

<https://doi.org/10.29289/259453942025V36S11181>

1181 – Cascade genetic testing and early detection in families with hereditary breast and ovarian cancer within the Brazilian public health system

Elisângela de Paula Silveira-Lacerda¹, Lis de Paula Lacerda¹, Luiz Henrique Alves Costa¹, Igor Marques Cesário Calassa¹, Amanda Santos Limongi Santos Limongi¹, Rasivel dos Reis Junior¹, Délio Marques Conde¹, Rosemar Macedo Sousa Rahal¹

¹Universidade Federal de Goiás – Goiânia (GO), Brazil.

Introduction: Approximately 5–10% of breast and ovarian cancers are hereditary, predominantly associated with pathogenic variants in BRCA1, BRCA2, and TP53. Cascade genetic testing in at-risk relatives is a cost-effective strategy to enable early detection, targeted prevention, and precision oncology. In the Brazilian public health system, the Goiás Todo Rosa Program has implemented a structured pathway integrating genetic testing into routine oncologic care, expanding access to hereditary cancer risk assessment.

Methods: This cross-sectional study included 308 patients who met the National Comprehensive Cancer Network (NCCN) criteria for hereditary breast and ovarian cancer syndromes and were referred to the Human Genetics Center at the Federal University of Goiás. Following genetic counseling and informed consent, peripheral blood samples were collected for next-generation sequencing using the Oncomine™ BRCA Expanded panel. First-degree relatives of index cases carrying pathogenic variants were invited for cascade testing and clinical follow-up.

Results: Pathogenic variants were identified in 5.8% of patients in BRCA1, 1.8% in BRCA2, and 3.9% in TP53. Five families underwent cascade investigation, revealing recurrent variants, including c.1010G>A and c.455C>T. Among the tested relatives, 47.05% were mutation carriers, demonstrating a high co-segregation rate. Notably, a cancer-related death was observed among affected carriers, highlighting the clinical relevance of early identification and surveillance.

Conclusion: Cascade genetic testing showed a high yield in identifying at-risk individuals, supporting its role in preventive oncology and personalized management. The Goiás Todo Rosa Program establishes an integrated care model linking primary and specialized services, enabling timely access to molecular diagnostics and structured follow-up. This approach strengthens public health strategies and demonstrates the feasibility of implementing precision oncology within a universal healthcare system, with the potential to reduce morbidity and mortality associated with hereditary cancers.

Ethics: Research Ethics Committees of Hospital das Clínicas, Federal University of Goiás (HC-UFG) (CAAE: 50626315.6.0000.5078; Opinion No. 2.901.648) and the Association for Cancer Control in Goiás (ACCG) (CAAE: 50626315.6.3001.0031; Opinion No. 4.093.502).

Funding: Goiás State Health Secretariat (SES-GO), Coordination for the Improvement of Higher Education Personnel (CAPES), National Council for Scientific and Technological Development (CNPq), Brazilian National Genomics and Precision Health Program (Genomas Brazil).

Keywords: prevention; Li-Fraumeni syndrome; germline genetic panel; screening.