

BIOGRAPHICAL SKETCH

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NAME: Antonio Percesepe

POSITION TITLE: Professor and chief of Medical Genetics Unit

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable. Add/delete rows as necessary.)*

INSTITUTION AND LOCATION	DEGREE (if applicable)	Start Date MM/YYYY	Completion Date MM/YYYY	FIELD OF STUDY
High school – Pescara	60/60	1977	1982	Classical High School
Catholic University of Rome	110/110 cum laude	1982	1988	Medicine
Catholic University of Rome	50/50 cum laude	1988	1992	Medical Specialty in Gastroenterology and endoscopy
Depts. of Internal Medicine, University of Modena and Medical Genetics, University of Helsinki	Ph.D	1994	1998	Gastrointestinal Oncology
University of Florence	50/50 cum laude	2003	2007	Medical Specialty in Medical Genetics

A. Personal Statement

Born in Pescara, 1964, where I have attended classical studies before moving to Rome to study Medicine at the Catholic University. After the board Certification in Gastroenterology at the same University I have moved to Modena for my PhD in Gastrointestinal Oncology, spending two years in Helsinki in Albert de la Chapelle laboratories of Medical Genetics to study Lynch syndrome. Back in Modena in 1998, after a two-year postdoc, I have started my university career in 2000 as Assistant Professor in Medical Genetics, working also as a clinical geneticist at the University Hospital of Modena after the second Board certification in the Discipline. In 2008 I have become head of the Medical Unit at the University Hospital of Modena and in 2015 I have moved to Parma for the upgrade to Associate Professor and Director of the Hospital Unit.

Over the past three decades, my research activities have been largely devoted to the study of the genotype/phenotype correlations in genetic disorders, including cancer predisposition syndromes like Lynch syndrome, Neurofibromatosis type I, rare cancers. With the study of the basic mechanisms of disease my research has been aimed to enable early patient diagnosis, risk stratifications and genetic counseling for patients and their families. I divide my daily activity between performing clinical diagnosis of genetic diseases as Chief of the Genetic Unit of the University Hospital of Parma and leading a research group together with one assistant professor with several PhD students and graduate students. Outside the research and clinical activities, I am a passionate Professor of Medical Genetics and President of the Course of Medicine and Surgery (in English) of the University of Parma.

I will oversee the recruitment of the eligible patients such as those affected by Neurofibromatosis I with plexiform neurofibromas which undergo surgery due to the ongoing neoplastic transformation to MPNST. NF1 is also predisposing to other rare tumors, which will be also recruited for the project. My Unit is member of the ERN GENTURIS (European Reference Network on Genetic Tumour Risk Syndromes) for Neurofibromatosis for which about 150 patients per year are under follow-up.

B. Positions, Scientific Appointments and Honors

2000: Winner of the Petzcoller travel award for the American Association of Cancer Research
 2018-2021: Regional (Emilia Romagna) Coordinator of the Italian Society of Human Genetics
 July 2016 to December 2017: Member of the Parma Ethics Committee for Genetics
 March 2021 - : Member of Ethics Committee of North Emilia-Romagna as Expert in Genetics
 Jan 2022 -: Representative of the Health Care Provider of the University Hospital of Parma for the European Reference Network for rare disease GENTURIS (Genetic Tumor Risk)
 Jan 2024 - : President of the Course of Medicine and Surgery of the University of Parma
 March 2024 - : Member of the Emilia Romagna Regional Molecular Tumor Board

C. Contributions to Science

Establishing a population-based incidence of an important disease like Lynch syndrome;
 Contributing to the identification of three new disease/genes;
 Refining the genotype/phenotype correlation in several human disorders like Lynch syndrome, limb anomalies, Neurofibromatosis I.

Specifically:

1. Clinical and biomolecular aspects of Lynch syndrome, in particular I contributed to the characterization of low-degree microsatellite instability (MSI) and to the early phases of this mechanism, by describing the MSI pattern of early adenomas (Oncogene, 1998) and of carcinomas, thus showing that MSI in the early phases can be interpreted as a tumor clock dating the starting point of the phenomenon. Moreover, in a population-based study of all the incident cases of colon cancer, he has established the frequency of Lynch syndrome in a high incidence area for colon cancer, setting the frequency in 0.3% of the total colorectal cancer burden (Journal of Clinical Oncology, 2001)
2. Skeletal dysplasias and upper limb malformations and related genes, in particular by contributing to the characterization of the genetic basis of a type IX syndactyly in collaboration with a group from Germany (American Journal of Human Genetics, 2014) and by describing a large series of VACTERL Association, which were ascertained by their upper limb anomaly (Journal of Pediatrics, 2014)
3. Genotype/phenotype correlation in Neurofibromatosis I, which were described in detail in a recent article published in Genes Chromosome and Cancer (2022)

D. Professional Experience

1988-1994	Resident at the Department of Internal Medicine and Gastroenterology of the Catholic University of Rome, Italy
1995	Resident at the Department of Internal Medicine of the University of Modena, Italy
1996-1997	Molecular biologist at the Dept. of Medical Genetics of the University of Helsinki, Finland
1998-2000	Postdoctoral fellow at the Dept. of Internal Medicine of the University of Modena
2000-2013	Assistant Professor at the Dept. of Medical Genetics of the University of Modena
2008-2015	Head of Medical Genetic Unit, University Hospital of Modena
11/2015	Director of the Unit of Medical Genetics at the Azienda Ospedaliero-Universitaria di Parma
12/2017	Associate Professor SSD MED/03 (Medical Genetics) at the University of Parma
01/2022	Chief representative of the University Hospital of Parma for the European Reference Network Genturis (Genetic Tumor Risk)
01/2024	President of the Course of Medicine and Surgery of the University of Parma
03/2024	Member of the Emilia Romagna Regional Molecular Tumor Board

E. Research activities

Author of 118 original articles <https://pubmed.ncbi.nlm.nih.gov/?term=percesepe+a&sort=pubdate&size=200>

Total H-INDEX: 35;
Total number of citations: 4252

Research activities have been developed along the following fields:

1. Genotype/phenotype correlation in Neurofibromatosis I
2. Skeletal dysplasias and upper limb malformations and related genes
3. Clinical and biomolecular studies of hereditary syndromes (current focus)
4. Non invasive prenatal diagnosis
5. Chromosomal syndromology
6. Clinical and biomolecular aspects of Hereditary Nonpolyposis Colorectal Cancer, HNPCC
7. Molecular studies of the mismatch repair genes and microsatellite instability

Grants of the last 15 years: Funding agency	Years	Title	Position	Amount (€)
Regione Emilia-Romagna. Research Program Region-University	2010-2012	Next-generation sequencing and gene therapy to diagnose and cure rare diseases in Regione Emilia Romagna (RER)	Partner/ Unit Coordinator	50.000
Associazione Emma ed Ernesto Rulfo per la Genetica Medica	2016-2022	Exome sequencing in intellectual disability	Coordinator	60.000
Rostock University International Parkinson's disease	2020-2024	<i>ROPAD: Rostock University International Parkinson's disease study (Co P.I.) (2020)</i>	Unit Co-PI	10.000
University of Parma	2022-2024	<i>ADVANCE Accelerated Diagnosis and non-inVasive strategies for AN early prediCtion of malignant transformation in hEreditary cancer predisposition syndrome: from NF1-associated benign precursor lesion to Malignant Peripheral Nerve Sheath Tumor (MPNST).</i>	- PI	72.000

PRIN-PNRR	2023-2025	<i>In-depth genetic study and clinical follow-up in fetuses with I and II trimester unspecific signs of increased risk to predict fetal prognosis and inform parental decision-making.</i>	Unit PI and Substitute PI of the Project	40.000
PNRR – rare tumors	2024-2026	<i>Liquid biopsy implementation in the management of rare tumors associated with hereditary syndromes</i>	Project PI	1.000.000