. In press.
30. **G.Gasparre**, A.M.Porcelli, G.Lenaz, G.Romeo. Relevance of Mitochondrial Genetics and Metabolism in Cancer Development. **Cold Spring Harb Perspect Biol**. 2013. Feb 1;5(2).
31. C.Calabrese, L.Iommarini, I.Kurelac, M.A.Calvaruso, M.Capristo, P.L.Lollini, P.Nanni, C.Bergamini, G.Nicoletti, C.De Giovanni, A.Ghelli, V.Giorgio, M.F.Caratozzolo, F.Marzano, C.Manzari, C.M.Betts, V.Carelli, C.Ceccarelli, M.Attimonelli, G.Romeo, R.Fato, M.Rugolo, A.Tullo, **G.Gasparre***, A.M.Porcelli*. Respiratory complex I is essential to induce a Warburg profile in mitochondria-defective tumor cells. **Cancer & Metabolism**. 2013. 1:11. *co-last authors.
32. G.Marucci, A.Maresca, L.Caporali, A.Farnedi, C.M.Betts, L.Morandi, D.de Biase, S.Cerasoli, M.P.Foschini, E.Bonora, M.Vidone, G.Romeo, E.Perli, C.Giordano, G.d'Amati, **G.Gasparre**, A.Baruzzi, V.Carelli, V.Eusebi. Oncocytic glioblastoma: a glioblastoma showing oncocytic changes and increased mtDNA copy number. **Hum Pathol**. In press.
33. A.Bartoletti-Stella, E.Mariani, I.Kurelac, A.Maresca, M.F.Caratozzolo, L.Iommarini, V.Carelli, L.H.Eusebi, A.Guido, L.Fuccio, M.Rugolo, A.Tullo, A.M.Porcelli, **G.Gasparre**. Gamma rays induce a p53-independent mitochondrial biogenesis that is counter-regulated by HIF1 α . **Cell Death & Disease**. In press.

C. Research Support

Ongoing Research Support

- | | |
|---|-----------------------|
| FIRB "Futuro in Ricerca" J31J10000040001 Gasparre, G. (PI) | 04/01/10-03/31/13 |
| Italian Ministry of Education, University and Research (MIUR) | |
| Significato funzionale delle mutazioni del DNA mitocondriale nel cancro. | |
| This study is a genetics and functional analysis of the tumorigenic potential of mitochondrial DNA mutations. | |
| Role: Principal Investigator | |
| | |
| Fondazione Umberto Veronesi Gasparre, G. (PI) | 01/01/2012-31/12/2013 |
| DISCO TRIP | |
| Disrupting mitochondrial complex I to trigger pseudonormoxia: an anticancer strategy | |
| Role: Principal Investigator | |
| | |
| Myrovlitys Trust Gasparre, G. (PI) | 01/01/2012-31/12/2013 |
| CHIC – Combined Haploinsufficiency in Inherited Cancers | |
| Role: Principal Investigator/supervisor of a grantee | |
| | |
| MiPEO Gasparre, G. (PI) | 01/01/2012-31/12/2013 |
| AIRC – Ass. Italiana Ricerca sul Cancro - Fellowships | |
| Mitochondria in Progression of Endometrial and Ovarian cancer | |
| Role: Principal Investigator/Supervisor of an AIRC fellow | |
| | |
| Ass. Italiana Ricerca sul Cancro Gasparre, G. (PI) | 01/01/2012-31/12/2014 |
| Mitochondrial dysfunction and low-proliferative tumor phenotype: Taking lessons from oncocytic tumors | |
| AIRC/FIRC Fellowships | |
| Role: Principal Investigator/Supervisor of an AIRC fellow | |

Liddy Shriver Sarcoma Initiative Rugolo, M. and Scotlandi, K. (PI's) 2011-2012
CD99 Engagement and Apoptosis in Ewing's Sarcoma: The Role of Mitochondria and Mitochondrial DNA Mutations
Role: researcher

IG8810 Romeo, G. (PI) 12/01/09-11/30/12
AIRC – Italian Association for Cancer Research
TRANSMIT – Translational significance of mitochondrial mutations in tumors.
The aim of this collaborative project is to define the determinants of the shift and selection of mitochondrial DNA mutations in cancer and correlate this process with the induction of a benign behavior of the tumor.
Role: researcher/grant manager

PERNO project for Neuro-Oncology Eusebi, V. (PI) 1/01/08-1/01/11
Regione Emilia Romagna
The project aims at characterizing tumors of the CNS, also in terms of mitochondrial mutations and metabolism.
Role: researcher

PRIN 2008SW44CS Pinton, P. (PI) 3/22/10-3/21/12
Italian Ministry of Education, University and Research (MIUR)
Ruolo dei mitocondri nella trasduzione dei segnali apoptotici nel cancro.
This study investigates the mechanisms of signal transduction that determine mitochondrial-dependent apoptosis in cancer cells.
Role: researcher

Completed Research Support

IG1157 Tallini, G. (PI) 12/01/05 -11/30/07
AIRC – Italian Association for Cancer Research
Functional genomics of thyroid oncocyoma.
The project studied the molecular determinants underlying oncocytic transformation in thyroid cancer.
Role: researcher

HERMIONE Mehlen, P. (PI) 9/01/06-8/31/09
European Union
The role of dependence receptors in cancer
The goal of this project was to study the interactions between dependence receptor and their ligands in determining apoptosis in cancer to provide novel therapeutic strategies.
Role: researcher

PRIN 2006064992_001 Romeo, G. (PI) 5/01/06-4/30/08
Italian Ministry of Education, University and Research (MIUR)
Variability and function of mitochondrial mutations in physiological and pathological conditions.
The project aimed at studying the variability of mitochondrial DNA mutations in correlation with their pathologic potential.
Role: researcher