P2 – IVB

ARGHIR Aurora, MD, PhD, Senior Researcher
Date of birth: 15th June 1970
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Gender: Female

Education:

2008	PhD in Medicine (Genetics) at "Carol Davila" University of Medicine and Pharmacy, Bucharest
2005	Board certified in Laboratory Medicine
1995	Medical Degree, "Carol Davila" University of Medicine and Pharmacy, Bucharest

Previous and current positions:

2010–present	Head of Medical Genetics Laboratory, Victor Babes National Institute of Pathology, Bucharest
2016–present	Lecturer, "Carol Davila" University of Medicine and Pharmacy, Bucharest
2001–2010	MD, Researcher, Medical Genetics Laboratory, Victor Babes National Institute of Pathology, Bucharest

Research interest: - identification of genetic changes in patient samples with neurodevelopmental disorders, with a main focus on intellectual disability, autism spectrum disorders and epilepsy syndromes, by microarrays and targeted NGS approaches.

Grants:

Participation in 23 national and international research projects (6 as PI, 17 as key member or researcher). Participation in COST action BM1004, "Enhancing the scientific study of early autism: A network to improve research, services and outcomes" and COST action CA16118 "European Network on Brain Malformation", as Management Committee member for Romania.
Previous projects : Genomic and postgenomic data analysis and data management – essential tools in precision medicine (Romanian MR - PN 16.22.05.01; 2016-2017); Genomic profile investigation by aCGH in children with cognitive and behavior problems (Romanian MR - PN 09.33.02.03; 2010-2015 – Principal Investigator); Bilateral Actions Romania-France – Structural genomic variants in ASDs (Romanian MR – Brancusi program; 2010-2012 - Principal Investigator); Clinical, citogenetic and molecular multidisciplinary research in mental retardation syndromes associated with congenital anomalies (Romanian MR – PN 42/130-1; 2008-2011). Current national research project focused on epilepsy genomics, Romanian Executive Agency for Higher Education and Research (PED 249/2017).

Awards:

Romanian Executive Agency for Higher Education and Research Prize (2016); ESHG Conference Mobility Awards (2005; 2008).

5 SELECTED PEER-REVIEWED PUBLICATIONS:

1. Lalli M.A., Jang J., Park J.C., Wang Y., Guzman E., Zhou H., Audouard M., Bridges D., Tovar K.R., Papuc S.M., Tutulan-Cunita A.C., Huang Y., Budisteanu M., **Arghir A**., Kosik K.S. - Haploinsufficiency of BAZ1B contributes to Williams syndrome through transcriptional dysregulation of neurodevelopmental pathways, Hum. Mol. Genet., 2016; 25(7):1294-306.
2. S.M. Papuc, K. Hackmann, J. Andrieux, C. Vincent-Delorme, M. Budisteanu, **A. Arghir**, E. Schrock, A.C. Tutulan-Cunita, N. Di Donato - Microduplications of 3p26.3p26.2 containing CRBN gene in patients with intellectual disability and behavior abnormalities, Eur. J. Med. Genet. (IF 1,46) 58(5): 319–323 (2015).
3. Khan NZ, Gallo LA, **Arghir A**, Budisteanu B, Budisteanu M, Dobrescu I, et al. Autism and the grand challenges in global mental health. Autism Res. 2012;5(3):156–9. doi: 10.1002/aur.1239.
4. Tutulan-Cunita AC, Papuc SM, **Arghir A**, Rötzer KM, Deshpande C, Lungeanu A, Budisteanu M. 3p Interstitial Deletion: Novel Case Report and Reiew. J Child Neurol. 2012; 27(8):1062-6.
5. Budisteanu M, Papuc SM, Tutulan-Cunita AC, Budisteanu B, Weis E, **Arghir A**, Zechner U, Bartsch O. De-novo Williams-Beuren and inherited Marfan syndromes in a patient with developmental delay and lens dislocation. Clin Dysmorphol. 2017 Jul;26(3):187-189.