PERSONAL INFORMATION

Simina Iulia-Elena (formerly Jurca-Simina)

simina.iulia@umft.ro

https://orcid.org/0000-0001-7768-2968
Scopus Author ID: 56388779200

OCCUPATIONAL FIELD

Academic and medical clinical genetics, pediatrics

PROFESSIONAL EXPERIENCE

Assistant Professor in Medical Genetics

03.10.2022- present — "Victor Babeș" University of Medicine and Pharmacy, Eftimie Murgu Square no. 2, Timișoara

Teaching in Romanian, English and French sections and research activities

10.2021- 06.2022
09.2016- 01.2020 — Associate Assistant Professor

Medical Genetics Specialist

01.04.2025- present — Timis Regional Center for Medical Genetics, part of the "Louis Țurcanu" Children's Emergency Clinical Hospital Timișoara, part of the European Reference Network for Rare Diseases ERN ITHACA

02.2023- 10.2025 — Oncomed Outpatient Clinic, Oncohelp Association, Str. C. Porumbescu No. 57-59, Timişoara

01.04.2021- present — "DR BABEU" Civil Medical Society, Timisoara, Liviu Rebreanu Blvd. no. 190

04.2025- present — Banatul Clinics - Regina Maria, Calea Martirilor 1989 1, Timișoara

Medical genetics and oncogenetic consultations

Pediatrics specialist

01.2022-30.03.2025 — “Louis Țurcanu” Children's Emergency Clinical Hospital, 2 Iosif Nemoianu Street, Timișoara

03.2021- 31.12.2021 — Administrative-territorial unit of Timisoara Municipality, 1 Bv. CD Loga Street, Timis County

01.04.2021- present — "DR BABEU" Civil Medical Society, Timisoara, Liviu Rebreanu Blvd. no. 190

01.2018- present — Banatul Polyclinic - Regina Maria, Calea Martirilor 1989 1, Timișoara

Type or sector of activity

Coordinator physician of the Pediatric Vaccination Center + on-call duty in the Emergency Department; Vaccination doctor - Incuboxx Vaccination Center, coordinating doctor of the Vaccination Center from 02.08.2021 to 31.12.2021; Outpatient, physical and telemedicine pediatric consultations

Resident physician Medical Genetics

University of Medicine and Pharmacy Oradea, "Dr. Gavril Curteanu" Municipal Hospital Oradea

01.2018- 08.2020 — Posted to: "Victor Babeș" University of Medicine and Pharmacy, Eftimie Murgu Square no. 2, Timișoara

“Louis Țurcanu” Children's Emergency Clinical Hospital, 2 Iosif Nemoianu Street, Timișoara

Type or sector of activity

Internships completed and sectors of activity: medical genetics, clinical genetics, cytogenetics, molecular genetics, informatics and biostatistics, obstetrics and gynecology, internal medicine, neurology, endocrinology + 1 year, 5 months and 2 weeks of internships considered from the first specialization in pediatrics

Resident physician Pediatrics

01.2012- 07.2017 "Louis Țurcanu" Children's Emergency Clinical Hospital, 2 Iosif Nemoianu Street, Timișoara

Type or sector of activity

Internships completed and sectors of activity: general pediatrics, pediatric nephrology, pediatric gastroenterology, pediatric cardiology, pediatric pulmonology-phthisiology, infectious diseases, pediatric surgery and orthopedics, toxicology, pediatric diabetes and nutrition, pediatric neurology, pediatric psychiatry, pediatric onco-hematology, pediatric emergencies, pediatric intensive care, neonatology, genetics, general ultrasound, research ethics.

Associate Practitioner in Pediatrics

09.2014-01.2016 "Raymond Poincaré" Hospital, Assistance Publique - Hôpitaux de Paris

Medical Genetics Department, Coordinator Professor Dominique P. GERMAIN

104, boulevard Raymond Poincaré, 92380 GARCHES, France

Type or sector of activity

Clinical genetics, research activities, participation in clinical trials

Volunteer at Save the Children - Timis Branch

10.2007-09.2014 Save the Children Organization, Timis Branch, Republicii Boulevard no. 1, Corp B, Ap 7, Timişoara, Timiș

Type or sector of activity

Organization and development of educational programs for health and prevention, as well as children's rights (participant and trainer)

EDUCATION AND TRAINING

2022-2023 **Postgraduate studies - Psychopedagogical Training Program Level I and II**

Department for Teaching Staff Training at the West University of Timişoara

10.2012- 24.10.2019 **Doctor of Medicine**

"Victor Babeş" University of Medicine and Pharmacy Timişoara

Confirmed by Order of the Ministry of Health OM-5748_13_10_2020

Thesis: Evaluation strategies of genetic pathology correlated with technological and information technology progress

Coordinator: Prof. Dr. Puiu Maria

Medical genetics

Thesis summary link: http://www.umft.ro/2019_789

Medical doctor

2005-2011 "Victor Babeş" University of Medicine and Pharmacy Timişoara

Faculty of General Medicine

2001-2005 **Bachelors**

"Horia, Cloșca and Crișan" High School in Abrud

Vocational-pedagogical profile; teacher-educator

PERSONAL COMPETENCES

Mother tongue: Romanian language

Other known foreign languages	UNDERSTANDING		SPEAKING		WRITING
	Obedience	Reading	Join the conversation	Oral speech	
English language	C1	C1	C1	C1	C1
French	C1	C1	C1	C1	C1

Levels: A1/A2: Elementary user - B1/B2: Independent user - C1/C2: Experienced user
Common European Framework of Reference for Languages

Communication skills

- Ease and openness in establishing interpersonal relationships and ability to establish good contacts with patients and their families, with specialists from various medical fields.
- Adherent to team spirit at work.

Organizational/managerial skills

Organizational culture and ability to write and coordinate projects adapted to NGOs working in the health field. Active involvement in educational projects, their coordination and organization of various extracurricular activities, direct support for patients, information and campaigns on rare diseases and training for students in various medical areas.

Digital competence

SELF				
Information processing	Communication	Content creation	Security	Problem solving
Experienced user	Experienced user	Independent user	Experienced user	Experienced user

Levels: Basic user - Independent user - Experienced user
Digital skills - Self-assessment grid

Other computer skills: a good command of the office suite (word processor, spreadsheet, presentation software), knowledge acquired in processing the bachelor's thesis, editing submitted posters and oral presentations, direct contribution to the writing of medical publications; good knowledge of photo editing, acquired as an amateur photographer.

Other skills

The multiple volunteer activities carried out during my educational trajectory have contributed to my formation as an active, ambitious personality, with a spirit of initiative, capable of involvement in demanding work and assuming a responsible role in the activities undertaken, interested in communication and child and adolescent psychology. The essential themes of the research carried out are in the area of genetics and pediatrics.

Driving license

Category B since 2004

ADDITIONAL INFORMATION

Research projects/clinical studies

1. **Development of genomic research in Romania, acronym: ROGEN,** Project code 324809, 17.12.2024 - 17.12.2029 - **member**
2. **MEDI.COM-RARE PROJECT** - Expanding medical and community services for people affected by genetic and rare diseases. AR 19076/26.10.2022 Call: Improving access to health services especially for vulnerable groups, including Roma. Program: Challenges in public health at European level, Main source of funding: Financial Mechanism of the European Economic Area 2014-2021 (26.10.2022 – 30.04.2024)- **member**
3. **PIR16183: A Prospective and Retrospective Cohort Study to Refine and Expand the Knowledge on Patients With Chronic Forms of Acid Sphingomyelinase Deficiency (ASMD)** , NCT04106544, Investigational Site Number 6420001, Timisoara, Romania, 300011, Study Director: Clinical Sciences & Operations Sanofi, Study Start Date: September 27, 2019, Estimated Primary Completion Date: March 2023- ***co-investigator*** between ***18.02.2020- 09.2023***
4. **Project Science and Technology in childhood Obesity Policy (STOP)** , Grant Agreement number 774548; Call: Horizon 2020-SFS-2016-2017; Topic: SFS-39-2017, Research Innovation Action 2018-2020. WP 8: T8.2 - Three-site RCT of an early childhood obesity intervention: design and tool development. Translation of material for intervention, Victor Babes University of Medicine and Pharmacy Timisoara, Romania – ***active member 31.08.2018- 2022***
5. **Professional training of medical personnel in medical GENetics – PROGEN (PROfessional training of medical personnel in medical GENetics)** Project financed through the Social European Fund through the Operational Program Human Capital 2014-2020. Priority axis 4- Social inclusion and poverty combat. Specific objective 4.8- Improvement of the competence level of professionals' in the medical field. SMIS107623; POCU contract: 91/4/8/107623/08.12.2017 (12.2017-12.2019) - ***trainer 2017-2019***
6. **Competitiveness Operational Program 2014-2020; priority axis 1 – Research, technological development and innovation (RD&I) to support economic competitiveness and business development action** 1.1.4 Attracting high-level

personnel from abroad in order to enhance the RD capacity; Title: Use of nutrigenomic models for the personalized treatment with medical foods in obese people (NutriGen) 2016-2019- ***volunteer 2017-2019***

7. **ACT1373: Evaluate the Safety, Pharmacodynamics, Pharmacokinetics, and Exploratory Efficacy of GZ/SAR402671 in Treatment-naïve Adult Male Patients With Fabry Disease** - NCT02228460 - ***co-investigator*** between December 2014 - January 2016, Garches, France
8. **Clinical, genetic and epigenetic aspects involved in the etiology of Prader Willi/ Angelman syndromes: multidisciplinary approach model of rare disorders in Romania - active member 2009-2011** CNMP PROJECT PARTNERSHIP, CONTRACT 41113, 2009-2011

Awards

Iulia E. Jurca-Simina, François Vialard, Karelle Benistan, Lucia Echevarria, Philippe de Mazancourt, Alessandro P. Burlina, Dominique P. Germain, "A 16 Mb deletion at Xp22.13 leads to highly skewed X chromosome inactivation and consequent severe phenotypic expression of Fabry disease in a heterozygous female"- 14th European Round Table on Fabry Disease - Fabry PhD Research Initiative, Paris, France- Abstract, poster and oral presentation- Fabry PhD Research Initiative- award- 13-14 March 2015

The best oral presentation: Iulia Simina, Iulia Perva, Catalin Munteanu, Adrian Trifa, " Lynch Syndrome- a case series in the experience of Oncogenetics Team from Oncohelp", The 13th Medical Genetics Conference with International Participation, Timișoara, 28th – 30th September 2023

Courses and conferences

- "EUROPLAN - European Project for Rare Diseases National Plans Development", Bucharest, Romania- 18th-19th June 2010
- The course of "Interdisciplinary approach of genetic rare diseases", Timisoara, Romania- 20th- 22nd September 2010
- "The Third National Medical Genetic Conference with International Participation", Timisoara, Romania- 22th-25th September 2010
- The Workshop "Gene Therapy", Timisoara, Romania- 3rd November 2010
- National Conference of Pediatrics, "Emergencies and chronic diseases", Bucharest, Romania March 2012
- European Society for Pediatric Nephrology- 45th Annual Scientific Meeting, Krakow, Poland- 6th-8th September 2012
- Course "Quality Management in Medical Sector and Institutional Communication", Oradea, Romania- 25th-27th April 2013
- The 7th German-Romanian Genetics Course: "Medical genetics, today", Oradea, Romania - 30th August- 1st September 2013
- The 7th Medical Genetics National Conference with international participation, Sibiu-Paltinis, Romania- 26th- 28th September 2013
- Fabry Masterclass VI, Prague, Czech Republic - 16th-17th May 2014
- 14th European Round Table on Fabry Disease, Paris, France – 13th-14th March 2015
- European Human Genetics Conference 2015, Glasgow, England, June 2015
- European Human Genetics Conference 2017, Copenhagen, Denmark, May 2017
- "Colloque international Les Territoire de la Sante: Production agroalimentaire, Nutrition, Securite alimentaire - PaNSaTS", Timisoara, Romania, October 2017
- "Personalised genomics in Pediatric Nephrology: from the lab bench to the bedside" - The International Pediatric Nephrology Association (IPNA) teaching course, Bucharest, Romania - 17th-18th November 2017.
- ICGEB Workshop on "Next Generation Diagnostics", Skopje, Republic of Macedonia, 22 -24 March 2018.
- Manchester Dysmorphology Course, Nowgen Centre, Manchester, UK, 17th - 19th April 2018.
- European Human Genetics Conference 2018, Milan, Italy, June 2018
- European Reference Networks Workshop, Zalau, Romania, 2018
- “Evocative signs in clinical genetics” Course, Gura Humorului, Romania, September 2018

- European Human Genetics Conference 2019, Gothenburg, Sweden, June 2019
- Pedipractic Pediatrics Conference, online 30.09-02.10.2020
- "Training on strategies to foster solutions of undiagnosed rare disease cases"- 12-14 April 2021, ISS Rome Italy
- European Human Genetics Conference, August 28–31, 2021 online edition
- National Pediatrics Congress, Craiova, September 15 – 18, 2021
- European Human Genetics Conference, June 11-14, 2022, Vienna, Austria
- Joint Symposium - 14-16th June, 2022, Brussels, Belgium
- European Human Genetics Conference, June 11-14, 2023, Glasgow
- European Human Genetics Conference, June 1-4, 2024, Berlin
- Hereditary Cancer Genetics Course, September 17-20, 2024, Bertinoro, Italy
- Rare Hematology Academy, September 4-5, 2024, Madrid, Spain
- Course in Genetic Counseling Skills, November 10-14, 2024, Muscat, Oman
- European Human Genetics Conference, May 24-27, 2025, Milan (virtual participation)

Member of medical societies

- Romanian Society of Medical Genetics SRGM
- European Society of Medical Genetics ESHG

PUBLICATION LIST

BOOK CHAPTERS

author	Title	Type	Date
Dominique P. Germain, **Iulia E. Jurca-Simina**	Principles of Human Genetics and Mendelian Inheritance, Neurometabolic Hereditary Diseases of Adults, Springer International Publishing AG, part of Springer Nature 2018(I):1-28, https://doi.org/10.1007/978-3-319-76148-0	International treaty chapter	2018
Cristina Gug, Maria Puiu, **Iulia Jurca-Simina**	Laboratory guides and guidelines: Génétique médicale- Travaux pratiques pour les étudiants en Médecine Générale, Victor Babeş Publishing House, Timişoara, CNCSIS: 324 © 2020, ISBN 978-606-786-212-6	Laboratory guides and tutors	2020
Nicoleta Ioana Andreescu, Adrian Pavel Trifa, Adela Chiriță – Emandi, Dorina Livia Stoicănescu, Cristina Rodica Gug, Simona Sorina Farcaș, Cristina Annemari Popa, Alexandra Mihăilescu, **Iulia Elena Simina** , Miruna Cristiana Gug	Genetics Course, "Victor Babeş" Publishing House, 2025, ISBN 978-606-786-490-8.	Course book	2025
Nicoleta Ioana Andreescu, Adrian Pavel Trifa, Adela Chiriță – Emandi, Dorina Livia Stoicănescu, Cristina Rodica Gug, Simona Sorina Farcaș, Cristina Annemari Popa, Alexandra Mihăilescu, **Iulia Elena Simina** , Miruna Cristiana Gug	Genetics - Practical Applications. "Victor Babeş" Publishing House 2025, ISBN 978-606-786-489-2	Laboratory guides and tutors	2025
Dorina Livia Stoicanescu, **Iulia Elena Simina.**	Applications of Genetics in Medicine. "Victor Babeş" Publishing House 2025, ISBN 978-606-786-494-6	Laboratory guides and tutors	2025

PUBLISHED ARTICLES

author	Title	Type	Date
Puiu, Maria; **Simina-Jurca, Iulia** ; Dumitriu, Simona; Arghirescu, Smaranda; Chirita-Emandi, Adela	Multiple hereditary exostoses – clinical features and management- Jurnalul Pediatrulu, Jan-Jun2012, vol XV, no 57-58, 2012, page 64-9 (B+)	Published article	June 2012
Maria Puiu, **Iulia Jurca Simina**	Angelman Syndrome, Medical Life, no. 29 (1175), ISSN 1583-8862 http://www.viata-medicala.ro/Sindromul-Angelman.html*articleID_5504-dArt.html	Published article	July 12, 2012
Corina Pienar, Maria Puiu, Adela Chiriță-Emandi, Simona Dumitriu, Cristina Popa, **Iulia Jurca-Simina** , Ioana Micle, Smaranda Arghirescu.	Childhood Obesity: between Nature and Nurture. Journal of the Pediatrician. Year XVI, Vol. XVI, No. 61-62, Counsel-June 2013; 3-8, ISSN 2065 – 4855, CNCSIS B+	Published article	June 2013
Mihai Gafencu, **Iulia Simina**	Overweight pathology in children form Timis County, Jurnalul	Published	August

Jurca , Laura Leahu, Andra Mitoceanu, Otilia Marginean, Gabriela Doros, Bogdan Korbuly	Pediatrulu – Year XVI, Vol. XVI, No. 63, July-September 2013, ISSN 2065 – 4855 http://www.jurnalulpediatrului.ro/pages/reviste/63.pdf#page=27	article	2013
Gafencu Mihai, **Jurca-Simina Iulia** , Butur Marius, Boruga Ovidiu, Doros Gabriela	Severe hypokalemia in HIV/AIDS Wasting Syndrome – a case report, Exp Clin Cardiol Vol 20 Issue1 pages 2275-2283 / 2014	Published article	January 2014
Gafencu M, **Jurca-Simina IE** , Costa R, Doros G	Distal renal tubular acidosis in AIDS young woman with wasting syndrome. Int Urol Nephrol. 2014 Dec;46(12):2423-7 PMID: 25298139. doi: 10.1007/s11255-014-0840-9 **. (FI/2014 1,293)**	Published article	December 2014
Adela Chirita-Emandi, Gabriela Doros, **Iulia Jurca Simina** , Mihai Gafencu, Maria Puiu	Head circumference references for school age children in western Romania, Rev. Med. Chir. Soc.Med. Nat., Iasi- 2015- Vol. 119, No. 4, page 1083-1091 ISSN 0048-7848	Published article	December 2015
Beth L. Thurberg, Dominique P. Germain, MD, Fernando Perretta, **Iulia E. Jurca-Simina** , Juan M. Politei	Fabry disease: Four case reports of meningioma and a review of the literature on other malignancies, Mol Genet Metab Rep. 2016 Oct 1;11:75-80. eCollection 2017 Jun. PMID: 28649509; PMCID: PMC5470937 doi: 10.1016/j.ymgmr.2016.09.005 **(FI/2018 1.354), 4 citations-02.07.2024**	Published article	October 2016
Jurca-Simina Iulia-Elena, Chirita Emandi Adela, Perva Iulia Teodora, Uhrová Mészárosová Anna, Corches Axinia, Doros Gabriela, Puiu Maria	Think about the founder effect in endogamous population - Congenital cataracts, Facial dysmorphism, and Neuropathy (CCFDN) Syndrome - two cases, Jurnalul Pediatrulu, 2018, XXI(81-82): 19-25 (B+)	Published article	June 2018
Jurca-Simina Iulia-Elena , Iulius Jugănaru, Mircea-Ştefan Iurciuc, Stela Iurciuc, Emil Ungureanu, Andreea Iulia Dobrescu, Adela Chiriţă-Emandi, Oana Raluca Voinescu, Ioana-Cristina Olariu, Maria Puiu, Doina Georgescu, Veronica-Mădălina Boruga	What if body fat percentage association with FINDRISC score leads to a better prediction of type 2 diabetes mellitus?, Rom J Morphol Embryol 2019, 60(1):205–210, PMID: 31263846 **(FI/ 2018 = 1.5); 2 citations-07.02.2024**	Published article	June 2019
Iulia E. Jurca-Simina , Adela Chirita-Emandi, Nicoleta Andreescu, Simona Farcaş, Alexandra Mihailescu, Anca-Maria Popa, Paul Tutac, Cristian Zimbru, Andreea I. Dobrescu, Iulia T. Perva, Amalia Murariu, Maria Puiu	Burden of rare genetic diseases –experience of Timis Regional Centre of Medical Genetics, Romania, Journal of Pediatrics, 2019 , XXII (85-86): 56-65 (B+)	Published article	June 2019
Roman Deiana, Gug Miruna, Gliga Petra, Chircă Corina, **Jurca-Simina Iulia** , Jurca Maria Claudia, Vaida Monica	Monogenic Cause For Renal Tubulopathies - Considerations Regarding Four Cases in Fertile Women. Proceedings of the 4th Congress of the Romanian Society for Minimally Invasive Surgery in Gynecology, pages 513-517	Published article	2019
João P. Oliveira, Albina Nowak, Frédéric Barbey, Márcia Torres, José P. Nunes, Fernando Teixeira-e-Costa, Fernanda Carvalho, Susana Sampaio, Isabel Tavaresj, Odete Pereira, Ana L. Soares, Cátia Carmona, Maria-Teresa Cardoso, **Iulia E. Jurca-Simina** , Marco Spada, Susana Ferreira, Dominique P. Germain	Fabry disease caused by the GLA p.Phe113Leu (p.F113L) variant: Natural history in males, Eur J Med Genet. 2020;63(2):103703. doi:10.1016/j.ejmg.2019.103703 (**FI/2020=2.368), 13 citations-07.02.2024**	Published article	February 2020
NCD Risk Factor Collaboration (NCD-RisC).. **IE Jurca-Simina**	Height and body-mass index trajectories of school-aged children and adolescents from 1985 to 2019 in 200 countries and territories: a pooled analysis of 2181 population-based studies with 65 million participants. Lancet. 2020 Nov 7;396(10261):1511-1524 **(IF 2019:14.554), 118 citations-02.07.2024**	Published article	November 2020
Sabau, I.M., Andreescu, N.I., Chiriță-Emandi, A., **Jurca-Simina, I.** , Bugi, M.A., & Puiu, M.	Genetics in anorexia nervosa, Journal of Pediatrics XXIV (93-94): 23-27	Published article	January 2021
Sabau, I.M., Andreescu, N.I., Chiriță-Emandi, A., **Jurca-Simina, I.** , Bugi, M.A., & Puiu, M.	Ketogenic diet and genetic disorders, Journal of Pediatrics XXIV (93-94): 28-33	Published article	January 2021
NCD Risk Factor Collaboration (NCD-RisC).. **IE Jurca-Simina**	Heterogeneous contributions of change in population distribution of body mass index to change in obesity and underweight. eLife 2021;10:e60060 DOI: 10.7554/eLife.60060 **(IF= 8.14), 39**	Published article	March 2021

	citations-02.07.2024		
Nowicka P, Ek A, **Jurca-Simina IE** , Bouzas C, Argelich E, Nordin K, García S, Vasquez Barquero MY, Hoffer U, Reijs Richards H, Tur JA, Chirita-Emandi A, Eli K.	Understanding the complex impact of the Covid-19 pandemic on children with overweight and obesity: a comparative ecological analysis of parents' perceptions in three countries. BMC Public Health. 2022 May 17;22(1):1000 **(IF on 2 years, 2022- 3,295), 7 quotes-02.07.2024**	Published article	May 2022
NCD Risk Factor Collaboration (NCD-RisC).. including **IE Jurca-Simina**	Diminishing benefits of urban living for children and adolescents' growth and development. Nature. 2023 Mar;615(7954):874-883. two: 10.1038/s41586-023-05772-8. Epub 2023 Mar 29. PMID: 36991188; PMCID: PMC10060164 **(IF=64.8), 7 citations-02.07.2024**	Published article	March 2023
Bugi MA, Jugănaru I, Isac R, **Simina IE** , Munteanu AI, Mang N, Brad GF, Nicoară DM, Cîrnatu D, Mărginean O.	Factors Impacting the Reduction in Neophobia Prevalence in Phenylketonuria Patients. Nutrients. 2024 Mar 7;16(6):768. two: 10.3390/nu16060768. PMID: 38542680; PMCID: PMC10975818 (**IF= 5.9)**	Published article	March 2024
NCD Risk Factor Collaboration (NCD-RisC).. including **IE Jurca-Simina**	Worldwide trends in underweight and obesity from 1990 to 2022: a pooled analysis of 3663 population-representative studies with 222 million children, adolescents, and adults. Lancet. 2024 Mar 16;403(10431):1027-1050. two: 10.1016/S0140-6736(23)02750-2. Epub 2024 Feb 29. PMID: 38432237; PMCID: PMC7615769 **(IF=168.9), 12 citations-02.07.2024**	Published article	March 2024
Bugi MA, Jugănaru I, **Simina IE** , Nicoară DM, Cristun LI, Brad GF, Boru C, Cîrnatu D, Mărginean O.	Exploring Adult Eating Behaviors and Food Neophobia: A National Study in Romania. Foods. 2024 Apr 24;13(9):1301. doi: 10.3390/foods13091301. PMID: 38731672; PMCID: PMC11083192 (**IF= 4.7)** .	Published article	April 2024
Bugi MA, Jugănaru I, **Simina IE** , Nicoară DM, Cristun LI, Brad GF, Huțanu D, Isac R, Kozma K, Cîrnatu D, Mărginean O.	Evaluating Therapy and Growth in Children with Phenylketonuria: A Retrospective Longitudinal Study from Two Romanian Centers. Medicine 2024, 60, 1185. https://doi.org/10.3390/medicina60071185 **(IF =2.4)**	Published article	July 2024
Perva IT, **Simina IE** , Bende R, Motofelea AC, Chirita Emandi A, Andreescu N, Sima A, Vlad A, Sporea I, Zimbru C, Tutac PC, Puiu M, Niculescu MD	Use of a Micronutrient Cocktail to Improve Metabolic Dysfunction-Associated Steatotic Liver Disease (MASLD) in Adults with Obesity: A Randomized, Double-Blinded Pilot Clinical Trial. Medicine (Kaunas). 2024 Aug 21;60(8):1366. two: 10.3390/medicina60081366. PMID: 39202647; PMCID: PMC11356300 **(IF =2.4)**	Published article	August 2024
Cavaloiu B, **Simina IE** , Vilciu C, Trăilă IA, Puiu M.	Nusinersen Improves Motor Function in Type 2 and 3 Spinal Muscular Atrophy Patients across Time. Biomedicines. 2024 Aug 6;12(8):1782. doi: 10.3390/biomedicines12081782. PMID: 39200246; PMCID: PMC11351209 **(IF =3.9)**	Published article	August 2024
Cavaloiu B, **Simina IE** , Chisavu L, Vilciu C, Traila IA, Puiu M.	Quality of Life Assessment in Romanian Patients with Spinal Muscular Atrophy Undergoing Nusinersen Treatment. Neurol Int. 2024 Aug 26;16(5):891-904. doi: 10.3390/neurolint16050067. PMID: 39311340; PMCID: PMC11417783 **(IF =3.2)**	Published article	August 2024

CONGRESS AND CONFERENCE ABSTRACTS

author	Title	Type	Date
M Puiu, M Gafencu, **I. Jurca Simina** et al.	"Prader Willi Syndrome- from research project to multicentre approach", the third National Congress of Medical Genetics, with international participation, Timisoara, Romania, 22-25 Sept. 2010. Romanian Journal of Rare Diseases 2010	Abstract, oral presentation	September 2010
Iulia Jurca Simina, Florin Jurca Simina, Ionela Moaca, Pop Norbert, Stefan Berci, Iulia Popa, Cristina Irimia, Oana Rosca, Graziella Ecob, Adrian Juverdeanu, Carmen Dumitranoiu, Narcis Dobre, Mihai Gafencu, Maria Puiu	"Save the children" with rare diseases"- Romanian Journal of Rare Diseases, Supplement 1/2010 ISSN 2068-5882	Abstract, oral presentation	2010
Ionela Moaca, **Iulia Jurca Simina,** Florin Jurca Simina, Pop Norbert, Stefan Berci, Iulia Popa, Cristina Irimia, Oana Rosca, Graziella Ecob, Adrian Juverdeanu, Carmen Dumitranoiu, Narcis Dobre, Mihai	"Special needs children's Day" - Romanian Journal of Rare Diseases, Supplement 1/2010 ISSN 2068-5882	Abstract, poster presentation	2010

Gafencu, Maria Puiu			
Oana Rosca, Ionela Moaca, **Iulia Jurca Simina** , Florin Jurca Simina, Pop Norbert, Stefan Berci, Iulia Popa, Cristina Irimia, Graziella Ecob, Adrian Juverdeanu, Carmen Dumitranoiu, Narcis Dobre, Mihai Gafencu, Maria Puiu	"Rare diseases Week in Timisoara" - Romanian Journal of Rare Diseases, Supplement 1/2010 ISSN 2068-5882	Abstract, poster presentation	2010
Jurca-Simina Iulia- Elena, Ecob Graziella, Bacalu Alina, Gafencu Mihai	"Child's obesity, a look from the perspective of age, sex and lifestyle"- Public prize at the National Congress for Students and Young Doctors- 14 th edition", Bucharest, Romania	Abstract, oral presentation	9- 12 December 2010
IE Jurca-Simina , M Gafencu, D Dan, M Puiu	Poster presentations: 731 "Prader Willi Syndrome (PWS) - Particular Molecular Profile and Diagnostic Protocol in Romania" *Arch Dis Child* 2012; 97:A211	Abstract, poster presentation	2012
I. Jurca-Simina , M. Gafencu, A. But, S. Hanini	"Child and adolescent obesity - Influence of diet and lifestyle" Book of abstracts pg. 89 National Conference of Pediatrics, 'Emergencies and chronic diseases', Bucharest, Romania.	Abstract, poster presentation	March 2012
Doros G., Popoiu A., Gafencu M., **Jurca-Simina IE** ., Leahu L., But A.	"Risk factors for cardiovascular disease in school age children and teenagers" - 46 th Annual Meeting of the AEPC, Istanbul, Turkey, 23-26 May 2012, Cardiology in the Young, Vol 22, Suppl. 1, p.111-112, ISSN 1047-9511, Impact factor 2012 0.948	Abstract, poster presentation	May 2012
Gafencu M **, Jurca-Simina IE** . et al.	"Neurological component in HIV with hypokalemia - case report" - poster presentation at Neonatology National Conference, Sibiu, 2012	Abstract, poster presentation	June 2012
M. Gafencu, R. Costa, G. Doros, K. Nilima, A. Schiller, **I. Jurca-Simina**	"Renal involvement in HIV infected children"- Pediatric Nephrology, vol 27 issue 9, pp 1605-1829(2012), The 45 th Annual Meeting on September 6 th - 8 th 2012, Krakow, Poland, ISSN online 1432-198X.	Abstract, poster presentation	September 2012
IE Jurca-Simina , M Gafencu, D Dan, M Puiu	"Prader Willi Syndrome (PWS) - Particular Molecular Profile and Diagnostic Protocol in Romania", Arch Dis Child 2012;97:A211 doi:10.1136/archdischild-2012-302724.0731- Poster presentations	Abstract, poster presentation	October 2012
IE Jurca-Simina, M. Puiu, M. Gafencu	"Renal disease's genetic counseling- a must for an affected family", ESHG 2013, European Journal of Human Genetics, Volume 21 Supplement 2, p 424, ISSN1018-4813, 2012 Impact Factor-4.319.pag 576	Abstract, poster presentation	June 2013
Jurca Simina I , Gafencu M, et al.	"Clinical and evolutionary aspects of Klippel Feil Syndrome", National Congress of Medical Genetics, with international participation, Paltinis, September 2013	Abstract, poster presentation	September 2013
Doros G, Gafencu M, **Jurca Simina I** et al.	"Clinical profile, evolution and side effects of specific therapy in a rare disease -PAH secondary to CHD", National Congress of Medical Genetics, with international participation, Paltinis, September 2013	Abstract, poster presentation	September 2013
M. Gafencu, G. Doros, D. Dan, **I. Jurca Simina,** LN Bogdan, M. Puiu	"Rare Diseases week in Timisoara - a campaign with a good start", European Journal of Human Genetics, Volume 22, Supplement 1, May 2014, Milan, page 346, P18.40-M	Abstract, poster presentation	May 2014
IE Jurca-Simina , RM Jurac, M. Cucuruz, C. Jinca, E. Boeriu, C. Popa, S. Arghirescu, M. Puiu	"Particularities of ATRA therapy in pediatric patients with acute promyelocytic leukemia", European Journal of Human Genetics, Volume 22, Supplement 1, May 2014, Milan, page 493, J15.17	Abstract in volume of abstracts	May 2014
DP Germain, J.-B. Riviere, I. Dabaj, J. Bataille, C. Jauny **, IE Jurca-Simina** , L. Faivre and I. Haegy	"Clove syndrome: a case report", Twenty-sixth European Meeting on Dysmorphology, 9 - 11 September 2015, Le Bischenberg	Abstract, presentation	September 2015
IE Jurca-Simina , J. Rabès, PA Richard, C. Jauny, F. Koraichi, R. Carlier, AA Hagège, P. de Mazancourt, M. Puiu, DP Germain	"Pitfalls in the diagnosis of Fabry disease: further evidence that p.Asp313Tyr is a non-pathogenic polymorphism", P06.09A, Metabolic and mitochondrial disorders session, European Human Genetics Conference 2017, Copenhagen, Denmark, May 2017	Abstract, poster presentation	May 2017
Iulia Jurca-Simina, Alexandra Mihăilescu, Iulia Perva, Alexandra Sima, Iulian Puiu Velea, Adela Chiriță-Emandi, Nicoleta Andreescu, Mihai Niculescu, Maria Puiu	"NutriGen. Utilization of nutrigénomic models to personalize dietary treatments in obesity ", Colloque international Les Territoires de la Sante: Agroalimentaire Production, Nutrition, Securite alimentaire - PaNSaTS, Timisoara, Romania	Oral presentation	October 2017
Jurca-Simina Iulia-Elena, Chirita-Emandi Adela, Nicoleta	"Autosomal Dominant Multicystic Dysplastic Kidney Phenotype – No Genotype Identified (Yet)", Abstract book of "Personalized	Abstract, poster	November 2017

Andreescu, Olariu Nicu, Isac Raluca, Jurca-Simina Florin-Ioan, Gafencu Mihai, Puiu Maria	genomics in Pediatric Nephrology: from the lab bench to the bedside"- The International Pediatric Nephrology Association (IPNA) teaching course and the VIIth Meeting of the Southeastern Europe Pediatric Nephrology Working Group (VIIth SEPNWG), Bucharest, Romania- 17th-18th of November 2017, ISBN 978-973-0-25841-7.	presentation	
Jurca-Simina Iulia-Eena , Chirita-Emandi Adela, Andreescu Nicoleta, Cristian Zimbru, Urtila Patricia, Ioana Micle, Puiu Maria	"Heterozygous known mutation in LPL gene causing Lipoprotein Lipase Deficiency with severe Hypertriglyceridemia in a child". Balkan Journal of Medical Genetics, vol 21,2018, supplement 1, ISSN1311-0160, p57 (ICGEB Workshop "Next Generation Diagnostics", Skopje, March 22-24,2018)	Abstract, poster presentation	March 2018
I. Jurca-Simina , A. Chirita-Emandi, N. Andreescu, N. Olariu, R. Isac, F. Farkas, Z. Andrei, M. Gafencu, M. Puiu	"Bilateral Multicystic Dysplastic Kidney in a three-generation family", Control No. 2018-A-2701-ESHG, European Human Genetics Conference (ESHG), Milan, Italy, June 2018	Abstract, E-poster	June 2018
T. Marcovici, M. Puiu, C. Bacos, **I. Jurca-Simina,** O. Belei, O. Marginean, A. Grozavu	"Classic Dravet Syndrome in an adolescent male. Case report", Control No. 2018-A-2605-ESHG, European Human Genetics Conference (ESHG), Milan, Italy, June 2018	Abstract, E-poster	June 2018
Iulia-Elena Jurca-Simina , Adela Chirita-Emandi, Nicoleta Andreescu, Florina Stoica, Adina Ionac, Maria Puiu	"Ocular manifestations in Anderson-Fabry disease", VIIIth Congress of the Romanian Society of Strabology and Pediatric Ophthalmology in conjunction with IInd Conference on Ophthalmogenetics with International Participation, Brasov, Romania	Abstract, oral presentation	June 2018
Iulia Jurca-Simina, Dorica Dan, Adela Chirita Emandi, Puiu Maria	"RO-NMCA ID Network- RoNetwork Multiple Congenital Abnormalities with ID", European Reference Networks Workshop, Zalau, Romania, 2018	Oral presentation	July 2018
Aniko Manea, **Jurca-Simina Iulia-Elena** , Daniela Cioboata, Oana Costescu, Florina Doandes, Nicoleta Lungu, Marioara Boia	Rapid and Optimal Diagnosis in Malformative Syndromes at newborns, European Human Genetics Conference (ESHG), Gothenburg, Sweden, June 2019	Abstract, E-poster	June 2019
IE Jurca-Simina , A. Chirita-Emandi, N. Andreescu, CL Șerban, C. Zimbru, M. Puiu1	Molecular genetic diagnosis in skeletal disorders - a Western Romanian delineation, European Human Genetics Virtual Conference 2020, taking place from June 6-9, 2020	Abstract, E-poster	June 2020
Nowicka P, Ek A, **Jurca-Simina IE** , Bouzas C, Argelich E, Nordin K, García S, Vasquez Barquero MY, Hoffer U, Reijs Richards H, Tur JA, Chirita-Emandi A, Eli K.	Understanding the complex impact of the Covid-19 pandemic on overweight children and obesity: a comparative ecological analysis of parents' perceptions in three countries - STOP Project WP8 - Joint Symposium - 14-16th June - Brussels	Poster	June 2022
Iulia-Elena Simina , Florin-Ioan Jurca, Florina Stoica, Meda Ada Bugi, Maria Puiu, Adela Chirita-Emandi	When a routine blood test can change it all- Hereditary hyperferritinemia cataract syndrome, European Human Genetics Conference, Glasgow, United Kingdom, June 10-13	Abstract, E-poster	June 2023
Iulia Simina	Molecular techniques for investigating genetic modifications, Oncogenetics Course within the "Cancer Immunotherapy" Conference, 6th edition, Timisoara, June 15-18, 2023	Oral presentation	June 2023
Iulia Simina	Types of genetic modifications. Somatic mutations vs. germline mutations, Oncogenetics Course within the “Cancer Immunotherapy” Conference, 6th edition, Timisoara, June 15-18, 2023	Oral presentation	June 2023
Iulia Simina	Genetic testing in other types of cancer. Rare genetic syndromes of predisposition to cancer, Oncogenetics Course within the "Cancer Immunotherapy" Conference, 6th edition, Timisoara, June 15-18, 2023	Oral presentation	June 2023
Iulia Simina , Iulia Perva, Catalin Munteanu, Adrian Trifa	Li-Fraumeni Syndrome a not so rare cause of hereditary breast cancers - a series of cases, National Forum of Gynecology-Oncology, 6th Edition, Conference of the Romanian Society of Breast Surgery and Oncology	Oral presentation	June 2023
Iulia Simina, Simona Cerbu, Irina Sarau, Maria Puiu	Acid Sphingomyelinase Deficiency Insights and a New Hope in the First Therapy to Treat It. "Inborn Errors of Metabolism in Child and Adult Pathology" conference, June 29-30, 2023, Cluj Napoca	Oral presentation	June 2023
Iulia Simina, Iulia Perva, Catalin Munteanu, Adrian Trifa	Lynch Syndrome- a case series in the experience of Oncogenetics Team from Oncohelp, The 13th Medical Genetics Conference With International Participation, Timișoara, 28th – 30th September 2023	Oral presentation	September 2023
Iulia Simina	Neuroblastoma – a rare disease, a rare cancer, Rare Disease Day Timișoara. "Multidisciplinarity in rare diseases", March 14, 2024	Oral presentation	March 2024

Iulia-Elena Simina, Iulia Teodora Perva, Catalin-Vasile Munteanu, Oana Cristina Voinea, Adrian Trifa	Is the obvious enough when considering Lynch Syndrome diagnosis? Rare cancers in index cases and diagnostic challenges, European Human Genetics Conference, Berlin, June 1-4	Abstract, E-poster	June 2024
Iulia Simina	Novelties in the Treatment of Niemann-Pick Type A/B and B Disease. Inherited Metabolic Diseases in pediatric and adult pathology, focus on therapy, 5 th ed., with the session "At the crossroads between metabolism and autoinflammatory disorders", 28-29 June 2024, Cluj-Napoca, Romania	Oral presentation	June 2024
Maria Puiu, **Iulia Simina**	Expanding Neonatal Screening: From Suspicion to Confirmed Diagnosis and Integrated Management. Inherited Metabolic Diseases in pediatric and adult pathology, focus on therapy, 5 th ed., with the session "At the crossroads between metabolism and autoinflammatory disorders", 28-29 June 2024, Cluj-Napoca, Romania	Oral presentation	June 2024
Maria Puiu, **Iulia Simina** , Ioana Streata, Dorica Dan	Interdisciplinarity in inborn errors of metabolism, 26th National Symposium on Psychoneuroendocrinology, with International Participation, September 26-28, 2024, Arad	Oral presentation	September 2024
Iulia Simina	Huntington's Disease- Roundtable- The Huntington's Puzzle: Science, Support, and Solutions- MEDIS, 02-06 April 2025	Oral presentation	April 2025
Iulia Simina	Interactive game on family dynamics and genetic testing - Roundtable - The Huntington's Puzzle: Science, Support, and Solutions - MEDIS, 02-06 April 2025	Oral presentation	April 2025
Iulia Simina	Genetics Clinical case, Guest speaker at MedMysteries Solved, EMSA Timisoara	Oral presentation	April 2025
Iulia Simina, Iulia Perva, Catalin Munteanu, Dorel Popovici, Adrian Trifa	Revisiting the impact of allele dropout on therapy/somatic testing: a case study of ovarian tumor misclassification as BRCA-proficient, European Human Genetics Conference, Milan, May 24-27	Abstract, E-poster	May 2025
Iulia-Elena Simina, Oana Voinescu, Nicoleta Andreescu, Adina Ionac, Adela Chiriță-Emandi	A rare genetic disease, a dramatic trajectory – vascular Ehlers-Danlos syndrome, 10-NefroCardDia 2025, Timisoara,	Oral presentation	October 16-18, 2025

REVIEWERS

Name/position	University/Hospital	Phone	E-mail
MD, PhD **Maria Puiu** **Primary care physician, pediatrics and medical genetics** **Professor Medical Genetics**	National Institute for Research and Development in Genomics 37 Dionisie Lupu Street, Sector 2, Bucharest, Romania	+40745138917	**maria.puiu@gmail.com**
MD, PhD **Dominique Germain** **Medical genetics specialist** **Professor of medical genetics**	Head of department UFR des Sciences de la Santé Simone Veil 2, allée de la source de la Bièvre, 78180 Montigny Center Hospitalier Universitaire Raymond Poincaré, 92380 Garches University of Versailles - St Quentin en Yvelines, France	+33147104438	**dominique.germain@inserm.fr** **dominique.p.germain@aphp.fr** **dominique.germain@uvsq.fr**

Date
02/17/2026