

RETURN OF RESULTS QUALITATIVE INTERVIEWS

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BACKGROUND

- As the number of people having exome sequencing grows, there is ongoing debate about whether and how to return results
 - + Share results out of respect for participants
 - + Share result to maximize benefits when risks are low
 - Don't share results in order to minimize harms to participants
 - Don't share results in order to minimize burden on medical system

BACKGROUND

- Several studies have reported that the general population and study participants discriminate between different result types
- Main motivations for desiring results are:
 - Having as much information as possible
 - Disease screening/early detection
 - Influence on diet and lifestyle decisions
 - Impact of results on personal planning, such as purchase of insurance
 - “a belief in the value of even uninterpretable information”

PERSONAL AND CLINICAL UTILITY

- Clinical utility: Possibility to influence or alter care and improve a patient's wellbeing
 - Often used as a standard for return of results
 - BUT, unclear to what degree any results actually impact care
- Personal utility: Value gained from results because of their impact on perceptions of oneself, relationships, or other factors
 - Unclear how much weight this deserves
 - Difficult to define

AIMS

- Describe experience of participants who have received individual genetic testing results
- Describe the psychological/emotional and communication impacts of return of results
- Explore the concept of personal utility with participants who have received individual results

CLINSEQ[®] STUDY POPULATION

- 1000+ individuals in the Washington DC area who agree to have exome sequencing
- Mostly healthy, but 25% are recruited specifically because of a personal history of heart disease
- Age: 45-65 years
- Highly educated
- Roughly 1:1 M:F ratio
- Consented from 2007 - present

CLINSEQ[®] RETURN OF RESULT PROCESS

- Phone contact to offer return of result
- In-person disclosure session with geneticist and genetic counselor
- Follow-up written summary & phone counseling if needed

ELIGIBILITY CRITERIA & RECRUITMENT

- Enrolled in ClinSeq[®] and received one genetic testing result
 - Have not participated in previous sociobehavioral studies on return of result experience
 - Willing to complete one, 30-minute phone interview
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- All eligible participants contacted 3 times via phone or postal mail and offered study participation

PARTICIPANTS & METHODS

- N=31 interviews completed
- N=29 interviews recorded, transcribed and coded
- Results returned included:
 - Hereditary cancer susceptibility
 - Cardiomyopathy and arrhythmia
 - Hypercholesterolemia
 - Myoclonus dystonia
 - Spherocytosis
 - Protein S deficiency
- Result Return Session Dates: 12/2012 – 5/2013

EXPERIENCE RECEIVING RESULTS

- Most participants were satisfied with the experience overall, or described more positive than negative parts of the process.

...the negative is negligible. I tend to think that the positive definitely outweighs any inconvenience. 100 times to 1.

-Female, 54, FLCN Result

- A small number of participants felt the sessions overemphasized the potential impact of the results

...the genetic counselor spent with me asking me how it felt and how I felt emotionally and all and you know I can understand that looking at the big picture if someone has been told they have something which could cause deformities in their children or something they would be very stressed out you know and how they see its become policy to do that for everyone but it just didn't, it wasn't all that stressful a thing so I didn't expect to be asked a lot about my emotions and how I felt

-Male, 63, BRCA1 Result

RESULT IMPACT: EMOTIONAL

- Most common response was that results “satisfy curiosity”, are “intellectually exciting”
- Most participants had neutral feelings about impact of results

I was surprised, maybe a little concerned at first, until I had an opportunity to digest all the information in the report. I have far more concerns about other aspects of my health than I do that.

-Female, 57, MSH6 Result

- Roughly half received results were expected based on personal and family history. Those who received expected results generally found them helpful, but sometimes surprising.

It wasn't a relief, it was just, it was... there was a... that's what I'm looking for; it's kind of like that... it's something I knew, but this was just the confirmation.

-Male, 70, APOB Result

RESULT IMPACT: COMMUNICATION OF RESULTS

- Generally people are sharing with family and healthcare providers
- Some impact on healthcare or health behaviors

Well, I got the report later on, which I passed to my GP and we talked about it and he did a breast exam, and, you know, it's part of my medical profile. So not only do I have the heightened level of awareness, but so does my doctor.

-Male, 65, BRCA1 Result

- Greatest impact on personal “vigilance” or self monitoring

I think knowing what I have, maybe I'm a little more in tune with my body and, you know, any symptoms or non-symptoms, you know. Maybe I'm a little more cautious.

-Female, 64, LDLR Result

DESIRES FOR FUTURE RESULTS

- Yes, still mostly want to know everything!

Give me everything you know.

-Male, 70, APOB Result

- Some discrimination between result types with prompting

The issue is, you know, whether people, or I, want to have information about issues that I can't control, and I still struggle with that.

-Female, 68, PROS1 Result

- Some desire for process to change

Would they be willing to download all of the results into a file? And basically I would take the results, downloaded in into a file if I could get them.

-Male, 55, CCR5 Result

CONCLUSIONS

- Most of our participants react well to result disclosure process
- Negative emotional impacts have been minimal
 - Optimism
 - Distance from results due to age? Personal family history?
 - “Cool factor”
- Attitudes about results are newly-formed and fluid
 - Counseling implications
- Personal utility derived from knowledge of results/self, better advocacy and vigilance around personal health

THANKS!

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