FUNDING APPLICATION

C.1 CURRICULUM VITAE

Personal information

Name, Surname:	Severin, Emilia		
Date of birth:	23.05.1954	Gender:	Female
Nationality:	Romanian		
Researcher unique identifier(s) (ORCID, Researcher ID etc.):	Web of Science ResearcherID: AAI-4341-2020 https://orcid.org/0000-0003-3901-615X ResearcherID: I-1692-2013 Scopus Author ID: 17535233100		
URL for personal website (if case):	U-1700-033G-3237		

Education

1977	Biology/Genetics – Bucharest University - Romania
1978	Master in Ecology
1994 (dissertation defended)	Ph.D. Genetic Approaches in Collagen Disorders
1994 - 1995	Fellowship: Mutagenesis (micronuclei, SCE) - Universita degli studi di Padova, Italy
1997	Fellowship: Molecular technique (PCR) - Consiglio Nazionale delle Ricerche - Milano, Italy

Positions - current and previous

(*Academic sector/research institutes/industrial sector/public sector/other)*

Year	Job title – Employer - Country
2022	Senior researcher at Academy of Medical Sciences
1999	Professor – Carol Davila University of Medicine and Pharmacy – Bucharest, Romania
1982-2024	All academic ranks from assistant to full professor

Career breaks (if case)

Year	Reason
yyyy-yyyy	N/A

Project management experience

(*Academic sector/research institutes/industrial sector/public sector/other. Please list the most relevant.)*

Year	Project title - Role – Funder – Budget – link to project webpage
2004-2006	Molecular Investigations of Pax9 gene mutations involved in tooth agenesis – research project coordinator – VIASAN 390 – 452,000 RON

2008-2011	Deafness in children - clinical and molecular diagnosis (CONEXIN) PNCDI 1 – 42-143, project coordinator – ANCS – 1,784,739.00 RON – http://www.conexin.ro
2015-2018	INNOVCare (Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions) -partner leader – EC – 1,994,414.72 Euro - https://innovcare.eu/wp-content/uploads/2017/06/Update-on-INNOVCare_Raquel-Castro.pdf
2018-2021	Rare2030 foresight study – partner leader - co-funded by the European Union Pilot Projects and Preparatory Actions Programme (2014- 2020), as pilot project PP-1-2-2018-Rare 2030 https://download2.eurordis.org/rare2030/poe/Membership%20List%20Sub-group%201.pdf
2004-2006	Project" "Testing the mutagenic effect of Paracetamol on a laboratory animal model" - scientific manager, VIASAN 390, link to the webpage
1994-1995	PRIN - Studio dell'attività mutagena e clastogenica di potenziali farmaci antitumorali e di agenti fotoattivi per la fotochemioterapia – University of Padua, Italy, Partner leader, link to the webpage

Other relevant professional experiences

(e.g. institutional responsibilities, organisation of scientific meetings, membership in academic societies, review boards, advisory boards, committees and major research or innovation collaborations, other commissions of trust in public or private sector)

Year	Description - Role
2002 – present time	Mentor for PhD training programme Carol Davila University of Medicine and Pharmacy, Bucharest
2013 - 2018	Member of European Commission Expert Group on Rare Diseases
2010 – present time	Vice-president of National Counsel for Rare Diseases, Romanian Ministry of Health
2013 – present time	Member of Medical Genetics Commission, Romanian MoH
2015 – 2016	Project evaluation expert: 2nd ERANet-LAC Transnational Joint Call for Research and Innovation projects on the thematic area "HEALTH (2016). RARE-Best practices Project (2015, 2016).
2015 – present time	Peer Reviewer: Journal of Cellular and Molecular Medicine, British Medical Journal Case Studies, Journal of Dental Research, Journal of Pediatric Genetics, Clinical Case Reports, Frontiers Journals, MDPI Journals.
2016 – present time	DNA DAY Essay Contest (ASHG) Judge

C.2 Track record of the last 10 years

1. Țiburcă, L.; Zaha, D.C.; Jurca, M.C.; **Severin, E**.; Jurca, A.; Jurca, A.D., ***The Role of Aminopeptidase ERAP1 in Human Pathology—A Review***, Curr. Issues Mol. Biol. 2024, 46, 1651-1667. https://doi.org/10.3390/cimb46030107

2. Jurca CM, Kozma K, Petchesi CD, Zaha DC, Magyar I, Munteanu M, Faur L, Jurca A, Bembea D, **Severin E**, Jurca AD, ***Tuberous Sclerosis, Type II Diabetes Mellitus and the PI3K/AKT/mTOR Signaling Pathways-Case Report and Literature Review***, Genes (Basel), 2023, Feb 8;14(2):433, 2023, doi: 10.3390/genes14020433. PMID: 36833359; PMCID: PMC9957184, ISSN: 2073-4425

3. Lupu M, Ioghen M, Perjoc RȘ, Scarlat AM, Vladâcenco OA, Roza E, Epure DA, Teleanu RI, **Severin EM**, ***The Importance of Implementing a Transition Strategy for Patients with Muscular Dystrophy: From Child to Adult-Insights from a Tertiary Centre for Rare Neurological Diseases***, Children (Basel). 2023

May 28;10(6):959. doi: 10.3390/children10060959. PMID: 37371191; PMCID: PMC10296957, 2023, doi: 10.3390/children10060959. PMID: 37371191; PMCID: PMC10296957, ISSN: 2227-9067

4. Magdalena Budisteanu, Florentina Linca, Lucia Emanuela Andrei, Laura Mateescu, Adelina Glangher, Doina Ioana, **Emilia Severin**, Sorin Riga and Florina Rad, ***Recognition of Early Warning Signs and Symptoms - the First Steps on the Road to Autism Spectrum Disorder Diagnosis***, Annali dell'Istituto Superiore di Sanitathis link is disabled, 2022, 58(3), pp. 183–191, 2022, Doi: 10.4415/ANN_22_03_07

5. Maria Claudia jurcă, Sânziana Iulia Jurcă, Filip Mirodot, Bogdan Bercea, **Emilia Maria Severin**, Marius Bembea, Alexandru Daniel Jurcă, ***Changes in skeletal dysplasia nosology***, Rom J Morphol Embryol. 2021 Jul-Sep; 62(3): 689–696, Published online 2022 Feb 24, 2021, doi: 10.47162/RJME.62.3.05

6. Budisteanu M, Papuc SM,Streata I, Cucu M, Pirvu A, Serban-Sosoi S, Erbescu A, Andrei E, Iliescu C, Ioana D, **Emilia Severin** , Ioana M and Arghir A., ***The Phenotypic Spectrum of 15q13.3 Region Duplications: Report of 5 Patients***, Genes. 2021; 12(7):1025 2021, https://doi.org/10.3390/genes12071025

7. Vesa CM, **Severin E**, Bembea M, Amorin Remus Popa AR, Sklerniacof A, Jurca C, Kozma K, Daina C, Jurca A, Petchesi C , Galusca D, Jurca S , Zaha DC, ***Congenital diaphragmatic hernia: a retrospective study regarding the clinical experience of Bihor County Genetics Center***, Rom Biotechnol Lett. 2021; 26(3): 2671-2678, 2021, doi: 10.25083/rbl/26.3/2671-2678

8. **Severin E**., De Santis M., Ferrelli RM., Taruscio D., ***Health systems sustainability in the framework of rare diseases actions. Actions on educational programmes and training for professionals and patients***, Ann Ist Super Sanita, Jul-Sep;55(3):265- 269. 2019, doi: 10.4415/ANN_19_03_12.

9. **Emilia Severin**, ***Rare diseases in Romania – a response to 'transposition and implementation of EU rare diseases policy*** in Eastern Europe'Expert Review of Pharmacoeconomics & Outcomes Research, 2018, DOI: 10.1080/14737167

10. Ionescu Adriana Maria, Dragomir Cristina, **Severin Emilia**, Schipor Sorina, Savu Lorand, ***The Noninvasive Fetal RHD Genotyping from Cell-Free Fetal DNA Circulating in Maternal Blood: A Feasible Tool in Clinical Practice of Mother-Fetus Rh Incompatibility in Romania***, Romanian Biotechnological Letters, Vol. 21, No. 4: 8152-59, 2016

C.3 Narrative CV

I have served as a Professor of Genetics at the "Carol Davila" University of Medicine and Pharmacy in Bucharest, Romania, for over three decades. During this time, I have cultivated a strong reputation within the academic community for my significant contributions to medical genetics education. In March 2018, I was honoured to receive the Diploma of Excellence from the Carol Davila University of Medicine for my exceptional contribution to the advancement of genetics and genomics. Starting from 2019 I am senior researcher at Academy of Medical Sciences, in Bucharest Romania.
As a researcher, the Executive Agency for Higher Education, Research, and Innovation Funding (UEFISCDI) awarded me the Diploma for Competence and Scientific Attire in the Evaluation Activity of Research in Higher Education in 2001. This diploma was granted by the Executive Unit for Financing Higher Education, Research, Development, and Innovation, under the Ministry of National Education.

As a senior member of the Romanian National Alliance for Rare Diseases (RONARD), I have played a collaborative role in co-authoring the Romanian National Plan for Rare Diseases, an integral component of the Romanian National Health Strategy.
Additionally, my involvement as a member of the European Commission for Rare Diseases (EUCERD) has led to my participation in the formulation of recommendations for cross-border genetic testing of rare diseases within the EU. Between 2014 and 2018, I undertook the responsibility of elaborating the annual reports on the State of the Art of Rare Diseases in Romania, highlighting activities within EU Member States and other European countries.
I am also honoured to be affiliated with esteemed scientific societies and professional organizations, including the American Society of Human Genetics and the American College of Medical Genetics and Genomics, where I hold the status of Correspondence Member).
I coordinated **6 national/international research projects and 12 clinical studies** in the fields of genetics, molecular genetics, and genomics, computational molecular evolution, viral genomics and bioinformatics, human immunology – genes and environment. In my research activities I coordinated the development of new multilayer approaches combining large data sets and high-throughput information at genomic, transcriptomics, methylomics, proteomics, metabolomics, and phenome level, for investigation and personalized treatment of rare monogenic diseases and common complex diseases. Together with my team I was focused on identification and characterization of genes, pathways, and molecular mechanisms converting human health to a disease, utilizing new and state-of-the-art computational, bioinformatics, and molecular genetics and genomics approaches in an integrative way. The results of my research activities were presented in 10 monographs, 8 courses tailored for medical students, and 23 book chapters featured in publications both within Romania and internationally. In 2019, the collective volume "Medical Genetics - Ed. Polirom" was honored with the "C.I. Parhon" Prize, bestowed by the Romanian Academy.
I also participated as a team member in other 18 projects in the fields of microbiology, pathogen surveillance, fungal pathogens, antimicrobial resistance, mitochondrial medicine, CRISPR gene editing.
In this project I will participate into the following tasks: T3.3 (Monitoring elderly or people with disabilities), T3.6 (Development of models and methods for validation in real environment), T4.1 (Design and fabrication of microsensors for use in multi-well microfluidic bioreactors), T4.2 (Development of a standardized multi-organ multi-well plate for modelling the gut and its microbiome), T4.4 (Data analysis for diagnosis and personalised treatment), T5.3. (Organ of chip platform – hardware and AI integration), T6.2 (New ideas and ground-breaking projects), 6.3 (Implementation of new projects), T7.1, T7.2, T7.3 (dissemination, communication and exploitation), T8.1, T8.2, T8.3 (management and Ethics), T1.1 – T1.4 (setting up and management of the Centre of Excellence).

Note: For each nominated person, please present the CV (uploaded as a single document of maximum 6 pages, saved with the name of the member, A4 format, Times New Roman font, 11-point font size, 1.15 line spacing and 2 cm margins).