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## AMERICAN COLLEGE of MEDICAL GENETICS

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### Statement on Population Screening for BRCA-1 Mutation in Ashkenazi Jewish Women

A major gene responsible for familial breast cancer, BRCA-1, recently has been identified. This discovery has resulted in a number of ongoing discussions regarding the ethical aspects of screening and counseling for the presence of this mutation. Of particular importance is the recent demonstration that about 1% of the Ashkenazi Jewish population not only carry a mutated form of BRCA-1, but the great majority of such carriers have a single mutation which can be detected easily by standard molecular techniques. This observation has led to the suggestion that a screening program to identify carriers of this mutation be instituted for women of Ashkenazi Jewish origin.

Currently, however, a primary lack of knowledge concerning the penetrance of this gene prevents the provision of accurate prognostic information to identified carriers. Even when this knowledge becomes available, the diverse issues concerning screening and counseling in this situation must be resolved in order to clarify the realistic options available to such carriers, for the risk of both breast cancer and ovarian cancer. For these reasons, testing for the BRCA-1 mutation in high risk families, and population screening of Ashkenazi Jewish individuals, should be performed only after discussion of test limitations and with appropriate informed consent. Further, at present, such population screening is best performed by investigators working under IRB-approved research protocols. Moreover, additional ethnic-group specific mutations are being identified in BRCA 1 and 2 and should be addressed in a similar fashion.

*This guideline is designed primarily as an educational resource for medical geneticists and other health care providers to help them provide quality medical genetic services. Adherence to this guideline does not necessarily assure a successful medical outcome. This guideline should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the geneticists should apply his or her own professional judgement to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from this guideline.*

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