CAROL DAVILA UNIVERSITY OF MEDICINE AND PHARMACY, BUCHAREST
DOCTORAL SCHOOL
DOMAIN: MEDICINE

GENOMIC MEDICINE IN OBSTETRICS AND PRECISION ONCOLOGY: FROM PRENATAL DIAGNOSIS TO POPULATION-BASED CANCER GENOMICS IN ROMANIA

HABILITATION THESIS ABSTRACT

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Over the past two decades, genomics has simultaneously transformed obstetrics and oncology, supplying new tools for prevention, diagnosis and personalized therapy. Romania, however, is only beginning the systematic integration of these technologies into routine care. This thesis shows—through a critical synthesis of the author's work—how the two fields can be harmonized to create a molecularly informed continuum of care that stretches from the fetus in utero to adult patients with solid tumours. The overarching goal is to demonstrate how the author's results can underpin a national platform for integrative genomic medicine aligned both with European trends and with local healthcare needs.

Objectives and Methodology

The thesis pursues four specific aims: (1). Critical appraisal of ten key ISI-indexed papers selected for clinical relevance, journal impact and author position. (2). Demonstration of clinical translation, ranging from optimized prenatal screening algorithms to individualized oncologic therapies. (3). Documentation of the multidisciplinary approach underpinning study design and data interpretation. (4). Quantification of scientific impact through bibliometric indicators and its incorporation into a long-term development strategy.
A narrative-integrative methodology is employed: each publication is contextualised within international literature, correlated with Romanian clinical data, and then synthesised into a unified life-course vision of genomic medicine.

Original Scientific Contributions

The first thematic axis—materno-fetal genomics—covers implementation of non-invasive prenatal testing (NIPT) for common aneuploidies in a Romanian cohort, correlation of high-resolution ultrasound with genomic confirmation of anomalies (e.g., micrognathia–trisomy 18) and profiling of polymorphisms linked to obstetric complications such as thrombo-embolism and recurrent pregnancy loss. These studies substantially reduced the need for invasive procedures and accelerated clinical decision-making, paving the way for NIPT reimbursement by the National Health Insurance House (CNAS) in 2023. The second axis—precision oncogenomics—includes national-scale characterisation of homologous-recombination-deficiency (HRD) scores, identification of germline Lynch variants in colorectal cancer, and exploration of molecular resistance mechanisms in ovarian and colorectal tumours. The data supported formal inclusion of HRD testing in Romanian oncology guidelines and redirected roughly 25 % of eligible patients toward targeted therapies in pilot trials. Synergy between the two result

sets underpins the concept of life-course precision medicine: a reproductive-age genomic profile can act as a predictor of later gyn-onc risk, while oncogenetic testing feeds back valuable information for family counselling and pregnancy planning.

Professional, Educational and Socio-Economic Impact

Through the Romanian Cancer Registry and VIASAN projects, the author helped to establish the first university-based NGS facility in south-eastern Europe and to embed genetic variables in national cancer coding. Technology-transfer efforts shortened average time to confirmed genetic diagnosis from 45 to 18 days and cut unnecessary invasive obstetric procedures by 28 %, yielding an estimated socio-economic return of €3.4 for every euro invested. Educationally, a spiral curriculum in Medical Genetics, an e-learning platform and an interactive Atlas of Fetal Malformations have trained more than 200 residents and 14 PhD candidates, building an interdisciplinary school able to drive genomic-medicine implementation nationwide.

Strategic Directions 2025–2030

The proposed research agenda comprises three operational packages:

1. **Expansion of the ReNaVaFM mother–fetus registry** beyond 30 000 genomes and universal reimbursement of NIPT.
2. **Creation of the RO-WOMCAN atlas** based on 2 500 female tumours and validation of an AI algorithm for HRD classification.
3. **Development of the ISO 13485-certified AUTO-VAR pipeline** and launch of the FAIR-RO-Omics portal, integrated into the ELIXIR-Europe network.

The estimated €16 million budget will be secured through Horizon Europe, PNCDI IV, PNRR and industrial partnerships.

Conclusion

The thesis demonstrates that merging genomic obstetrics with precision oncogenomics is not merely feasible but essential for addressing Romania’s demographic and oncologic challenges. By combining original research, educational leadership and policy impact, the author proves the capacity to steer complex programmes and to train the generation that will embed genomic medicine as a cornerstone of public health. The 2025–2030 roadmap will move the country from genomic-technology consumer to data generator, ensuring long-term sustainability and competitiveness within the European healthcare landscape.