PERSONAL INFORMATION

Ina Ofelia Focșa

TITLE APPLIED FOR

PhD in Medical Science

WORK EXPERIENCE

Since 2020	Clinical Genetics MD Cytogenomic Medical Laboratory, 35 Floreasca Street, Bucharest ▪ Genetic counseling, clinical genetic examination, cytogenetics, molecular genetics/ NGS data clinical curator
Since 2016	Assistant Professor Medical Genetics Department, Faculty of Medicine, "Carol Davila" University of Medicine and Pharmacy, 19-21 Gerota Street, Bucharest ▪ Teaching in medical genetics field ▪ Research in medical genetics
2013-2015	Research assistant "Victor Babes" National Institute of Pathology, 99-101 Splaiul Independentei Avenue, Bucharest, Romania ▪ Research in medical genetics – neurodevelopmental disorders; cell cultures, cytogenetic, molecular genetics (PCR-based genetic tests, aCGH)
2010-2015	Medical Genetics resident „Sf. Ioan" Clinical Emergency Hospital, Bucharest, 14 Vitan Barzesti Av ▪ Clinical dysmorphology, Clinical Genetics, Genetic Counseling, Cytogenetic, Molecular genetics
2000-2009	Medical representative, senior medical representative, key account representative Glaxo-Wellcome; Pfizer HCP Corporation and Boehringer Ingelheim Pharma ▪ Promotion of medicines; medical group presentations, organizing workshops, seminars and symposia

EDUCATION AND TRAINING

2021

Senior Medical Geneticist EQF level 8
Ministry of health
- Clinical genetics, Genetic Counseling, Cytogenetics, Molecular genetics

Since 2015.10

PhD Student EQF level 8
"Carol Davila" University of Medicine and Pharmacy, 37 Dionisie Lupu Street, Bucharest,
- Research in clinical and genetic aspects of ciliopathies; doctoral thesis named "Connections between genetic spectrum and clinical picture in ciliopathies"

2010-2015

Board-certified in Medical Genetics EQF level 7
Ministry of health
- Clinical genetics (perinatal genetics, genetics of cancer), Genetic Counseling, Cytogenetics, Molecular Genetics

1999-2000

Intern EQF level 6
Ministry of health
- Advance training focused on perinatal medicine/gynecology and General Medicine performed in "Panait Sarbu" Obstetrics-Gynecology Clinical Hospital, Bucharest

1992-1998

Medical doctor EQF level 6
"Carol Davila" University of Medicine and Pharmacy Bucharest, Romania
- General medicine

PERSONAL SKILLS

Mother tongue(s): Romanian

Other language(s)

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken interaction	Spoken production	
English	C1	C1	B1	B1	C1
	"Carol Davila" University of Medicine certificate/no.779/08.09.2015				
French	B1	B2	B2	B2	B2
Russian	A1	A2	A1	A1	A1

Communication skills

Excellent communication skills and team working gained due to my experience as medical representative and due to the training in Assertive Communication and Business Negotiation Courses;

Organisational / managerial skills

Very good adaptability to various working environments based on my personal ability to be flexible, to easy interact and create connection with people, acquired during my experience to work in different teams and fields (clinical, research, pharmaceutical fields);
Very good deadline compliance based on my capacity to be organized and due to attending of Time management training;
Workshops and scientific symposia organizing;

Job-related skills

Competences:
- Board-certified in Medical Genetics

Skills:

- Genetics of neurodevelopmental disorders, imprinting disorders and ciliopathies
- Dysmorphology
- Genetic counseling
- Genetic laboratory techniques (blood cell cultures; DNA, RNA, plasmid DNA isolation; classical cytogenetic, FISH technique; PCR-based technologies; array CGH; NGS technologies)

Digital competence

SELF-ASSESSMENT

Information processing	Communication	Content creation	Safety	Problem solving
Independent user	Independent user	Independent user	Independent user	Independent user

Levels: Basic user - Independent user - Proficient user
Digital competences - Self-assessment grid

- Good command of Office suite
- Good command of Corel Draw
- Good command of software for genetic laboratory applications
- Good command of software for computerized diagnosis

Driving licence

B category

ADDITIONAL INFORMATION

Fellowships

- Fellowship for participation at Basic and Advance Course in Genetic Counseling- Bertinoro 2016
- Fellowship for participation at Manchester dysmorphology Course 2015
- Scholarship for attendance at 2nd European Imprinting Disorders School-Paris 2015
- Scholarship for participation at Lab techniques in imprinting disorders- Aachen, 2015

Memberships

SRGM (Romanian Society of Medical Genetics) member;
ESHG; (European Society of Human Genetics) member;
ASHG (American Society of Human Genetics) member;
COST BM1208 (European Network for Human Congenital Imprinting Disorders) member
COST CA16118 European Network on Brain Malformation (Neuro-MIG)

Courses

- **"Fifth European Society of Human Genetics training: Course in Cardiogenetics"**, October 2-5, 2021, Antwerp, Belgium
- **"Eighth European Course in Clinical Dysmorphology "What I Know Best"** - Web-seminar, October 1-3, 2020, Rome, Italy
- **Clinical management of Brain Malformation Training school** COST Action CA16118: European Network on Brain Malformations, March 5-6 2020
- The European Society of Human Genetics (ESHG) and European School of Genetic Medicine (ESGM)- "**Clinical Genomics and NGS**" Web-seminar, April 30- May 1, 2018, Bertinoro, Italy
- Universita Cattolica de Sacro cuore, Instituto di Genetica Medica- **Seventh European Course in Clinical Dysmorphology "What I Know Best"**, November 6-8, 2017, Roma, Italy

- The European Society of Human Genetics (ESHG) and European School of Genetic Medicine (ESGM)- "**Clinical Genomics and NGS**" Web-seminar, April 30- May 5, 2017, Bertinoro, Italy
- American Society of Human Genetics- **Genetic Testing in Children Webinar,** May 6, 2017
- American Society of Human Genetics- **Undergraduate Genetics Education Workshop,** October 18 -22, 2016, Vancouver, Canada
- The European Molecular Biology Organization (EMBO)- **3th International cilia meeting- "Cilia 2016",** October 4-7, 2016Amsterdam, The Netherlands
- The European Society of Human Genetics (ESHG) and European School of Genetic Medicine (ESGM)- **Basic and Advance Course in Genetic Counseling**- April 28th – May 3rd 2016 – Bertinoro di Romana, Italy
- European Network of Human Congenital Imprinting Disorders and Biomedicine and Molecular Biosciences COST Action BM1208- **2nd European Imprinting Disorders School-Paris 2015**;
- The European Society of Human Genetics (ESHG) and Manchester Centre for Genomic Medicine- **Manchester Dysmorphology Training Course 2015,** April 21-23, 2015, Manchester, UK
- European Network of Human Congenital Imprinting Disorders and Biomedicine and Molecular Biosciences COST Action BM1208- **Lab techniques in imprinting disorders**- Aachen, 2015;
- Universita Cattolica de Sacro cuore, Instituto di Genetica Medica- **Fifth European Course in Clinical Dysmorphology "What I Know Best"**, November 14-16, 2013, Roma, Italy
- The European Society of Human Genetics (ESHG), European School of Genetic Medicine (ESGM) and Centro Residenziale Universitario Bertinoro- **The 26th Course in Medical Genetics** – May 12-16, 2013, Bertinoro di Romagna, Italy
- V. Babes National Institute of Pathology- **Genetic diagnostic technologies** – May-September, 2013, Bucharest, Romania
- Romanian Society of Human Genetics and University of Medicine Oradea- **7th International Course in Medical Genetics "Medical Genetics Today"** – August 30- September 2, 2013, Oradea, Romania
- Romanian Society of Human Genetics and University of Medicine "Gr.T.Popa" Iasi **"Suggestive Sign in Medical Genetics"**-October 4, 2012, Iasi, Romania

Projects

2013-2015 research team member "V. Babeș" National Institute of Pathology
PN II. No 42-130-1
Multidisciplinary research- clinic, cytogenetic and molecular- on syndromes characterized by intellectual disability associated with congenital malformation: contribution to knowledge and management of rare diseases

Book chapters

First-Tier Array CGH in Clinically Variable Entity Diagnosis: 22q13.3 Deletion Syndrome
M. Budisteanu, A. Tutulan Cunita, **I O Focsa,** S. M. Papuc, A. Arghir; Chapter in book Chromosomal Abnormalities (October 9th 2019) DOI: 10.5772/intechopen.89399
Available from: https://www.intechopen.com/books/chromosomal-abnormalities/first-tier-array-cgh-in-clinically-variable-entity-diagnosis-22q13-3-deletion-syndrome

Publications

Clinical aspects of a rare disease: Bardet Biedl syndrome
Ina Ofelia Focșa, Magdalena Budișteanu, Cristina Stoica, Florina Nedelea, Claudia Jurcă, Lavinia Caba, Lăcrămioara Butnariu, Monica Pânzaru, Cristina Rusu, and Mihaela Bălgrădean: Modern Medicine 2022, Vol. 29, No. 1
A case of Bardet-Biedl syndrome caused by a recurrent variant in BBS12: A case report
Ina Ofelia Focșa, Magdalena Budișteanu, Carmen Burloiu, Sheraz Khan, Laurențiu C. Bohîlțea, Erica E. Davis and Mihaela Bălgrădean: A case report. Biomedical Reports, 15, 103. https://doi.org/10.3892/br.2021.1479

Clinical and genetic heterogeneity of primary ciliopathies (Review)
Focșa, I.O., Budișteanu, M., & Bălgrădean, M. (2021). International Journal of Molecular Medicine, 48, 176. https://doi.org/10.3892/ijmm.2021.5009
Bardet Biedl Syndrome: A Reference for Ciliopathies
Focsa I. O., Toader D. O., Budisteanu M., Bohiltea L. C., Suciu N., Balgradean M.: Proceedings of the Annual Days of the National Institute for Mother and Child Health "Alessandrescu-Rusescu" March 2020, *ISBN 978-88-85813-86-1*
Treatment of Epilepsy Associated with Common Chromosomal Developmental Diseases
M. Budisteanu, C. Jurca, Sorina Mihaela Papuc, **I. Focsa**, D. Riga, S. Riga, A. Jurca, A. Arghir; *Open Life Sciences*, vol. 15, no. 1, 2020, pp. 21-29. https://doi.org/10.1515/biol-2020-0003
Neurofibromatosis type 1 associated with moyamoya syndrome. Case report and review of the literature
M. Budisteanu, C Burloiu, S.M. Papuc, **I.O. Focsa**, D. Riga, S. Riga, A. Arghir; Rom J Morphol Embryol. 2019;60(2):713-716. PMID: 31658349; ISSN 2066-8279

Presentations at Congress and Conferences

Clinical and molecular characterization of a rare case diagnosed with Bardet Biedl Syndrome
I. O. Focsa, M. Budisteanu, C. Burloiu, L. C. Bohiltea, M. Balgradean; 53rd European Society of Human Genetics (ESHG) Virtual Conference, June 6-9, 2020; *Eur J Hum Genet* **28,** 798–1016 (2020). https://doi.org/10.1038/s41431-020-00741-5; *ISSN 1476-5438*
Genomic assay in autism spectrum disorders: a study of 200 cases
I. O. Focsa, S.M. Papuc, A. Tutulan Cunita, F. Rad, L. Mateescu, R. Grozavescu , B. Budisteanu, C. Iliescu , D. Birca , C. Anghelescu, D. Ioana, D. Riga, S. Riga, I. Dobrescu, L.C. Bohiltea, A. Arghir M. Budisteanu, *2019 Annual Meeting of the American Society of Human Genetics*, Houston, Texas on October 15-19, 2019
Molecular cytogenetic and clinical assessment in a new case of partial trisomy 9
I. O. Focșa, D. Ioana, C. Anghelescu, L. C. Bohilțea, M. Budișteanu, 52rd *European Society of Human Genetics Conference 2019,* June 15-18, 2019 in Gothenburg, Sweden, *Eur J Hum Genet* **27,** 1174–1813 (2019). https://doi.org/10.1038/s41431-019-0494-2; *ISSN 1476-5438*
From cilia to complex disorders: The ciliopathies
I. O. Focsa, L. C. Bohiltea, M. Balgradean, *Annual Scientific Meeting of Victor Babeș Institute;* The 11th National Pathology Symposium, November 22 - 24, 2018, Bucharest, Romania
Clinical approach of Angelman Syndrome: a study of 23 cases
I. O. Focsa, L. C. Bohiltea, D. Craiu, C. Iliescu, D. Birca, A.C. Tutulan Cunita, S.M. Papuc, A. Arghir, Magdalena Budisteanu, *American Society of Human Genetics Annual Meeting*, October 17-21, 2018, San Diego, USA
The past, the present and the future in ciliopathies
Ina Ofelia Focsa,
The 5th National Congress of Medical Genetics, September 26 - 28, 2018, Gura Humorului, Romania
Romanian Jurnal of Rare Disease, supl. 1/2018, pg 49, ISSN 2068-5882, www.rjrd.ro
A new case involving 2q13 microduplication associated with autism spectrum disorder, intellectual disability and dysmorphic features
I. O. Focsa, L.C.Bohiltea, I. Streata, S. Sosoi, M. Ioana,M.Budisteanu, 51st *European Society of Human Genetic Conference, June 16-19, 2018, Milan, Italy, Eur J Hum Genet* **27,** 1–688 (2019). https://doi.org/10.1038/s41431-019-0404-7; *ISSN 1476-5438*
Clinical and molecular variability in Niemann-Pick type B
I. Focsa, S. Macovei, Dobrescu, L. Bohiltea, M. Budisteanu *American Society of Human Genetics Annual Meeting*, October 17-21, 2017, Orlando, USA
The impact of Moyamoya syndrome in a patient with neurofibromatosis type I
IO Focsa, LC Bohiltea, C Burloiu, M Budisteanu, XXIII World Congress of Neurology, September 16-21, 2017, Kyoto, Japan, *Journal of the Neurological Sciences,* Volume 381, Supplement, 15 October 2017, Pages 381–382, Doi http://dx.doi.org/10.1016/j.jns.2017.08.3292 ISSN 0022-510X

Genomic variants in intellectual disability in siblings
Magdalena Budisteanu, Sorina Mihaela Papuc, Raluca Colesniuc, Raluca Grozavescu, Diana Barca, Bogdan Budisteanu, Carmen Burloiu, **Ina Ofelia Focsa**, Iuliana Dobrescu, Andreea-Cristina Tutulan-Cunita, Aurora Arghir
11th European Cytogenetics Conference 1-4 July, 2017, Florence, Italy
Molecular cytogenetics 2017 10(Suppl 1):20, https://doi.org/10.1186/s13039-017-0319-3
ISSN: 1755-8166
ACSL4 intragenic deletion in a boy with complex phenotype
I.O. Focsa, L.C.Bohltea, B.Wollnik, M.Budisteanu
50th European Society of Human Genetics Conference, May 27–30, 2017, Copenhagen,Denmark, Eur J Hum Genet 26, 820 1023 (2019). https://doi.org/10.1038/s41431-018-0248-6; ISSN 1476 5438
Clinical characterization of a new case with chromosome 3 terminal microdeletion, involving CHL1 gene
I.O Focsa, M Ioana, I Streata, S Serban Sosoi, A Pirvu, LC Bohiltea, M Budisteanu
8th Europaediatrics Congress, 7-10 June, 2017, Bucharest, Romania
BMJ Journals: Archives of Disease in Childhood Jun 2017, 102 (Suppl 2) A88; DOI: 10.1136/archdischild-2017-313273.228; ISSN 1468-2044
Challenges in clinical interpretation of gfap gene variant in a child with with Alexander disease
Magdalena Budisteanu, Sorina Mihaela Papuc, **Ina Ofelia Focsa**, Dana Craiu
8th Europaediatrics Congress, 7-10 June, 2017, Bucharest, Romania BMJ Journals: Archives of Disease in Childhood Jun 2017, 102 (Suppl 2) A85; DOI: 10.1136/archdischild-2017-313273.219 ISSN 1468-2044
Clinical findings in a girl with 8p23.1-p22 duplication
I.O Focsa, I. Streata, S. Serban Sosoi, L.C Bohiltea, M. Ioana, M. Budisteanu
American Society of Human Genetics Annual Meeting, October 18-22, Vancouver, Canada
Epileptic seizures associated with chromosomal abnormalities
M. Budisteanu, A. Tutulan-Cunita, S. Papuc, C. Iliescu, C. Burloiu, **I. Focsa**, D.Craiu, I. Minciu, D. Barca, B. Budisteanu, M. Cristea, I. Borcan, A. Arghir
The European Human Genetic Conference 2016, May 21-24, 2016, Barcelona, Spain
European Journal of Human Genetics 24(1) 191, 2016, ISSN 1018-4813
Copy number variants in a cohort of Romanian patients with neurodevelopmental disorders
Arghir, S. Papuc, C. Iliescu, D. Craiu, D. Barca, B. Budisteanu, **I. Focsa**, M. Cristea, I. Borcan[1], M. Budisteanu, A. Tutulan-Cunita
The European Human Genetic Conference 2016, May 21-24, 2016, Barcelona, Spain
European Journal of Human Genetics 24(1): 439-440,2016, ISSN 1018-4813
Chromosome 8p abnormalities associated with severe global developmental delay: report of two cases
I. O. Focsa, M. Budisteanu, A. Tutulan-Cunita, S.M. Papuc, C. Burloiu, A. Arghir
The 13th International Congress of Human Genetics 2016, April 3-7, 2016, Kyoto, Japan
Duplication of 1q21.3-q22 in a child with developmental delay, dysmorphic features and obesity
I. O. Focsa, A. Tutulan-Cunita, S.M. Papuc, M. Budisteanu, D. Le Tessier, A. Lebar, J-M. Dupont, A. Arghir
American Society of Human Genetics Annual Meeting, 6-10.10.2015, Baltimore, USA
1p36 deletion syndrom – a rare cause of intellectual disability in children
M. Budisteanu, A. Tutulan-Cunita, S.M. Papuc, **I. Focsa**, D. Barca, O. Tarta-Arsene, C. Iliescu, I. Acsinte, N. DiDonato, K. Hackmann, A. Arghir
2nd National Conference of Romanian Paediatric Neurology Society, November 5-7,2015, Bucharest, Romania
Management of pediatric patients with neuropsychiatric disorders: genetic and genomic techniques as diagnostic tools
Tutulan-Cunita, A. Arghir, **I. O. Focsa**, S.M. Papuc, D.Craiu C. Iliescu, I. Borcan, M. Cristea, M. Budisteanu
The XIIth Paediatric National Congress National,September 2-5 2015, Timisoara,
Paediatrician Journal XVIII (S2): 85 (2015), ISSN 2360-4557.
Diagnosis and management of Romanian children with Williams-Beuren syndrome
I. O. Focsa, A. Arghir, S. M. Papuc, A. C. Tutulan-Cunita, C. Iliescu, D. Craiu, C. Burloiu, M. Boer, B. Budisteanu, M. Budisteanu
The European Human Genetic Conference 2015, June 6-9, Glasgow, Scotland, United Kingdom

European Journal of Human Genetics 23(1): 195-196, 2015, ISSN 1018-4813
Genomic technologies – investigative tools in neurodevelopmental disorders
A. Tutulan-Cunita, M. Budisteanu, **I.O. Focsa**, S.M. Papuc, C. Pomeran, C. Burloiu, I. Minciu, B. Budisteanu, D. Craiu, A. Arghir
X^th International Congress of Public Association of geneticists and breeders of Republic of Moldova, June 28- july 1, 2015, Chisinau, Moldova
Phelan-McDermid syndrome in two siblings with complex phenotype
Dr. Magdalena BUDIŞTEANU, Dr. Andreea-Cristina TUTULAN-CUNIŢA, Drd. Sorina Mihaela PAPUC, Dr. **Ina Ofelia FOCŞA**, DR. Silviu VULTUR, Dr. Aurora ARGHIR, Dr. Raluca Grozavescu
12th European Paediatric Neurology Society Congress, May 27-30, 2015, Vienna, Austria
European Journal of Paediatric Neurology, 19:S76–S77, 2015, ISSN 1090-3798
Comparative genomic hybridization microarray-based in neurodevelopmental disorders investigation in children
Dr. Aurora ARGHIR, Dr. Magdalena BUDIŞTEANU, **Dr. Ina Ofelia FOCŞA**, Drd. Sorina Mihaela PAPUC, Dr. Carmen BURLOIU, Dr. Catrinel ILIESCU, Dr. Diana BARCA, Dr. Marioara CRISTE A, Dr. Ioana BORCAN, Dr.Andreea-Cristina TUTULAN-CUNIŢA
Paediatric National Conference 2015, April 1-4, 2016, Bucharest, Romania
Angelman Syndrome - imprinting disorder with severe clinical impairment: diagnosis and management *(Oral presentation)*
Dr. Ina FOCŞA, Dr. Magdalena BUDIŞTEANU, Drd. Sorina Mihaela PAPUC, Dr. Andreea-Cristina TUTULAN CUNIŢA, Dr. Catrinel ILIESCU, Dr. Diana BARCA, Dr. Bogdan BUDIŞTEANU Dr. Dana CRAIU, Dr. Monica BOER, Dr. Aurora ARGHIR
2nd European Imprinting Disorders School, May 4-6, 2015, Paris, Fra

ANNEXES

April, 2022