curriculum vitae

Personale Information

Name	**Novelli Antonio**
Nationality	Italian
Date of birth	18/02/1969
Gender	M

Present job position

Date	✓ Since march 2015
Position	✓ Chief of Medical Genetics department
Institute	✓ Children Hospital Bambino Gesu', IRCCS ✓ Viale di San Paolo,15 ✓ 00146 Rome, Italy

Learn more current and recent information

- ✓ Associate Professor school of Medicine at the UNICAMILLUS University since august 2024
- ✓ He is qualified as full Professor for the competitive sector 06 / A1 - Medical Genetics, sector scientific disciplinary MED03. VALID FROM 07/11/2023 TO 07/11/2034
- ✓ He is qualified as associate Professor for the competitive sector 06 / A1 - Medical Genetics, sector scientific disciplinary MED03. VALID FROM 07/01/2020 TO 07/01/2029
- ✓ He is the scientific coordinator of the II level InterUniversity Specialising Master's Degree Tor Vergata and Bambino Gesu' Chldren Hospital in Cytogenetics and Cytogenomics since 2011 to today (IX edition)
- ✓ He is faculty member at the School of Specialization in Medical Genetics at the University of Tor Vergata from 01 January 2019 to today
- ✓ He is a professor of genetics at the Postgraduate School of Clinical Pathology and Clinical Biochemistr from the 2013-14 academic year to today. Biomedical Campus University of Rome

Professional Experience

Date	October 2005 - February 2015
Position	Biologist
Competence	Person in charge of Citogenetics Department
Institute	Mendel Institute, "Casa Sollievo della Sofferenza" Hospital Viale Regina Margherita, 261 - Rome

Date	Since 2007
Position	Consultant of Medical Genetic Department
Institute	San Pietro Fatebenefratelli Hospital, Via Cassia 600, 00100 Rome

Date	July 1994 - December 2006
Position	Consultant of Cytogenetics Department
Institute	BIOS S.p.a., Via D. Chelini, 39 - Rome

Date	August 2002 - November 2005
Position	Consultant of Medical Genetics Department
Institute	S. Camillo Forlanini Hospital- Rome

Date	June 1994 - July 2000
Position	Fellow researcher. Fellow researcher. New technologies Development in human cytogenetics
Institute	Tor VergataUniversity - Rome

Date	January 1994 - December 1995
Position	Consultant. Legal-medicine tests involving DNA polymorphisms
Institute	Criminal department of scientific police

Education

Date	December 2001
Biology Specialty	**Medical Genetics**
University	University "G. D'Annunzio" - Chieti

Date	April 1994
Degree	National Certification in Biology n. 047995

Date	Since March 1993
Degree	Biology
University	Urbino University

Personal Competence

Mather tongue **Italian**

Other Languages

Auto evaluation	**Comprehension**		**Spoken**		**Written**
European Level	Listening	reading	Interaction	Oral Production	Written Production
English	**good**	**good**	**good**	**good**	**Sufficient**

Further information

Position and competences	Member of Italian Society of Human Genetics (SIGU) National coordinator of SIGU Cytogenetics working group
Position and competences	Prenatal Assessor for CEQA (Cytogenetic European Quality Assessment) Eurogentest
Position and competences	SIGU Assessor Cytogenetics external quality control. National cytogenetics quality control (National Institute of Health)
Position and competences	Professor at Pediatrics and Genetics Specialty School, Messina University
Position and competences	Professor at Medical Genetics Specialty School, La Sapienza University (Rome)
Position and competences	Professor at Clinical Pathology Specialty School, Campus Biomedico University (Rome)

- ✓ Participation in writing Prenatal Non-Invasive Screening guide lines (Non Invasive Prenatal Testing – NIPT), published on 03/07/2015 and 09/03/2021
- ✓ Participation in the drafting of the Ministerial Recommendations "Transfer of Omic Techniques in Clinical Practice" published in September 2021 as an expert member of the Section I Working Group of the Consiglio Superiore di Sanita' - Ministry of Health
- ✓ Acknowledgement of merits, Medicina Italia Prize 7th edition, Pescara 20 November 2022
- ✓ Participation at the Genetics Prescription Pertinence meeting of National Health System on 04.09.2015
- ✓ From 2021 to date Member of the Advisory Board NIPT Prenatal Screening, Revvity
- ✓ From 2023 to present Member of the Advisory Board Retina XLRP, Johnson & Johnson
- ✓ Participation as speaker/moderator at congresses and conferences of national and international interest
- ✓ Participation in the creation of the GenomeUp srl entrepreneurial project within the programme of LUISS EnLabs (winter 2018 edition - batch XII) for innovative start-ups and research spin-offs.
- ✓ Patent for industrial invention entitled "Method for the calculation of the adverse prognoses risk score in respiratory viral disease infections, using host genom DEA Ref.: 23AP24I (ex22B591). Patent No. 10202300000680

Published in international scientific journals:

Antonio Novelli is author and co-author of more than 400 scientific articles in international journals with medium-high impact factor, among the main topics: genomics, mental retardation, copy number variations, genetic and genomic syndromes.

Scopus H-index: 38
ORCID: 0000-0002-9037-4297
Scopus Author ID: 7005749853
Web of Science ResearcherID: K-4035-2016

Total Publications: 430 (*scopus source*)

The 30 most recent scientific publications in the year 2024 in international scientific journals

Prenatal identification of a pathogenic maternal *FGFR1* variant in two consecutive pregnancies with fetal forebrain malformations.
Graziani L, Nuovo S, Pisaneschi E, Carriero ML, Baghernajad Salehi L, Nardone AM, Manganaro L, Novelli A, **D'Apice MR, Mappa I, Novelli G.**
J Matern Fetal Neonatal Med. 2024 Dec;37(1):2344718. doi: 10.1080/14767058.2024.2344718. Epub 2024 Apr 28.
PMID: 38679587

Massive pericardial effusion in an infant with Aymé-Gripp syndrome: A case report and review of the literature.
Esposito A, Niceta M, Novelli A, **Magliozzi M, Parlapiano G, Baban A, Perrone MA.**
Am J Med Genet A. 2024 Sep;194(9):e63586. doi: 10.1002/ajmg.a.63586. Epub 2024 May 6.
PMID: 38709155 Review.

Genetic Variability of *SOX10*-Related Disorders within an Italian Family: Straddling the Line between Kallmann and Waardenburg Syndrome.
Graziani L, Carriero ML, Pozzi F, Minotti C, Andreadi A, Bellia A, Ruta R, Bengala M, Novelli A, **Lauro D, Novelli G.**
Mol Syndromol. 2024 Aug;15(4):339-346. doi: 10.1159/000536574. Epub 2024 Mar 22.
PMID: 39119450

Loss-of-function variants in ERF are associated with a Noonan syndrome-like phenotype with or without craniosynostosis.
Dentici ML, Niceta M, Lepri FR, Mancini C, Priolo M, Bonnard AA, Cappelletti C, Leoni C, Ciolfi A, Pizzi S, Cordeddu V, Rossi C, Ferilli M, Mucciolo M, Colona VL, Fauth C, Bellini M, Biasucci G, Sinibaldi L, Briuglia S, Gazzin A, Carli D, Memo L, Trevisson E, Schiavariello C, Luca M, Novelli A, **Michot C, Sweertvaegher A, Germanaud D, Scarano E, De Luca A, Zampino G, Zenker M, Mussa A, Dallapiccola B, Cavé H, Digilio MC, Tartaglia M.**
Eur J Hum Genet. 2024 Aug;32(8):954-963. doi: 10.1038/s41431-024-01642-7. Epub 2024 Jun 1.
PMID: 38824261

Lack of association between classical HLA genes and asymptomatic SARS-CoV-2 infection.
Marchal A, Cirulli ET, Neveux I, Bellos E, Thwaites RS, Schiabor Barrett KM, Zhang Y, Nemes-Bokun I, Kalinova M, Catchpole A, Tangye SG, Spaan AN, Lack JB, Ghosn J, Burdet C, Gorochov G, Tubach F, Hausfater P; COVID Human Genetic Effort; COVIDeF Study Group; French COVID Cohort Study Group; CoV-Contact Cohort; COVID-STORM Clinicians; COVID Clinicians; Orchestra Working Group; Amsterdam UMC COVID-19 Biobank; NIAID-USUHS COVID Study Group; Dalgard CL, Zhang SY, Zhang Q, Chiu C, Fellay J, Grzymski JJ, Sancho-Shimizu V, Abel L, Casanova JL, Cobat A, Bolze A.
HGG Adv. 2024 Jul 18;5(3):100300. doi: 10.1016/j.xhgg.2024.100300. Epub 2024 Apr 26.
PMID: 38678364 **Free PMC article.**

De Novo *DNM1L* Mutation in a Patient with Encephalopathy, Cardiomyopathy and Fatal Non-Epileptic Paroxysmal Refractory Vomiting.
Berti B, Verrigni D, Nasca A, Di Nottia M, Leone D, Torraco A, Rizza T, Bellacchio E, Legati A, Palermo C, Marchet S, Lamperti C, Novelli A, **Mercuri EM, Bertini ES, Pane M, Ghezzi D, Carrozzo R.**

Int J Mol Sci. 2024 Jul 16;25(14):7782. doi: 10.3390/ijms25147782.
PMID: 39063023 Free PMC article.

Movement disorder phenotype in CTNNB1-syndrome: A complex but recognizable phenomenology.
Garone G, Innocenti A, Grasso M, Mandarino A, Capuano A, Della Bella G, Frascarelli F, Diodato D, Onesimo R, Zampino G, Novelli A, Digilio MC, Bartuli A, Dentici ML, Parisi P, Galosi S, Tonduti D, Bertini E, Sinibaldi L, Specchio N.
Parkinsonism Relat Disord. 2024 Jul 9;126:107057. doi: 10.1016/j.parkreldis.2024.107057. Online ahead of print.PMID: 39067319

Partial atrioventricular canal defect and aortic coarctation associated with variants in GDF1 and NOTCH1 genes: A case report.
Putotto C, Masci M, Magliozzi M, Novelli A, Marino B, Digilio MC, Toscano A.
Birth Defects Res. 2024 Jul;116(7):e2382. doi: 10.1002/bdr2.2382.
PMID: 38975735

Usmani-Riazuddin syndrome can have a recognizable phenotype: Report of a novel AP1G1 variant.
Gnazzo M, Pascolini G, Parlapiano G, Petrizzelli F, Perrino D, Porco L, Bartuli A, Novelli A, Baban A.
Clin Genet. 2024 Jul;106(1):109-113. doi: 10.1111/cge.14531. Epub 2024 Apr 25.
PMID: 38665048

DNA methylation profiling in Kabuki syndrome: reclassification of germline KMT2D VUS and sensitivity in validating postzygotic mosaicism.
Niceta M, Ciolfi A, Ferilli M, Pedace L, Cappelletti C, Nardini C, Hildonen M, Chiriatti L, Miele E, Dentici ML, Gnazzo M, Cesario C, Pisaneschi E, Baban A, Novelli A, Maitz S, Selicorni A, Squeo GM, Merla G, Dallapiccola B, Tumer Z, Digilio MC, Priolo M, Tartaglia M.
Eur J Hum Genet. 2024 Jul;32(7):819-826. doi: 10.1038/s41431-024-01597-9. Epub 2024 Mar 25.
PMID: 38528056

Clinical and molecular cytogenetic studies of five new patients with 20q11q12 deletion and review of the literature: Proposition of two critical regions.
Bensaid S, Bendahmane M, Loddo S, Poke G, Januel L, Nicolle R, Malan V, Chatron N, Ottombrino S, Dentici ML, Novelli A, Digilio MC, Sanlaville D.
Am J Med Genet A. 2024 Jul;194(7):e63580. doi: 10.1002/ajmg.a.63580. Epub 2024 Mar 21.
PMID: 38511524 Review.

A Case of CDKL5 Deficiency Due to an X Chromosome Pericentric Inversion: Delineation of Structural Rearrangements as an Overlooked Recurrent Pathological Mechanism.
Lombardo A, Sinibaldi L, Genovese S, Catino G, Mei V, Pompili D, Sallicandro E, Falasca R, Liambo MT, Faggiano MV, Roberti MC, Di Donato M, Vitelli A, Russo S, Giannini R, Micalizzi A, Pietrafusa N, Digilio MC, Novelli A, Fusco L, Alesi V.
Int J Mol Sci. 2024 Jun 24;25(13):6912. doi: 10.3390/ijms25136912.
PMID: 39000022 Free PMC article.

A De Novo *CaSR* Missense Variant in Combination with Two Inherited Missense Variants in *CFTR* and *SPINK1* Detected in a Patient with Chronic Pancreatitis.
Bontempo P, Surace C, Menale L, Alicata C, D'Elia G, Tomaiuolo AC, Minervino D, Lorefice E, Novelli A.
Biomedicines. 2024 Jun 9;12(6):1278. doi: 10.3390/biomedicines12061278.
PMID: 38927485 Free PMC article.

A novel mutation in GAS8 gene associated with chronic rhinosinusitis with nasal polyposis in a case of primary ciliary dyskinesia: a case report.
Artesani MC, Santarsiero S, Sitzia E, Lepri FR, Magliozzi M, Majo F, Ullmann N, Stracuzzi A, Novelli A, Cristalli G, Fiocchi A.
Front Pediatr. 2024 May 30;12:1345265. doi: 10.3389/fped.2024.1345265. eCollection 2024.
PMID: 38873586 Free PMC article.

Congenital Hyperinsulinism of a Large Italian Cohort: A Retrospective Study.
Tagliaferri F, Iannuzzi R, Canciani G, Bernabei SM, Campana C, Caviglia S, Greco B, Lepri FR, Novelli A, Pizzoferro M, Garganese MC, Spada M, Francalanci P, Dionisi-Vici C, Maiorana A.
Horm Res Paediatr. 2024 May 28:1-13. doi: 10.1159/000538943. Online ahead of print.
PMID: 38806014

Diagnostic accuracy of cell-free DNA in maternal blood in detecting chromosomal anomalies in twin pregnancies: systematic review and meta-analysis.
Della Valle L, Piergianni M, Khalil A, Novelli A, Rizzo G, Mappa I, Prasad S, Matarrelli B, Gatta V, Stuppia L, Pagani G, Flacco ME, D'Antonio F.
Ultrasound Obstet Gynecol. 2024 May 22. doi: 10.1002/uog.27698. Online ahead of print.
A Novel *COL4A5* Pathogenic Variant Joins the Dots in a Family with a Synchronous Diagnosis of Alport Syndrome and Polycystic Kidney Disease.
Graziani L, Minotti C, Carriero ML, Bengala M, Lai S, Terracciano A, Novelli A, Novelli G.
Genes (Basel). 2024 May 8;15(5):597. doi: 10.3390/genes15050597.
PMID: 38790225 Free PMC article.
Expanding the Phenotype of the CACNA1C-Associated Neurological Disorders in Children: Systematic Literature Review and Description of a Novel Mutation.
Cipriano L, Piscopo R, Aiello C, Novelli A, Iolascon A, Piscopo C.
Children (Basel). 2024 Apr 30;11(5):541. doi: 10.3390/children11050541.
PMID: 38790536 Free PMC article. Review.
Spectrum of *ERCC6*-Related Cockayne Syndrome (Type B): From Mild to Severe Forms.
Sartorelli J, Travaglini L, Macchiaiolo M, Garone G, Gonfiantini MV, Vecchio D, Sinibaldi L, Frascarelli F, Ceccatelli V, Petrillo S, Piemonte F, Piccolo G, Novelli A, Longo D, Pro S, D'Amico A, Bertini ES, Nicita F.
Genes (Basel). 2024 Apr 18;15(4):508. doi: 10.3390/genes15040508.
PMID: 38674442 Free PMC article.
A new variant in the GATA6 gene associated with tracheoesophageal fistula, pulmonary vein stenosis and neonatal diabetes.
Pugnaloni F, Martini L, De Rose DU, Landolfo F, Giliberti P, Ruta R, Novelli A, Rapini N, Barbetti F, Toscano A, Conforti A, Bagolan P, Capolupo I, Dotta A.
Horm Res Paediatr. 2024 Apr 4. doi: 10.1159/000536621. Online ahead of print.
PMID: 38574486
Unique Features of Cardiovascular Involvement and Progression in Children with Marfan Syndrome Justify Dedicated Multidisciplinary Care.
Baban A, Parlapiano G, Cicenia M, Armando M, Franceschini A, Pacifico C, Panfili A, Zinzanella G, Romanzo A, Fusco A, Caiazza M, Perri G, Galletti L, Digilio MC, Buonuomo PS, Bartuli A, Novelli A, Raponi M, Limongelli G.
J Cardiovasc Dev Dis. 2024 Apr 3;11(4):114. doi: 10.3390/jcdd11040114.
PMID: 38667733 Free PMC article. Review.
A second case report of medulloblastoma in a patient carrying biallelic pathogenic MUTYH germline variants.
Cipri S, Del Baldo G, Carai A, Cacchione A, Agolini E, Novelli A, Rossi S, Colafati GS, Boccuto L, Mastronuzzi A.
Neuropathol Appl Neurobiol. 2024 Apr;50(2):e12968. doi: 10.1111/nan.12968.
PMID: 38477379 No abstract available.
Case report: Long term response to growth hormone in a child with Silver-Russell syndrome-like phenotype due to a novel paternally inherited IGF2 variant.
Ventresca S, Lepri FR, Criscuolo S, Bottaro G, Novelli A, Loche S, Cappa M.
Front Endocrinol (Lausanne). 2024 Mar 26;15:1364234. doi: 10.3389/fendo.2024.1364234. eCollection 2024.
PMID: 38596219 Free PMC article.
Structural rearrangements as a recurrent pathogenic mechanism for SETBP1 haploinsufficiency.
Alesi V, Genovese S, Roberti MC, Sallicandro E, Di Tommaso S, Loddo S, Orlando V, Pompili D, Calacci C, Mei V, Pisaneschi E, Faggiano MV, Morgia A, Mammì C, Astrea G, Battini R, Priolo M, Dentici ML, Milone R, Novelli A.
Hum Genomics. 2024 Mar 22;18(1):29. doi: 10.1186/s40246-024-00600-0.
PMID: 38520002 Free PMC article.
Congenital Heart Defects in Patients with Molecularly Confirmed Sotos Syndrome.
Calcagni G, Ferrigno F, Franceschini A, Dentici ML, Capolino R, Sinibaldi L, Minotti C, Micalizzi A, Alesi V, Novelli A, Baban A, Parlapiano G, Coviello D, Versacci P, Putotto C, Chinali M, Drago F, Bartuli A, Marino B, Digilio MC.
Diagnostics (Basel). 2024 Mar 11;14(6):594. doi: 10.3390/diagnostics14060594.
PMID: 38535015 Free PMC article.

The changing landscape of neonatal diabetes mellitus in Italy between 2003-2022.
Rapini N, Delvecchio M, Mucciolo M, Ruta R, Rabbone I, Cherubini V, Zucchini S, Cianfarani S, Prandi E, Schiaffini R, Bizzarri C, Piccini B, Maltoni G, Predieri B, Minuto N, Di Paola R, Giordano M, Tinto N, Grasso V, Russo L, Tiberi V, Scaramuzza A, Frontino G, Maggio MC, Musolino G, Piccinno E, Tinti D, Carrera P, Mozzillo E, Cappa M, Iafusco D, Bonfanti R, Novelli A**, Barbetti F; Diabetes Study Group of Italian Society for Pediatric Endocrinology and Diabetes (ISPED).**
J Clin Endocrinol Metab. 2024 Feb 26:dgae095. doi: 10.1210/clinem/dgae095. Online ahead of print.
PMID: 38408297

Case report: A new *de novo* 6q21q22.1 interstitial deletion case in a girl with cerebellar vermis hypoplasia and developmental delay and literature review.
Minotti C, Graziani L, Sallicandro E, Digilio MC, Falasca R, Alesi V, Novelli G, Dentici ML, Loddo S, Novelli A.
Front Genet. 2024 Feb 6;14:1315291. doi: 10.3389/fgene.2023.1315291. eCollection 2023.
PMID: 38380230 **Free PMC article.**

Non-Invasive Prenatal Test Analysis Opens a Pandora's Box: Identification of Very Rare Cases of SRY-Positive Healthy Females, Segregating for Three Generations Thanks to Preferential Inactivation of the XqYp Translocated Chromosome.
Politi C, Grillone K, Nocera D, Colao E, Bellisario ML, Loddo S, Catino G, Novelli A**, Perrotti N, Rodolfo I, Malatesta P.**
Genes (Basel). 2024 Jan 16;15(1):103. doi: 10.3390/genes15010103.
PMID: 38254992 Free PMC article.

PATZ1-Rearranged Tumors of the Central Nervous System: Characterization of a Pediatric Series of Seven Cases.
Rossi S, Barresi S, Colafati GS, Genovese S, Tancredi C, Costabile V, Patrizi S, Giovannoni I, Asioli S, Poliani PL, Gardiman MP, Cardoni A, Del Baldo G, Antonelli M, Gianno F, Piccirilli E, Catino G, Martucci L, Quacquarini D, Toni F, Melchionda F, Viscardi E, Zucchelli M, Dal Pos S, Gatti E, Liserre R, Schiavello E, Diomedi-Camassei F, Carai A, Mastronuzzi A, Gessi M, Giannini C, Novelli A**, Onetti Muda A, Miele E, Alesi V, Alaggio R.**
Mod Pathol. 2024 Feb;37(2):100387. doi: 10.1016/j.modpat.2023.100387. Epub 2023 Nov 23.
PMID: 38007157

Patient preferences in genetic newborn screening for rare diseases: study protocol.
Martin S, Angolini E, Audi J, Bertini E, Bruno LP, Coulter J, Ferlini A, Fortunato F, Frankova V, Garnier N, Grauman Å, Gross E, Hauber B, Hansson M, Kirschner J, Knieling F, Kyosovksa G, Ottombrino S, Novelli A, Raming R, Sansen S, Saier C, Veldwijk J.
BMJ Open. 2024 Apr 19;14(4):e081835. doi: 10.1136/bmjopen-2023-081835.PMID: 38643010 Free PMC article.

Scientific Books and Manuals

Reproductive biology and techniques
Chapter 14 pages 303-320 Genetic Factors of Infertility
edi-ermes 2021

Arch of Janus
journal in Italy of Medical Humanities N°102 winter 2019
Screening and prenatal diagnosis
Sce Sc Publisher - Cooperative Society

Reproductive Medicine: The common clinical and
diagnostic pathway
Gynaecology: Chapter 7 Pages 341-350
Genetic testing in the infertile couple
Editeam Publishing 2018

Handbook of Fetal Neurosonology;
Genetic Bases of Congenital Malformations of the Central Nervous System and Related Diagnostic Tests
Chapter 1 p. 1-27
SIEOG (Italian Society of Obstetric and Gynaecological Sonography)
Editeam, may 2017

Manual of Fetal Echocardiography;
Genetics of Congenital Heart Disease and Diagnostic Tests; Chapter 15 p. 214-238.
SIEOG (Italian Society of Obstetric and Gynaecological Sonography)
Editeam, november 2016

Trombophilia in Obstetrics and Gynaecology
Haematological Parameters in Pregnancy Chapter 3 p. 73-91
Verduci Publisher 2015

Data: 10/08/2024 **firma**