PERSONAL INFORMATION

Enrico Pierantozzi

University of Siena, Siena, Italy

Department of Developmental and Molecular Medicine
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Sex Male | *Date of birth* 21/09/1975 | *Nationality* Italian

Enterprise	University	EPR
☐ Management Level	☐ Full professor	☐ Research Director and 1st level Technologist / First Researcher and 2nd level Technologist / Principal Investigator
☐ Mid-Management Level	☒ Associate Professor	☐ Level III Researcher and Technologist
☐ Employee / worker level	☐ Researcher and Technologist of IV, V, VI and VII level / Technical collaborator	☐ Researcher and Technologist of IV, V, VI and VII level / Technical collaborator

WORK EXPERIENCE

2021, Dec – to date

Associate Professor, Human Histology and Embryology

University of Siena, Department of Developmental and Molecular Medicine

- Histology and Embryology teaching
- Main topics of research activity: 1) role of obscurin and sAnk1.5 proteins in the structure, function and metabolism of skeletal muscle fibers; 2) Molecular mechanisms involved in triaddevelopment and organization; 3) study of the structural, contractile and molecular alterations induced by mutations in skeletal muscle specific proteins that are causative for diseases with skeletal muscle aetiology such as MH and CCD (RYR1), and TAM (CASQ1),

Business or sector Cell Biology, Histology

2018, Dec – 2021, Nov

Senior Researcher

University of Siena, Department of Developmental and Molecular Medicine

- Histology and Embryology teaching
- Main topic of research activity: role of Obscurin, small-Ankyrin1.5, Junctophilin1, Calsequestrin1, RyR1 proteins in the structure, function and metabolism of skeletal muscle fibers

Business or sector : Cell Biology, Histology

2017, Mar – 2018, Nov

Assistant Researcher

University of Siena, Department of Developmental and Molecular Medicine

- Histology and Embryology teaching
- Main topic of research activity: morphological and structural alterations in human congenital myopathies and cardiomyopathies, using patients biopsies, cell lines and genetically modified mice as model systems.

Business or sector: Cell Biology, Histology

2023, Oct – 2025, Sep

Research Unit Responsible

University of Siena, Department of Developmental and Molecular Medicine

- Scientific Head of the Siena Unit within the BANDO PRIN 2022 project " MiR-486 as a novel therapeutic target for RYR1-related core myopathies ".

Business or sector Cell Biology, Histology

2014, Mar – 2017, Feb

Research Unit Responsible

University of Siena, Department of Developmental and Molecular Medicine

- Scientific Head of the Siena Unit within the FIR 2013 project "Structural and functional alterations in Central Core Disease (CCD): understanding of molecular mechanisms and genetic bases aimed at the development of therapeutic strategies".

Business or sector Cell Biology, Histology

2011, Jan – 2014, Feb

Assistant Researcher

University of Siena, Department of Neurosciences

- Main topics of research activity: 1) histological and molecular characterization of skeletal muscle fibers of either sAnk1KO and obscurinKO mice. 2) characterization of proliferative and differentiation potential of human multipotent pericytes isolated from skeletal and smooth muscle tissues, and from adipose tissue.

Business or sector Cell Biology, Histology, Stem Cell Biology

2006, Jul – 2010, Dec

Post-Doc position

University of Siena, Department of Neurosciences

- Main topic of research activity: isolation and characterization of multipotent stem cells from myocardium, adipose tissue, smooth and skeletal muscle tissue.

Business or sector Cell Biology, Histology, Stem Cell Biology

EDUCATION AND TRAINING

2006

PhD, Medical Embriology

University of Tor Vergata, Rome, Italy

2002

Master's degree in Biological Sciences

University of La Sapienza, Rome, Italy

WORK ACTIVITIES

Awards

Editorial activity

Invited presentations

2023, “20th IIM meeting” (Assisi, Italy)
2018, “37th annual meeting of the European Malignant Hyperthermia Group" (Ferrara, Italy);
2017, “2017 Spring Padua Muscle Days” (Padova, taly)

Grants

Patents

ADDITIONAL INFORMATION

Publications
Updated 2025, January

total number of publications in peer-review journals: 33
total number of citations: 932
H index: 19

Selected Papers

1. Pioner et al. (2025). Obscurin deficiency leads to compensated dilated cardiomyopathy and increased arrhythmias. J Gen Phys. Volume 157, Issue 4. IF 3,30.
2. Serano et al. (2025). Intracellular Membrane Contact Sites in Skeletal Muscle Cells. Membranes. 15(1), 29. IF 3,30.
3. Gamberucci et al. (2024). TAM-associated CASQ1 mutants diminish intracellular Ca2+ content and interfere with regulation of SOCE. J Muscle Res Cell Motil. Epub ahead of print. IF 1,81
4. Pierantozzi et al. (2023). Skeletal muscle overexpression of sAnk1.5 in transgenic mice does not predispose to type 2 diabetes. Sci Rep, 13(1):8195. IF 4,99
5. Rossi D, Lorenzini S, Pierantozzi E, Van Petegem F, Osamwonuyi Amadsun D, Sorrentino V. (2022). Multiple regions within junctin drive its interaction with calsequestrin-1 and its localization to triads in skeletal muscle. J Cell Sci 135:jcs259185. IF 5,28
6. Pierantozzi E, Szentesi P, Paolini C, Dienes B, Fodor J, Oláh T, Colombini B, Rassier DE. Rubino EM, Lange S, Rossi D, Csernoch L. Impaired Intracellular Ca2+ Dynamics, M-Band and Sarcomere Fragility in Skeletal Muscles of Obscurin KO Mice. Int J Mol Sci 23:1319. IF 5,923
7. Rossi D, Gigli L, Gamberucci A, Bordoni R, Pietrelli A, Lorenzini S, Pierantozzi E, Peretto G, De Bellis G, Della Bella P, Ferrari M, Sorrentino V, Benedetti S, Sala S, Di Resta C. (2020). A novel homozygous mutation in the TRDN gene causes a severe form of pediatric malignant ventricular arrhythmia. Heart Rhytm, 17:296-304. IF 5,225
8. Rossi D, Scarcella AM, Liguori E, Lorenzini S, Pierantozzi E, Kutchukian C, Jacquemond V, Messa M, De Camilli P, Sorrentino V. (2019). Molecular determinants of homo- and heteromeric interactions of Junctophilin-1 at triads in adult skeletal muscle fibers. PNAS, 116:15716-15724. IF 9,58
9. Pierantozzi E, Szentesi P, Al-Gaadi D, Oláh T, Dienes B, Sztretye M, Rossi D, Sorrentino V, Csernoch L. (2019). Calcium Homeostasis Is Modified in Skeletal Muscle Fibers of Small Ankyrin1 Knockout Mice. Int J Mol Sci, 20:1-13. IF 4,60
10. Barone V, Del Re V, Gamberucci A, Polverino V, Galli L, Rossi D, Costanzi E, Toniolo L, Berti G, Malandrini A, Ricci G, Siciliano G, Vattemi G, Tomelleri G, Pierantozzi E, Spinozzi S, Volpi N, Fulceri R, Battistutta R, Reggiani C, Sorrentino V. (2017) Identification and characterization of three novel mutations in the CASQ1 gene in four patients with tubular aggregate myopathy. Human Mutation, 38:1761-1773 IF 5,49
11. Randazzo D, Blaauw B, Paolini C, Pierantozzi E, Spinozzi S, Lange S, Chen J, Protasi F, Reggiani C, Sorrentino V. (2017) Exercise-induced alterations and loss of sarcomeric M-line organization in the diaphragm muscle of obscurin knockout mice. Am J Physiol Cell Physiol, 312: c16-c28 IF 3,59
12. Pierantozzi E, Vezzani B, Badin M, Curina C, Severi FM, Petraglia F, Randazzo D, Rossi D, Sorrentino V. (2016) Tissue-Specific Cultured Human Pericytes: Perivascular Cells from Smooth Muscle Tissue Have Restricted Mesodermal Differentiation Ability. Stem Cells Dev. 25:674-686 IF 4,01
13. Pierantozzi E, Badin M, Vezzani B, Curina C, Randazzo D, Petraglia F, Rossi D, Sorrentino V. (2015) Human pericytes isolated from adipose tissue have better differentiation abilities than their mesenchymal stem cell counterparts. Cell Tiss Res, 361: 769-778 IF 3,56

14. Rossi D, Vezzani B, Galli L, Paolini C, Toniolo L, Pierantozzi E, Spinozzi S, Barone V, Pegoraro E, Bello L, Cenacchi G, Vattemi G, Tomelleri G, Ricci G, Siciliano G, Protasi F, Reggiani C, Sorrentino V. (2014) A mutation in the CASQ1 gene causes a vacuolar myopathy with accumulation of sarcoplasmic reticulum protein aggregates. Human Mutation, 35: 1163-1170 IF 6,15
15. Randazzo D, Giacomello E, Lorenzini S, Rossi D, Pierantozzi E, Blaauw B, Reggiani C, Lange S, Peter AK, Chen J, Sorrentino V. (2013) Obscurin is required for ankyrinB-dependent dystrophin localization and sarcolemma integrity. J Cell Biol, 200: 523-536 IF 9,68
16. Pierantozzi E, Gava B, Manini I, Roviello F, Marotta G, Chiavarelli M, Sorrentino V. (2011) Pluripotency regulators in human mesenchymal stem cells: expression of NANOG but not of OCT-4 and SOX-2. Stem Cell Dev, 20:915-923 IF 5,17