

NIKOLAOS M. MARINAKIS, MSc, PhD, ErCLG

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Assistant Professor of Genetics, Clinical Laboratory Geneticist

CLINICAL AND LABORATORY WORK	RESEARCH WORK
<ul style="list-style-type: none">Standardization of molecular analysis methodologies and their application at service providing levelMolecular diagnosis of rare genetic diseases (Neurodevelopmental, skeletal, congenital anomalies, skin, nephrological, cardiovascular, ophthalmological, etc.) in more than 2000 cases (2017-2025)	<ul style="list-style-type: none">Study and identification of molecular pathogenicity mechanism in various monogenic diseasesDevelopment and application of molecular analysis methods for diagnostic and prenatal purposes

WORK EXPERIENCE

Laboratory of Genetics, Medical School, Democritus University of Thrace **Current position** **Assistant Professor of Genetics**

Current composition of the laboratory: 1 PhD student
Projects: Molecular investigation of rare genetic disorders

Laboratory of Medical Genetics, Medical School, St. Sophia's Children Hospital, NKUA **04/2022 – 07/2025** **Scientific Research Associate – Genome analyst:** Supervisor of Clinical Next Generation Sequencing: Clinical Whole Exome Sequencing data analysis, Variant interpretation in rare diseases

Genome Diagnostics, Laboratory of Genome Bioinformatics, Radboud University Medical Center, Nijmegen **08/2023** **Post-doc Researcher – ESHG observership programme awardee:** “WES, WGS data analysis, variant interpretation, CNV analysis, Long Read sequencing, bioinformatic pipelines, novel genetic mechanisms”, Laboratory of Genome Bioinformatics, Prof. Christian Gilissen (Christian.Gilissen@radboudumc.nl)

Laboratory of Medical Genetics, Medical School, St. Sophia's Children Hospital, NKUA **11/2017 – 04/2022** **PhD candidate:** “Next Generation Sequencing as a diagnostic-laboratory tool in rare genetic diseases and evaluation of the findings with bioinformatics and laboratory methods”. (<https://pergamos.lib.uoa.gr/uoa/dl/object/3197923>), Professor J. Traeger-Synodinos (jtraeger@med.uoa.gr)

Techniques:

Bioinformatic analysis of NGS data, Variant interpretation, clinical application of NGS, RNA sequencing

Laboratory of Medical Genetics, Medical School, St. Sophia's Children Hospital, NKUA **09/2016 – 10/2017** **Master student:** “Pilot application of Next Generation Sequencing (NGS) in all exons of the genome (Whole Exome Sequencing) for standardization and evaluation methodology for diagnostic use”. (<https://pergamos.lib.uoa.gr/uoa/dl/object/2019565>), Professor J. Traeger-Synodinos (jtraeger@med.uoa.gr)

Techniques:

Next Generation Sequencing (Library preparation, Data analysis), PCR, Sequencing, MLPA, DNA extraction.

Molecular Genetics Laboratory, Molecular Biology and Genetics Dept, DUTH **09/2012 – 10/2014** **Bachelor Thesis:** “Functional study of polymorphisms p.Asp115Tyr and p.Phe175Leu of the NAT1 enzyme in the primate *Macaca mulatta* (Rhesus)”, Assistant Professor S. Boukouvala (sboukouval@mbg.duth.gr).

Techniques:

Recombinant DNA (Cloning), Site-Directed Mutagenesis, Primer Design, PCR, Agarose gel electrophoresis, Bacterial Transformation, Miniprep, Protein Expression and Purification, SDS-PAGE, Enzymatic activity assay, Western Blotting.

EDUCATION

PhD in Medical Genetics - HFRI PhD Scholarship **2017 – 2022** Laboratory of Medical Genetics, Medical School, St. Sophia's Children's Hospital, National and Kapodistrian University of Athens

MSc in Clinical Biochemistry and Molecular Diagnostics. **2015 – 2017** Department of Biology, National and Kapodistrian University of Athens

BSc Molecular Biology & Genetics **2010 – 2014** Department of Molecular Biology and Genetics, Democritus University of Thrace

SHOLARSHIPS - FELLOWSHIPS

- **European Society of Human Genetics**, Conference National Fellowship, European Human Genetics Conference 2025, Milan, Italy, 2025
- **European Society of Human Genetics**, Basics in Human Genetics ErCLG course, Figuera Da Foz, Portugal, 04-08 September 2023
- **European Society of Human Genetics, Observership programme 2023**, Genome Diagnostics, Laboratory of Genome Bioinformatics, Radboud University Medical Center, Nijmegen, the Netherlands
- **European Society of Human Genetics**, Conference Fellowship, European Human Genetics Conference 2022, Austria, Vienna, 11-14 June 2022
- **Hellenic Foundation for Research and Innovation**, Scholarships to PhD Candidates, 2019-2022
- **European Society of Human Genetics**, 30th course jointly organized by ESGM, ESHG and CEUB - Clinical Genomics and NGS, Bertinoro (Italy), 30 April – 5 May 2017

RESEARCH PROGRAMMES

- **National research network to elucidate the genetic basis of Alzheimer's and Parkinson's neurodegenerative diseases, detect reliable biomarkers, and develop innovative computational technologies and therapeutic strategies underpinning precision medicine**, MIS 5149305, TAEDR-0534767: Scientific Research Associate, Next Generation Sequencing data analysis and variant interpretation, 2023-2024
- **3billion Exome Sequencing Research Study 2022**: Principal Investigator, End the diagnostic odyssey grant
- **Erasmus+ 2018-2020**: Researcher, Connecting Genes to Rare Diseases through New Generation Sequencing (NGS) Technology and Advanced Teaching Methods, University of Athens (Greece), University of Antwerpen (Belgium), Istituto Ortopedico Rizzoli (Italy). <https://www.ngenes.eu/>

TEACHING

Undergraduate Medical Students, DUTH

2025-Today

- Genetics
- Biology
- Clinical and Applied Genetics

E-learning: “Next Generation Sequencing - NGS: Bioinformatic analysis, variant interpretation and clinical application”, NKUA 2024

- Academic director of the e-learning program

MSc “Medical Genetics: Clinical and Laboratory Direction”, Medical School, NKUA

2017-Today

- Next Generation Sequencing: Clinical application, Bioinformatic analysis of data
- Variant evaluation, Databases and in-silico analysis
- Next Generation Sequencing: Bioinformatic analysis of data, cases investigation

Master Thesis supervisor

- Study of the frequency of carriers of pathogenic variants in autosomal recessive genes with the aim of using them for reproductive selection
- Molecular investigation of type I and II collagens and pathogenic mechanisms of the respective collagenopathies

Master Thesis examination committee

- Analysis of genome-wide exon sequencing data in a child with psychomotor retardation and brachydactyly
- Secondary findings in clinical next generation sequencing

PhD students guidance

F-N. Tilemis, Laboratory of Medical Genetics (NKUA), Optimizing the diagnostic performance of Next Generation Sequencing (NGS) in Medical Genetics, through bioinformatics and laboratory procedures (2024)

SCIENTIFIC ACTIVITIES	LANGUAGES
<ul style="list-style-type: none"> ○ ClinGen Retina Gene Biocurator ○ PanelApp Reviewer, panelapp.genomicsengland.co.uk ○ Journal Reviewer, Molecular Syndromology (Karger publications), Molecular Diagnosis and Therapy (Springer publications) 	<p>Greek English</p>

BIBLIOGRAPHY - PEER-REVIEW PAPERS IN JOURNALS

1. Kekou K, Papadopoulos C, Svingou M, Chrysanthou-Piterou M, Nitsa E, Veltra D, **Marinakis N**, Tilemis FN, Dimitrios P, Arnaoutoglou M, Moschou M, Xirou S, Bakirtzis C, Tsvigoulis G, Papadimas GK, Sofocleous C. **Insights into the heterogeneity of oculopharyngeal muscular dystrophy**. *Neurogenetics*. 2025 Sep 24;26(1):68. doi: 10.1007/s10048-025-00849-0.
2. Argyropoulou OD, **Marinakis NM**, Charalampopoulos G, Tilemis FN, Liasis N, Manoussakis MN. **IL-1R/IL-6R blockade for null NLRP12 variant-carrying Takayasu arteritis**. *Rheumatology (Oxford)*. 2025 Oct 25:keaf567. doi: 10.1093/rheumatology/keaf567.
3. Chapman KA, Ullah F, Yahiku ZA, Khan S, Kodiparthi SV, Kellaris G, White HG, Powell AT, Correia SP, Stödtberg T, Sofocleous C, **Marinakis NM**, Fryssira H, Tsoutsou E, Traeger-Synodinos J, Accogli A, Scirucchio V, Salpietro V, Striano P, Muss C, Keren B, Heron D, Berger SI, Pond KW, Sirimulla S, Davis EE, Bhattacharya MRC. **Pathogenic variants in TMEM184B cause a neurodevelopmental syndrome associated with alteration of metabolic signaling**. *Am J Hum Genet*. 2025 Oct 2;112(10):2381-2401. doi: 10.1016/j.ajhg.2025.08.004.
4. Ni, C., Wei, Y., Vona, B., Park, D., Wei, Y., Schmitz, D. A., Ding, Y., Sakurai, M., Ballard, E., Li, L., Liu, Y., ..., Veltra D., **Marinakis NM**, ... Buszczak, M. (2025). **A programmed decline in ribosome levels governs human early neurodevelopment**. *Nature cell biology*, 10.1038/s41556-025-01708-8. Advance online publication. <https://doi.org/10.1038/s41556-025-01708-8>
5. **Marinakis, N. M.**, Kampouraki, A., Veltra, D., Tilemis, F. N., Vasilopoulou, M., Dokou, A., Georgiadou, E., Karavergou, E., Christolouka, M., Alexopoulos, A., Kirillidi, D., Goudesidou, M., Kosma, K., Sofocleous, C., & Makrythanasis, P. (2025). **Unraveling the Role of WDR91: Case Report of a Previously Unrecognized Clinical Entity**. *Clinical genetics*, 10.1111/cge.70012. Advance online publication. <https://doi.org/10.1111/cge.70012>
6. Zacharogianni, M., Papandreou, N. C., **Marinakis, N. M.**, Tilemis, F. N., Traeger-Synodinos, J., & Palioura, S. (2025). **Clinical and Structural Characterization of a Novel TGFBI Mutation Linked to a Lattice Corneal Dystrophy Variant in a Greek Family**. *American journal of ophthalmology*, 274, 112–121. <https://doi.org/10.1016/j.ajo.2025.03.003>
7. Kollia, E., Kokkinou, E., Outsika, C., Koltsida, G., Zouvelou, V., Vontzalidis, A., Dalivigka, Z., Veltra, D., Sofocleous, C., **Marinakis, N. M.**, Tilemis, F. N., Yapijakis, C., Anagnostopoulou, K. K., Loukas, Y. L., Spanou, M., Dinopoulos, A., Nikaina, E., Skiathitou, A. V., Siahianidou, T., Georgiadou, E., ... Pons, R. (2025). **Motor phenotyping in a Greek cohort of patients with neonatal and infantile onset developmental and epileptic encephalopathy**. *European journal of paediatric neurology: EJPn: official journal of the European Paediatric Neurology Society*, 55, 1–8. <https://doi.org/10.1016/j.ejpn.2025.03.001>
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9. **Marinakis, N. M.**, Tilemis, F. N., Veltra, D., Svingou, M., Sofocleous, C., Kekou, K., Kosma, K., Kampouraki, A., Kontse, C., Fylaktou, I., Sertedaki, A., Kanaka-Gantenbein, C., Traeger-Synodinos, J., & Makrythanasis, P. (2025). **Estimating at-risk couple rates across 1000 exome sequencing data cohort for 176 genes and its importance relevance for health policies**. *European journal of human genetics: EIJHG*, 33(1), 65–71. <https://doi.org/10.1038/s41431-024-01726-4>
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2. Tilemis F.N., Selenti N., **Marinakis N.**, Kosma K., Christodoulaki V., Makrythanasis P., Sofocleous C., **Phelan-McDermid 22q13.31 deletion originating from a maternal balanced translocation**, European Human Genetics Conference (ESHG), Milan, Italy, 2025
3. Kakourou G., Mamas T., Chatzirvasanis S., Kosma K., **Marinakis N.**, Veltra D., Kampouraki A., ... Sofocleous C., Vrettou C., Makrythanasis P., **Experience with clinical and technical complexities of Preimplantation Genetic Testing for monogenic disease (PGT-M)**, European Human Genetics Conference (ESHG), Milan, Italy, 2025
4. Tsentemidou E., Kosma K., **Marinakis N.**, Tilemis F.N., Christodoulaki V., Kampouraki A., Saranti S., Kostaridou S., Traeger-Synodinos J., Makrythanasis P., **The burden of combined sensory impairment and two distinct genetic diseases in a single patient**, European Human Genetics Conference (ESHG), Milan, Italy, 2025
5. Brantzos T., Mitrakos A., Tzetsis M., Kosma K., **Marinakis N.**, Tilemis F.N., Sofocleous C., Makrythanasis P., **Genetic profile of Wilson's disease in a Greek cohort**, European Human Genetics Conference (ESHG), Milan, Italy, 2025
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7. Kosma K., Kampouraki A., Liakopoulou-Tsitsipi T., Veltra D., Marinakis N., Tilemis F.N., Christodoulaki V., Tsentemidou E., Selenti N., Mitrakos A., Tzetsis M., Solokleous C., Makrythanasis P., **From VUS to pathogenic: reclassifying a KDM6B variant through clinical and genomic collaboration**, European Human Genetics Conference (ESHG), Milan, Italy, 2025
8. Kekou K., Papadopoulos C., Svingou M., Chrysanthou-Piterou M., Nitsa E., Veltra D., **Marinakis N.**, Tilemis F.N., Parissis D., Arnaoutoglou M., Moschou M., Xirou S., Bakirtzis C., Tsigvoulis G., Papadimas G.K., Makrythanasis P., Sofocleous C., **Mitochondria-based anticipation effect. Insights from an Oculopharyngeal Muscular Dystrophy Cohort**, European Human Genetics Conference (ESHG), Milan, Italy, 2025
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10. Christodoulaki V., Kosma K., Stergiou N., Chronis C., Filippou O., **Marinakis N.**, Tilemis F.N., Kampouraki A., Karava V., Botsa E., Traeger-Synodinos J., Kanaka-Gantenbein C., Makrythanasis P., **Case report of a pediatric patient with acute kidney injury and tubular proteinuria**, European Human Genetics Conference (ESHG), Milan, Italy, 2025
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12. Tilemis F.N., **Marinakis N.**, Veltra D., Svingou M., Kekou K., Mitrakos A., Tzetsis M., Kosma K., Makrythanasis P., Sofocleous C., Traeger-Synodinos J., **Increasing the diagnostic yield of Whole Exome Sequencing (WES) through CNV detection**, European Human Genetics Conference (ESHG), 2023
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14. Hufnagel B. R., Goldstein J., Hankey W., Sergouniotis P., Lee K., Arno G., on behalf of the **ClinGen Retina Gene Curation Expert Panel**, **A gene-centric nomenclature and classification system for inherited retinal disorders**, Association for Research in Vision and Ophthalmology, Inc. (ARVO) Annual Meeting, New Orleans, La, 2023
15. Loukas Y., Spanou M., Anagnostopoulou K.,...Kosma K., **Marinakis N.**, Voudris K., Korona A., Dragoumi P., Katerelos A., Niotakis G., Dinopoulos A., **RARE-ID: An Eastern European project for the identification of genetic causes of pediatric autism and Undiagnosed Neurologic Disorders via WGS; preliminary results.** **European Structural and Investment Funds 2014-2020**, 15th European Paediatric Neurology Society Congress, 2023
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