Are Individuals Pursuing Healthy Genome Sequencing Really Healthy?

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INTRO

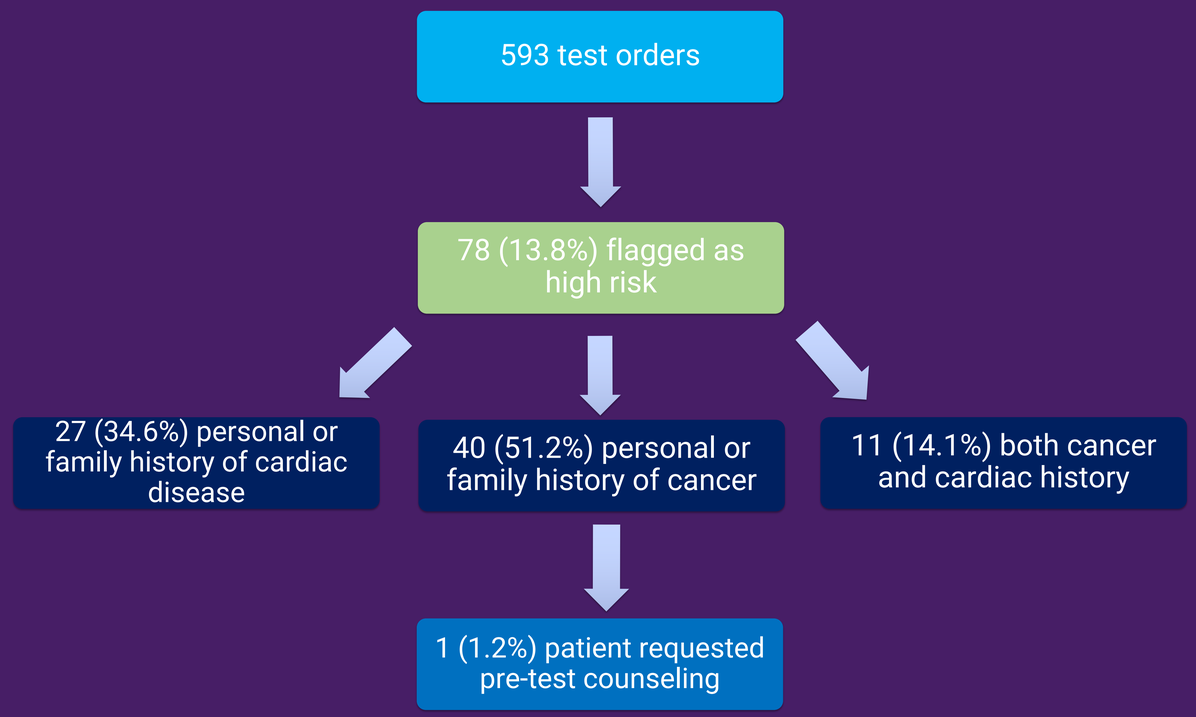
- Proactive genomic screening is increasingly being explored by reportedly healthy individuals.
- Since proactive screening is not guided by clinical history, many genes associated with hereditary forms of disease may be missed.
- Because of the limitations associated with healthy genome screening, we evaluated the percentage of patients who ordered healthy whole genome sequencing to determine how many people that pursue this testing would qualify for indication based diagnostic testing.

METHODS

- During Nov-Dec of 2018 we received 593 test orders for healthy genome sequencing, acting as the authorizing medical practice.
- In order to ensure patient's were informed of the possible limitations in the setting of a concerning high risk history, the self reported family and personal history of all orders were reviewed by a genetic counselor prior to testing approval.
- Those patients who reported a personal or family history meeting NCCN criteria for consideration of genetic testing or cardiac society guideline recommendations for counseling and/or genetic testing were sent outreach via email to set expectations and offer the option of pre-test genetic counseling, at no additional cost, to discuss indication based testing.
- Of those patients found to be high risk we queried:
  - Personal and family history indications
  - Those who elected to pursue pre and/or post testing genetic counseling
  - Results of those who had received their sequencing results thus far

Patients who participated in consumer directed, proactive genome sequencing were found to have a high risk personal or family history warranting consideration of diagnostic testing 13.8% of the time.

Of these high risk individuals, there was very low uptake of pre-test genetic counseling (1.2%) services offered at no additional cost.



RESULTS:

- 31 (39.7%) results back, with no medically actionable pathogenic variant identified thus far. (59 medically actionable genes as determine by the ACMG)
- Avg patient age: 44 years

DISCUSSION

- There is an increasing interest in proactive genetic services driven by patients themselves, via consumer initiated testing.
- We found that approximately 14% of patients who pursue proactive testing would likely qualify for diagnostic testing. This number is likely higher, as this was self reported history and many patients may have opted to not complete.
- The uptake of pre-testing genetic counseling services was low. One reasons for low uptake may be that, though these individuals had a high risk personal or family history, this was not the intended purpose of pursuing testing.
- Additionally, the testing was offered at a promotional price of $200. This price point might have influenced individuals' interests in other testing options, as the may be largely early adopters, or individuals eager to have access to their sequencing and did not see the price as high a burden.
- Further research is need to assess patient's motivation for testing and their decisions for not further exploring diagnostic testing options, as well as their knowledge of healthy screening versus diagnostic testing.

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