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Summary of theses and dissertations

Genetics and nursing: nurses' role in the oncogenetics counseling and in the molecular study of the hereditary breast and ovary cancer syndrome

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ABSTRACT

Genetic counseling has appeared in the nursing literature since the early 1960, and it became part of nursing standardized language when it was included in the Nursing Interventions Classification, being defined as an interactive helping process, a communication and educational intervention, focusing on assisting an individual, family or group, manifesting or at risk for developing or transmitting a birth defect or genetic condition. During genetic counseling clients receive information and education about genetic tests, benefits, risks, and meaning of tests results. This study proposed, for the first time in Latin America, the implementation of the nursing process, through nursing consultations, for clients in the oncogenetics counseling process for Hereditary Breast and Ovary Cancer Syndrome (HBOC). The conceptual model of Wanda Horta was adopted as theoretical framework and the case study method was used for 11 women with HBOC selected among a sample of 1168 individuals, during 2001-2003. Genetic tests for mutation detection in gene *BRCA1* were performed by DHPLC (Denaturing High Performance Liquid Chromatography), PTT (Protein Truncation Test), and direct sequencing. Nursing diagnosis of knowledge deficit was observed in all clients, and the strategy of teaching and learning was used in the phases of planning, implementation and evaluation of the nursing process. All 11 clients presented alterations in *BRCA1* gene. Six individuals presented IVS8-64delT variant and S1436S, four presented IVS16-68A>G variant, and case 11 was the only one who also presented other three alterations, IVS8-57delT, IVS18+66C>A, and the S1613G. These results suggest that these families could share the same haplotypes, sharing the same ancestral group. These techniques can

identify a Brazilian panel of mutations. Incorporating molecular data, and genetic counseling as a part of the nursing process can provide important components of health care for families with hereditary breast cancer at oncogenetics services in Brazil.

Key-words: genetic counseling, nursing consult, molecular biology techniques

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