5-2018

An exploration of factors influencing patient outcomes of psychiatric genetic counseling

Sarah Gerrard
Sarah Lawrence College

Angela Inglis
Sarah Lawrence College

Emily Morris
Sarah Lawrence College

Jehannine Austin
Sarah Lawrence College

An exploration of factors influencing patient outcomes of psychiatric genetic counseling

Sarah Gerrard[1], Angela Inglis[2,3], Emily Morris[2,3], Jehannine Austin[2,3]

1) Joan H. Marks Graduate Program in Human Genetics, Sarah Lawrence College, Bronxville, NY
2) Department of Psychiatry, University of British Columbia, Vancouver, Canada
3) Department of Medical Genetics, University of British Columbia, Vancouver, Canada

Submitted in partial completion of the Master of Science Degree at Sarah Lawrence College, May 2018

ABSTRACT
Though understanding how different characteristics of the patient and session influence outcomes of genetic counseling (GC) is important, little research data currently exits on this topic. We conducted a retrospective review of charts from patients who attended a specialist psychiatric GC clinic between February 1, 2012 and January 31, 2017. We extracted data to explore the effects of patient and session-related variables on Genetic Counseling Outcome Scale scores (GCOS, validated instrument that measures empowerment). We used ANOVA to analyze the pre-, to one-month post-GC change in GCOS scores in relation to eleven variables. 307 charts were included in analysis. Overall, GCOS scores significantly increased after GC ($p<0.0005$). No significant differences in GCOS change scores were identified with respect to: sex, ethnicity, diagnosis, mode of referral, type of appointment, genetic counseling student involvement, presence of observers or personal/family history of mental illness. Significant relationships were found between GCOS change scores and mode of delivery of GC ($p=0.048$, $\eta^2 = 0.020$) and primary indication for the appointment (understanding recurrence risk versus other, $p=0.001$, $\eta 2 = 0.037$). This exploratory study provides the first data on how a number of characteristics of the patient and session influence outcomes of genetic counseling. Understanding the patient and session-related factors that do seem to influence outcomes may allow for adjustment of service delivery strategies to promote the best possible outcomes.

Key Words
Genetic counseling, empowerment, Genetic Counseling Outcome Scale, patient outcomes, patient variables, session variables

INTRODUCTION

It has become increasingly important for the genetic counseling profession to demonstrate the value of genetic counseling through research evaluating patient outcomes. The National Society of Genetic Counselors (NSGC) has prioritized the identification of outcomes unique to genetic counseling, for use in outcome research and clinical intervention (Redlinger-Grosse et al., 2016). Since outcomes in the field of genetic counseling have yet to be well defined, a recent study aimed to elucidate and categorize outcomes defined by diverse groups of practicing genetic counselors (Zierhut, Shannon, Cragun, & Cohen, 2016). The most common outcome themes involved appropriate ordering of genetic tests and accurate interpretation of results, adherence to appropriate medical management, psychosocial outcomes and patient and provider knowledge (Zierhut et al., 2016). An additional unique outcome identified was the impact of genetic counseling on family member outcomes (Zierhut et al., 2016).

Important patient related outcomes such as satisfaction, knowledge and empowerment have been identified and validated through a growing body of studies. The majority of genetic counseling outcome studies have focused on cancer genetic counseling (Burke et al., 2000; Cabrera, Blanco, Yagüe, & Zabalegui, 2010; Cragun et al., 2015; Oberguggenberger et al., 2016). These studies demonstrated that after genetic counseling patients had more accurate perceptions of cancer risk, they were more knowledgeable about cancer and were less anxious about their personal cancer risk (Burke et al., 2000; Cabrera et al., 2010). Additional research to date indicates that genetic counseling increases the level of patient knowledge and positive health behaviors, while decreasing anxiety and decisional conflict (Madlensky et al., 2017). There is increasing evidence that the most important outcomes of genetic counseling are psychosocial, which is in accordance to a psychotherapeutically oriented approach in this field, and demonstrates the validity of these outcome measures (Austin, Semaka, & Hadjipavlou, 2015). Empowerment has been identified as a patient benefit from clinical genetic services, and can be defined as a set of beliefs that enable an individual to feel that they have some control over and hope for the future (Marion Mcallister, Dunn, & Todd, 2011).

In addition to the lack of research evaluating patient outcomes, limited research has analyzed the effects of patient or session-related variables on genetic counseling outcomes. Several studies have addressed potential differences between modes of genetic counseling; specifically telephone genetic counseling versus traditional in-person genetic counseling. A randomized non-inferiority trial comparing pre- and post-test telephone *BRCA1/2* genetic counseling to standard in-person genetic counseling showed no significant differences between knowledge, satisfaction, decision conflict and cancer distress two-weeks and three-months post genetic counseling (Schwartz et al., 2014). There isn't sufficient evidence to support the telephone genetic counseling model as a comparative alterative to in-person counseling with respect to all patient outcomes. However, research does indicate many positive and comparable outcomes. A recent study demonstrated that method of obtaining family history information had a significant impact on patient-reported self-efficacy (Slomp, Morris, Inglis, Lehman, & Austin, 2017). Specifically, there was a significant increase in IMSES scores, one-month after genetic counseling, for individuals who had their family history taken before their appointment, and a non-significant decrease in IMSES scores, one-month post genetic counseling, for individuals who had their family history taken during the appointment (Slomp et al., 2017). The IMSES is an instrument that measures confidence in managing psychiatric illness (Slomp et al., 2017).

While the effects of referral type have not been researched in the context of a genetic counseling appointment, some preliminary studies suggest a difference in outcomes between individuals who self-refer and those who are referred by a health care provider in clinical genetics and healthcare services (Christensen et al., 2015; Snyder et al., 2008). In a study examining the behavioral impact of genetic risk information for Alzheimer's disease, self-referred participants were more likely than actively recruited participants to make behavioral changes, such as changes in mental activities and diet, based on their genetic risk assessment results (Christensen et al., 2015). In another study, cancer survivors who self-referred to a study testing the efficacy of diet and exercise interventions had greater increases in

weekly exercise minutes and fruit and vegetable consumption compared to those who were referred, and displayed greater motivation to respond to educational materials (Snyder et al., 2008). Investigating the potential differences between self-referrals and referrals from a health care provider to genetic counseling services, and their relationship with outcome measures, may provide insight into future modes of referral for genetic counselors.

In a first-year evaluation of a specialized psychiatric genetic counseling clinic in Vancouver, BC there were significant increases shown, one-month post genetic counseling, in the baseline levels of empowerment ($p<0.0001$) and self-efficacy ($p=0.011$) (Inglis, Koehn, Mcgillivray, Stewart, & Austin, 2015). There is little research evaluating the impact of different patient and session variables on levels of empowerment, and limited research on other outcomes. While some research has evaluated modes of genetic counseling service delivery and method of family history taking, no research has examined the effects of other variables such as sex, ethnicity, diagnosis, modes of referral, primary indication for referral, type of appointment, genetic counseling student involvement, presence of observers, special group designation, and personal and family histories of mental illness on patient outcomes of genetic counseling. We aim to perform an exploratory story that examines the change (from before, to one-month post genetic counseling) in levels of empowerment with respect to several variables. Understanding factors that influence outcome measures may allow for adjustment of service delivery strategies to promote the best possible outcomes for different types of patients attending genetic counseling sessions.

MATERIALS AND METHODS

Participants and Procedure

We conducted a retrospective chart review using data from patients who received genetic counseling at a specialist psychiatric genetic counseling clinic in Vancouver, BC, between February 1, 2012 and January 31, 2017, and were entered into a clinical database. Typically, all English-speaking

patients who attend this clinic complete the Genetic Counseling Outcome Scale (GCOS) prior to genetic counseling (T1) and again at the standard one-month follow-up time point (T2). This clinical instrument is used at T1, along with the Illness Management Self Efficacy Scale (ISMES) for patients with a personal lived experience of mental illness, to assist the genetic counselor in establishing pertinent discussion points for the session. At T2, these clinical instruments aid in assessing how the patient is doing and what additional topics may need to be addressed. We did not assess ISMES scores in this study. For a further description on the psychiatric genetic counseling clinic see Inglis et al., 2014. The following patient data, stored in a de-identified clinical database includes, but is not limited to: demographic information, diagnosis, mode of referral (self-referral or referral from a health care provider), mode of genetic counseling (in-person, telephone or telehealth), primary indication for referral, type of appointment (family or individual), genetic counseling student involvement (yes or no), presence of observers (yes or no), special group designation (referrals from BC Children's Hospital OCD clinic, referrals for inpatients at Burnaby Centre for Mental Health and Addictions, or no special group designation), personal and family histories of mental illness and GCOS scores. We extracted patient data that met the following criteria: (1) they attended their first appointment between February 1, 2012 and January 31, 2017, (2) the patient was the primary individual attending the appointment, and (3) the patient had completed the GCOS prior to (T1) and one-month post (T2) genetic counseling. The primary individual attending the appointment is defined as the original patient referred, and not a family member or additional individual attending the appointment. Institutional Review Board approval was received from the BC Children and Woman's Research Ethics Board (H15-02632).

Quantitative Instrument

The GCOS is a validated, clinical genetics-specific PROM (patient reported outcome measure) (M. Mcallister, Wood, Dunn, Shiloh, & Todd, 2011). This instrument measures levels of empowerment, incorporating components such as perceived personal control, emotional regulation, benefits to

other relatives, and hope for the future (M. Mcallister et al., 2011). All 24 items are rated on a 7-point Likert scale (1= strongly disagree, 7 = strongly agree). Scores range from 24 to 168 with higher scores indicating higher levels of empowerment. All patients attending the psychiatric genetic counseling clinic, including those with personal and/or family histories of mental illness, completed the GCOS whenever it was appropriate. Instances where patients would not complete the GCOS at one or both of the time points would include when: (1) the patient is actively experiencing symptoms of psychosis, (2) time constraints of appointment, or (3) the patient declines.

Data Analyses

Descriptive statistics were applied to the demographic data, and GCOS total scores at T1 and T2 were calculated according to instrument-specific instructions. In accordance to these, any patients who declined to answer 6 or more questions on the instrument were removed from analysis. We conducted a paired sample *t* test to compare the overall change in GCOS scores from T1 to T2. We used a significance threshold of $p<0.05$. Next, we conducted one-way between-group analyses of variance (ANOVAs) for each variable using mean GCOS change scores (T2 – T1), and a significance threshold of $p<0.05$. We excluded any group where the sample size was equal to one, due to constraints of the analysis parameters. For the special groups designation variable, there were 5 patients categorized as 22q11.2 referrals who were excluded since they will be assessed in another study. For this variable, we conducted separate ANOVAs comparing referrals from BC Children's Hospital OCD clinic to patients with no special group designation, and referrals from inpatients at Burnaby Centre for Mental Health and Addictions to patients with no special group designation. For two variables, primary indication for referral and personal history of mental illness, patients in the clinical database were present in more than one group. For this reason, an ANOVA was conducted separately for each group within the primary indication for referral variable. A composite variable was created for the personal history of mental illness variable, and we conducted an ANOVA for individuals with a personal history of schizophrenia, bipolar disorder or schizoaffective disorder.

Any additional diagnoses were described. Assumptions for continuity, independence of observations, and normality were met. The homogeneity of variance assumption was met for all variables except the Burnaby Centre referrals group in the special group designation variable. For that analysis, we used a Welch test instead of ANOVA, and a significance threshold of $p<0.05$. For variables with a significant ANOVA ($p<0.05$), we performed the Tukey's HSD post hoc analysis, with a significance threshold of $p<0.05$. These analyses were performed using IBM SPSS Statistics 24.

RESULTS

There were 318 patients in the clinical database that met the inclusion criteria. Of those, 307 had no more than 5 unanswered items in the GCOS instrument at either T1 or T2 time points. The average age of patients analyzed were 41 years old, the majority were female (83.4%), and the most common ethnicity was European (67.8%). Comprehensive demographic data is shown in Table 1.

Table 1 Demographic information

	All patients (included in analysis) N = 307
Age [mean(sd)]	41.13 (12.09)
Sex [n (%)]	
Male	50 (16.3)
Female	256 (83.4)
Other	1 (0.3)
Ethnicity [n (%)]	
European	208 (67.8)
Asian	46 (15.0)
Aboriginal	1 (0.3)
African	3 (1)
Mixed	34 (11.1)
Other	3 (1)
Unknown	12 (3.9)
Personal History of Mental Illness [n][1]	
Schizophrenia	17
Bipolar disorder	55
Schizoaffective	6
OCD	12
Depression	168
Anxiety	118
Eating disorder	13
ADD/ADHD	11
Autism/ASD	1
PTSD	21
Borderline Personality disorder	6
Addiction	28

Other	19
Family History Only	48

[1]Patients may have more than one diagnosis

Table 2 Assessment of GCOS scores using paired sample *t* tests

	All patients (included in analysis) N = 307
GCOS T1 scores [mean (sd)]	111.09 (17.68)
GCOS T2 scores [mean (sd)]	127.17 (18.20)
Change (T2-T1) [mean (sd)]	16.08 (14.63)
p value	<0.0005
Cohen's *d*	1.10

Overall, GCOS scores significantly increased from T1 to T2 ($p<0.0005$, $d=1.10$) (see Table 2). There was no significant difference in GCOS change scores (T2 – T1) for individuals of different sex ($F(1, 304) = 2.158$, $p=0.143$) or ethnicity ($F(4, 289 = 0.727)$, $p=0.574$). There were no significant differences in GCOS change scores with respect to: mode of referral ($F(1, 305) = 1.266$, $p=0.261$), types of appointment ($F(1, 305) = 0.326$, $p=0.568$), genetic counseling student involvement ($F(1, 299) = 0.036$, $p=0.851$) or presence of observers ($F(1, 167 = 0.061$, $p=0.805$). Additionally, there were no significant differences in GCOS change scores comparing referrals from BC Children's hospital OCD clinic patient to referrals with no special group designation ($F(1, 289) = 0.099$, $p=0.754$), or comparing referrals from Burnaby Centre for Mental Health and Addictions to referrals with no special group designation (Welch's $F(1, 10.328) = 2.553$, $p=0.140$).

A significant relationship was found between GCOS change scores and mode of genetic counseling ($F(2, 304) = 3.067$, $p=0.048$). The effect size was small ($\eta^2 = 0.020$), and Tukey's post hoc analysis identified no significant differences between groups. There was a quantitative increase in GCOS change scores from the telephone genetic counseling group ($M = 12.49$, $SD = 13.35$) to the in-person counseling group ($M = 17.11$, $SD = 14.84$), a mean increase of 4.62, $SE = 2.29$, which was not statistically significant ($p=0.111$). There was also a quantitative increase in GCOS change scores from the telehealth genetic counseling group ($M = 10.80$, $SD = 12.90$) to the in-person genetic counseling

group (M = 17.11, SD = 14.84), a mean increase of 6.31, SE = 3.87, but it was not statistically significant (p=0.234). There was a significant increase in GCOS change scores for patients who stated that recurrence risk was a primary indication for referral, compared to those who did not (F (1, 305) = 11.624, p=0.001). The effect size was small to medium (η^2 = 0.037). There were no significant differences in GCOS change scores for patients who stated other primary indications, including: understanding causes of mental illness (F (1, 305) = 1.149, p=0.285), information regarding protective factors (F (1, 305) = 0.618, p=0.411), previous genetic testing (1, 305) = 0.005, p=0.942), pregnancy related (F (1, 305) = 0.925, p=0.337), other primary indications (F (1, 305) = 2.034, p=0.155, or unsure about primary indication (F (1, 305) = 3.368, p=0.067), compared to those who didn't. There were no significant differences in GCOS change scores comparing individuals with a personal history of mental illness, to those with a family history only (F (1, 305) = 1.233, p=0.268). Finally, there was no significant difference in GCOS change scores between individuals with a diagnosis of schizophrenia, bipolar disorder or schizoaffective disorder (F (2, 77) = 2.422, p=0.096).

Table 3 Comparison of GCOS change scores using a one-way between groups ANOVA

	n	T1 mean (sd)	T2 mean (sd)	Change (sd) (T2-T1)	ANOVA p value	η^2
Sex						
Male	50	111.65 (17.15)	125.05 (19.50)	13.40 (17.26)	0.143[2]	0.007
Female	256	110.93 (17.82)	127.63 (17.97)	16.70 (13.97)		
Other	1	124.00	114.78	-9.22		
Ethnicity						
European	208	111.38 (17.42)	128.30 (17.69)	16.92 (14.60)	0.574[2]	0.010
Asian	46	113.48 (15.73)	126.93 (17.83)	13.45 (14.05)		
Aboriginal	1	111.00	110.00	-1.00		
African	3	95.29 (17.28)	111.29 (29.65)	16.00 (12.62)		
Mixed	34	107.22 (22.22)	124.46 (17.18)	17.23 (13.56)		
Other	3	108.57 (25.75)	118.49 (43.85)	9.93 (19.13)		
Mode of referral						
Self-referral	114	109.62 (16.38)	124.48 (18.12)	14.86 (15.61)	0.261	0.004
Health care provider	193	111.96 (18.38)	128.76 (18.11)	16.80 (14.01)		
Mode of GC						
Telephone	48	111.08 (19.54)	123.57 (18.47)	12.49 (13.35)	0.048	0.020
In-person	244	110.89 (17.22)	128.00 (18.13)	17.11 (14.84)		
Telehealth	15	114.37 (19.64)	125.17 (18.15)	10.80 (12.90)		
Primary Indication						
Recurrence risk	147	110.87 (17.56)	129.87 (16.04)	19.00 (13.83)	0.001	0.037
Understanding causes	189	110.47 (17.80)	127.26 (18.58)	16.79 (14.72)	0.285	0.004
Protective factors	81	114.76 (16.98)	129.69 (18.11)	14.93 (12.52)	0.411	0.002
Had genetic testing	4	93.39 (18.03)	110.00 (23.76)	16.61 (17.17)	0.942	0.000

Pregnancy related	17	121.74 (15.41)	134.50 (15.50)	12.76 (9.37)	0.337	0.003
Other	6	98.00 (10.55)	122.50 (13.03)	24.50 (10.03)	0.155	0.007
Unsure	20	117.19 (13.98)	127.49 (21.56)	10.30 (17.84)	0.067	0.011
Type of appointment						
Family	89	110.72 (16.16)	127.54 (16.63)	16.83 (15.60)	0.568	0.001
Individual	218	111.24 (18.29)	127.02 (18.84)	15.78 (14.24)		
GC student involvement						
Yes	72	112.45 (17.23)	128.51 (20.62)	16.06 (13.45)	0.851	0.000
No	229	110.59 (17.70)	127.03 (17.23)	16.44 (14.99)		
Presence of observer						
Yes	38	116.31 (15.20)	132.45 (17.18)	16.13 (11.82)	0.805	0.000
No	131	111.51 (17.92)	127.04 (18.11)	15.53 (13.75)		
Special Groups						
OCD clinic	16	118.22 (19.93)	133.63 (16.95)	15.41 (9.92)	0.754	0.000
Burnaby Centre	11	116.68 (16.82)	122.25 (25.45)	5.57 (22.62)	0.140[3]	-
None	275	110.20 (17.48)	126.76 (18.00)	16.56 (14.44)		
History of Mental Illness						
Personal History	259	111.16 (18.10)	127.64 (18.35)	16.48 (14.61)	0.268	0.004
Family History Only	48	110.73 (18.10)	124.66 (18.35)	13.93 (14.72)		
Personal History						
Schizophrenia[1]	17	114.65 (16.78)	123.85 (21.82)	9.20 (18.15)	0.096	0.061
Bipolar disorder[1]	55	112.03 (20.38)	129.04 (18.19)	17.01 (16.00)		
Schizoaffective[1]	6	118.49 (14.61)	123.17 (30.94)	4.68 (23.18)		

[1]Includes all individuals regardless of an additional diagnosis
[2]Excluded groups where n=1 for the purposes of this analysis
[3]Welch analysis performed (homogeneity of variance assumption not met)

DISCUSSION

In order to assess the effectiveness of genetic counseling it is important to identify and measure specific outcomes. While there is prior research focusing on these two goals, and limited research assessing the effect of variables on outcomes, this is the first study to examine the influence of multiple patient and session-related variables on outcomes in genetic counseling. Overall, there was a significant increase in empowerment following genetic counseling ($p<0.0005$). This result was expected based on previous research (Inglis et al., 2015; Slomp et al., 2017). The patient variables sex and ethnicity had no impact on levels of empowerment. This finding demonstrates that patients with different sexes and ethnicities benefit from psychiatric genetic counseling. While the patient variable sex captures sex assigned at birth, we didn't assess the gender identity of each patient which would capture an assessment of gender experience and outcomes in genetic counseling. There was one patient, who met inclusion criteria, who

identified as transgender however we had to exclude them when performing the ANOVA due to constraints of the analysis parameters.

Four additional variables assessed had no impact on levels of empowerment: mode of referral (self-referral or referral from a health care provider), type of appointment (individual or family), genetic counseling student involvement (yes or no) and presence of observers (yes or no). It's possible that smaller differences in levels of empowerment between these groups were masked by large increases in levels of empowerment for all patients. While the GCOS change scores were quantitatively larger for those who were referred by health care provider, it was not statistically significant. This quantitative difference could be explained by the hypothesis that individuals who self-refer have higher baseline levels of empowerment, however the T1 GCOS scores were not significantly different between the two groups (t (305) = -1.119, p=0.264). Individuals who self-refer may have stronger personal or family histories of disease, higher levels of anxiety, or other psychosocial variables that may play a role in their response to treatment (Audrain et al., 1998; Henrikson, Harris, & Bowen, 2007). In order to better elucidate these contributing factors, it would be important to further evaluate these two groups regarding their main concerns and personal or family histories of mental illness. The mode of referral may have no impact on genetic counseling outcomes, however these results raise the importance of educating health care providers about the benefits and outcomes of psychiatric genetic counseling. The referring health care providers involved in this study included psychiatrists, general practitioners, genetic counselors and mental health workers. Patients experienced increased levels of empowerment, regardless of additional individuals attending the appointment, including other family members, genetic counseling students who participate in the session and observers who don't participate. In fact, the effect sizes are remarkably low ($\eta 2 < 0.001$). This demonstrates that the effects of genetic counseling, for primary patients, are not affected by additional attendees such as their children, parents, or spouses. It was not expected that the presence of genetic counseling students or silent observers would impact the effects of psychiatric genetic counseling. No significantly different effect of genetic

counseling with respect to special group designation was detected. Importantly, patients referred from the Burnaby Centre had quantitatively lower GCOS change scores (M = 5.57, SD = 22.62) compared to patients who weren't referred from a special group (M = 16.56, SD = 14.44), however the differing sample sizes in these two groups and non-homogeneity of variance did not allow for elucidation of this finding. This warrants further investigation.

The mode of genetic counseling did have a significant impact on the patient-reported measure of empowerment (p=0.048). While Tukey's post hoc analysis was not significant, patients who received in-person genetic counseling had quantitatively larger GCOS change scores compared to patients who received genetic counseling over the phone, or through videoconference. Only 48 patients received telephone genetic counseling, and 15 patients were seen through telehealth, compared to 244 patients who came in-person to their genetic counseling appointment. The trend demonstrated is larger increases in empowerment, after genetic counseling, for individuals who receive in-person genetic counseling, and equivalent samples sizes would lend support to this observation. A randomized trial comparing telegenetics to in-person cancer genetic counseling reported patient satisfaction did not differ by group on either satisfaction scale, but also identified the need for further randomized trials to compare longer-term psychosocial and behavioral outcomes (Buchanan et al., 2015). Telephone genetic counseling has the potential to increase access to comprehensive genetic services and decrease costs, however the concerns arise when considering the ability to translate knowledge and provide adequate patient support (Schwartz et al., 2014). It's important to assess alternative delivery modes, and their relationship with patient outcomes, given the increased demand for genetic counseling and genetic testing. Although previous literature has demonstrated comparable outcome between in-person genetic counseling and telephone genetic counseling, these results suggest that patients benefit from in-person genetic counseling, with respect to levels of empowerment.

Primary indications for referral impacted genetic counseling outcomes, although these results are interpreted with caution. There was a significant increase in levels of empowerment for patients who stated that understanding recurrence risk was a primary indication for referral, compared to patients who did not (p=0.001). The effect size was small to medium ($\eta^2 = 0.037$), indicating that increases in levels of empowerment can be moderately attributed to that primary indication. The effects of genetic counseling may be greater when patients are interested in understanding the recurrence risk estimates, related to mental health conditions, for themselves or their family members. However, some patients within the understanding recurrence risk group stated additional primary indications for referral that could be confounding these results. Our approach to this analysis is an appropriate first step, but more in-depth investigation will better elucidate how patients with different primary concerns benefit from genetic counseling. No effect on levels of empowerment were found when assessing the other primary indications individually, including: understanding the causes of mental illness, information regarding protective factors, the presence of previous genetic testing, pregnancy related factors, other indications and patients who were unsure of their primary indication.

History of mental illness had no impact on levels of empowerment when comparing individuals with a personal history of mental illness to individuals with a family history of mental illness only. Additionally, there were no significant differences in levels of empowerment when comparing individuals with a personal history of schizophrenia, bipolar disorder or schizoaffective disorder. Previous research has demonstrated that genetic counseling improves knowledge and risk perception accuracy for individuals with these serious mental illnesses (Hippman et al., 2016). Therefore, we chose to expand on this and assess the impact of these specific diagnoses on levels of empowerment after genetic counseling. Our results demonstrate that individuals with different diagnoses of a serious mental illness benefit from genetic counseling. Individuals were included in this analysis regardless of additional diagnoses (anxiety, depression or others).

Study limitations

This exploratory study was conducted using a convenience sample, and therefore there was no control group. All client data was accessed retrospectively, and all patients included received genetic counseling. Additional limitations include the patient demographics. The majority of patients were female, European and had a personal history of mental illness (Personal history n = 259, Family history only n = 48). The psychiatric diagnoses were per patient report, and not confirmed. Furthermore, GCOS scores were measured one-month after genetic counseling, but longer-term effects were not assessed. While this study was conducted in a specialist clinic, we predict that these results translate to all areas of the genetic counseling profession although validation is warranted.

Practice implications

These findings highlight the importance of educating genetic counselors about specific patient outcomes, and indicates a need to delineate the influence that patient and session-related variables have on outcomes in genetic counseling. Evaluating outcomes in clinical genetics has been difficult because traditional measures of health status are not applicable in chronic genetic conditions (M. Mcallister et al., 2011). This study utilizes the validated GCOS-24, that captures the construct of empowerment as an outcome of genetic counseling. GCOS-24 includes aspects of perceived personal control, which is considered a valid outcome measure for genetic counseling and extends beyond traditionally accepted educational outcomes (Berkenstadt, Shiloh, Barkai, Katznelson, & Goldman, 1999). It also captures emotional regulation and hope for the future, which has not been included in previous questionnaires (M. Mcallister et al., 2011). Our data demonstrates that patients with different ethnicities, sexes and diagnoses benefit from psychiatric genetic counseling, and that patient and session-related variables do seem to influence genetic counseling outcomes. As an exploratory study these findings provide an initial framework for future studies to expand on, and highlight the implications that evidence based research may have on genetic counseling practice.

Research recommendations

This research lends support to future areas of research, including other factors that may influence patient outcomes such as coping style. In studies analyzing the effect of coping style on emotional outcomes after testing and cancer genetic counseling, researchers found that individuals with a "high monitoring" coping style have a greater desire for information regarding their illness and may have a higher need for certainty with respect to test results (Nordin, K., Liden, A., Hansson, M., Rosenquist, R., Berglund, 2002; Shiloh, S., Koehly, L., Jenkins, J., Martin, J., Hadley, 2008). One common genetic counseling outcome theme identified by Zierhut et al., 2016 was adherence to medical management, which could be explored in future research assessing adherence to medication, or the number of presentations to clinics or hospitals. It is important to acknowledge that themes involving genetic testing and adherence to appropriate medical management are not common in genetic counseling outcome literature, while psychosocial outcomes and levels of knowledge are very commonly discussed. This could represent changes in the field, such as an increasing number of variants of uncertain significance (VUS), as we are implementing testing ahead of our ability to explain all genetic variation. Additionally, there are limited studies looking at long-term health outcomes, and the diverse practice settings in which genetic counselors work (Madlensky et al., 2017). An additional outcome, that we were unable to assess in this study, was the impact of genetic counseling on family members. Future research could assess levels of empowerment for family members of primary patients, as measured by the GCOS. Furthermore, we did not assess family history of mental illness with respect to different diagnoses. Finally, the primary indications for referral variable can be divided further to compare whether this concern is related to the patient themselves, or to others.

CONCLUSION

This exploratory study provides the first data on how a number of characteristics of the patient and session influence outcomes of genetic counseling. Patients benefit from psychiatric genetic counseling, regardless of sex, ethnicity, diagnosis, history of mental illness, students, observers and additional

attendees. We demonstrate that in-person genetic counseling may lead to greater outcomes for patients, and that variables such as the primary indications for referral, specifically understanding recurrence risk, may predict different outcomes of genetic counseling for patients. Furthermore, it adds continued support to the value in psychiatric genetic counseling for individuals with a personal or family history of mental illness. There is a need for more evidence based research evaluating patient outcomes in genetic counseling, which is useful in order to better understand how current services meet patient's needs, and which changes in service delivery will be the most effective.

ACKNOWLEDGEMENTS

The authors thank Dr. Arianne Albert for her assistance regarding the statistical analyses and Caitlin Slomp for her insight throughout the research process. This research was supported by Janelle Villiers, and the Joan H. Marks Graduate Program in Human Genetics at Sarah Lawrence College.

Conflict of interest statement

AI and EM provide genetic counseling in the context of the clinic described. SG and JA declare no potential conflicts of interest.

REFERENCES

Audrain, J., Schwartz, M. D., Lerman, C., Hughes, C., Peshkin, B. N., & Biesecker, B. (1998). Psychological distress in women seeking genetic counseling for breast-ovarian cancer risk: The contributions of personality and appraisal. *Annals of Behavioral Medicine*, *19*(4), 370–377.

Austin, J., Semaka, A., & Hadjipavlou, G. (2015). Conceptualizing genetic counseling as psychotherapy in the era of genomic medicine, *Journal of Genetic Counseling, 23*(6), 903–909. https://doi.org/10.1007/s10897-014-9728-1.Conceptualizing

Berkenstadt, M., Shiloh, S., Barkai, G., Katznelson, M. B. M., & Goldman, B. (1999). Perceived personal control (PPC): A new concept in measuring outcome of genetic counseling. *American Journal of Medical Genetics*, *82*(1), 53–59. https://doi.org/10.1002/(SICI)1096-8628(19990101)82:1<53::AID-AJMG11>3.0.CO;2-#

Buchanan, A. H., Datta, S. K., Celette, &, Skinner, S., Hollowell, G. P., Beresford, H. F., … Adams, M. B. (2015). Randomized Trial of Telegenetics vs. In-Person Cancer Genetic Counseling: Cost, Patient Satisfaction and Attendance. *Journal of Genetic Counseling*, 24, 961-970. https://doi.org/10.1007/s10897-015-9836-6

Burke, W., Culver, J. O., Bowen, D., Lowry, D., Durfy, S., McTiernan, a, & Andersen, M. R. (2000). Genetic counseling for women with an intermediate family history of breast cancer. *American Journal of Medical Genetics*, *90*(5), 361–8. https://doi.org/10.1002/(SICI)1096-8628(20000228)90:5<361::AID-AJMG4>3.0.CO;2-8

Cabrera, E., Blanco, I., Yagüe, C., & Zabalegui, A. (2010). The impact of genetic counseling on knowledge and emotional responses in Spanish population with family history of breast cancer. *Patient Education and Counseling*, *78*(3), 382–388. https://doi.org/10.1016/j.pec.2009.10.032

Christensen, K. D., Roberts, J. S., Zikmund-Fisher, B. J., Kardia, S. L. R., McBride, C. M., Linnenbringer, E., & Green, R. C. (2015). Associations between self-referral and health behavior responses to genetic risk information. *Genome Medicine*, *7*(1), 0–11. https://doi.org/10.1186/s13073-014-0124-0

Cragun, D., Camperlengo, L., Robinson, E., Caldwell, M., Kim, J., Phelan, C., … Pal, T. (2015). Differences in BRCA counseling and testing practices based on ordering provider type. *Genetics in Medicine : Official Journal of the American College of Medical Genetics*, *17*(1), 51–7. https://doi.org/10.1038/gim.2014.75

Henrikson, N. B., Harris, J. N., & Bowen, D. J. (2007). Predictors of self-referral into a cancer genetics registry. *Cancer Epidemiology Biomarkers and Prevention*, *16*(7), 1387–1392. https://doi.org/10.1158/1055-9965.EPI-06-0009

Hippman, C., Ringrose, A., Inglis, A., Cheek, J., Albert, A. Y. K., Remick, R., … Austin, J. C. (2016). A Pilot Randomized Clinical Trial Evaluating the Impact of Genetic Counseling for Serious Mental Illnesses. *The Journal of Clinical Psychiatry*, *77*(2), e190–e198. https://doi.org/10.4088/JCP.14m09710

Inglis, A., Koehn, D., Mcgillivray, B., Stewart, S. E., & Austin, J. (2015). Evaluating a unique, specialist psychiatric genetic counseling clinic: uptake and impact PubMed Central CANADA. *Clinical Genetics*, *87*(3), 218–224. https://doi.org/10.1111/cge.12415

Madlensky, L., Trepanier, A. M., Cragun, D., Lerner, B., Shannon, K. M., & Zierhut, H. (2017). A Rapid Systematic Review of Outcomes Studies in Genetic Counseling. *Journal of Genetic Counseling*, 1–18. https://doi.org/10.1007/s10897-017-0067-x

Mcallister, M., Dunn, G., & Todd, C. (2011). Empowerment: qualitative underpinning of a new clinical genetics-specific patient-reported outcome. *European Journal of Human Genetics*, *19160*(10), 125–130. https://doi.org/10.1038/ejhg.2010.160

Mcallister, M., Wood, A., Dunn, G., Shiloh, S., & Todd, C. (2011). The Genetic Counseling Outcome Scale: A new patient-reported outcome measure for clinical genetics services. *Clinical Genetics,* 79, 413-424. https://doi.org/10.1111/j.1399-0004.2011.01636.x

Nordin, K., Liden, A., Hansson, M., Rosenquist, R., Berglund, G. (2002). Coping style, psychological distress, risk perception, and satisfction in subjects attending genetic counseling for hereditary cancer. *Journal of Medical Genetics*, (38), 689–694. https://doi.org/10.1136/jmg.2005.034710

Oberguggenberger, A., Sztankay, M., Morscher, R. J., Sperner-Unterweger, B., Weber, I., Hubalek, M., … Meraner, V. (2016). Psychosocial outcomes and counselee satisfaction following genetic counseling for hereditary breast and ovarian cancer: A patient-reported outcome study. *Journal of Psychosomatic Research*, *89*, 39–45. https://doi.org/10.1016/j.jpsychores.2016.08.005

Redlinger-Grosse, K., Veach, P. M. C., Cohen, S., LeRoy, B. S., MacFarlane, I. M., & Zierhut, H. (2016). Defining Our Clinical Practice: The Identification of Genetic Counseling Outcomes Utilizing the Reciprocal Engagement Model. *Journal of Genetic Counseling*, 25, 239-257. https://doi.org/10.1007/s10897-015-9864-2

Schwartz, M. D., Valdimarsdottir, H. B., Peshkin, B. N., Mandelblatt, J., Nusbaum, R., Huang, A.-T., … King, L. (2014). Randomized Noninferiority Trial of Telephone Versus In-Person Genetic Counseling for Hereditary Breast and Ovarian Cancer. *Journal of Clinical Oncology*, *32*, 618–626. https://doi.org/10.1200/JCO.2013.51.3226

Shiloh, S., Koehly, L., Jenkins, J., Martin, J., Hadley, D. (2008). Monitoring coping style moderates emotional reations to genetic testing for hereditary nonpolyposis colorectal cancer: a longitudinal study. *Psycho-Oncology*, *17*, 746–755. https://doi.org/10.1002/pon

Slomp, C., Morris, E., Inglis, A., Lehman, A., & Austin, J. (2017). Patient outcomes of genetic counseling: assessing the impact of different approaches to family history collection. *Clinical Genetics*, (October), 1–7. https://doi.org/10.1111/cge.13176

Snyder, D. C., Sloane, R., Lobach, D., Lipkus, I. M., Peterson, B., Kraus, W., & Demark-Wahnefried, W. (2008). Differences in baseline characteristics and outcomes at 1- and 2-year follow-up of cancer survivors accrued via self-referral versus cancer registry in the FRESH START Diet and exercise trial. *Cancer Epidemiology, Biomarkers & Prevention : A Publication of the American Association for Cancer Research, Cosponsored by the American Society of Preventive Oncology*, *17*(5), 1288–94. https://doi.org/10.1158/1055-9965.EPI-07-0705

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*, *25*(5), 993–1001. https://doi.org/10.1007/s10897-015-9930-9