

Genetic Counselors' Perceptions and Experiences of Counseling and Testing for Low Penetrance Alleles

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This study explores genetic counselors' experiences, knowledge, and opinions surrounding genetic testing and available guidelines for testing of five genes with low penetrance: APOE (Alzheimer's disease), F5 (thrombophilia), GBA (Parkinson's disease), HFE (hemochromatosis), and MTHFR (indications unrelated to folate metabolism). The study examined genetic counselors' perceptions of "Direct to Consumer" (DTC) testing for low penetrance risk alleles and their knowledge of available guidelines for testing of specific alleles. We found that genetic counselors are more willing to order testing for an allele with low penetrance when they believe there is strong evidence to support the association with disease. The likelihood to order testing also increases when there are actions that can be taken to mitigate the effects of the condition (e.g. impact to clinical care or lifestyle). Of the genes we covered, the most commonly ordered tests were for variants in HFE and F5, which were also the genes participants considered to be the most evidence-based and actionable. Additionally, genetic counselors reported they are more likely to order testing for these alleles if there is a known family history of the condition. Our results show that there is often discordance between the number of patients requesting testing for these low penetrance alleles and the reported number of times clinical testing has been ordered. For each allele, less than 35% of counselors reported existence of institutional guidelines for genetic testing, with only 5.0% reporting guidelines for GBA. Less than 75% of counselors surveyed were aware of the American College of Medical Genetics and Genomics (ACMG) practice guideline for MTHFR testing. Finally, while this study shows that most genetic counselors would not recommend DTC testing for any of these genes, it is possible that patients will pursue testing independent of their clinical care.