STATEMENT

REGARDING: competition for the academic position of "Associate Professor"—one 0.5-FTE position in Higher Education Field 7. "Health Care and Sports," in Professional Area **7**.1. "Medicine" for the needs of the Department of Medical Genetics at the Faculty of Medicine, Medical University of Varna, and one full-time position for the Laboratory of Medical Genetics at the University Hospital "St. Marina," announced in the "State Gazette" - No. 6 / January 16, 2026

OPINION PREPARED BY: Prof. Savina Petrova Hadzhidekova, MD, PhD, MF, MU – Sofia, appointed as a member of the Scientific Jury pursuant to Order No. R109-130/March 16, 2026, of the Rector of MU – Varna, and a decision under Protocol No. 55/March 4, 2026, of the Faculty Council of the Faculty of Medicine; selected to draft an opinion at the 1st meeting of the Scientific Jury.

CANDIDATE: Dr. Mari Ara Khachmerian-Andreeva, MD

I. General presentation of the procedure and the candidate

The opinion was prepared in accordance with Order R109-130/16.03.2026** of **the Rector of Medical University – Varna.

The submitted set of materials complies with the Regulations for the Implementation of the Law on the Development of Academic Staff in the Republic of Bulgaria, Article 137, Paragraph 2 of the Regulations on the Conditions and Procedures for the Award of Academic Degrees and the Holding of Academic Positions at Medical University (MU) – Varna:

In preparing this review, the requirements of the Law on the Development of Academic Staff in the Republic of Bulgaria (ZRASRB) and the Regulations for its implementation have been observed, as well as the specific criteria for the award of academic degrees and the holding of academic positions at MU-Varna.

Brief summary of the candidate's professional and academic background

Mari Ara Khachmerian-Andreeva completed her higher education with a Master's degree in Medicine between 2005 and 2011, after which she focused her professional and scientific development on the field of medical genetics. From 2012 to 2017, she was an independent PhD student at the Department of Medical Genetics at Medical University – Varna, where she defended a dissertation on maternal biochemical screening as a method for prenatal genetic prevention. She subsequently continued her research as a PhD student in the same department (2024–2025), developing a topic related to genetic counseling for patients with probable tumor-predisposing syndromes.

In 2018, she completed a specialty in medical genetics, which solidified her professional profile as a clinician and researcher in this highly specialized field. Her professional career is closely linked to the University Hospital "St. Marina" in Varna, where she began working as a physician in the Medical Genetics Laboratory in 2012, and since 2019 has held a leadership position as head of the laboratory.

Her academic career has developed in parallel with her clinical practice. From 2012 to 2018, she served as an assistant professor, and since 2018, she has been a senior assistant professor in the Department of Medical Genetics at the Medical University of Varna. In this role, she actively participates in the training of medical students (in Bulgarian and English), as well as other healthcare professionals, including medical laboratory technicians, midwives, and nurses.

Her research activity is intensive and consistent, including a significant number of publications—52 scientific articles and 71 conference abstracts. She participates in a number of scientific forums and is part of research teams on four projects under the "Science" Fund of the Medical University of Varna, as well as on a national project aimed at implementing innovative technologies in the management of oncological and rare diseases.

II. Scientific Activity and Contributions

Habilitation Thesis

Dr. Mari Hachmerian Andreeva's publication record, as presented in the list of equivalent publications accompanying her habilitation thesis, can be assessed as systematic, sufficient in volume, and thematically sound, in line with her scientific specialization in medical genetics.
For the purposes of habilitation, 10 scientific publications are presented, which form the corpus of equivalent works. They cover the period from 2013 to 2021 and reflect a consistent development of the candidate's scientific interests.
Regarding the type of scientific output, a significant portion of the publications consists of clinical cases and case reports, supplemented by analytical and comparative studies. This profile is characteristic of medical genetics and reflects the direct link between the candidate's scientific and diagnostic-consultative activities.
Some of the works have been published in international peer-reviewed scientific journals, which contributes to enhancing the candidate's scientific visibility and integration into the international scientific community. In parallel, there are also publications in Bulgarian scientific journals, demonstrating a commitment to the development of national medical science.
The report covers the candidate's scientific output, based on 10 equivalent publications produced between 2013 and 2021, clearly defining the main thematic areas and their relevance. The research is concentrated in the field of medical genetics, with a focus on rare diseases, genomic medicine, and reproductive health.
The introduction emphasizes alignment with international scientific priorities, including WHO policies and current trends in genomic medicine, which lends strategic significance to the presented research.

Main Research Areas and Contributions

A. Diagnosis and clinical characterization of rare genetic syndromes

This is the leading area of the candidate's scientific work. The contributions are expressed in:

- an in-depth analysis of diagnostic challenges in rare syndromes;
- expanding knowledge of phenotypic variability;
- support for early and accurate diagnosis.

Specific syndromes (Kabuki, Cornelia de Lange, Bardet-Biedl, 16p11.2 duplication) are examined, with an emphasis on overcoming the so-called "diagnostic odyssey" in these patients. This contribution has high practical value, especially in the context of clinical genetics.

B. Genomics and Medical-Genetic Counseling

The second area is related to contemporary aspects of genetic counseling. Contributions include:

- analysis of the transformation of counseling in the context of genomic medicine;
- formulation of approaches for interpreting and communicating complex genetic results (including variants of unknown significance—VUS);
- adaptation of international models to Bulgarian practice.

This aspect demonstrates theoretical and applied depth, as well as consideration of the ethical and social dimensions of modern medicine.

C. Reproductive Genetics

The third area includes research on genetic factors in recurrent spontaneous abortions. The contributions are expressed in:

- providing original data on the Bulgarian population;
- studying immunogenetic markers (HLA-G, TNF-alpha);
- analysis of thrombophilic genetic polymorphisms.

These results have direct application in clinical practice, supporting a personalized approach to the management of pathological pregnancy.

III. Publication Activity

In addition to the habilitation thesis comprising 10 publications and the scientific publications used to obtain the educational and scientific degree PhD," the candidate presents the following publications:

- 6 full-text publications
- 14 abstracts from presentations at international scientific conferences, published in scientific journals, peer-reviewed, and indexed in world-renowned scientific databases
- 1 published book based on a defended dissertation for the award of the academic degree of "PhD"

IV. Research Areas and Contributions

The candidate's contributions are multifaceted and combine a clinical focus, a laboratory-diagnostic approach, and practical algorithms for clinical management. They are organized into several interrelated areas: oncogenetics and hereditary tumor-predisposing syndromes, prenatal diagnosis and genomic technologies, cytogenetics and molecular diagnostics in oncohematology, clinical dysmorphology and rare genetic diseases, as well as ethical aspects, telegenetics, and education.

In terms of content, the contributions focus on improving diagnostic accuracy, introducing and evaluating modern genetic technologies, optimizing genetic counseling, and raising awareness among at-risk patients and families. This determines their significance for both the academic field and daily medical practice.

1. Oncogenetics and Hereditary Tumor-Predisposing Syndromes

This area is central to the candidate's habilitation profile. A significant original contribution is the development of guidelines for improving medical-genetic counseling for patients with a probable hereditary cancer-predisposing syndrome. The emphasis on detailed family history, referral criteria, DNA analysis, and cascade counseling for at-risk relatives is directly relevant to the early identification of individuals with an increased risk of cancer.

The scientific and practical value is also reflected in the assessment of the diagnostic performance of targeted panel testing using NGS technologies in hereditary breast and ovarian cancer and Lynch syndrome. The importance of expanded panels for detecting pathogenic variants in genes with moderate penetrance is highlighted, which provides a basis for more precise clinical management. An additional applied contribution is the dissemination of reliable and accessible information to patients through the book "The Genetic Code of Cancer."

2. Prenatal Diagnosis and Genomic Technologies

Advances in the field of prenatal diagnosis reflect the technological shift from biochemical screening to non-invasive prenatal testing. The analysis of a large volume of clinical data —over 17,000 cases over an eight-year period—has high practical value for optimizing algorithms for mass prenatal screening and for evaluating the effectiveness of combined biochemical screening during the first trimester.

An original scientific contribution is the socio-psychological analysis of Bulgarian women's attitudes toward NIPT. Identifying unrealistic expectations regarding the diagnostic capabilities of the tests and highlighting the need for mandatory pre-test counseling demonstrate a mature and responsible approach to the implementation of new genomic technologies. The applied aspect is complemented by an analysis of the change in the structure of invasive diagnostics following the introduction of NIPT.

3. Cytogenetics and Molecular Diagnostics in Oncohematology

The candidate presents contributions related to improving the diagnosis and prognostic assessment of hematologic neoplasms. The combined approach of conventional cytogenetics and interphase FISH is considered clinically significant, particularly in multiple myeloma, where a low mitotic index may limit the diagnostic value of classical cytogenetic methods.

The application of iFISH for detecting prognostically significant aberrations such as del17p13 and IGH rearrangements is important for risk stratification and clinical follow-up. The analysis of incidentally detected constitutional chromosomal abnormalities, which involves important ethical, diagnostic, and family-reproductive aspects, provides additional value.

4. Clinical dysmorphology and rare genetic diseases

In this area, the contributions demonstrate clinical expertise in complex and rare genetic disorders. The presented phenotype-genotype correlations in Kabuki syndrome, Cornelia de Lange syndrome, RASopathies, neurofibromatosis type 1, Noonan syndrome, and 16p11.2 duplication are significant for improving diagnostic pathways and implementing a multidisciplinary approach.

Separately, the innovative contribution related to the application of facial recognition software in the diagnostic algorithm should be emphasized. This points toward modernizing daily clinical practice and toward earlier recognition of rare syndromes in patients with dysmorphic features and complex symptoms.

5. Ethical Issues, Telegenetics, and Education

Contributions in this area are timely and reflect the contemporary challenges facing medical communication. The study of the transformation of genetic counseling during COVID-19 and the transition to remote work is unique to Bulgarian practice. The distinction between cases suitable for remote consultation and situations where in-person contact remains indispensable is well-founded.

Assessing patients' understanding of genetic information also has scientific and practical value. Findings regarding the risk of overinterpreting results without qualified post-test counseling are particularly important for reducing unnecessary anxiety and inappropriate medical interventions.

The contributions presented can be considered original, as they include original research, analyses of real clinical and laboratory data, the introduction and evaluation of modern diagnostic approaches, and the formulation of practical guidelines for medical genetics. Of particular significance is the link between the scientific results and their practical applicability in clinical counseling, prevention, diagnosis, and follow-up of at-risk patients and families.

V. Citation and Scientific Recognition

The academic record submitted lists a total of 7 citations, reflecting the candidate's research activity. The citations provided yield 105 points, with a required minimum of 80 points. Therefore, the criteria for groups D10–D12 have been met. The cited references are in scientific sources that reflect the visibility of the candidate's scientific output in the international scientific literature.

VI. Participation in Scientific Forums and Projects

The candidate reports 34 participations with poster presentations. These include national forums on rare diseases, pediatrics, and medical genetics, as well as international conferences focused on human genetics, prenatal diagnosis, cytogenomics, and dysmorphology.

Dr. Khachmerian-Andreeva has 11 recorded oral presentations. The topics of her presentations demonstrate a practical focus on genetic counseling, personalized medicine, hereditary thrombophilia, oncogenetics, and the interdisciplinary application of medical genetics.

The report lists 9 creative assignments, specializations, and qualification courses abroad. These contribute to the candidate's professional development in the fields of medical genetics, clinical cytogenetics, hereditary oncogenetics, dysmorphology, and reproductive and prenatal genetic diagnostics.

VII. Teaching and Educational Activities

The data presented demonstrate Dr. Mari Ara Khachmerian-Andreeva's significant and consistent teaching activity in the field of medical genetics. The candidate participates in the instruction of both Bulgarian-speaking and English-speaking students, which is a significant indicator of academic and teaching adaptability.

For the period 2019/2020–2024/2025, the candidate's total reported teaching load is 1,209 hours. This includes 29 hours of lectures and 1,180 hours of seminars. The breakdown by language of instruction is 663 hours in Bulgarian and 546 hours in English.

The majority of the teaching load consists of conducting laboratory sessions. This reflects direct involvement in the students' practical training and in the acquisition of applied knowledge and skills in the subject. The inclusion of recorded lectures complements the candidate's profile as an instructor who is also involved in theoretical instruction.

VIII. Conclusion

Based on the academic report on the publications, citations, and research profiles of Dr. Mari Ara Khachmerian-Andreeva, prepared by the Library of the Medical University – Varna, it can be concluded that the candidate meets the minimum scientometric requirements for the academic position of "associate professor" in Field 7, "Healthcare and Sports," Professional Area 7.1, "Medicine."

The data presented show compliance with the required indicators across the main categories, including the submission of a dissertation for the award of a PhD degree, a habilitation thesis in the form of scientific publications, publication activity in peer-reviewed and indexed journals, as well as the required number of citations.

Group of indicators	Reported result	Minimum requirement	Conclusion
A1	50 points	Required dissertation	Completed
B4	101.74 points; 10 full-text publications in peer-reviewed and indexed journals	Habilitation thesis / scientific publications	Completed
G5–G9	201.26 pages	Minimum 200 credits	Completed
D10–D12	7 citations; 105 points	Minimum 80 points	Fulfilled

Dr. Marie Ara Khachmerian-Andreeva meets the minimum national and institutional scientometric requirements for the academic position of “Associate Professor” in the field of “Medical Genetics.” The presented scientific output, habilitation thesis, publication activity, and citation index meet the required indicators; therefore, the candidate can be evaluated positively in terms of compliance with the minimum requirements for participation in the habilitation procedure.

Based on the foregoing, I give a positive review for the conferral of the academic rank of “Associate Professor” to Dr. Mari Ara Khachmerian-Andreeva, MD, in the field of higher education 7. Health Care and Sports, professional field 7.1. Medicine, specialty “Medical Genetics.”

May 25, 2026 Opinion prepared by: …….

Sofia Prof. Savina Hadzhidekova, MD