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Genetic Counselors' Assessment of Videos to Augment Whole Exome Sequencing Patient Education

Rebecca Hernan
Sarah Lawrence College, rhernan@gm.slc.edu

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Rebecca Hernan

Joan H. Marks Graduate Program in Human Genetics

Sarah Lawrence College

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Abstract

Patient education for basic genetics as well as genetic testing options is a key role of a genetic counselor (GC). As whole exome sequencing (WES) becomes more commonly used in clinical care, an increased burden of genetic education is placed on both the GC and the patient due to the complexity of this test. One method to help alleviate this burden is the use of educational tools, these tools need to be assessed for their efficacy. There is a lack of research eliciting GCs' opinion of educational tools intended for WES.

The parents of minor patients evaluated at Columbia University Medical Center who were having WES as part of clinical care were randomized to be invited to watch educational videos before their visit or to receive routine care. Treating GCs were blinded to the randomization. GCs' impressions of the session, including the effectiveness of the session, parent genetic knowledge, and time allocation were assessed by a survey following the session. A trend of GC-reported greater parent genetic knowledge and less time spent on education for those who were invited to watch the videos was observed, although it was not statistically significant. GCs reported that they spent less time on psychosocial discussion with parents in this group as opposed to those who had routine care ($p=0.02$).

The results of this study suggest that WES educational tools may augment traditional, in person, genomic education. Somewhat surprisingly, they also apparently resulted in less time spent on psychosocial counseling. Although these results suggest that WES educational tools may be useful for GCs, they need to be further studied to better understand how GCs perceive the efficacy of these tools, the impact of them on psychosocial care, and how best to incorporate them into clinical practice.

Introduction

A key role of a genetic counselor is to educate their patients about genetic testing to facilitate informed decision making (Ormond et al., 2010; Ropers, 2012). To aid them in this endeavor, genetic counselors often use supplemental educational tools such as visual aids or printed fact sheets (e.g. frequently asked questions). As testing continues to grow in complexity, the concurrent educational tools need to evolve as well. This is especially apparent as whole exome sequencing (WES) becomes more frequently used in clinical settings. The dramatic reduction of the price of WES in the past 10 years and its continual decline has resulted in increased availability and access to the test. This has led to more genetic counselors providing WES education to patients and families, increasing the education burden on both the counselor and patient. To aid both groups in this endeavor, the need for effective educational tools has become more immediate.

Educational tools need to be studied to determine effectiveness not only for the patient, but also for genetic counselors. These types of tools have been studied in the past with traditional genetic testing but there is a lack of research and educational tools available for WES. Furthermore, much of the information for current educational tools relates to a patient's use of those tools and how they aid in the understanding and retention of genetic information. Understanding how educational tools affect the session from the counselor's perspective is important in assessing the effectiveness of the tool and potentially identifying ways in which a genetic counseling session can be adapted to better serve the patient.

One method by which these tools can be evaluated is to examine how they aid in the improvement of the counseling session as assessed by genetic counselors. Outcomes of counseling sessions include appropriateness of testing and accuracy of results interpretation, psychosocial outcomes, adherence to or receipt of appropriate medical management, and patient and provider knowledge (Zierhut, Shannon, Cragun, & Cohen, 2016). Each of these outcomes could potentially be influenced by the use of educational tools.

Counselors must allocate the limited time within a session to educate patients on genetics and genetic testing as well as address the delicate and sometimes emotionally charged issues that are inherent to genetic testing. Psychosocial outcomes are an important part of a genetic counselor's role in patient care. Patients experience better medical outcomes as well as better information retention when counseling is emphasized over teaching within a session (Austin, Semaka, & Hadjipavlou, 2014; Overby, Chung, Hripcsak, & Kukafka, 2013; Roter, Erby, Lori H., Larson, Susan, & Ellington, Lee, 2007). Despite the value placed on psychosocial goals within a genetic counseling session, educational goals often take precedence when counselors feel that they have a limited amount of time with their patients (Hartmann, Veach, MacFarlane, & LeRoy, 2015). Interestingly, researchers have also found that educational goals are more difficult to achieve in the absence of a personal connection with the patient (Ellington, Kelly, Reblin, Latimer, & Roter, 2011; Meiser, Irle, Lobb, & Barlow-Stewart, 2008). Several reviews and opinions note that while genetic counseling students are taught to counsel with psychosocial goals in mind, once they enter clinic, they are encouraged to focus on the

educational aspects of a session (Austin et al., 2014; Bernhardt et al., 2009; Biesecker, 2003; Hartmann et al., 2015). This educational focus is exacerbated in the setting of consenting for complicated testing such WES (Mills & Haga, 2014; Wynn, 2015).

Education for WES has several specific challenges that include an increasing frequency of uncertain results, secondary findings, managing patient expectations, and a difficult and time-consuming pre-test counseling experience (Amendola et al., 2015; Hooker, Ormond, Sweet, & Biesecker, 2014; Tomlinson et al., 2016). Following a complicated education session, patients may struggle to retain and apply the information to their decision-making process (Glanz, Rimer, & Viswanath, 2008; Machini, Douglas, Braxton, Tsipis, & Kramer, 2014). Educational tools that are developed to address this need must be evaluated to determine their effectiveness. Prior studies examining the effectiveness of supplemental educational tools have been mostly in the realm of counseling for cancer genetics education and panel testing (Axilbund, Hamby, Thompson, Olsen, & Griffin, 2005; Cull et al., 1998; Wakefield et al., 2008). These studies have found that those who receive supplemental educational tools prior to the counseling session have a higher initial level of genetics knowledge and are able to retain more of the information provided during the session. Analysis of pre-counseling educational tools for breast cancer genetics found the tools to result in shorter sessions and improved the effectiveness of a given session (Green et al., 2005). This study also found that counselors used their time more effectively by focusing their educational content and better addressing each patient's individual priorities. These results suggest that pre-session educational interventions have the ability to not only shorten a genetic counseling consultation, but allow the counselor to focus more on

individualized risks and patient concerns, a key component of an effective genetic counseling session.

The many potential positive effects of a more personalized and less information-focused genetic counseling session include more efficient use of time and greater satisfaction overall for both the patient and the counselor. The development of appropriate educational tools has also been colloquially encouraged by genetic counselors (Axilbund et al., 2005; Green et al., 2005; Machini et al., 2014) but, to date, has not been extensively studied with respect to the counselors' viewpoint on their effectiveness. This study is intended to ascertain the effect of pre-counseling video education for WES on a genetic counseling session as appreciated by the genetic counselor.

Methods & Materials

This study was approved by the Columbia University IRB and the Sarah Lawrence College IRB.

Educational Videos

Six educational videos reviewing genes, inheritance, chromosome and microarray analysis, WES, and the benefits and limitations of genetic testing were developed for this study. The videos were created by a team that included genetic counselors, geneticists, psychologists, medical students, and genetic research coordinators over a period of eighteen months.

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Instruments

Genetic Counselor Survey

A survey to assess the treating counselor's impressions of the session, including genetic knowledge of the patient, the effectiveness of the session, and the time spent on patient education and psychosocial conversation, was developed specifically for this study (appendix A). The survey, administered through REDCap, consisted of eight multiple choice questions and a single comments section. Informed consent was the first question of the survey.

Participant Questionnaire

The parent or legal guardian of the patient completed a survey following the appointment. The analysis of this survey will be addressed in a separate publication.

Procedures

Minors or patients not mentally capable of providing consent

Potential participants were identified from the clinical population at Columbia University Medical Center from April 2016 through January 2017. Inclusion criteria for the study were as follows: 1) patient was a minor or adult with a medical guardian, 2) patient was being seen for an initial appointment for an indication of autism or developmental delay or was being seen for a follow up appointment regardless of indication, 3) patient had not previously had WES and did not have a molecular diagnosis, 4) parent/guardian was able to speak and read English. Eligible participants were randomized to either watch the educational videos (Video cohort) or to receive standard care (No Video cohort); those in the Video cohort were invited to view the videos prior to the appointment and on the day

of the appointment, but were not required to do so. The treating counselor and physician were not informed of the patient's video cohort status. Patients who were offered WES as part of their clinical evaluation and elected to have the test were invited, by a research coordinator, to participate in the TEECH study. Following enrollment of the patient, the survey invitation was emailed to the genetic counselor.

Data Analysis

Descriptive statistics of the patient sessions are presented in frequencies. The associations between video cohort status and genetic counselor survey response to each survey question was analyzed by Chi-squared test. Six of the eight survey questions responses were a three-level Likert scale but responses were not normally distributed and therefore these questions were analyzed as a binary variable. A p-value of <0.05 was considered statistically significant.

Results

Patients evaluated at the Columbia University Medical Center (CUMC), Division of Medical Genetics were screened and 649 patients were identified as potential TEECH participants. Of those, 94 had clinical WES and were consented and enrolled in the study; 47 in the Video cohort and 47 in the No Video cohort. Genetic counseling surveys were completed for 87 of the 94 (93%) enrolled patients (Figure 1).

Table I shows the breakdown of treating physicians and counselors. A single counselor saw 27 of the 94 patients (29%) with care for the remaining patients evenly distributed among ten other counselors. Two physicians treated a majority of the patients enrolled (79%) while three additional physicians were involved in the care of the other

study participants. The most common indications for genetic evaluation were developmental delay (30%), autism (19%), and birth defects/dysmorphic features (16%), however, patients were seen for over 14 different types of medical indications (Table II). The average age of the patients was 8.66 years (range 0.4-35.03). The average time between a genetic counseling session and completion of the survey was 3.85 days (range 0.13 – 32.72, median 1.77).

The results of the genetic counselor survey were analyzed with comparison between the video cohorts. A majority of counselors reported the session to be effective or very effective with no difference between the Video and No Video cohorts. Only one counselor reported an ineffective session, the patient was in the No Video cohort. Counselors also reported that most parents of the enrolled patients had an average or greater understanding of general genetics (Video: 88%, No Video: 80%) at the beginning of the session (Figure 2a). Additionally, counselors reported most more parents in the Video cohort had an average or greater understanding of WES (71%) as compared to the No Video cohort (57%), but this difference was not statistically significant (p=0.1584). Overall, counselors did not report a difference in whether or not parent questions regarding WES conveyed a deeper understanding of the test between the two cohorts (Video: 79%, No Video: 75%). Figure 2b shows that counselors rarely reported skipping any parts of genetic education within the session and there was no significant difference between the two cohorts (Video: 19%, No Video: 16%).

Counselors were also asked about allocation of time within a session (Figure 2c). Generally, counselors reported spending an average or longer amount of time on genetic

education and secondary findings education in both cohorts. In sessions with parents who had the option to watch educational videos, counselors more frequently reported spending less than the average amount of time on psychosocial issues as compared to the No Video cohort (Video: 50%, No Video: 26%, p=0.0202).

The results of the GC survey were also stratified by visit type and analyzed separately. More counselors in follow up visits reported that their counseling sessions were very effective, but there was no difference between the Video and No Video cohorts in either visit type. Trends already observed in the overall analysis were similar to trends seen in the stratified analysis with the exception of parent understanding of WES and time spent on psychosocial issues. While counselors reported that parent understanding of WES in new visits was similar to the overall data (a majority had an average or greater understanding), a different trend was seen in the follow-up visits (Figure 3a). Counselors more frequently reported that parents in the Video cohort had an average or greater understanding (75%) of WES compared to parents in the No Video cohort (38%) (p=0.066).

Discussion

We surveyed genetic counselors' perceptions of a genetic counseling sessions and parent knowledge for a cohort of patients randomized to have the option to watch educational videos before their counseling session and those who did not have the option (current routine care). In all but one of the areas assessed by the survey, there was no difference between the cohorts. When asked to assess the parents' knowledge of general genetics and WES, genetic counselors answered that a majority of parents had an average or higher level of knowledge at the beginning of the session. Counselors also spent an

average or longer amount of time on various educational aspects of the session regardless of whether or not the parents had the option to watch the videos. Following this trend, counselors reported that they did not skip any of the usual educational aspects of a session.

The only domain in which a significant difference was observed between the two cohorts was in the time spent discussing psychosocial issues. In sessions where parents had the option to watch the videos counselors reported spending less time on psychosocial issues than in sessions where the parents hadn't seen the videos. One reason for this finding may be that parents who had watched the videos had already had the opportunity to consider their own views and concerns about testing and therefore took less time to discuss these issues with their counselors. Previous studies have shown that patients who have received education before a counseling session are able to use the intervening time to consider their own values and beliefs on genetic testing and make decisions that are more in line with those values (Mancini et al., 2006; Wakefield et al., 2008). Another possibility is that parents who watched the videos had more questions about the technical aspects of WES due to their earlier exposure to the test, leading counselors to spend more time on WES education and less time focusing on the parents' beliefs around genetic testing. This explanation does not seem to be overly likely as the counselors also reported that parent questions did not convey a deeper understanding of WES for either cohort. Additionally, this was the only area in which counselors indicated less time than average was spent, suggesting that these sessions overall were shorter and less likely to include a more in depth or lengthy explanation of WES.

The results of this study were surprising as previous studies have shown that genetic counselors do perceive positive changes in their sessions after a patient has used a pre-session educational tool. These changes include shorter sessions, a better initial understanding of genetics, and a greater focus on psychosocial rather than educational issues (Cull et al., 1998; Green et al., 2005). However, one of the previously studied tools was an interactive program that required an hour for the patient to complete. The videos studied here were deliberately designed to be shorter and more accessible to families with young children and busy schedules (6 videos, 2-4 minutes long). The difference in findings may be due in part to the different levels of involvement required by each tool.

It should be noted that in previous studies, a portion of the counselors surveyed indicated that although the pre-session tools were helpful, they did not adjust the educational component of their sessions due to the fact that they felt an obligation to cover all of the necessary genetics topics regardless of the baseline knowledge of the patient (Green et al., 2005). This leads us to ask if the educational videos studied here may have actually been helpful for the patients, but the counselors did not appreciate that effectiveness. Counselors may be unwilling to skip over educational aspects due to a concern about the true level of patient/parent understanding, or for other reasons such as liability issues or wanting to give their patients equal care.

Although the videos did not appear to help the counselors to shorten or abbreviate the educational aspects of their sessions, the counselors also did not report any negative effects. Sessions were not longer than typical and were not associated with a less effective session. These tools may still be offered to patients as an additional educational method,

although we cannot say that they augment genetic education as perceived by the counselors. A previous study has also shown that educational videos such as these have the ability to reinforce genetics knowledge for patients and aid them in retention of information (Axilbund et al., 2005).

Limitations

A limitation of this study is that the genetic counselor survey relied on self-reported measures. Counselors were asked to estimate the amount of time spent on several different aspects of their sessions, but were not actually timed. They were then asked to compare these estimates to a typical session, something that is dependent on an individual counselor's style and experience. Additionally, the scaled responses failed to have a normal distribution, suggesting that the responses available may have been too restrictive.

Another limitation is that the parents were not observed while watching the videos. Instead, the videos were emailed to the parents before they came to clinic or they were invited to watch the videos in the waiting room. It is possible a portion of those in the Video cohort did not watch some, or all, of the videos. Additionally, the counselors were blinded to which cohort their patient had been randomized. While this method was used to ensure that any difference observed between the two groups was not influenced by the counselors' knowledge of the patient's cohort, it might have affected the comfort level the counselors had for modifying the session based on the parent's knowledge from the videos. For example, they were not able to contract with the parent about what they had learned or what questions they had from the videos which may have influenced the counselors' perceptions of the session with and without the video.

The study sample is modest. The data showed several trends of decreased time spent on education and discussion of secondary findings as well as a greater understanding of genetics and WES for the video cohort. These trends may be significant when powered by a larger sample size.

Conclusions

Although the data generally did not show a significant difference in session components or outcomes for those who watched the videos, there was an overall trend of better parent knowledge and less time spent on education. This suggests that these types of educational tools may have beneficial uses for genetic counselors and should continue to be developed and studied for efficacy in the clinic. Additionally, this study only analyzed the genetic counselor viewpoint. Understanding the parent perception of educational tools may influence how counselors choose to utilize these tools. To better understand this aspect of patient education, parents in this study were also surveyed regarding their experiences with the educational videos. Future analysis will include a comparison of parent views of the effectiveness of the videos and the counselor views. It would be interesting to ascertain if the parents believed they had a solid understanding of WES before discussing the test with their counselors and if they found the videos to be helpful. The results of the parent survey will be addressed in a future publication.

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References

Amendola, L. M., Lautenbach, D., Scollon, S., Bernhardt, B., East, K., Everett, J., … Victoria, M. (2015). Illustrative case studies in the return of exome and genome sequencing results, *12*(3), 283–295. http://doi.org/10.2217/pme.14.89.Illustrative

Austin, J., Semaka, A., & Hadjipavlou, G. (2014). Conceptualizing genetic counseling as psychotherapy in the era of genomic medicine. *Journal of Genetic Counseling*, *23*(6), 903–9. http://doi.org/10.1007/s10897-014-9728-1

Axilbund, J. E., Hamby, L. A., Thompson, D. B., Olsen, S. J., & Griffin, C. A. (2005). Assessment of the use and feasibility of video to supplement the genetic counseling process: A cancer genetic counseling perspective. *Journal of Genetic Counseling*, *14*(3), 235–243. http://doi.org/10.1007/s10897-005-4065-z

Bernhardt, B. A., Rushton, C. H., Carrese, J., Pyeritz, R. E., Kolodner, K., & Geller, G. (2009). Distress and Burnout among Genetic Service Providers. *Genetics in Medicine*, *11*(7), 527–535. http://doi.org/10.1097/GIM.0b013e3181a6a1c2.Distress

Biesecker, B. B. (2003). Back to the future of genetic counseling: Commentary on "Psychosocial genetic counseling in the post-nondirective era." *Journal of Genetic Counseling*, *12*(3), 213–217. http://doi.org/10.1023/A:1023280118962

Cull, A., Miller, H., Porterfield, T., Mackay, J., Anderson, E. D., Steel, C. M., & Elton, R. A. (1998). The use of videotaped information in cancer genetic counselling: a randomized evaluation study. *British Journal of Cancer*, *77*(5), 830–837. Retrieved from http://www.pubmedcentral.nih.gov/articlerender.fcgi?artid=2149970&tool=pmcentrez&rendertype=abstract

Ellington, L., Kelly, K. M., Reblin, M., Latimer, S., & Roter, D. (2011). Communication in Genetic Counseling: Cognitive and Emotional Processing. *Health Communication*, *26*(7), 667–675. http://doi.org/10.1080/10410236.2011.561921

Glanz, K., Rimer, B. K., & Viswanath, K. (2008). *Health Behavior and Health Education; Theory, Research and Practice* (4th ed.). San Francisco: Jossey-Bass.

Green, M. J., Peterson, S. K., Baker, M. W., Friedman, L. C., Harper, G. R., Rubinstein, W. S., … Mauger, D. T. (2005). Use of an educational computer program before genetic counseling for breast cancer susceptibility: effects on duration and content of counseling sessions. *Genetics in Medicine*, *7*(4), 221–229. http://doi.org/10.1097/01.GIM.0000159905.13125.86

Hartmann, J. E., Veach, P. M., MacFarlane, I. M., & LeRoy, B. S. (2015). Genetic Counselor Perceptions of Genetic Counseling Session Goals: A Validation Study of the Reciprocal-Engagement Model. *Journal of Genetic Counseling*, *24*(2), 225–237. http://doi.org/10.1007/s10897-013-9647-6

Hooker, G. W., Ormond, K. E., Sweet, K., & Biesecker, B. B. (2014). Teaching genomic counseling: Preparing the genetic counseling workforce for the genomic era. *Journal of Genetic Counseling*, *23*(4), 445–451. http://doi.org/10.1007/s10897-014-9689-4

Machini, K., Douglas, J., Braxton, A., Tsipis, J., & Kramer, K. (2014). Genetic counselors' views and experiences with the clinical integration of genome sequencing. *Journal of Genetic Counseling*, *23*(4), 496–505. http://doi.org/10.1007/s10897-014-9709-4

Mancini, J., Noguès, C., Adenis, C., Berthet, P., Bonadona, V., Chompret, A., … Julian-Reynier, C. (2006). Impact of an information booklet on satisfaction and decision-

making about BRCA genetic testing. *European Journal of Cancer*, *42*(7), 871–881. http://doi.org/10.1016/j.ejca.2005.10.029

Meiser, B., Irle, J., Lobb, E., & Barlow-Stewart, K. (2008). Assessment of the Content and Process of Genetic Counseling: A Critical Review of Empirical Studies. *Journal of Genetic Counseling*, *17*(5), 434–451. http://doi.org/10.1007/s10897-008-9173-0

Mills, R., & Haga, S. B. (2014). Genomic counseling: Next generation counseling. *Journal of Genetic Counseling*, *23*(4), 689–692. http://doi.org/10.1007/s10897-013-9641-z

Ormond, K. E., Wheeler, M. T., Hudgins, L., Klein, T. E., Butte, A. J., Altman, R. B., … Greely, H. T. (2010). Challenges in the clinical application of whole-genome sequencing. *The Lancet*, *375*, 1749–1751. http://doi.org/10.1016/S0140-6736(10)60599-5

Overby, C. L., Chung, W. K., Hripcsak, G., & Kukafka, R. (2013). Cancer Genetic Counselor Information Needs for Risk Communication: A Qualitative Evaluation of Interview Transcripts. *Journal of Personalized Medicine*, *3*(3), 238–250. http://doi.org/10.3390/jpm3030238

Ropers, H.-H. (2012). On the future of genetic risk assessment. *Journal of Community Genetics*, *3*(3), 229–236. http://doi.org/10.1007/s12687-012-0092-2

Roter, D. L., Erby, Lori H., P., Larson, Susan, M., & Ellington, Lee, P. (2007). Assessing the Oral Literacy Burden in Genetic Counseling Dialogue. *Social Science & Medicine*, *65*(7), 1442–1457. http://doi.org/10.1038/nature13314.A

Tomlinson, A. N., Skinner, D., Perry, D. L., Scollon, S. R., Roche, M. I., & Bernhardt, B. A. (2016). “Not Tied Up Neatly with a Bow”: Professionals’ Challenging Cases in Informed Consent for Genomic Sequencing. *Journal of Genetic Counseling*, *25*(1), 62–72.

http://doi.org/10.1007/s10897-015-9842-8

Wakefield, C. E., Meiser, B., Homewood, J., Peate, M., Taylor, A., Lobb, E., … Group, Ag. C. (2008). A randomized controlled trial of a decision aid for women considering genetic testing for breast and ovarian cancer risk. *Breast Cancer Research and Treatment*, *107*(2), 289–301. http://doi.org/10.1007/s10549-007-9539-2

Wynn, J. (2015). Genomic Testing: a Genetic Counselor's Personal Reflection on Three Years of Consenting and Testing. *Journal of Genetic Counseling*. http://doi.org/10.1007/s10897-015-9868-y

Zierhut, H. A., Shannon, K. M., Cragun, D. L., & Cohen, S. A. (2016). Elucidating Genetic Counseling Outcomes from the Perspective of Genetic Counselors. *Journal of Genetic Counseling*, 1–9. http://doi.org/10.1007/s10897-015-9930-9

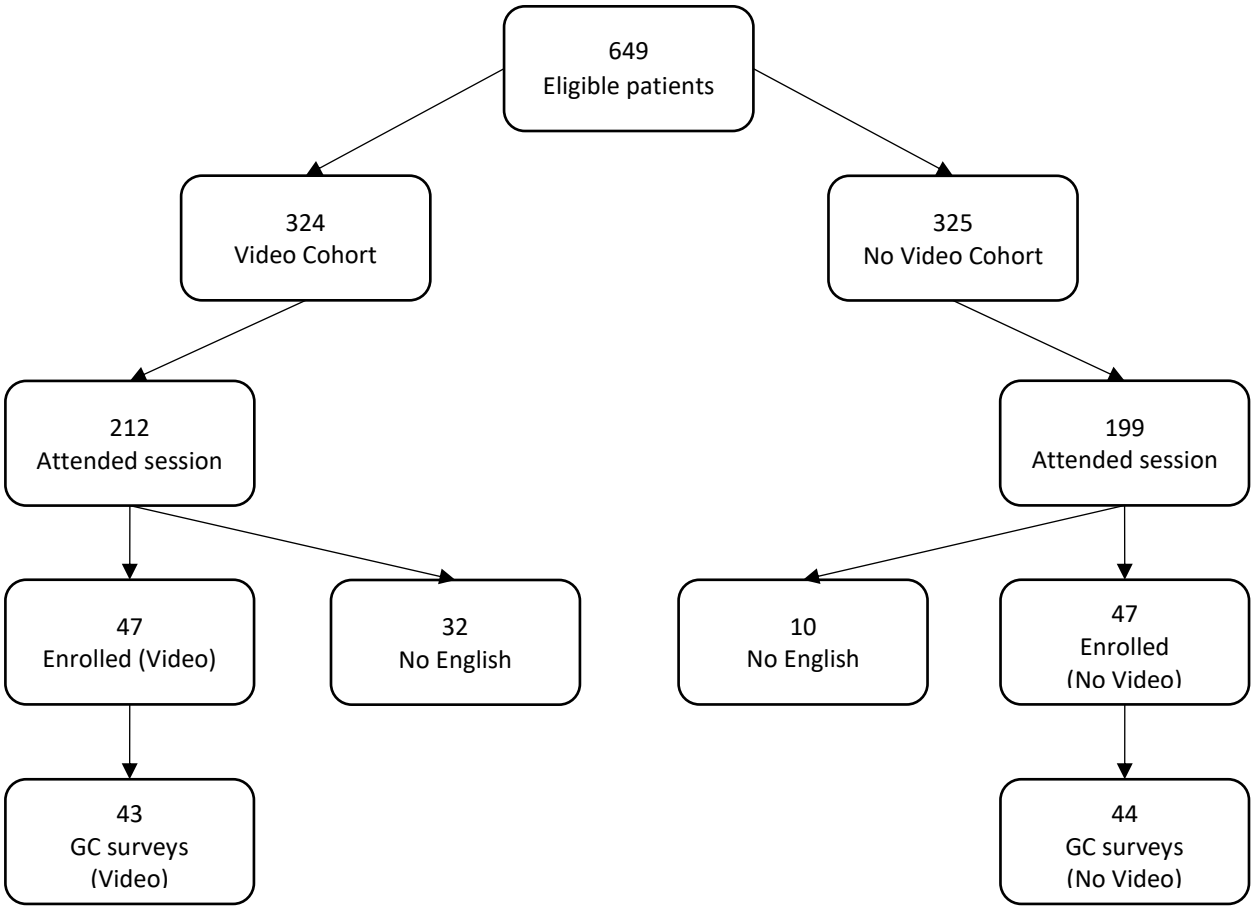

Figure 1: Summary of patient enrollment and genetic counselor survey completion.

		Video Cohort	No Video Cohort	Total	Percent
	ID #	47	47	94	
Counselor	1	6	4	10	11%
	2	4	2	6	6%
	3	1	3	4	4%
	4	4	5	9	10%
	5	3	1	4	4%
	6	1	1	2	2%
	7	8	6	14	15%
	8	11	16	27	29%
	9	6	5	11	12%
	10	2	2	4	4%
	11	1	2	3	3%
Physician	1	19	17	36	38%
	2	5	5	10	11%
	3	3	2	5	5%
	4	1	3	4	4%
	5	19	20	39	41%

Table I: Distribution of patients seen by genetic counselors and physicians.

	Video Cohort	No Video Cohort	Total	Percent
New Visit	**34**	**34**	**68**	**72%**
Developmental Delay	10	10	20	29%
Autism	6	8	14	21%
Seizures	3	2	5	7%
Birth Defects & Dysmorphic Features	5	4	9	13%
Dermatological	4	3	7	10%
Other*	6	7	13	19%
Follow-Up & In Patient Follow-Up	**13**	**13**	**26**	**28%**
Developmental Delay	4	4	8	31%
Autism	3	1	4	15%
Seizures	2	1	3	12%
Birth Defects & Dysmorphic Features	2	4	6	23%
Dermatological	1	0	1	4%
Other*	1	3	4	15%

Table II: Summary of patient indications for WES. *Other includes failure to thrive, metabolic disorders, hearing loss, skeletal dysplasia, myopathy, retinal disease, and connective tissue disorders.

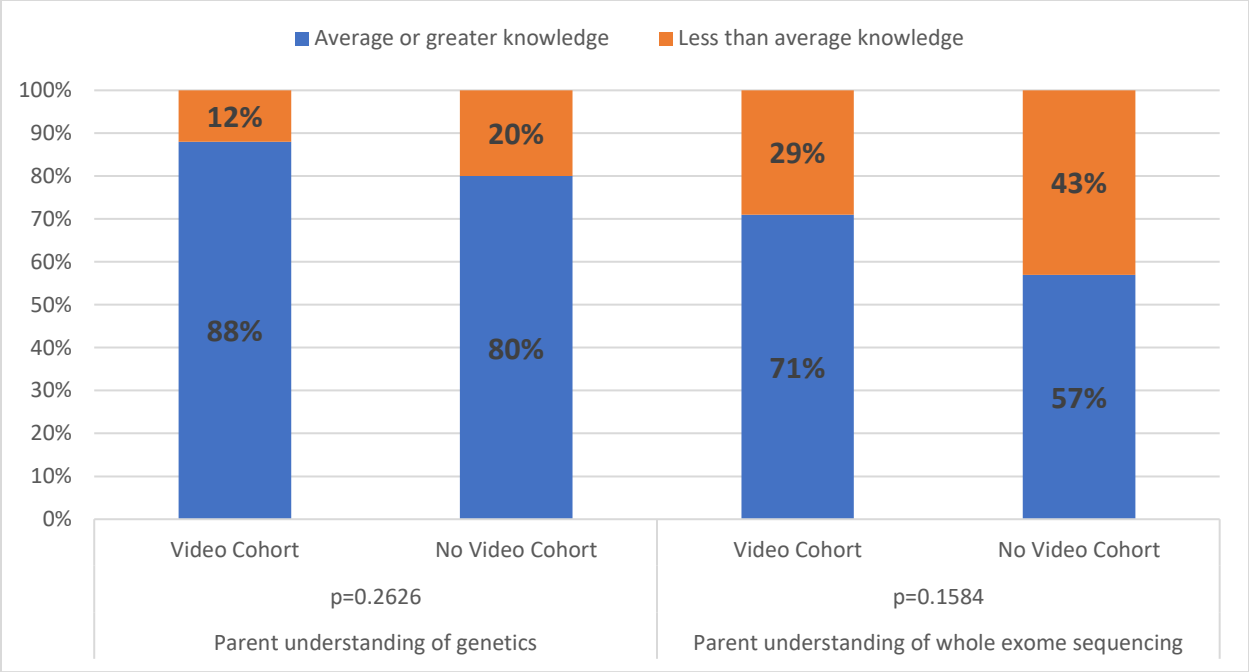

Figure 2a: Comparison of parent knowledge at the beginning of genetic counseling session between Video and No video cohorts.

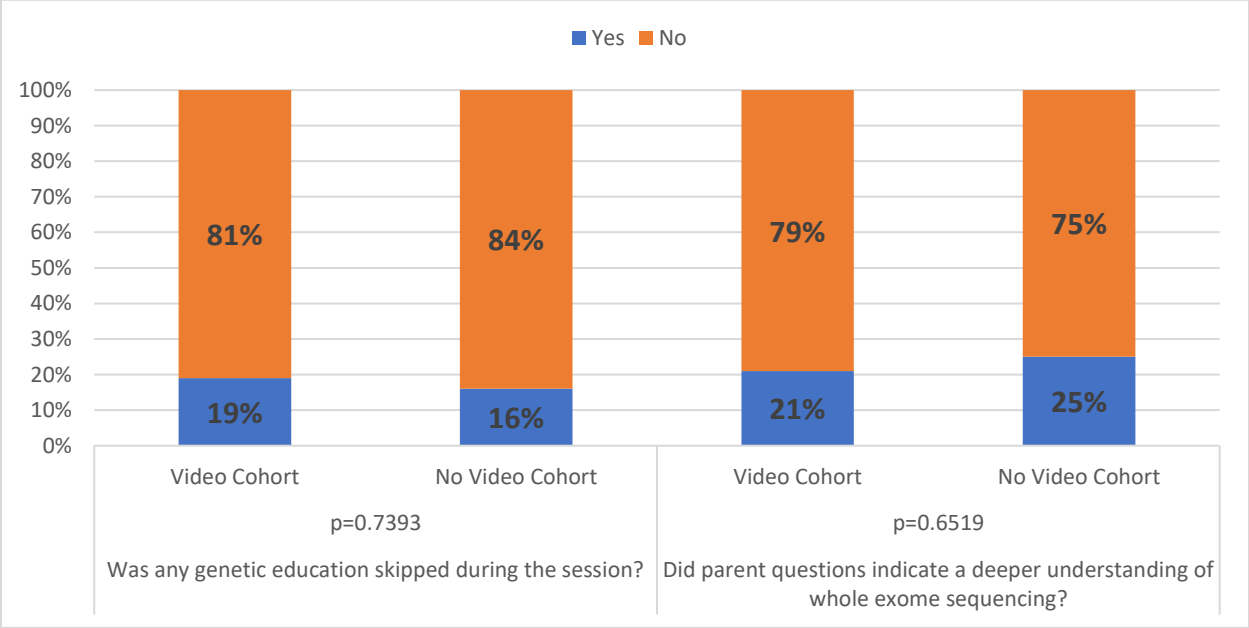

Figure 2b: Comparison of counselor acknowledgement of parent understanding during a session between Video and No video cohorts.

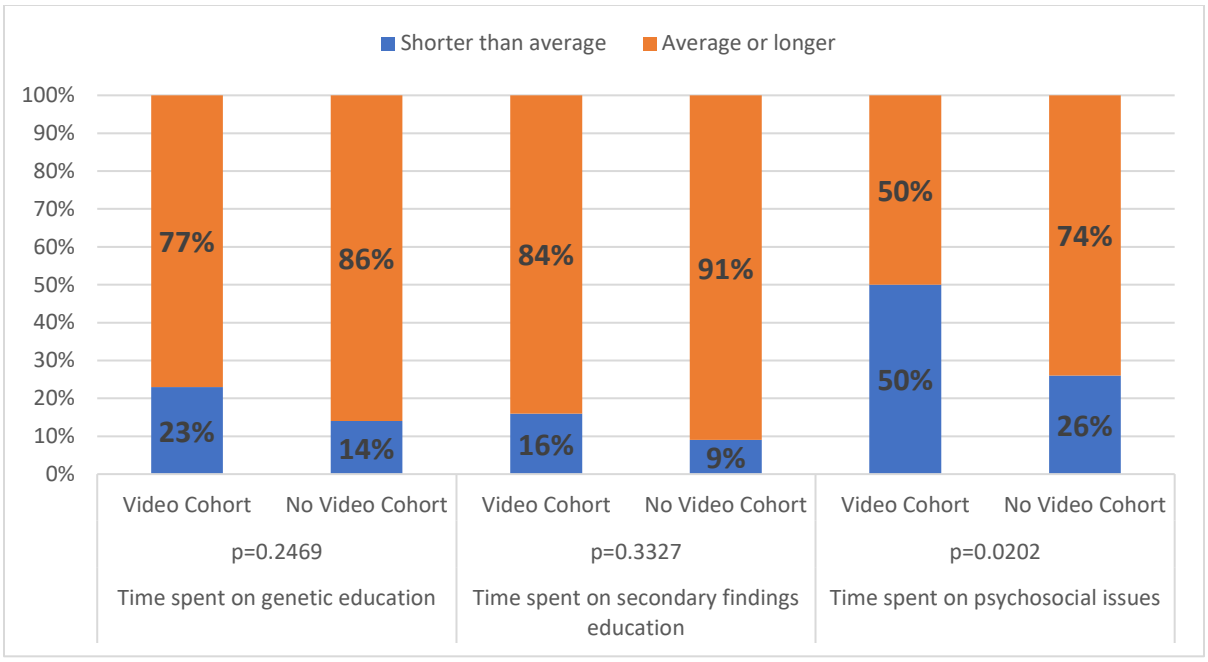

Figure 2c: Comparison of time spent on various components of genetic counseling session between Video and No video cohorts.

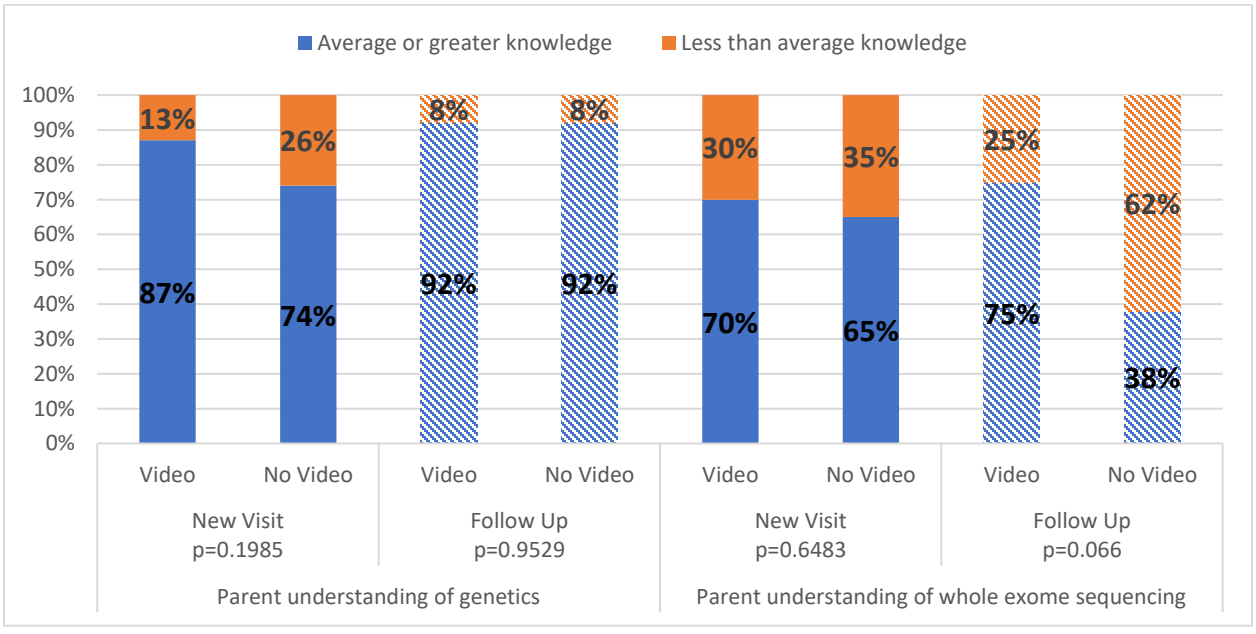

Figure 3a: Comparison of parent knowledge at the beginning of genetic counseling session. Data is stratified according to new visit or follow-up visit. Note: in-patient follow up visits were combined with regular clinic follow ups.

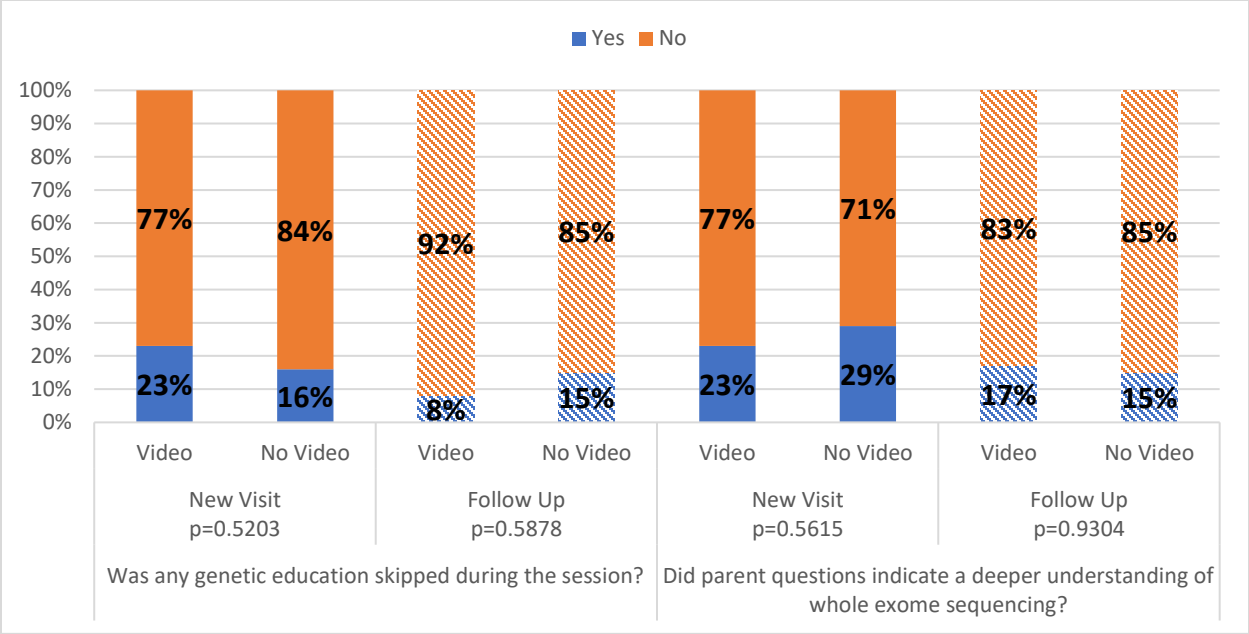

Figure 3b: Comparison of counselor acknowledgement of parent understanding during a session. Data is stratified according to new visit or follow-up visit. Note: in-patient follow up visits were combined with regular clinic follow ups.

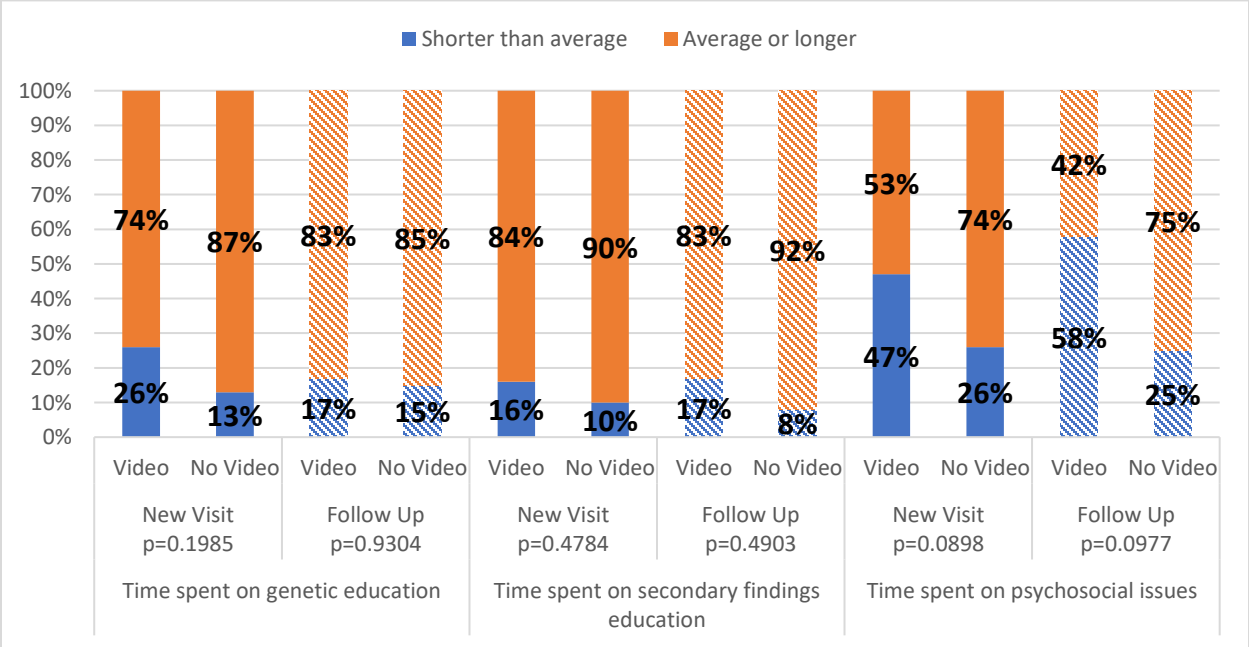

Figure 3c: Comparison of time spent on various components of genetic counselling session. Data is stratified according to new visit or follow-up visit. Note: in-patient follow up visits were combined with regular clinic follow ups.

Appendix A: Genetic counselor survey

Genetic Counselor Survey

Please complete the following questions about your appointment with [Patient Name] on [Date of Appointment].

1) Please rate the over all effectiveness of the genetic counseling session.
   Very Effective Effective Ineffective

2) How would you rate your patient's understanding of genetics at the start of the appointment?
   Very Knowledgeable Somewhat Knowledgeable No Knowledge

3) How would you rate your patient's understanding of exome sequencing at the start of the appointment?
   Very Knowledgeable Somewhat Knowledgeable No Knowledge

4) Did you skip any information pertaining to genetic education that you normally discuss?
   Yes No

5) Were the patient's questions about exome sequencing more involved and/or convey a deeper understanding of the material than the average patient?
   Yes No

6) How much time did you spend on genetic education during the session?
   Longer than average Average Shorter than average

7) How much time did you spend on secondary findings education during the session?
   Longer than average Average Shorter than average

8) How much time did you spend discussing psychosocial issues during the session?
   Longer than average Average Shorter than average

9) Please provide any additional comments about the session