GIUSEPPE NOVELLI

BRIEF CURRICULUM VITAE

NAME	Giuseppe Novelli
DATE AND PLACE OF BIRTH	February 27, 1959, Rossano (CS), Italy
CITIZENSHIP	Italian
MARITAL STATUS	Married, three children
POSITION	Rector of the University of Rome Tor Vergata
INSTITUTIONAL ADDRESS	University of Rome Tor Vergata Via Cracovia 50 00133 Rome, Italy Tel +39 06 7259.8753 E-mail rettore@uniroma2.it

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EDUCATION AND TRAINING

1983 to 1985: Postgraduate Speciality in Medical Genetics. University "La Sapienza", Rome, Italy
1981 to 1987: Didactic and Research Activity, University Researcher, Faculty of Chemistry, University of Urbino "Carlo Bo", Urbino, Italy
1977 to 1981: Biological Science Degree, with honours, University of Urbino "Carlo Bo", Urbino, Italy
Medical Genetics, Pharmacogenetics, Genomics

SELECTED MEMBERSHIPS AND BOARDS

2016 to present: Member and Coordinator of Genetics Sub-group, National Committee for Biosafety, Biotechnologies and Life Sciences (CNBBSV), Italian Presidency of the Council of Ministers, Italy
2016 to present: Chairman, National Observatory for Healthcare Professions, Ministry of Education, University and Research, Italy
2016 to present: Expert, European Medicines Agency – EMA, London
2016 to present: Member of the Undiagnosed Rare Disease Network, Italy
2012 to present: Genetics Consultand, Research Centre "IRCCS Neuromed", Pozzilli (IS), Italy
2018: Delegate (Healthcare issues), Conference of Italian Universities Rectors – CRUI, Italy
2016 to 2018: Chairman, Medical Genetics Committee for National Scientific Certification, Ministry of Education, University and Research, Italy
2013 to 2018: Member, Health Council of the Italian Ministry of Health, Rome, Italy
2009 to 2018: President of the Italian College of Professors of Medical Genetics
2016 to 2017: Member, Genome Project National Committee, Italy at Ministry of Health
2014 to 2017: Vice President, Conference of Italian Universities Rectors – CRUI, Italy
2002 to 2016: Member, Ethical Committee University Hospital Tor Vergata – PTV, Rome, Italy
2008 to 2015: Member, Pharmacogenomics Working Party (PgWP) of European Medicines Agency - EMA, London, UK
2011 to 2013: Member of Governing Board, ANVUR -National Agency for Quality Assessment of University and Research, Italy
2010 to 2013: Member of European Science Foundation (ESF)
2008 to 2011: Dean, Faculty of Medicine and Surgery, University of Rome Tor Vergata, Rome, Italy
2006 to 2007: Member of Working Group on "Expert of Advanced Therapies", Italian Medicines Agency (AIFA)
2006: Member of the Committee Rare Diseases Italian Ministry of Health and Delegate for the Lazio Region

2000: Member, Study Committee on the Use of Stem Cells at the Italian Ministry of Health
1998 to 2000: Member, Research Committee, University of Rome Tor Vergata, Rome, Italy
1999 to 2000: Member, Scientific Council of Experimental Institute of National Council of Research Italy (University Research Committee) – CNR, Rome, Italy
1998 to 1999: Member, Working Group on "Cloning", Italian Presidency of the Council of Ministers, Italy
1996 to 1998: Member, Ethical Committee School of Medicine, University of Rome Tor Vergata, Rome, Italy
1992 to 1995: Consultand, Italian Ministry of Interior (Scientific Police)

OTHER ASSIGNMENTS

He is currently extern expert at the "Agencie d'évaluation de la recherche et de l'enseigneraient supérieur (AERES)" France; has been Member of the National Commission for the Post-Genome Programme at the Italian Ministry of University and Research (MIUR); has been member and Italian delegate for genetic testing programme at the OECD (Organisation for Economic Co-Operation and Development); has been member of the "Groupe d'experts en Genetique moleculaire", at the Ministère de la Santé, de la Famille et des Personnes Handicapées (Paris); he is a Reviewer of the National Research Agency (ANR), France since 2009. He is member of the "Board of Trustees" of the Biagio Agnes Foundation.

RESEARCH EXPERIENCE

2001 to present: Director, U.O.C. Medical Genetics Laboratory, Policlinico Universitario "Tor Vergata", Rome, Italy
1999 to present: Full Professor, Medical Genetics, Faculty of Medicine and Surgery, University of Rome Tor Vergata, Rome, Italy
2016 to present: Adjunct Professor, University of Nevada, School of Medicine, Reno, USA
2003 to present: Adjunct Professor, University of Arkansas for Medical Sciences, Little Rock, USA
2011 to 2015: Scientific Director, Research Centre Fatebenefratelli, Ospedale San Pietro, Rome, Italy
1998 to 2011: Director, Postgraduate Speciality in Medical Genetics, University of Rome Tor Vergata, Rome, Italy
1995 to 1999: Associate Professor, Human Genetics**,** University of Rome Tor Vergata, Rome, Italy
1996 to 1997: Visiting Professor "MiniSabbatical", University of Southern California (USC), Los Angeles, USA
1992 to 1995: Associate Professor, Molecular Genetics, Faculty of Medicine and Surgery, University Cattolica di Milano, seat of Rome - Policlinico Gemelli, Italy
1990: Associate, Groupe de Génétique Moléculaire INSERM U.91, Créteil, France
1983 to 1992: University Researcher, Molecular Genetics, University of Urbino "Carlo Bo", Urbino, Italy
1983 to 1984: Visiting Researcher, Unité de Recherches de Biologie Prénatale INSERM U.73, Paris, France

SELECTED HONORS

2018: SIMI Medal of Italian Interior Medicine Society
2017: Commitment Honor of the Order of Merit of the Italian Republic
2017: Premio Nazionale Medicina, Pescara
2015: Premio Gaetano Conte per Disordini Neuromuscolari
2015: Premio Alvaro per Scienza e Cultura
2011: Premio Scanno per la Medicina (XXXIX Edizione)
2011: Premio Nazionale Gentile di Fabriano per la Scienza e l'Innovazione, XV Edizione
2009: Premio Internazionale La Calabria nel Mondo
2009: Premio "Vittorio Aprile", Roma

2004: Premio Pericle D'Oro per la Ricerca Scientifica
2003: Premio "Brutium" scienza
2002: Premio "Ferrari" Società Italiana Genetica Umana (SIGU)
1984: Premio Associazione Italiana Ricerca e Cura Handicap

SCIENTIFIC LEADERSHIP PROFILE

Mapping, Identification and Cloning of Human Genes
He started his research activity in the field of Genetic and Biochemistry in 1980.
His primary focus was the mapping, identification and characterization of human-disease genes (Laron dwarfism, cystic fibrosis, DiGeorge syndrome, Mandibuloacral dysplasia, Friedrich ataxia vitamin-E-deficiency, spinal muscular atrophy, hypoplastic glomerulocystic kidney disease, myotonic dystrophy, psoriasis, galactosemia, hereditary nonspherocytic haemolytic anemia, atherosclerosis and myocardial infarction, vacuolar neuromyopathy, patella aplasia hypoplasia). The Spectrum of clinical features associated with interstitial chromosome 22q11 deletions: a European collaborative study (J. Med. Genet. : 34 Issue: 10 Pages: 798-804, 1997) provided the scientific evidence showing that patients with 22q11 deletion provide a large spectrum of symptoms and phenotypes.
This landmark study (cited more 398 times) provided the first of the complexity of the phenotype associated to this syndrome and suggest the involvement of different genes mapping in the 22q11 region. In the same year, Giuseppe Novelli started in searching for genes mapping in the region and isolate and characterized a novel one, UFD1L, a developmentally expressed ubiquitination gene, which is deleted in 22q11 syndrome (Hum Mol Genet., 6, 259-265, 1997). After the isolation, Novelli studied the structure, the expression, the conservation during evolution and its role in the syndrome. For these studies (in total 24 peer-reviewed articles), Novelli wrote two editorials (Trends Genet. 1999 Jul;15(7):251-4 and Mol. Med. Today, 2000 Jan;6(1):10-1). The results obtained during this period, have allowed his participation to a EU consortium (chaired by P. Scambler) and facilitated collaboration with basic researchers and geneticists, trying to find the pathophysiological pathways and molecular mechanisms underlying some of the observations that Novelli had made in his first studies.
In collaboration with Dr. Meisterernst M (Munich, Germany), Novelli published the cloning of a novel gene, PCQAP (PC2 glutamine/Q-rich-associated protein), that maps to the DiGeorge typically deleted region and encodes a protein identified as a subunit of the large multiprotein complex PC2 (Genomics, 2001 Jun 15;74(3):320-32).
He continued researches in this field focusing his attention studying the regulatory effects of 22q11 haploinsufficiency during development by analyzing the expression pattern of the orthologous MM16 genes in mouse embryos at different stages of development (Gene. 2007, 391(1-2):91-102) and studying the morphogenetic mechanisms in a mouse model for this disorder (Cardiovasc Pathol. 2006 Jul-Aug;15(4):194-202). They also demonstrated that folic acid and methionine periconceptional supplementations may influence the incidence of congenital defects and may probably induce negative selection of embryos presenting developmental anomalies (Cardiovasc Pathol. 2008 Apr 14).

In 2002 publication (Am J. Hum Genet., Aug;71(2):426-31), Novelli demonstrated for the first time that a single nucleotide mutation in the LMNA gene is responsible of a progeroid syndrome, the mandibuloacral dysplasia (MAD) and suggest that this protein is actively involved in premature aging. Mutations in the LMNA gene have been found at present in about 26 different diseases called as "laminopathies" which include muscular dystrophy, cardiomyopathy, lipodystrophy, insulin-resistance, diabetes, and premature aging. The involvement of his research group in this field is documented at present by 11 peer reviewed papers appeared on prestigious journals (i.e. Hum Mol Genet., Exp Cell Res., Aging Cell, J Clin Endocrinol Metab, Physiol Genomics) and the establishment of an European Network funded by an EU grant FP6 "Euro-laminopathies" no. 018690 (http://www.projects.mfpl.ac.at/euro-laminopathies/php/index.php).

Identification and characterization of a splicing isoform of the endothelial receptor for oxidized low-density lipoprotein (ox-LDLs): LOXIN, encoded by the OLR1 gene. They demonstrated a protective role of LOXIN in pathologies correlated with LOX-1 overexpression such as atherosclerosis and tumors (Rev. In Int J. Mol. Sci.2017).

Gene Therapy Research
In collaboration with D. Gruenert (San Francisco, USA), Novelli developed an innovative technique of gene targeting strategy based on oligonucleotide design and delivery to restore a normal gene function by homologous recombination using small DNA fragment (SFHR) (Small Fragment Homologous Replacement). In a series of papers published on Hum Mole Genet., Mol Therapy, Biotechniques, Hum Gene Ther., J. Clin Invest) they demonstrated the efficiency of this techniques to correct mutated human cells in vitro and in vivo. Recent advances in gene targeting and novel strategies have led to the suggestion that SFHR might be used as clinical therapy for genetic disease and used in the cellular genetic therapy approach. This technique is the prototype of the currently gene-editing approach.

Stem Cells Research
Using immunohistochemistry and FACS analyses they isolated and characterized Human Cytotrophoblastic-derived Multipotent Cells (hCTMCs) from Human Chorionic villi (CVS). These cells may be a safe and convenient source of cells for cell-based therapy, as well as an ideal target for in utero fetal gene therapy (Cloning Stem Cells. 2009).
They developed an original treatment for pulmonary fibrosis in a murine model. This study has opened a new avenue along which AECII differentiated from HUES may come along as a knight in shining armour to help us to combat a still life-threatening and poorly manageable lung disease (Eur Resp J. 2012)
They have developed a cancer stem cell model starting from normal human stem cells derived from amniotic and chorionic placenta membranes. These cells are able to differentiate into neural cell lineages and to undergo spontaneous transformations and acquire an NB-like phenotype (Stem Cell Res Ther. 2014);
They have developed an advanced protocol to derive human induced pluripotent stem cells (hiPSCs) from patients affected by genetic diseases. hiPSCs represent a major impact on patient' health. hiPSCs represent a valid model for the in vitro study of monogenic diseases, together with a better comprehension of the pathogenic mechanisms of the pathology, for both cell and gene therapy protocol applications (Cell Reprogram. 2015).
Recently, they investigated in vitro the volatile compounds (VOCs) released during human induced pluripotent stem cells (hiPSCs) reprogramming using electronic sensors to control the process of differentiation of pluripotent stem cells (Sci Reports, 2017).

Personalized Medicine, Pharmacogenetics and Pharmacogenomics
Personalized medicine provides to physicians a molecular makeup of each patient. Looking at the patient on this level helps the physician get a profile of the patient's genetic distinction, or mapping. By investigating this genetic profile, medical professionals are then able to select patients, and use the found information to plan out a course of treatment that is much more in step with the way their body works. Personalized medicine is a direct extension of the genomic medicine that use genetic information to prevent or treat disease in adults or their children. They developed original protocol and identified novel genomic biomarkers for drug efficacy and drugs adversal effects (Pharmacogenomics 2014,2015, 2016, 2017). Recent studies were addressed related to Stevens-Johnson syndrome/toxic epidermal necrolysis associated to specific drugs (Plos One 2016, Pharmacogenomics 2017).
They discovered that genetic variations in candidate microRNA (miRNA or miR) genes could contribute to susceptibility to complex diseases like diabetes, lupus, and Chron’s disease (Acta Diabetol, 2016, Molec Diagn Ther, 2017).

Forensic DNA Analysis
Novelli introduced for the first time in Italy the DNA analysis for forensic use (Nature 1991). He and his group developed many protocols and platforms for DNA analysis at the crime scene.

Contribution to public awareness of science
Giuseppe Novelli has been actively involved in the fostering of science and scientific policy in Italy at various levels, with a focus on Biochemical Genetics, Human Genetics, Medical Genetics and Molecular Genetics, taking public stands on several issues.
He regularly gives interviews and contributes to the most authoritative organs of the italian press, such as daily newspapers, magazines, web magazines, scientific and general radio and television.

MEMBERSHIP OF SCIENTIFIC SOCIETIES

2019: Academia Europaea
2010: Oligonucleotide Therapeutics Society (OTS)
2008: Accademia Medica di Roma
2007: African Society of Human Genetics (AfSHG)
2005: Board Committee, American Society of Gene Therapy (ASGT)
1997: Founder Member, Italian Society of Human Genetics (SIGU)
1990: Human Genome Organization (HUGO)
1989: European Society of Human Genetics, Board (ESHG)
1988: American Society of Human Genetics (ASHG)

DIRECTION AND MEMBERSHIP OF EDITORIAL BOARDS

2019 to present: Diabetes Monitor Journal (co-editor)
2019 to present: Human Genomics
2010 to present: Genetics Research International
2009 to present: Academic Editor, Plos One
2005 to present: Journal of Pharmacogenomics & Pharmacoproteomics
2004 to present: BMC Medical Genetics, since 2004
2004 to present: Encyclopedia of Life Science for Genetics and Molecular Biology
2004 to present: Journal Inflammation & Allergy – Drug Targets (IADT)
2003 to present: Associate Editor, Journal of Cardiovascular Medicine
2002 to present: Journal of Cardiovascular Medicine
2000 to present: La Clinica Terapeutica
2001 to present: Co-Editor, Acta Myologica
1999 to 2013: Clinical Genetics, 1999-2013
1999 to 2003: Neuromuscular Disorders

IMPACT

Giuseppe Novelli holds four International patents.
He has mainly focused his research activity on issues concerning Biochemical Genetics, Human Genetics, Medical Genetics and Molecular Genetics.
He has contributed to identifying numerous genes causing human genetic diseases.
He has found the cause of: Laron disease; Acral mandibular dysplasia; Psoriasis and Psoriasis arthitis.
He is currently studying the genetic basis of complex diseases, the characterization and iPS cell lines, and the identification of novel genomic biomarkers for pharmacogenetics.
He has set up a Centre of Excellence for the study of genomic, complex and multifactorial diseases at Università degli Studi di Roma "Tor Vergata", funded by MIUR (Ministry of Education University and Research) in 2001.
He has set up the Bioscience Genomics, a spin-off of the Tor Vergata University of Rome.
He has coordinated several research projects funded by MIUR, CNR, Ministero della Salute, Telethon, AFM, EU FP5, EU FP6 e EU FP7, Ministero degli Esteri, Fondazione Veronesi, AIRC, AIFA, ISS. He has been advisor for the spin-off Onconetics (USA).

The broad impact of the contribution of Giuseppe Novelli is testified by citations. **As of July 2019 he has over 14.593 citations (Scopus) or 21.579 (Google Scholar) and an H-index of 53 (Scopus),** or **69 (Google Scholar).**
A bibliometric analysis indicates that he is one of the most quoted geneticist among the TIS (Top Italian Scientists)

SELECTED PUBLICATIONS (2015-2018)

1. Spitalieri P, Talarico RV, Murdocca M, Fontana L, Marcaurelio M, Campione E,Massa R, Meola G, Serafino A, Novelli G, Sangiuolo F, Botta A. Generation and Neuronal Differentiation of hiPSCs From Patients With Myotonic Dystrophy Type 2. Front Physiol. 2018 Jul 27;9:967. doi: 10.3389/fphys.2018.00967.
2. Ferese R, Scala S, Biagioni F, Giardina E, Zampatti S, Modugno N, Colonnese C, Storto M, Fornai F, Novelli G, Ruggieri S, Gambardella S. Heterozygous PLA2G6 Mutation Leads to Iron Accumulation Within Basal Ganglia and Parkinson's Disease. Front Neurol. 2018 Jul 10;9:536. doi: 10.3389/fneur.2018.00536. eCollection 2018.
3. Capuano R, Spitalieri P, Talarico RV, Catini A, Domakoski AC, Martinelli E, Scioli MG, Orlandi A, Cicconi R, Paolesse R, Novelli G, Di Natale C, Sangiuolo F. Volatile compounds emission from teratogenic human pluripotent stem cells observed during their differentiation in vivo. Sci Rep. 2018 Jul 23;8(1):11056.
4. Brancati F, Camerota L, Colao E, Vega-Warner V, Zhao X, Zhang R, Bottillo I, Castori M, Caglioti A, Sangiuolo F, Novelli G, Perrotti N, Otto EA; Undiagnosed Disease Network Italy. Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. Eur J Hum Genet. 2018 Sep;26(9):1266-1271.
5. Gargiuli C, Schena E, Mattioli E, Columbaro M, D'Apice MR, Novelli G, Greggi T, Lattanzi G. Lamins and bone disorders: current understanding and perspectives. Oncotarget. 2018 Apr 27;9(32):22817-22831. doi: 10.18632/oncotarget.25071.
6. Ciccacci C, Perricone C, Alessandri C, Latini A, Politi C, Delunardo F, Pierdominici M, Conti F, Novelli G, Ortona E, Borgiani P. Evaluation of ATG5 polymorphisms in Italian patients with systemic lupus erythematosus: contribution to disease susceptibility and clinical phenotypes. Lupus.
7. Dinarelli S, Girasole M, Spitalieri P, Talarico RV, Murdocca M, Botta A, Novelli G, Mango R, Sangiuolo F, Longo G. AFM nano-mechanical study of the beating profile of hiPSC-derived cardiomyocytes beating bodies WT and DM1. J Mol Recognit. 2018 Oct;31(10):e2725. doi: 10.1002/jmr.2725.
8. Maroofian R, Murdocca M, Rezaei-Delui H, Nekooei A, Mojarad M, Sangiuolo F, Novelli G, Superti-Furga A, D'Apice MR. A novel in-frame deletion in ZMPSTE24 is associated with autosomal recessive acrogeria (Gottron type) in an extended consanguineous family. Clin Dysmorphol. 2018 Jul;27(3):88-90.
9. 9: Spitalieri P, Talarico RV, Caioli S, Murdocca M, Serafino A, Girasole M,
10. Dinarelli S, Longo G, Pucci S, Botta A, Novelli G, Zona C, Mango R, Sangiuolo F. Modelling the pathogenesis of Myotonic Dystrophy type 1 cardiac phenotype through human iPSC-derived cardiomyocytes. J Mol Cell Cardiol. 2018 May;118:95-109.
11. Politi C, Ciccacci C, Novelli G, Borgiani P. Genetics and Treatment Response in Parkinson's Disease: An Update on Pharmacogenetic Studies. Neuromolecular Med. 2018 Mar;20(1):1-17. doi: 10.1007/s12017-017-8473-7. Epub 2018 Jan 5. Review.
12. Santoro M, Fontana L, Maiorca F, Centofanti F, Massa R, Silvestri G, Novelli G, Botta A. Expanded [CCTG]n repetitions are not associated with abnormal methylation at the CNBP locus in myotonic dystrophy type 2 (DM2) patients. Biochim Biophys Acta Mol Basis Dis. 2018 Mar;1864(3):917-924.
13. Cenni V, D'Apice MR, Garagnani P, Columbaro M, Novelli G, Franceschi C, Lattanzi G. Mandibuloacral dysplasia: A premature ageing disease with aspects of physiological ageing. Ageing Res Rev. 2018 Mar;42:1-13.
14. Cascella R, Strafella C, Longo G, Manzo L, Ragazzo M, De Felici C, Gambardella S, Marsella LT, Novelli G, Borgiani P, Sangiuolo F, Cusumano A, Ricci F, Giardina E. Assessing individual risk for AMD with genetic counseling, family history, and genetic testing. Eye (Lond). 2017 Sep 15.

15. Latini A, Ciccacci C, Novelli G, Borgiani P. Polymorphisms in miRNA genes and their involvement in autoimmune diseases susceptibility. Immunol Res. 2017 Aug;65(4):811-827.
16. Ciccacci C, Latini A, Politi C, Mancinelli S, Marazzi MC, Novelli G, Palombi L, Borgiani P. Impact of glutathione transferases genes polymorphisms in nevirapine adverse reactions: a possible role for GSTM1 in SJS/TEN susceptibility. Eur J Clin Pharmacol. 2017 Jul 8.
17. Rufini S, Ciccacci C, Novelli G, Borgiani P. Pharmacogenetics of inflammatory bowel disease: a focus on Crohn's disease. Pharmacogenomics. 2017 Jul;18(11):1095-1114.
18. Foo FL, McEniery CM, Lees C, Khalil A; International Working Group on Maternal Haemodynamics. Assessment of arterial function in pregnancy: recommendations of the International Working Group on Maternal Haemodynamics. Ultrasound Obstet Gynecol. 2017 Jul 1.
19. Capuano R, Spitalieri P, Talarico RV, Domakoski AC, Catini A, Paolesse R, Martinelli E, Novelli G, Sangiuolo F, Di Natale C. A preliminary analysis of volatile metabolites of human induced pluripotent stem cells along the in vitro differentiation. Sci Rep. 2017 May 9;7(1):1621.
20. Gambardella S, Ferese R, Biagioni F, Busceti CL, Campopiano R, Griguoli AMP, Limanaqi F, Novelli G, Storto M, Fornai F. The Monoamine Brainstem Reticular Formation as a Paradigm for Re-Defining Various Phenotypes of Parkinson's Disease Owing Genetic and Anatomical Specificity. Front Cell Neurosci. 2017 Apr 18;11:102.
21. Ferradini V, Cassone M, Nuovo S, Bagni I, D'Apice MR, Botta A, Novelli G, Sangiuolo F. Targeted Next Generation Sequencing in patients with Myotonia Congenita. Clin Chim Acta. 2017 Jul;470:1-7.
22. Bruno V, Rizzacasa B, Pietropolli A, Capogna MV, Massoud R, Ticconi C, Piccione E, Cortese C, Novelli G, Amati F. OLR1 and Loxin Expression in PBMCs of Women with a History of Unexplained Recurrent Miscarriage: A Pilot Study. Genet Test Mol Biomarkers. 2017 Jun;21(6):363-372.
23. Mizzi C, Dalabira E, Kumuthini J, Dzimiri N, Balogh I, Başak N, Böhm R, Borg J, Borgiani P, Bozina N, Bruckmueller H, Burzynska B, Carracedo A, Cascorbi I, Deltas C, Dolzan V, Fenech A, Grech G, Kasiulevicius V, Kádaši Ľ, Kučinskas V, Khusnutdinova E, Loukas YL, Macek M Jr, Makukh H, Mathijssen R, Mitropoulos K, Mitropoulou C, Novelli G, Papantoni I, Pavlovic S, Saglio G, Sertić J, Stojiljkovic M, Stubbs AP, Squassina A, Torres M, Turnovec M, van Schaik RH, Voskarides K, Wakil SM, Werk A, Del Zompo M, Zukic B, Katsila T, Lee MT, Motsinger-Rief A, Mc Leod HL, van der Spek PJ, Patrinos GP. Correction: A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS One. 2017 Feb 16;12(2):e0172595.
24. Rizzacasa B, Morini E, Pucci S, Murdocca M, Novelli G, Amati F. LOX-1 and Its Splice Variants: A New Challenge for Atherosclerosis and Cancer-Targeted Therapies. Int J Mol Sci. 2017 Jan 29;18(2). pii: E290.
25. Conigliaro P, Ciccacci C, Politi C, Triggianese P, Rufini S, Kroegler B, Perricone C, Latini A, Novelli G, Borgiani P, Perricone R. Polymorphisms in STAT4, PTPN2, PSORS1C1 and TRAF3IP2 Genes Are Associated with the Response to TNF Inhibitors in Patients with Rheumatoid Arthritis. PLoS One. 2017 Jan 20;12(1):e0169956.
26. Pucci S, Polidoro C, Joubert A, Mastrangeli F, Tolu B, Benassi M, Fiaschetti V, Greco L, Miceli R, Floris R, Novelli G, Orlandi A, Santoni R. Ku70, Ku80, and sClusterin: A Cluster of Predicting Factors for Response to Neoadjuvant Chemoradiation Therapy in Patients With Locally Advanced Rectal Cancer. Int J Radiat Oncol Biol Phys. 2017 Feb 1;97(2):381-388.
27. Campione E, Botta A, Di Prete M, Rastelli E, Gibellini M, Petrucci A, Bernardini S, Novelli G, Bianchi L, Orlandi A, Massa R, Terracciano C. Cutaneous features of myotonic dystrophy types 1 and 2: Implication of premature aging and vitamin D homeostasis. Neuromuscul Disord. 2017 Feb;27(2):163-169.
28. Carr DF, Bourgeois S, Chaponda M, Takeshita LY, Morris AP, Castro EM, Alfirevic A, Jones AR, Rigden DJ, Haldenby S, Khoo S, Lalloo DG, Heyderman RS, Dandara C, Kampira E, van Oosterhout JJ, Ssali F, Munderi P, Novelli G, Borgiani P, Nelson MR, Holden A, Deloukas P, Pirmohamed M. Genome-wide association study of nevirapine hypersensitivity in a sub-Saharan African HIV-infected population. J Antimicrob Chemother. 2017 Apr 1;72(4):1152-1162.
29. Santoro M, Masciullo M, Silvestri G, Novelli G, Botta A. Myotonic dystrophy type 1: role of CCG, CTC and CGG interruptions within DMPK alleles in the pathogenesis and molecular diagnosis. Clin Genet. 2016 Dec 19.
30. Gambardella S, Biagioni F, Ferese R, Busceti CL, Frati A, Novelli G, Ruggieri S, Fornai F. Vacuolar Protein Sorting Genes in Parkinson's Disease: A Re-appraisal of Mutations Detection Rate and Neurobiology of Disease. Front Neurosci. 2016 Nov 24;10:532. eCollection 2016. Review.
31. Ciccacci C, Perricone C, Politi C, Rufini S, Ceccarelli F, Cipriano E, Alessandri C, Latini A, Valesini G, Novelli G, Conti F, Borgiani P. A polymorphism upstream MIR1279 gene is associated with pericarditis development in

Systemic Lupus Erythematosus and contributes to definition of a genetic risk profile for this complication. Lupus. 2017 Jul;26(8):841-848.
32. Botta A, Rossi G, Marcaurelio M, Fontana L, D'Apice MR, Brancati F, Massa R, G Monckton D, Sangiuolo F, Novelli G. Identification and characterization of 5' CCG interruptions in complex DMPK expanded alleles. Eur J Hum Genet. 2017 Feb;25(2):257-261.
33. Romeo F, Novelli G, Ferrari M, Talamo M. Beyond the cardiovascular risk charts: the new way of hybrid profiles. J Cardiovasc Med (Hagerstown). 2016 Dec;17(12):851-854.
34. Ciccacci C, Politi C, Biancone L, Latini A, Novelli G, Calabrese E, Borgiani P. Polymorphisms in MIR122, MIR196A2, and MIR124A Genes are Associated with Clinical Phenotypes in Inflammatory Bowel Diseases. Mol Diagn Ther. 2017 Feb;21(1):107-114.
35. Cassone M, Ferradini V, Longo G, Sarchielli P, Murasecco D, Romoli M, Pasquini E, Novelli G, Prontera P, Sangiuolo F. Genotype-phenotype correlation of F484L mutation in three Italian families with Thomsen myotonia. Muscle Nerve. 2017 Jun;55(6):E24-E25.
36. Mizzi C, Dalabira E, Kumuthini J, Dzimiri N, Balogh I, Başak N, Böhm R, Borg J, Borgiani P, Bozina N, Bruckmueller H, Burzynska B, Carracedo A, Cascorbi I, Deltas C, Dolzan V, Fenech A, Grech G, Kasiulevicius V, Kádaši Ľ, Kučinskas V, Khusnutdinova E, Loukas YL, Macek M Jr, Makukh H, Mathijssen R, Mitropoulos K, Mitropoulou C, Novelli G, Papantoni I, Pavlovic S, Saglio G, Setric J, Stojiljkovic M, Stubbs AP, Squassina A, Torres M, Turnovec M, van Schaik RH, Voskarides K, Wakil SM, Werk A, Del Zompo M, Zukic B, Katsila T, Lee MT, Motsinger-Rief A, Mc Leod HL, van der Spek PJ, Patrinos GP. A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS One. 2016 Sep 16;11(9):e0162866.
37. Bianchi L, Costanza G, Campione E, Ruzzetti M, Di Stefani A, Diluvio L, Giardina E, Cascella R, Cordiali-Fei P, Bonifati C, Chiricozzi A, Novelli G, Ensoli F, Orlandi A. Biomolecular index of therapeutic efficacy in psoriasis treated by anti-TNF alpha agents. G Ital Dermatol Venereol. 2016 Sep 14.
38. Politi C, Ciccacci C, D'Amato C, Novelli G, Borgiani P, Spallone V. Recent advances in exploring the genetic susceptibility to diabetic neuropathy. Diabetes Res Clin Pract. 2016 Oct;120:198-208.
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