Removing the barrier of cost for family variant testing in cancer predisposition genes significantly increases uptake among relatives

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BACKGROUND

- Family variant cascade testing (FVT) uptake is often limited by a variety of barriers, one of which is cost.
- In July of 2017, Invitae began offering FVT at no additional charge for first-degree relatives (FDR) of probands who test positive for a pathogenic or likely pathogenic (P/LP) variant within 90 days after the initial test report is released.
- We examined the number of relatives and the percentage of families that participated in FVT before and after this program implementation.

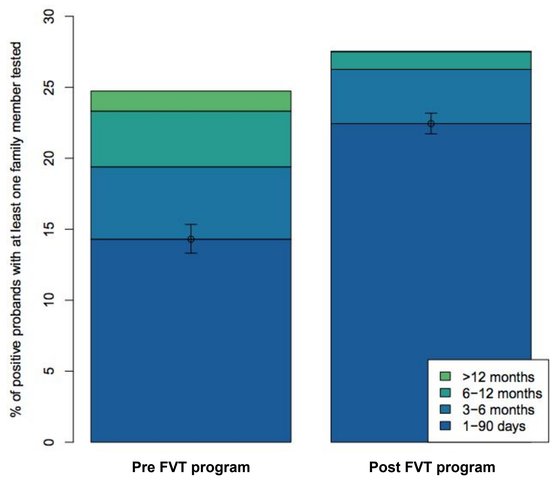
METHODS

- Probands with P/LP variants in hereditary cancer genes were studied
  - One cohort of 4,617 probands had their P/LP variants identified in Jan-July of 2017, at which time the charge for FVT was $200 for FDR.
  - A second cohort of 12,566 probands had their P/LP variants identified in July 2017-May 2018, during which time there was no additional charge for testing for FDR of probands.
- We examined how many relatives had testing during different time intervals including <90 days, <6 months, 6 months-1 year, and >1 year after the initial proband positive result was returned.

RESULTS

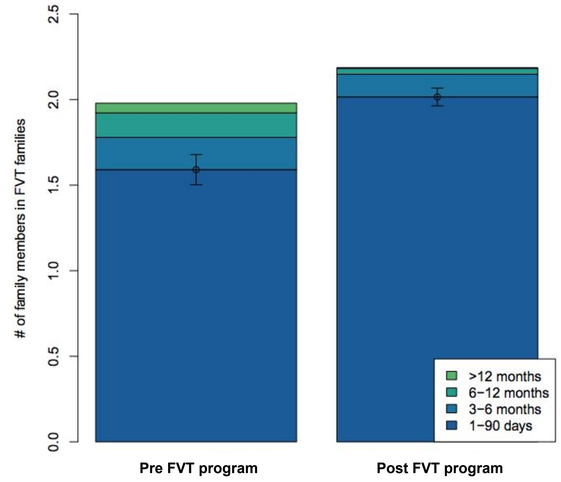
- Comparing the percentage of probands that had at least one relative tested, we observed a statistically significant ($p<0.001$) increase in testing during the no-additional charge time frame.

Figure 1: Probands with at least one relative being tested



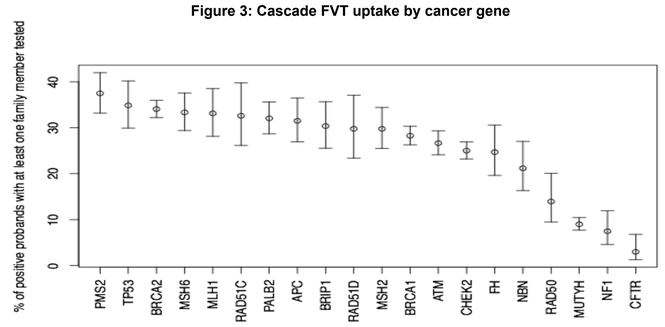
- There was also a small, but statistically significant ($p<0.001$) increase in the number of relatives receiving cascade testing per family within 3 months of a proband's positive result.

Figure 2: Number of family members per family that had FVT testing



- FVT uptake tended to be higher for greater risk genes with established management guidelines.

Figure 3: Cascade FVT uptake by cancer gene



CONCLUSIONS

- These data indicate that there was a significant increase in uptake of FVT after the removal of the cost barrier. There was an increase in the percentage of families where at least one relative had testing, and also an increase in the number of relatives per family that had testing.
- Other clinical areas including cardiology, pediatrics, and neurology have also seen a significant uptake in FVT since the policy change.
- Removing the cost barrier as an obstacle appeared to enable more at-risk family members to obtain valuable information potentially relevant to their own healthcare management. This is reflected with the high uptake of genes associated with Hereditary Breast and Ovarian Cancer, Li-Fraumeni Syndrome, and Lynch Syndrome.
- Our data also reveals that there are still a large percentage of family members that are not getting cascade testing. This suggests that there are additional barriers that warrant further study.

FUTURE DIRECTIONS

- Possible other barriers that warrant further study include:
  - shortage and accessibility of genetics professionals
  - PCPs unfamiliar with ordering/acting on genetic test results
  - family dynamics and communication barriers
  - insurance coverage for genetic counseling
  - logistics of test ordering

Disclosures: All authors are stockholders and employees of Invitae.