Personalized genomic testing (PGT) reporting preferences among over 2,500 individuals and healthcare providers (HCPs): Healthy individuals deem medical risk factors as the most important reason to pursue genetic testing

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Background: The use of genomic testing has expanded from the diagnosis of affected individuals to personalized risk assessment for disease prevention in healthy individuals. A unique study was performed to assess the level of interest and potential uptake of personalized genomic testing (PGT) among healthy individuals and healthcare providers (HCPs). This study assessed the motivations, preferences for results, and concerns about personalized genomic testing in healthy individuals among 2,500 individuals as well as genetic and non-genetic HCPs. **Methods:** A mixed methodology including qualitative interviews and quantitative surveys were used to assess the reporting preferences among healthy individuals and HCPs. Surveys and interviews were conducted between January and June 2017. Six two-hour focus groups were conducted among 35 healthy individuals and a 15-minute quantitative survey was conducted among 2,511 healthy individuals over 18yo, all recruited in partnership with Ipsos Healthcare. Additionally, 8 non-genetics HCPs (PCP, OBGyn, NPs, DOs) underwent 1-hour telephone interviews and 60 non-genetics HCPs completed a 15-minute online survey. An additional 23 HCPs specializing in genetics (Geneticists, GCs) were assessed separately through a 15-minute survey and 2-hour focus group from May through June 2017. **Results**: Healthy individuals are generally concerned about their health. Overall, 47% (1190/2511) of healthy individuals surveyed were interested in a genetic test that includes medical risk factors for themselves, risk for their children (carrier status), pharmacogenomics and ancestry. When asked to rank these categories, 85% of individuals considered medical risk factors as the most important feature of genetic testing, followed by carrier status and pharmacogenomics. Among diseases to be reported, cancer and heart disease were ranked the highest. Over 90% of healthy individuals perceived treatable and preventable diseases as the most important aspects of genetic testing, and interestingly ~50% also thought it would be important to test for non-treatable or non-preventable diseases. Healthy individuals were mostly concerned about the accuracy of testing (45%) and genetic discrimination (45%). Among non-genetics HCPs surveyed, carrier status (90%) was ranked as the most important reason for prescribing personalized genomic testing. Genetics HCPs also ranked carrier status (96%) as the primary reason for testing their patients, followed by medical risk factors for the proband (92%) and pharmacogenomics (88%). Non-genetic HCPs were mostly concerned about the accuracy of testing, while genetic HCPs main concern was regarding the return of results. **Discussion:** Results from this survey provide a framework for offering personalized genomic testing to healthy individuals and inform genetic counseling issues unique to this group. As the industry expands genetic testing to healthy/unaffected individuals, this study highlights the need to work together to responsibly bridge the current gaps of personalized genomic testing.