Direct-to-consumer genetic testing for factor V Leiden and prothrombin 20210G>A: the consumer experience

Sarah L. Elson1, Nicholas A. Furlotte1, Bethann S. Hromatka1, Catherine H. Wilson1, Joanna L. Mountain1, Helen M. Rowbotham1, Elizabeth A. Varga2, Uta Francke1,3
123andMe, Inc., Mountain View, CA - research.23andme.com; 2Nationwide Children’s Hospital, Columbus, OH; 3Stanford University Department of Genetics, Stanford, CA
*elsons@23andme.com

Introduction

Venous thromboembolism (VTE) is a condition characterized by the formation of blood clots in veins. VTE affects 300,000 – 600,000 individuals in the U.S. each year, can cause long-term complications, and is associated with high morbidity and mortality (1, 2). Variants in clotting factor genes are responsible for inherited predisposition (thrombophilia). The two most common genetic risk factors are factor V Leiden (c.1691G>A) in the F5 gene (1), and prothrombin 20210G>A in the F2 gene (4).

Since VTE risk is compounded by numerous acquired and situational factors, known thrombophilia status may influence clinical recommendations, including decisions about contraception and hormone replacement therapy (HRT), and may prompt increased vigilance and/or prophylaxis during pregnancy and postpartum (4–8).

There is limited consensus about which individuals should be tested for F5 and F2 variants to inform treatment and prevention decisions. Clinical guidelines recommend testing only in circumstances where test results are likely to influence clinical management and discourage routine testing for patients with VTE and their family members (4, 9–11). However, there are indications of consumer interest in testing (12), and there have been public health efforts to raise awareness of VTE (1).

Given the gap between clinical guidelines and consumer and public health organizations’ interest in greater awareness, it is useful to assess the impact of direct-to-consumer (DTC) testing for thrombophilia. Here we report data from a nine-month study on consumer response to DTC genetic testing for variants in F5 and F2.

Methods

Study participants were customers of 23andMe who obtained 23andMe’s personal genomic services, including a VTE genetic risk report, between November 2007 and November 2013. The study was conducted according to 23andMe’s IRB-approved research protocol.

We defined cases as participants whose results indicated having one or more F5 or F2 variant, and controls as having neither of these variants. Study participation consisted of responding to at least one of two online surveys. Survey 1 addressed participants’ recall of test results and subsequent perception of VTE risk, personal and family history of VTE, sharing of results with health care providers (HCPs) and family, behavioral and emotional responses to testing, and demographics. Survey 2, sent nine months later, further interrogated participants’ health outcomes, attitudes, and understanding of the causes and consequences of VTE. Tables 1, 3-5, 6A and 6C include data from Survey 1; Tables 2, 4, and 6B include data from Survey 2. 8,536 mutation-positive cases and 11,353 mutation-negative controls were invited to participate in the study. 1,244 cases and 1,110 controls responded to Survey 1; 751 cases and 574 controls took both surveys (Table 1).

Results

Risk perception: Cases demonstrated awareness of VTE risk (Table 2).

Survey 1 asked participants to indicate having one or more of the following diagnoses: hypertension (8.8%), diabetes (14.6%), myocardial infarction (11.4%), and cancer (15.4%). Further, cases reported positive impacts of receiving DTC VTE test results (Table 3).

Outcomes of receiving 23andMe genetic test results for VTE: Cases reported moderate rates of sharing test results with family members and HCPs (Table 3). HCPs offered risk-reducing recommendations to approximately one third of cases (Table 4). A majority of cases reported making changes based on results of their VTE reports (Table 5).

Discussion

This study builds on previous investigations into consumer reactions to DTC testing for disease risk (13), in this instance providing insight into outcomes of testing for VTE risk. We observed moderate rates of cases taking action following receipt of test results (e.g., sharing results with a medical provider or making behavioral changes) and high rates of satisfaction with test results. Further, cases reported positive impacts of receiving test results, including ability to take preventative measures, to inform medical providers, and to recognize signs and symptoms, as well as increased knowledge of VTE and personal risk (data not shown). Taken together, these results suggest that consumers may experience personal benefit from receiving DTC genetic results for VTE risk, even if clinical actionability is limited.

Acknowledgments

We thank the employees and research participants of 23andMe who made this research possible.

References