## PERSONAL INFORMATION


| | |
|---|---|
| Name | Arcangela De Nicolo, M.D., Ph.D. |
| Institute and Work address | IRCCS San Raffaele Scientific Institute<br>Via Olgettina 60<br>20132 Milan<br>Italy |
| Nationality | Italian |


## EDUCATION

| | |
|---|---|
| 2020 | *Master (second level),* Cancer Genetics<br>University of Pavia, Pavia, Italy |
| 2010 | *Certificate*, Research Synthesis and Meta-Analysis in Public Health and Clinical Medicine<br>Harvard T.H. Chan School of Public Health, Boston, MA, USA |
| 2003 | *Ph.D.*, Oncology and Surgical Oncology<br>University of Padua, Padua, Italy |
| 2000 | *Certificate of Advanced Study Course*, Clinical and Experimental Oncology<br>University of Bari, Bari, Italy |
| 1998 | *M.D. summa cum laude*<br>University of Bari, Bari, Italy |

### Licensure

| | |
|---|---|
| 1999 | Enrollment in the Italian Registry of Medical Doctors, Surgeons, and Dentists, District of Bari |
| 1999 | Italian Medical License |

## WORK EXPERIENCE

2022-, *Coordinator*, Translational Genetics Research Area/Center for Omics Sciences, IRCCS San Raffaele Scientific Institute, Milan, Italy

2019-2022, *Visiting Scientist* and *Collaborating Expert*/Department of Research–Unit of Molecular Bases of Genetic Risk and Genetic Testing, Fondazione IRCCS Istituto Nazionale dei Tumori, Milan, Italy

2018-2022, *Investigator* (2018-), *Head of Program Section* (2019-)/Cancer Genetics Program (formerly Cancer Genomics Program), Veneto Institute of Oncology IRCCS, Padua, Italy

2016-2018, *Physician Scientist*/Cancer Genomics Program, Veneto Institute of Oncology IRCCS, Padua, Italy

2015-2023*, Adjunct Faculty*/Ph.D. Course in Clinical and Experimental Oncology and Immunology*,* University of Padua, Padua, Italy

2015 (1 month), *Collaborating Researcher*/Department of Immunology and Diagnostic Molecular Oncology, Veneto Institute of Oncology IRCCS, Padua, Italy

2013-2015*, Collaborating Guest*/Department of Biological Chemistry and Molecular Pharmacology*,* Harvard Medical School, Boston, MA, USA

2011-2013*, Clinical Observer*/Breast Oncology Center and Center for Cancer Genetics and Prevention/Susan F. Smith Center for Women's Cancers*,* Dana-Farber Cancer Institute, Boston, MA, USA

2009-2013*, Faculty Academic Appointment - Instructor in Medicine*/Departments of Cancer Biology and Medicine*,* Dana-Farber Cancer Institute/Harvard Medical School/Brigham and Women's Hospital, Boston, MA, USA

2003-2009*, Research Fellow in Genetics*/Departments of Cancer Biology and Genetics*,* Dana-Farber Cancer Institute/Harvard Medical School, Boston, MA, USA

2002 (6 months)*, Visiting Ph.D. Student*/Departments of Cancer Biology and Genetics*,* Dana-Farber Cancer Institute/Harvard Medical School, Boston, MA, USA

## LANGUAGES

First: Italian

Other: English (full professional proficiency); Spanish (elementary proficiency)

## ADDITIONAL INFORMATION

**Student Supervision**
2015-2023, (co)supervised two Ph.D. students, one medical student, and one Erasmus exchange program student at the University of Padua, Padua, Italy

**Teaching of Students in Doctoral Programs**
2016-2021, served as a lecturer in the Ph.D. Course in Clinical and Experimental Oncology and Immunology, University of Padua, Padua, Italy. Seminar series - Genetic testing for cancer susceptibility in the NGS era: challenges and opportunities

**Professional Societies (National and International)**
2018-, Società Italiana di Genetica Umana (SIGU)
2014-, European Society for Medical Oncology (ESMO)
2012-, American Society of Clinical Oncology (ASCO)
2011-, European Association for Cancer Research (EACR)
2002-, American Association for Cancer Research (AACR)

**Editorial Activities**
2023-, Cancers (*Guest Editor)*
2008-, Breast Cancer Research and Treatment, Cancer Epidemiology, Biomarkers and Prevention, Clinical Cancer Research, European Journal of Human Genetics, Familial Cancer, Frontiers in Oncology, JCO Precision Oncology, Journal of Medical Genetics, Mutation Research (*Ad hoc Reviewer)*

**Grant review activities**
2013, Italian Ministry of Health (*Foreign Reviewer*)
2020, Mia Neri Foundation Onlus Association (*Reviewer*)

**Guidelines and Recommendations**
SIGU/Pannelli multigenici per la diagnosi genetica di forme di predisposizione ai tumori: linee di indirizzo per l'utilizzo nella pratica clinica - under evaluation (*Compiler*)

SIGU/Linee di indirizzo sull'analisi dei geni *BRCA1* e *BRCA2* in ambito clinico: criteri di accesso al test, aggiornamento sulle piattaforme diagnostiche e interpretazione del test somatico (*Compiler*)

**Scientific Interests**
Hereditary Breast and Ovarian Cancer, *BRCA1* and *BRCA2* Genes, Variant Classification and Reporting, Variants of Uncertain Significance, Overlapping and Atypical Cancer Phenotypes, Molecular Mechanisms of Carcinogenesis, Cell Division and DNA Repair

**Poster and Oral Presentations at Scientific Meetings**
(Co)author in 52 poster or oral presentations at scientific meetings (including 21 presentations as first or last author at 36 international meetings)

**Invited Presentations and Organized and Chaired Sessions** (2016-)
Invited speaker and/or session chair and organizer at 19 national and international scientific meetings

## Selected Peer-Reviewed Publications (2019-)


1. Block I, Mateu-Regué À, Do TTN, Miceikaite I, Sdogati D, Larsen MJ, Hao Q, Nielsen HR, Boonen SE, Skytte AB, Jensen UB, Høffding LK, **De Nicolo A**, Viel A, Tudini E, Parsons MT, Hansen TVO, Rossing M, Kruse TA, Spurdle AB, Thomassen M. Male with an apparently normal phenotype carrying a *BRCA1* exon 20 duplication *in trans* to a *BRCA1* frameshift variant. *Breast Cancer Res 2024;26(1):6*.

2. Zanti M, O'Mahony DG, Parsons MT, Li H, Dennis J, Aittomäki K, Andrulis IL, Anton-Culver H, Aronson KJ, Augustinsson A, Becher H, Bojesen SE, Bolla MK, Brenner H, Brown MA, Buys SS, Canzian F, Caputo SM, Castelao JE, Chang-Claude J, GC-HBOC study Collaborators, Czene K, Daly MB, **De Nicolo A**, Devilee P, Dörk T, Dunning AM, Dwek M, Eccles DM, Engel C, Evans DG, Fasching PA, Gago-Dominguez M, García-Closas M, García-Sáenz JA, Gentry-Maharaj A, Geurts–Giele WRR, Giles GG, Glendon G, Goldberg MS, Gómez Garcia EB, Güendert M, Guénel P, Hahnen E, Haiman CA, Hall P, Hamann U, Harkness EF, Hogervorst FBL, Hollestelle A, Hoppe R, Hopper JL, Houdayer C, Houlston RS, Howell A, ABCTB Investigators, Jakimovska M, Jakubowska A, Jernström H, John EM, Kaaks R, Kitahara CM, Koutros S, Kraft P, Kristensen VN, Lacey JV, Lambrechts D, Léoné M, Lindblom A, Lubiński J, Lush M, Mannermaa A, Manoochehri M, Manoukian S, Margolin S, Martinez ME, Menon U, Milne RL, Monteiro AN, Murphy RA, Neuhausen SL, Nevanlinna H, Newman WG, Offit K, Park SK, Paul J, Peterlongo P, Peto J, Plaseska-Karanfilska D, Punie K, Radice P, Rashid MU, Rennert G, Romero A, Rosenberg EH, Saloustros E, Sandler DP, Schmidt MK, Schmutzler RK, Shu XO, Simard J, Southey MC, Stone J, Stoppa-Lyonnet D, Tamimi RM, Tapper WJ, Taylor JA, Teo SH, Teras LR, Terry MB, Thomassen M, Troester MA, Vachon CM, Vega A, Vreeswijk MPG, Wang Q, Wappenschmidt B, Weinberg CR, Wolk A, Zheng W, Feng B, Couch FJ, Spurdle AB, Easton DF, Goldgar DE, Michailidou K, on behalf of the Breast Cancer Association Consortium and the Evidence-Based Network for the Interpretation of Germline Mutant Alleles. A likelihood ratio approach for utilizing case-control data in the clinical classification of rare sequence variants: application to *BRCA1* and *BRCA2. Hum Mutat 2023*; https://doi.org/10.1155/2023/9961341.

3. Stolarova L, Kleiblova P, Zemankova P, Stastna B, Janatova M, Soukupova J, Achatz MI, Ambrosone C, Apostolou P, Arun BK, Auer P, Barnard M, Bertelsen B, Biobank Japan, Blok MJ, Boddicker N, Brunet J, Burnside ES, Calvello M, Campbell I, Chan SH, Chen F, Chiang JB, Coppa A, Cortesi L, Crujeiras-González A, Consortium CZECANCA, De Leeneer K, De Putter R, DePersia A, Devereux L, Domchek S, Efremidis A, Engel C, Ernst C, Evans DGR, Feliubadaló L, Fostira F, Fuentes-Ríos O, Gómez-García EB, González S, Haiman C, van Overeem Hansen T, Hauke J, Hodge J, Hu C, Huang H, Ishak NDB, Iwasaki Y, Konstantopoulou I, Kraft P, Lacey J, Lázaro C, Li N, Lim WK, Lindström S, Lori A, Martinez E, Martins A, Matsuda K, Matullo G, McInerny S, Michailidou K, Montagna M, Monteiro ANA, Mori L, Nathanson K, Neuhausen SL, Nevanlinna H, Olson JE, Palmer J, Pasini B, Patel A, Piane M, Poppe B, Radice P, Renieri A, Resta N, Richardson ME, Rosseel T, Ruddy KJ, Santamariña M, Santana dos Santos E, Teras L, Toland AE, Trentham-Dietz A, Vachon CM, Volk AE, Weber-Lassalle N, Weitzel JN, Wiesmuller L, Winham S, Yadav S, Yannoukakos D, Yao S, Zampiga V, Zethoven M, Zhang ZW, Zima T, Spurdle AB, Vega A, Rossing M, Del Valle J, **De Nicolo A**, Hahnen E, Claes KBM, Ngeow J, Momozawa Y, James PA, Couch FJ, Macurek L, and Kleibl Z. ENIGMA *CHEK2*gether project: a comprehensive study identifies functionally impaired *CHEK2* germline missense variants associated with increased breast cancer risk. *Clin Cancer Res 2023; 29(16):3037-50*.

4. Figlioli G, **De Nicolo A**, Catucci I, Manoukian S, Peissel B, Azzollini J, Beltrami B, Bonanni B, Calvello M, Bondavalli D, Pasini B, Vignolo Lutati F, Ogliara P, Zuradelli M, Pensotti V, De Vecchi G, Volorio S, Verderio P, Pizzamiglio S, Matullo G, Aneli S,

Birolo G, Zanardi F, Tondini C, Zambelli A, Livraghi L, Franchi M, Radice P, Peterlongo P. Analysis of Italian *BRCA1/2* pathogenic variants identifies a private spectrum in the population from the Bergamo province in Northern Italy. *Cancers (Basel) 2021;13(3):532*.

5. Del Bianco P, Stagni C, Giunco S, Fabozzi A, Elefanti L, Pellegrini S, Vecchiato A, Pigozzo J, Zamuner C, De Rossi A[§], **De Nicolo A[§]**, Menin C[§]. TERT promoter mutations differently correlate with the clinical outcome of MAPK inhibitor-treated melanoma patients*. Cancers (Basel) 2020;12(4):946.* [§]Shared senior authorship.

6. Landi MT, Bishop DT, MacGregor S, Machiela MJ, Stratigos AJ, Ghiorzo P, Brossard M, Calista D, Choi J, Fargnoli MC, Zhang T, Rodolfo M, Trower AJ, Menin C, Martinez J, Hadjisavvas A, Song L, Stefanaki I, Scolyer R, Yang R, Goldstein AM, Potrony M, Kypreou KP, Pastorino L, Queirolo P, Pellegrini C, Cattaneo L, Zawistowski M, Gimenez-Xavier P, Rodriguez A, Elefanti L, Manoukian S, Rivoltini L, Smith BH, Loizidou MA, Del Regno L, Massi D, Mandala M, Khosrotehrani K, Akslen LA, Amos CI, Andresen PA, Avril M-F, Azizi E, Soyer HP, Bataille V, Dalmasso B, Bowdler LM, Burdon KP, Chen WV, Codd V, Craig JE, Dębniak T, Falchi M, Fang S, Friedman E, Simi S, Galan P, Garcia-Casado Z, Gillanders EM, Gordon S, Green A, Gruis NA, Hansson J, Harland M, Harris J, Helsing P, Henders A, Hočevar M, Höiom V, Hunter DJ, Ingvar C, Kumar R, Lang J, Lathrop GM, Lee JE, Li X, Lubiński J, Mackie RM, Malt M, Malvehy J, McAloney K, Mohamdi H, Molven A, Moses EK, Neale RE, Novaković S, Nyholt DR, Olsson H, Orr N, Fritsche LG, Puig-Butille JA, Qureshi AA, Radford-Smith GL, Randerson-Moor J, Requena C, Rowe C, Samani NJ, Sanna M, Schadendorf D, Schulze H-J, Simms LA, Smithers M, Song F, Swerdlow AJ, van der Stoep N, Kukutsch NA, Visconti A, Wallace L, Ward SV, Wheeler L, Sturm RA, Hutchinson A, Jones K, Malasky M, Vogt A, Zhou W, Pooley KA, Elder DE, Han J, Hicks B, Hayward NK, Kanetsky PA, Brummett C, Montgomery GW, Olsen CM, Hayward C, Dunning AM, Martin NG, Evangelou E, Mann GJ, Long G, Pharoah PDP, Easton DF, Barrett JH, Cust AE, Abecasis G, Duffy DL, Whiteman DC, Gogas H, **De Nicolo A**, Tucker MA, Newton Bishop JA, GenoMEL Consortium, Q-MEGA and QTWIN Investigators, ATHENS Melanoma Study Group, 23andMe, The SDH Study Group, IBD Investigators, Essen-Heidelberg Investigators, AMFS Investigators, MelaNostrum Consortium, Peris K, Chanock SJ, Demenais F, Brown KM, Puig S, Nagore E, Shi J, Iles MM, Law MH. Genome-wide association meta-analysis combining multiple risk phenotypes provides insights into the genetic architecture of cutaneous melanoma susceptibility. *Nat Genet 2020;52(5):494-504*.

7. Joukov V and **De Nicolo A**. The centrosome and the primary cilium: the yin and yang of a hybrid organelle. *Cells 2019;8(7):701*. Review.

8. Parsons MT, Tudini E, Li H, Hahnen E, Wappenschmidt B, Feliubadaló L, Aalfs CM, Agata S, Aittomäki K, Alducci E, Alonso-Cerezo MC, Arnold N, Auber B, Austin R, Azzollini J, Balmaña J, Barbieri E, Bartram CR, Blanco A, Blümcke B, Bonache S, Bonanni B, Borg Å, Bortesi B, Brunet J, Bruzzone C, Bucksch K, Cagnoli G, Caldés T, Caliebe A, Caligo MA, Calvello M, Capone GL, Caputo S, Carnevali I, Carrasco E, Caux-Moncoutier V, Cavalli P, Cini G, Clarke EM, Concolino P, Cops EJ, Cortesi L, Couch FJ, Darder E, de la Hoya M, Dean M, Debatin I, Del Valle J, Delnatte C, Derive N, Diez O, Ditsch N, Domchek SM, Dutrannoy V, Eccles DM, Ehrencrona H, Enders U, Evans DG, Faust U, Felbor U, Feroce I, Fine M, Galvao HCR, Gambino G, Gehrig A, Gensini F, Gerdes AM, Germani A, Giesecke J, Gismondi V, Gómez C, Gómez Garcia EB, González S, Grau E, Grill S, Gross E, Guerrieri-Gonzaga A, Guillaud-Bataille M, Gutiérrez-Enríquez S, Haaf T, Hackmann K, Hansen TVO, Harris M, Hauke J, Heinrich T, Hellebrand H, Herold KN, Honisch E, Horvath J, Houdayer C, Hübbel V, Iglesias S, Izquierdo A, James PA, Janssen LAM, Jeschke U, Kaulfuß S, Keupp K, Kiechle M, Kölbl A, Krieger S, Kruse TA, Kvist A, Lalloo F, Larsen M, Lattimore VL, Lautrup C, Ledig S, Leinert E, Lewis AL, Lim J, Loeffler M, Lopez-Fernández A, Lucci-Cordisco E, Maass N, Manoukian S, Marabelli M, Matricardi L, Meindl A, Michelli RD, Moghadasi S, Moles-Fernández A, Montagna M, Montalban G, Monteiro AN, Montes E, Mori L, Moserle L, Müller CR, Mundhenke C, Naldi N, Nathanson KL, Navarro M, Nevanlinna H, Nichols CB, Niederacher D, Nielsen HR, Ong KR, Pachter N, Palmero EI, Papi L, Pedersen IS, Peissel B, Pérez-Segura P, Pfeifer K, Pineda M, Pohl-Rescigno E, Poplawski NK, Porfirio B, Quante AS, Ramser J, Reis RM, Revillion F, Rhiem K, Riboli B, Ritter J, Rivera D, Rofes P, Rump A, Salinas M, Sánchez de Abajo AM, Schmidt G, Schoenwiese U, Seggewiß J, Solanes A, Steinemann D, Stiller M, Stoppa-Lyonnet D, Sullivan KJ, Susman R, Sutter C, Tavtigian SV, Teo SH, Teulé A, Thomassen M, Tibiletti MG, Tognazzo S, Toland AE, Tornero E, Törngren T, Torres-Esquius S, Toss A, Trainer AH, van Asperen CJ, van Mackelenbergh MT, Varesco L, Vargas-Parra G, Varon R, Vega A, Velasco Á, Vesper AS, Viel A, Vreeswijk MPG, Wagner SA, Waha A, Walker LC, Walters RJ, Wang-Gohrke S, Weber BHF, Weichert W, Wieland K, Wiesmüller L, Witzel I, Wöckel A, Woodward ER, Zachariae S, Zampiga V, Zeder-Göß C, KConFab Investigators, Lázaro C, **De Nicolo A**, Radice P, Engel C, Schmutzler RK, Goldgar DE, Spurdle AB. Large scale multifactorial likelihood analysis of *BRCA1* and *BRCA2* variants: An ENIGMA resource to support clinical variant classification. *Hum Mutat 2019;40(9):1557-78*.

13/02/2024