

Andrea Mafficini

Curriculum Vitae

University of Verona
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Personal Data

Place and date of birth **San Bonifacio (VR - Italy), December 5th 1975.**
Citizenship **Italian.**

Education

- 2007 **Ph.D. in gastroenterological and surgical sciences of inflammatory and neoplastic hepatobiliary and pancreatic diseases**, Università degli Studi di Verona, Verona, Italy, Department of Surgical and Gastroenterological Sciences.
- 2001 **M.Sc. in Biotechnologies**, Università degli Studi di Verona, Faculty of Mathematical, Physical and Natural Sciences, Verona, Italy.

Further Courses

- 2018 **Course for BRCA variants interpretation**, Fondazione IRCCS Istituto Nazionale dei Tumori, Milano, Italy, 20 giugno.
- 2013 **CSAMA bioinformatics/biostatistics course**, R and Bioconductor for Genome Science, Università di Padova, branch of Brixen, Italy, June 23-28th.
- 2009 **Advanced cytometry course**, SIICA & Università degli studi di Modena, Modena, Italy, March 10-13th.
- 2008 **Theoretical and practical course of 3D fluorescence microscopy**, Università degli studi di Verona, Verona, Italy, February 29th.
- 2002 **E.S.F. course "Expert in GMO traceability"**, Università Cattolica di Piacenza, branch of Cremona, Italy February-July.
- 2002 **Course "Internal auditor of quality systems"**, Università Cattolica di Piacenza, Piacenza, Italy, June 17-18th.

Professional experience

- 2019–now **Assistant professor**, ARC-Net research centre, Università degli studi di Verona, Verona, Italy.
Project: "Translational aspects of the research for molecular anomalies in neoplasms, with potential applicable developments in the clinical, diagnostic and prognostic field".
- 2018 **Independent research collaborator**, ARC-Net research centre, Università degli studi di Verona, Verona, Italy.
Project: "Clinically Applicable biomarkers to early diagnosis, patient risk stratification and therapy response in pancreatic cancer".

- 2012–2018 **Post-doctoral fellow**, ARC-Net research centre, *Università degli studi di Verona, Verona, Italy.*
 Project: "The genome of exocrine and endocrine pancreatic tumors as the Italian State contribution to the International Cancer Genome Consortium".
- 2009–2011 **Post-doctoral fellow**, ARC-Net research centre, *Università degli studi di Verona, Verona, Italy.*
 Project: "Identification of autoantibodies as specific markers in pancreatic cancer".
- 2007–2008 **Post-doctoral fellow**, Department of Pathology, *Università degli studi di Verona, Verona, Italy.*
 Project: "Development of new reagents for the study of FAM3C protein in pancreatic cancer".
- 2004–2006 **Ph.D. Student**, Department of Surgical and Gastroenterological Sciences, *Università degli studi di Verona, Verona, Italy.*
 Project: "New proteomics approaches for the identification of potential markers of pancreatic neoplasia".
- 2003 **Research fellow**, Department of Pathology, *Università degli studi di Verona, Verona, Italy.*
 Project: cloning, production and purification of human recombinant proteins.

Scientific Expertise

- Technical Statistic and bioinformatic analysis of scientific, clinicopathological and genomic/transcriptomic/proteomic data. Molecular biology and biochemistry wet lab, including: molecular cloning, expression and analysis of DNA/RNA/proteins, Immunoassays, flow cytometry, Next-generation sequencing.
- Conceptual Design of scientific projects and assays, peer reviewer for the scientific journals *PLoS One, BMC Cancer, Cancer Cell International, Aging, Therapeutic Advances in Gastroenterology, Expert Review of Molecular Diagnostics Chemotherapy, and GENE*

Computer Skills

- Overview Since 2012 I have maintained and developed a pipeline for Next-generation sequencing data annotation at the ARC-Net applied research centre. The activity includes tutoring users of the pipeline and performing customized analyses.
- Platforms GNU/Linux, Windows, MacOS X.
- Bioinformatics R, Bioconductor, Torrent Suite/Ion Reporter, SAMtools, VCFtools, BEDtools, SNPeff, SNPsift, Ensembl VEP, Integrative Genomics Viewer, Medcalc, GraphPad PRISM, SoftGenetics NextGENe, CLC genomics workbench, GITools.
- Scientific BioEdit, SerialCloner, Cytoscape, gProfiler, DAVID, BioMART, NCBI and UCSC databases, Aperio ImageScope, FIJI-ImageJ, FACSDiva, FlowJo.
- Office/graphics Microsoft and Libre Office, EndNote and Zotero, Adobe Photoshop and GIMP, Adobe Illustrator and Inkscape.
- programming BASH / SED / AWK, HTML and L^AT_EX.
- networking ftp, ssh, NFS, VNC, rsync.

Awards

- 2016 Jeremy Jass Prize for Research Excellence in Pathology 2014, awarded for the publication "Targeted next-generation sequencing of cancer genes dissects the molecular profiles of intraductal papillary neoplasms of the pancreas", *J Pathol* 2014;233: 217–227, DOI:10.1002/path.4344.

Teaching

- 2019 Teaching: "Diagnostic methodologies for Biochemistry and Molecular Biology". Course: Bachelor's degree in Biomedical Laboratory Techniques.

Languages

Italian **Native speaker**

English **Intermediate-Advanced**

Scientific Production

Publications: 53

Impact factor total: 380.042 – average: 7.308

Google Scholar Citations: 2154 – average citations per publication: 40.6 – h-index: 21

Web of Science Citations: 1455 – average citations per publication: 33.1 – h-index: 18

Scopus Citations: 1607 – average citations per publication: 32.1 – h-index: 19

Recent Publications

1. Barresi V, Simbolo M, **Mafficini A**, Piredda ML, Caffo M, Cardali SM, Germanò A, Cingarlini S, Ghimienti C, Scarpa A. *Ultra-Mutation in IDH Wild-Type Glioblastomas of Patients Younger than 55 Years is Associated with Defective Mismatch Repair, Microsatellite Instability, and Giant Cell Enrichment*. *Cancers (Basel)*. 2019 Aug 30;11(9). pii: E1279. doi: 10.3390/cancers11091279. PMID: 31480372 **IF:5.326**
2. Simbolo M, Barbi S, Fassan M, **Mafficini A**, Ali G, Vicentini C, Sperandio N, Corbo V, Rusev B, Mastracci L, Grillo F, Pilotto S, Pelosi G, Pelliccioni S, Lawlor RT, Tortora G, Fontanini G, Volante M, Scarpa A, Bria E. *Gene Expression Profiling of Lung Atypical Carcinoids and Large Cell Neuroendocrine Carcinomas Identifies Three Transcriptomic Subtypes with Specific Genomic Alterations*. *J Thorac Oncol*. 2019 Sep;14(9):1651-1661. doi: 10.1016/j.jtho.2019.05.003. PMID: 31085341 **IF:12.460**
3. **Mafficini A**, Scarpa A. *Genetics and Epigenetics of Gastroenteropancreatic Neuroendocrine Neoplasms*. *Endocr Rev*. 2019 Apr 1;40(2):506-536. doi: 10.1210/er.2018-00160. PMID: 30657883 **IF:15.745**
4. Amato E, **Mafficini A (shared first authorship)**, Hirabayashi K, Lawlor RT, Fassan M, Vicentini C, Barbi S, Delfino P, Sikora K, Rusev B, Simbolo M, Esposito I, Antonello D, Pea A, Sereni E, Ballotta M, Maggino L, Marchegiani G, Ohike N, Wood LD, Salvia R, Klöppel G, Zamboni G, Scarpa A, Corbo V. *Molecular alterations associated with metastases of solid pseudopapillary neoplasms of the pancreas*. *J Pathol*. 2018 Oct 10. doi: 10.1002/path.5180. [Epub ahead of print] PMID: 30306561 **IF:6.253**
5. **Mafficini A**, Scarpa A. *Genomic landscape of pancreatic neuroendocrine tumours: the International Cancer Genome Consortium*. *J Endocrinol*. 2018 Mar;236(3):R161-R167. doi: 10.1530/JOE-17-0560. Epub 2018 Jan 10. Review. PMID: 29321190 **IF:4.012**
6. Luchini C, Pea A, Lionheart G, **Mafficini A**, Nottegar A, Veronese N, Chianchiano P, Brosens LA, Noë

- M, Offerhaus GJA, Yonescu R, Ning Y, Malleo G, Riva G, Piccoli P, Cataldo I, Capelli P, Zamboni G, Scarpa A, Wood LD. *Pancreatic undifferentiated carcinoma with osteoclast-like giant cells is genetically similar to, but clinically distinct from, conventional ductal adenocarcinoma*. J Pathol. 2017 Jul 19. doi: 10.1002/path.4941. [Epub ahead of print] PMID: 28722124 **IF:6.253**
7. Scarpa A, Mafficini A. *Non-coding regulatory variations: the dark matter of pancreatic cancer genomics*. Gut. 2017 Jun 28. pii: gutjnl-2017-314310. doi: 10.1136/gutjnl-2017-314310. [Epub ahead of print] PMID: 28659348 **IF:17.016**
 8. Vezzalini M, Mafficini A (shared first authorship), Tomasello L, Lorenzetto E, Moratti E, Fiorini Z, Holyoake TL, Pellicano F, Krampera M, Tecchio C, Yassin M, Al-Dewik N, Ismail MA, Al Sayab A, Monne M, Sorio C. *A new monoclonal antibody detects downregulation of protein tyrosine phosphatase receptor type γ in chronic myeloid leukemia patients*. J Hematol Oncol. 2017 Jun 21;10(1):129. doi:10.1186/s13045-017-0494-z. PMID: 28637510 **IF:7.333**
 9. Scarpa A, Chang DK, Nones K, Corbo V, Patch AM, Bailey P, Lawlor RT, Johns AL, Miller DK, Mafficini A, Rusev B, Scardoni M, Antonello D, Barbi S, Sikora KO, Cingarlini S, Vicentini C, et al. *Whole-genome landscape of pancreatic neuroendocrine tumours*. Nature. 2017 Mar 2;543(7643):65-71. doi:10.1038/nature21063. Pubmed PMID: 28199314 **IF:41.577**
 10. Simbolo M, Mafficini A (shared first authorship), Sikora KO, Fassan M, Barbi S, Corbo V, Mastracci L, Rusev B, Grillo F, Vicentini C, Ferrara R, Pilotto S, Davini F, Pelosi G, Lawlor RT, Chilos M, Tortora G, Bria E, Fontanini G, Volante M, Scarpa A. *Lung neuroendocrine tumours: deep sequencing of the four WHO histotypes reveals chromatin remodelling genes as major players and a prognostic role for TERT, RB1, MEN1 and KMT2D*. J Pathol. 2016 Nov 22. doi: 10.1002/path.4853. [Epub ahead of print] Pubmed PMID: 27873319 **IF:6.253**
 11. Mafficini A, Amato E, Cataldo I, Rusev BC, Bertoncello L, Corbo V, Simbolo M, Luchini C, Fassan M, Cantù C, Salvia R, Marchegiani G, Tortora G, Lawlor RT, Bassi C, Scarpa A. *Ampulla of Vater Carcinoma: Sequencing Analysis Identifies TP53 Status as a Novel Independent Prognostic Factor and Potentially Actionable ERBB, PI3K, and WNT Pathways Gene Mutations*. Ann Surg. 2016 Sep 8. [Epub ahead of print] Pubmed PMID: 27611608 **IF:9.203**
 12. Mafficini A, Simbolo M, Parisi A, Rusev B, Luchini C, Cataldo I, Piazzola E, Sperandio N, Turri G, Franchi M, Tortora G, Bovo C, Lawlor RT, Scarpa A. *BRCA somatic and germline mutation detection in paraffin embedded ovarian cancers by next-generation sequencing*. Oncotarget. 2016 Jan 12;7(2):1076-83. doi: 10.18632/oncotarget.6834. Pubmed PMID: 26745875 **IF:5.168**
 13. Sadanandam A, Wullschleger S, Lyssiotis CA, Grotzinger C, Barbi S, Bersani S, Korner J, Wafy I, Mafficini A, Lawlor RT, Simbolo M, Asara JM, Blaker H, Cantley LC, Wiedenmann B, Scarpa A, Hanahan D. *A cross-species analysis in pancreatic neuroendocrine tumors reveals molecular subtypes with distinctive clinical, metastatic, developmental, and metabolic characteristics*. Cancer Discov 2015 Oct 7, epub ahead of print. Pubmed PMID: 26446169 **IF:24.373**
 14. Simbolo M, Mafficini A (shared first authorship), Agostini M, Pedrazzani C, Bedin C, Urso ED, Nitti D, Turri G, Scardoni M, Fassan M, Scarpa A. *Next-generation sequencing for genetic testing of familial colorectal cancer syndromes*. Hered Cancer Clin Pract. 2015 Aug 21;13(1):18. Pubmed PMID: 26300997 **IF:2.027**
 15. Melotti P, Mafficini A (shared first authorship), Lebecque P, Ortombina M, Leal T, Pintani E, Pepermans X, Sorio C, Assael BM. *Impact of MIF gene promoter polymorphism on F508del cystic fibrosis patients*. PLoS One. 2014 Dec 12;9(12):e114274. Pubmed PMID: 25503271 **IF:2.766**
 16. Mafficini A, Amato E, Fassan M, Simbolo M, Antonello D, Vicentini C, Scardoni M, Bersani S, Gottardi M, Rusev B, Malpeli G, Corbo V, Barbi S, Sikora KO, Lawlor RT, Tortora G, Scarpa A. *Reporting tumor molecular heterogeneity in histopathological diagnosis*. PLoS One. 2014 Aug 15;9(8):e104979. Pubmed PMID: 25127237 **IF:2.766**

17. Amato E, Molin MD, **Mafficini A (shared first authorship)**, Yu J, Malleo G, Rusev B, Fassan M, Antonello D, Sadakari Y, Castelli P, Zamboni G, Maitra A, Salvia R, Hruban RH, Bassi C, Capelli P, Lawlor RT, Goggins M, Scarpa A. *Targeted next-generation sequencing of cancer genes dissects the molecular profiles of intraductal papillary neoplasms of the pancreas*. J Pathol. 2014 Jul;233(3):217-27. Pubmed PMID: 24604757 **IF:6.253**
18. Jiao Y, Pawlik TM, Anders RA, Selaru FM, Streppel MM, Lucas DJ, Niknafs N, Guthrie VB, Maitra A, Argani P, Offerhaus GJ, Roa JC, Roberts LR, Gores GJ, Popescu I, Alexandrescu ST, Dima S, Fassan M, Simbolo M, **Mafficini A**, Capelli P, Lawlor RT, Ruzzenente A, Guglielmi A, Tortora G, de Braud F, Scarpa A, Jarnagin W, Klimstra D, Karchin R, Velculescu VE, Hruban RH, Vogelstein B, Kinzler KW, Papadopoulos N, Wood LD. *Exome sequencing identifies frequent inactivating mutations in BAP1, ARID1A and PBRM1 in intrahepatic cholangiocarcinomas*. Nat Genet. 2013 Dec;45(12):1470-3. Pubmed PMID: 24185509 **IF:27.125**
19. Sorio C, **Mafficini A (shared first authorship)**, Furlan F, Barbi S, Bonora A, Brocco G, Blasi F, Talamini G, Bassi C, Scarpa A. *Elevated urinary levels of urokinase-type plasminogen activator receptor (uPAR) in pancreatic ductal adenocarcinoma identify a clinically high-risk group*. BMC Cancer. 2011 Oct 14;11:448. PubMed PMID: 21999221. **IF:3.288**
20. **Mafficini A**, Ortombina M, Sermet-Gaudelius I, Lebecque P, Leal T, Iansa P, Reyhler G, Dahan K, Pepermans X, Lenoir G, Leonard A, Sorio C, Assael B, Melotti P. *Impact of polymorphism of Multidrug Resistance-associated Protein 1 (ABCC1) gene on the severity of cystic fibrosis*. J Cyst Fibros. 2011 Jul;10(4):228-33. PubMed PMID: 21435954. **IF:3.858**

Verona, October 10th, 2019

Mafficini Andrea

