Awareness of Genetic Counselling Services Amongst Allied-Healthcare Professionals

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A research report (in the format of a "submissible" paper) submitted to the Faculty of Health Sciences, University of the Witwatersrand, Johannesburg, in partial fulfilment of the requirements for the degree of Master of Science in Medicine (Genetic Counselling)

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Declaration

I, Jamey Thom, declare this research report (in the format of a "submissible" paper) is my own, unaided work. It is being submitted for the Degree of Master of Science in Medicine at the University of the Witwatersrand, Johannesburg. It has not been submitted before for any degree or examination at any other University.

(Signature of candidate)

27th day in February 2020, in Johannesburg

Contribution of the candidate to the paper

Declaration: Student's contribution to article(s) and agreement of co-author

I, Jamey Thom, student number 808293, declare that this Research Report is my own work and that I contributed significantly towards research findings presented in the paper intended for publication below.

Signature of Student

Date: 27/02/2020

Name of Primary Supervisor: Ms Tabitha Haw

Signature of Primary Supervisor

Date: 27/02/2020

Agreement by co-authors: By signing this declaration, the co-authors listed below agree to the use of the article(s) by the student as part of her Research Project.

Article Title: Awareness of Genetic Counselling Services Amongst Allied-Healthcare Professionals

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1st author	Jamey Thom		27/02/2020
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This research report is dedicated to my grandparents, Gran and Bimps. Thank you for always being my number one supporters, always taking the time to listen and for encouraging me to chase every opportunity.

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Presentations arising from this research

1. SASHG Biennial Congress 2019, 3rd-6th August, Cape Town, Western Cape (poster presentation)

2. University of the Witwatersrand Cross Faculty Postgraduate Symposium, 22nd October, School of Public Health, Wits (poster presentation)

3. Division of Human Genetics Departmental Seminar, 30th October, National Health Laboratory Service (oral presentation)

Abstract

Due to the lack of awareness of what genetic counselling entails, its availability and benefit, many patients are not referred for genetic counselling and genetic counsellors are underutilised. Individuals with genetic disorders present with a variety of symptoms and are frequently referred to allied-healthcare professionals for management. Allied-healthcare professionals are in a unique position to identify appropriate patients and refer them to genetic counselling. This study aimed to determine if allied-healthcare professionals, including physiotherapists, occupational therapists and speech and language therapists, were aware of genetic counselling services, had referred patients to these services and to identify barriers and facilitators to referral. This information allowed recommendations to be made to increase the number of patients referred by allied-healthcare professionals to genetic services. A paper-copy questionnaire that was adapted from Hayflick *et al.*'s study was administered to allied-healthcare professionals working at one of three state hospitals in Johannesburg. The questionnaire consisted of demographic and multiple choice questions. The multiple choice questions aimed to assess the allied-healthcare professional's knowledge of genetic services, whether they understood the role of genetic counselling and if they were interested in furthering their genetics education. 57 questionnaires were completed and their data analysed. Results indicated that 29/57, 50.9% knew that genetic counselling services were available. Of those that were aware of the services, 15/29, 51.7% had referred patients to these services in the last year. Barriers to referral included misconceptions about the role of genetic counsellors, insufficient knowledge of which patients would benefit from genetic counselling and being unable to get appropriate information from the genetic clinic for patient referral. Facilitators identified included allied-healthcare professionals recognising the benefit of genetic counselling and wanting to increase their knowledge in genetics. Therefore, genetics education of allied-healthcare professionals and improved information regarding the referral procedure may improve the number of patients referred to genetic counselling.

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List of Abbreviations

CHBAH	Chris Hani Baragwanath Academic Hospital
HPCSA	Health Professions Council of South Africa
NHLS	National Health Laboratory Service
Wits	University of the Witwatersrand
No.	Number
NRF	National Research Foundation

Research report in the format of a “submissible” paper

This work has been written up as a paper for submission to The Journal of Genetic Counselling. The author guidelines such as abstract, referencing and word count has been adhered to and can be found in Appendix D. Tables and Figures have been included in the test for ease of reference.

Awareness of Genetic Counselling Services Amongst Allied-healthcare Professionals

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Abstract for Paper Submission

Due to the lack of awareness of what genetic counselling entails, its availability and benefit, many patients are not referred for genetic counselling. Individuals with genetic disorders present with a variety of symptoms and are frequently referred to allied-healthcare professionals for management. Allied-healthcare professionals are in a unique position to identify appropriate patients and refer them to genetic counselling. This study aimed to determine if allied-healthcare professionals, including physiotherapists, occupational therapists and speech and language therapists, were aware of genetic counselling services, had referred patients to these services and to identify barriers and facilitators to referral. This information allowed recommendations to be made to increase the number of patients referred by allied-healthcare professionals to genetic services. A paper-copy questionnaire that was adapted from Hayflick *et al.*'s study was administered to allied-healthcare professionals working at one of three state hospitals in Johannesburg. The questionnaire consisted of demographic and multiple choice questions. The multiple choice questions aimed to assess the allied-healthcare professional's knowledge of genetic services, whether they understood the role of genetic counselling and if they were interested in furthering their genetics education. 57 questionnaires were completed and their data analysed. Results indicated that 29/57, 50.9% knew that genetic counselling services were available. Of those that were aware of the services, 15/29, 51.7% had referred patients to these services in the last year. Barriers to referral included misconceptions about the role of genetic counsellors, insufficient knowledge of which patients would benefit from genetic counselling and being unable to get appropriate information from the genetic clinic for patient referral. Facilitators identified included allied-healthcare professionals recognising the benefit of genetic counselling and wanting to increase their knowledge in genetics. Therefore, genetics education of allied-healthcare professionals and improved information regarding the referral procedure may improve the number of patients referred to genetic counselling.

Keywords: Allied-Healthcare Professionals**,** awareness, genetic counselling, genetic literacy, knowledge

Introduction

Genetic counselling has been found to empower patients by increasing feelings of control, self-efficacy, active decision-making and participation in self-management (Mcallister, Payne and Macleod, 2016). Genetic counselling is available in the state system in four cities in South Africa including Johannesburg, Cape Town, Stellenbosch and Bloemfontein and is only provided at a tertiary care level (Kromberg, Sizer and Christianson, 2013c). Genetic counselling services involve providing information and emotional support for prenatal genetic diagnoses, genetic screening, pre-symptomatic diagnostic testing, predictive testing, carrier testing and for patients with a confirmed genetic diagnosis (Kromberg and Krause, 2013a). In addition, due to failures in the South African healthcare system, genetic counsellors also refer patients for management of the symptoms of the genetic condition (Kromberg, Wessels and Krause, 2013b).

Due to the lack of awareness of what genetic counselling entails, its availability and benefit in both the public and amongst healthcare professionals, many patients are not referred for genetic counselling (Delikurt *et al.,* 2015). The percentage of patients that can benefit from genetic counselling and who are referred to the service is not known in South Africa. However, it has been reported that a minimum of 6.8% of births are affected by a congenital disorder and that of these 80.5% have a genetic or partially genetic cause (Malherbe *et al.,* 2015). This is in agreement with the birth prevalence of genetic disorders estimated by Christianson *et al*. (2006) which was 53.4 per 1000 live births. In 2017, approximately 17 000 babies were born at Chris Hani Baragwanath Academic Hospital (CHBAH) (Chris Hani Baragwanath Hospital, 2017) in Johannesburg. Based on the estimate of the prevalence of genetic disorders in live births determined by Christianson et al. (2006), approximately 923 babies would have been born with a genetic disorder at CHBAH in 2017. However, only 425 new patients were seen at the genetic clinic at CHBAH and many of these patients were not born in 2017. Therefore, more than half [498/923 (54%)] of babies born with a genetic condition in 2017 did not receive referral to genetic services, indicating the need to increase the referral rate.

There have been several studies internationally (e.g. Aalfs et al., 2003; Baars, Henneman & ten Kate, 2005; Claybrook et al., 2010; Freedman et al., 2003; Hayflick et al., 1998; Hunter et al., 1998; Klitzman et al., 2013) and one conducted in South Africa (van Wyk, 2008) which have evaluated doctors knowledge, perception and utilization of genetic services. The

international studies were conducted 10 years and more ago in developed countries, namely USA, the Netherlands and Canada. In all of these studies a questionnaire was administered to various types of physicians including oncologists, gynaecologists and paediatricians. The results indicated that health professionals' knowledge of genetic conditions and available genetic services is limited and doctors often do not address the psychosocial or ethical issues surrounding genetic conditions (de Abrew, Dissanayake & Korf, 2014). van Wyk administered a questionnaire to general practitioners in South Africa and found that there was a need to educate them about basic genetic concepts to improve the identification of at-risk patients that would benefit from genetic counselling (van Wyk, 2008).

Many individuals with genetic disorders present with developmental delay, speech delay, feeding problems and low muscle tone. Thus, they are frequently referred to allied-healthcare professionals for management. It has been reported that of the allied-healthcare professionals, physiotherapists, occupational therapists and speech-language therapists and audiologists are most frequently referred to (Lapham *et al.*, 2000). Thus, Allied-healthcare professionals are in a unique position to refer patients to genetic counselling as many of their patients have genetic diagnoses.

To date, there has only been one published study done 20 years ago, that evaluated referral to genetic services by allied- health professionals. The study was conducted in the USA and recruited dieticians, occupational therapists, physiotherapists, psychologists, speech-language therapists and social workers. In this study, a questionnaire was completed by 2052 participants. The results indicated that less than 20 % of allied-healthcare professionals referred patients for genetic counselling (Lapham *et al.*, 2000). The fact that the only study that has examined the number of patients referred to genetic counselling from allied health professionals was done internationally, indicates that there is a lack of literature surrounding this topic and a need to determine the rate of referral to genetic clinics from allied-healthcare professionals in the current South African context.

Purpose of this study

Aim

To determine whether allied-healthcare professionals' were aware of genetic counselling services in the hospitals in which they work, whether they have referred patients for genetic counselling and to identify barriers and facilitators to referral. This allowed recommendations to be made to increase the number of patients referred to genetic counselling.

Methods

Participants

Ethics clearance was obtained from the Human Research Ethics Committee (Medical) of the University of the Witwatersrand (Ethics Clearance Certificate no. M190246). Allied-healthcare professionals were recruited from three different state hospitals in Johannesburg. The inclusion criteria for this study were physiotherapists, occupational therapists and speech and language therapists who were registered with the Health Professions Council of South Africa and had been practicing for a year or more. The researcher contacted the head of the departments of physiotherapy, occupational therapy and speech and language therapy at the three hospitals and arranged to attend their departmental meeting where participants were informed about the study and given an information document, informed consent form and three-page questionnaire (Appendix C). These were then completed anonymously by each of the participants and placed in a sealed box which the researcher collected at an agreed upon time after the meeting. There are approximately 111 allied-healthcare professionals in total at the three hospitals but it is uncertain if all of them attended the departmental meetings.

Instrumentation

A three-page questionnaire was adapted from a survey used by Hayflick *et al*. (1998). In the Hayflick *et al*. study, a three-page validated survey was mailed to all members of the American Academy of Paediatrics, The American College of Obstetrics and Gynaecology and the American College of Physicians to determine their awareness and utilisation of genetic services. The questions that were chosen from Hayflick *et al.*'s study were selected and modified as needed based on their ability to provide information that fulfils the aims of this study (Table 1).

The first part of the questionnaire contained seven demographic questions including participant profession, qualifications, age, how long they have been practicing, how many patients they see per month and in which hospital they worked. The second section consisted of multiple-choice questions that were designed to assess the professional's knowledge of genetic services and if they had referred patients for genetic counselling in the last year. The questionnaire also assessed if participants understood the role of genetic counselling and if they were interested in furthering their genetics education (Tables 4 and 5).

Table 1: Research aims and corresponding questions from the administered questionnaire

Study Aims	Questions pertaining to each objective
Knowledge of genetic counselling	Q1; Q1.1; Q4; Q6
Knowledge of conditions to refer to genetic counselling	Q3
Knowledge of referral procedure to genetic counselling	Q2; Q5
Barriers and facilitators to genetic counselling referral	Q5, Q4
Suggestions for genetics education	Q7; Q8; Q9

Knowledge of which conditions should be referred for genetic counselling was evaluated by asking participants to indicate which conditions from a list should be referred for genetic counselling. The list was made up of six conditions that have a clear genetic aetiology, and eight which could be linked to a genetic syndrome but in isolation are usually not (Table 2).

Table 2: Diagnoses with a clear genetic aetiology and those that are isolated clinical features used to assess if allied-healthcare professionals know who to refer to genetic counselling.

Genetic Diagnosis	*Isolated Clinical Presentation*
Cystic Fibrosis	Autism
Down Syndrome	Cerebral Palsy
Charcot Marie Tooth Disorder	Low muscle tone
Prader Willi Syndrome	Failure to thrive
Spinal Muscular Atrophy	Hypermobility
Huntington Disease	Attention Deficit Hyperactivity Disorder
Duchene Muscular Dystrophy	Dyslexia
	Cleft lip and/or palate

Allied-healthcare professionals are not expected to make a diagnosis of a genetic syndrome. Therefore they would only be expected to refer patients who already have a genetic diagnosis. Thus, selecting only diagnosed genetic conditions was classified as a correct answer for the data analysis.

When knowledge of genetic counselling was assessed, participants were asked to select what they felt the responsibility of a genetic counsellor was. If all the correct answers and no incorrect answers were selected, the allied-healthcare professional was counted as understanding what genetic counsellors do.

Data Analyses

Outcomes of statistical tests and descriptive statistics (medians, percentages) were calculated using the Excel statistical package for responses to demographic items. The normality of the data was assessed using a Shapiro-Wilk test and none of the variables were found to be normally distributed. To determine whether the three professional groups of participants were similar and could be analysed together a Kruskal-Wallis test was used to determine if significant differences existed between any of the demographic details.

Descriptive statistics (percentages) were used to analyse the knowledge and awareness section of the questionnaire. Although certain questions in this section provided a five-point Likert-scale, the answers were analysed by grouping them into two categories that represented answers from either side of the scale and thus data from the middle or neutral option was excluded.

Results

There are approximately 111 allied health professionals in total practicing at the 3 hospitals. 80 participants completed the questionnaire. A total of 57 questionnaires were completed and analysed as questionnaires from 23 participants were excluded as they had not been practicing for a year or more. Occupational therapists made up the majority of the participants (22/57, 38.6%), followed by physiotherapists (18/57, 31.6%), and then speech and language therapists (17/57, 29.8%).

The demographic details were similar for the three groups of allied-healthcare professionals except for the number of patients seen per month. Physiotherapists saw significantly more patients than the other two groups (Table 3) ($P<0.01$).

Table 3: Demographic details of allied-healthcare professionals who participated in the study.

	Physiotherapists	Occupational Therapists	Speech and Language Therapists
Number of respondents	18	22	17
Median age and age range of participants	28.5 (23-49)	26.5 (23-50)	25 (24-37)
Median number and range of years practiced	5 (1-19)	3.5 (1-24)	3.5 (1-12)
Median number and range of patients per month per participant	170 (55-400)	78 (6-210)	47.5 (12-200)

Knowledge of genetic conditions that would benefit from referral to genetic services

Three of the respondents (3/57, 5.3%) were able to correctly identify all six genetic conditions that should be referred for genetic counselling. The majority of therapists knew that Down Syndrome (48/55, 87.3%), Huntington Disease (34/55, 61.8%) and Prader-Willi Syndrome (34/55, 61.8%) require referral. Charcot Marie Tooth Disorder was frequently missed (39/55, 70.9%) as a diagnosis requiring referral and the isolated clinical presentations with unclear aetiology that was incorrectly included most often was autism (20/55, 36.4%), cerebral palsy (14/55, 25.5%) and low muscle tone (14/55, 25.5%)

Knowledge of Genetic Counselling

Of the allied-healthcare professionals 29/57, 50.9% knew that genetic counselling was available in state healthcare facilities in Johannesburg. Of the 29 that knew the service was available, 15/29, 51.7% had referred patients in the last 12 months.

In order to determine whether the participants understood the role of genetic counsellors, they were asked to select options that they felt were the responsibility of a genetic counsellor. A total of 18/57, 31.6% correctly identified all 5 statements that described the role that genetic counsellors' have. The most frequently selected incorrect choice selected for the duty of a genetic counsellor was *To provide treatment of genetic conditions* (23/57, 40.4%).

Barriers and Facilitators to Genetic Counselling Referral

Participants were asked to rate how important certain factors were in influencing their decision to refer patients to genetic services (Table 4).

Table 4: Factors influencing allied-healthcare professionals' decision to refer patients for genetic counselling.

Factors influencing the decision to refer	Very Important/Important	Of Little Importance/Unimportant
Desire for management recommendations	49/55 (89.1%)	1/55 (1.8%)
Patients' interest in genetic counselling	48/55 (87.3%)	1/55 (1.8%)
Severity of the disorder	46/55 (83.6%)	4/55 (7.3%)
Availability of treatment for the disorder	36/55 (65.5%)	9/55 (16.4%)

***The sample size is reduced as neutral data was excluded.**

Table 5: Factors that prevent allied-healthcare professionals' from referring patients for genetic counselling.

Factors influencing the decision not to refer	Very Frequently/Frequently	Never/Rarely
Unaware of how to refer patients	33/54 (61.1%)	18/54 (33.3%)
Unaware of genetic services	26/54 (48.1%)	21/54 (38.9%)
Do not understand what genetic counsellors do	17/55 (30.9%)	25/55 (45.5%)
Unable to get appropriate information from the genetic clinic	14/54 (25.9%)	27/54 (50.0%)
Do not feel it is my responsibility to refer	12/55 (21.8%)	38/55 (69.1%)
Do not see much benefit for the patient	6/55 (10.9%)	46/55 (83.6%)

***The sample size is smaller than the total number of responses as neutral data was excluded.**
**** Further sample size reduction is due to sections not being completed.**

The most common factor which influence an allied-healthcare professional to provide referral to genetic counselling was if a family had expressed that they wanted to know the chance of a particular genetic condition occurring again.

Suggestions for Genetics Education

The majority (53/57, 93%) of participants agreed that they would be interested in furthering their knowledge in genetics. Most allied-healthcare professionals (51/56, 91.1%) said they would be interested in an overview of genetic conditions commonly seen by allied-healthcare professionals. The majority also indicated that they would like information on genetic resources for clients (49/57, 86%), information about the referral procedure to clinics (46/57, 80.7%), the role of genetic counselling (42/57, 73.7%) and information on new genetic technologies and advances (36/57, 63.2%).

Of the platforms that were suggested to increase genetic knowledge, 21/56, 37.5% of participants felt that discussion groups would be helpful, 29/56, 51.8% opted for written information, 38/56, 67.9% chose online resources and the majority (41/56, 73.2%) felt that academic lectures would be the most beneficial.

Discussion

Many allied healthcare professionals in our study were aware of genetic services in their hospitals but several barriers were found to hinder referral. These include insufficient knowledge of how to refer patients, which patients to refer, uncertainty around what genetic counsellors do and having little understanding of which patients with genetic conditions would benefit from referral. We also showed that there are facilitators which will improve referrals if they are utilised correctly.

Awareness of genetic counselling services

Suther and Goodson (2003) conducted a systematic review of the literature which explored barriers to the provision of genetic services amongst primary care physicians. This review highlighted that there was inadequate knowledge of genetic services amongst the medical professionals. We had similar findings as only half of the participants knew that genetic counselling services were available within the hospital in which they were practicing. To date there have been no other studies that have looked at allied-healthcare professional's awareness of genetic services. Because the allied-healthcare professionals are based within the hospitals where genetic services are offered, we would have expected a greater number to be aware of genetic services. .

Barriers preventing referral for genetic counselling

Number of patients referred to genetic services

Half of the allied-healthcare professionals in this study knew of genetic services but of the half that knew of the services. This indicates that insufficient knowledge of genetic services is one of the major barriers that prevent patient referral. Therefore, there is a need to determine which barriers are inhibiting referral. This is particularly important as there are only 13 genetic counsellors practicing in the state healthcare sector in South Africa. The small service has limited reach and there is a need to improve the number of patients referred to ensure that genetic counselling services have the best possible impact.

Allied-healthcare professionals did not refer as they said they were unable to get appropriate information from the genetic clinic. This is an important finding as it highlights that the way information is disseminated from the genetic clinics regarding the referral procedure needs to be improved. Awareness programs and marketing should be directed at allied-healthcare

professionals to increase the number of patients referred. In addition, the genetic clinic needs to be easily contactable.

Insufficient knowledge of which patients to refer

In addition to a lack of knowledge around how to refer patients, results indicated that the majority of allied healthcare professionals were unable to correctly identify all genetic conditions that would benefit from referral to genetic services. Certain conditions such as Down Syndrome, Huntington Disease and Prader-Willi Syndrome were identified by most allied-healthcare professionals as benefitting from referral. However, very few allied-healthcare professionals selected Charcot Marie Tooth Disease. This is possibly because Down Syndrome, Huntington disease and Prader-Willi Syndrome are well known genetic conditions whereas Charcot Marie Tooth Disease is not. The isolated clinical presentations that were incorrectly included most by the allied-healthcare professionals were autism, cerebral palsy and low muscle tone. These are unlikely to benefit from genetic counselling because the aetiology is unlikely to be genetic.

The uncertainty that allied-healthcare professionals have about which patients to refer is also likely to limit referrals. This indicates a need for allied-healthcare professionals to be informed about confirmed genetic diagnoses that would benefit from genetic counselling, regardless of presentation.

Misconceptions surrounding the genetic counselling profession

Findings indicated that there are misconceptions amongst allied-healthcare professionals about the role of genetic counsellors and this hinders referral. The majority of the respondents indicated that they understood what genetic counsellors do. However, when they were asked to select which factors influenced referral to genetic counselling, the option of *not understanding what genetic counsellors do* was not selected by the majority. This lead the researcher to infer that the participants felt that they understood what the role of a genetic counsellor was. However, even though the majority had indicated that they knew what genetic counsellors do in question 5, the low number of participants who selected the correct options for the role of a genetic counsellor in question 6, indicated that there were many misconceptions about the profession. The most common misconception that was incorrectly selected in question 6 was that genetic counsellors provided treatment of genetic conditions.

The availability of treatment for genetic conditions was selected as an important consideration by allied-healthcare professionals when deciding whether to refer a patient or not. This may hinder referral to genetic services as many genetic conditions are not treatable but patients can still benefit from genetic counselling because they gain information and emotional support (Resta *et al.*, 2006).

The majority of allied-healthcare professionals also said that the severity of the condition is important when considering referral to genetic counselling. The severity of a genetic condition is a subjective interpretation and thus the patient may consider the condition to be severe, even if the allied-healthcare professional does not. Therefore, the severity of a particular condition or whether or not there is treatment available does not detract from the benefit of genetic counselling.

The final barrier that was identified in this study was that allied-healthcare professionals felt that the patient's interest in genetic counselling was an important factor in their decision to refer them to genetic services. However, most patients are unaware of genetic counselling and will therefore not request it (Condit, 2010). Patients who could benefit from referral may therefore not be referred. Regardless of interest, allied-healthcare professionals should be advised to discuss the option of genetic counselling with their patient so that appropriate patients can be referred.

Facilitators of referral to genetic counselling

In this study the majority of allied-healthcare professionals said that they saw the benefit of genetic counselling for patients and agreed that it was their responsibility to refer patients. In addition, the majority recognised genetic counselling as being an important part of the management of patients. This indicates that should the allied-healthcare professional be able to recognise that a condition is genetic and know how to refer patients to the genetic clinic, the number of patients referred from allied-healthcare professionals may improve.

The vast majority (53/57, 93.0%) of allied-healthcare professionals said they would like to increase their genetic knowledge. This finding is similar to those of Lapham *et al.* (2000) who reported that of their participants, nearly 80% reported that they had taken no formal genetics courses in either their graduate of undergraduate programs and thus wanted to better their genetic education. This is encouraging as it suggests that there is an opportunity to increase allied-healthcare professional's knowledge of genetics, the role of the genetic counsellor and which patients would benefit from referral. The most favoured way of

disseminating this information would be through academic lectures that could be given by a genetic counsellor.

Study Limitations

Further investigation is required to confirm if the results of this study can be applied more generally to the allied-healthcare professionals in other parts of South Africa. However, it is likely that allied-healthcare professionals in the rest of South Africa have even less knowledge of genetic counselling because most genetic counsellors in South Africa work in Johannesburg. In addition, the questionnaire used in this study has not been validated However, as the questions asked were simple opinions and knowledge, we think that the data collected is valuable. It is possible that the sample may be biased as allied-healthcare professionals who already have knowledge of genetic counselling may have been more willing to complete the questionnaire. However, as 72.1% of allied-healthcare professionals completed the questionnaire it is unlikely that a bias would have had a major effect on the results.

Practice Implications

Despite the study limitations, practical suggestions can be made from our results. These suggestions will hopefully improve referral from allied-healthcare professionals to genetic counselling services. Allied-healthcare professionals indicated that they would like an overview of the genetic disorders commonly seen in their practice and information referral. In addition, information pertaining to the referral procedure to genetic clinics as well as contact information needs to be disseminated to the allied-healthcare professionals. Genetic counsellors should therefore arrange lectures at allied-healthcare professional's departmental meetings to educate allied-healthcare professionals in these topics. Distributing pamphlets to allied-healthcare professionals about genetic services may also be helpful. This may increase the number of patients with genetic conditions who are referred.

Research Recommendations

Future research should be conducted on a larger sample of allied-healthcare professionals to include those that are based in the four cities in South Africa where genetic counselling is available. A larger group will determine whether there is a lack of awareness across South Africa. Further research should be conducted to determine whether education of the allied-healthcare professionals increases the number of patients referred to genetic services and if

the aforementioned suggestions are useful. In addition, qualitative research could be utilised to gain a greater understanding of the opinions of the allied-healthcare professionals.

Conclusions

Genetic counselling has been shown to increase patient empowerment leading to positive outcomes. However, in Johannesburg State Healthcare many patients do not receive referral for genetic counselling. This study found that allied-healthcare professionals see the benefit of genetic counselling and consider it within their scope of practice to refer patients, however, there are barriers which hinder referral. These include a lack of knowledge of genetic services and which conditions to refer for genetic counselling. There are also misconceptions amongst allied-healthcare professionals as to what the genetic counselling profession entails and thus patients who would benefit from genetic counselling are often not referred. Both genetics education of allied-healthcare professionals and making contact information easily available may improve the number of patients referred to genetic counselling.

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Appendices

Appendices

Appendix A: Approved research protocol

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG

FACULTY OF HEALTH SCIENCES

CANDIDATE'S SURNAME: Thom	FIRST NAME/S: Jamey Emma	STUDENT NUMBER: 808293
CURRENT QUALIFICATIONS: BSc Biological Sciences; BHSc (Hons) Human Genetics		
TEL: 011 489 9216	CELL: 083 647 0601	E-MAIL: jameythom@gmail.com / FAX:
DEGREE FOR WHICH PROTOCOL IS BEING SUBMITTED: MSc (Med) Genetic Counselling		
PART-TIME OR FULL-TIME: Full-time		
FIRST REGISTERED FOR THIS DEGREE:	TERM: 1	YEAR: 2018
DEPARTMENT: Human Genetics		
TITLE OF PROPOSED RESEARCH: Awareness of Genetic Counselling Services Amongst Allied-Healthcare Professionals		
CANDIDATE'S SIGNATURE:		DATE: 30/01/2019
SUPERVISOR 1 (NAME & SURNAME): Tabitha Haw		% Supervision: 100
SUPERVISOR'S QUALIFICATIONS: BSc, BA (Hons), BSc (Hons), MSc (Med) Genetic Counselling		
SUPERVISOR'S DEPARTMENT: Human Genetics		
SUPERVISOR'S ADDRESS / TEL / E-MAIL: Room 2, First Floor, Jack Metz Building, Cnr Hospital & De Korte Str, Braamfontein/ Tel 011 489 9338/ Tabitha.Haw@nhls.ac.za		
SUPERVISOR 2 (NAME & SURNAME):		% Supervision
SUPERVISOR'S QUALIFICATIONS		
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SUPERVISOR'S QUALIFICATIONS		
SUPERVISOR'S ADDRESS / TEL / E-MAIL:		

SYNOPSIS OF RESEARCH: Please see next page.

ETHICS PENDING: (Y) / ETHICS APPROVED: Y (circle appropriate symbol)* To be submitted: 7/02/19	IF Y SUPPLY ETHICS CLEARANCE No:
***Please note human ethics clearance/waiver certificate or animal ethics certificate must be in the student's name**	
As supervisor, I confirm that I have read the protocol which has been submitted for assessment.	

SIGNATURE OF SUPERVISOR/S:	TSHew	UNIVERSITY OF THE WITWATERSRAND
SIGNATURE PG OFFICE STAFF	REGISTERED YES✓ NO.....	STAMP

SYNOPSIS OF RESEARCH

Introduction

Genetic counselling aims to ensure that genetic information is disseminated in a meaningful way that increases knowledge and provides psychosocial support. In South Africa, many individuals that have genetic conditions and who would benefit from genetic counselling are not receiving referrals to such services. Allied-healthcare professionals see patients that have been referred to them with confirmed genetic diagnoses. In addition, it is within their scope of practice to refer patients to other supplementary healthcare services. Therefore, they are in a unique position to note whether individuals have been seen for genetic counselling and refer them to the genetic clinic if necessary.

Aim

This study aims to determine whether allied-healthcare professionals are aware of genetic counselling services, whether they refer patients to genetic counselling and to identify barriers that may inhibit referral.

Method

This study is a cross-sectional study that will involve the administration of a self-report questionnaire to physiotherapists, occupational therapists and speech therapists based at Charlotte Maxeke Johannesburg Academic Hospital, Chris Hani Baragwanath Academic Hospital and Rahima Moosa Mother and Child Hospital who meet the inclusion criteria. The expected sample size is 89.This number is based on the number of allied-healthcare professionals reported to be working at the aforementioned hospitals and accounts for 20% of individuals who may be absent or choose not to participate.

Data Analysis

Information regarding demographic details, allied-healthcare professionals' knowledge of genetic conditions, genetic services, the referral procedure, barriers that prevent referrals and suggestions to improve genetics education and referral rate will be analysed quantitatively.

Potential Outcome

The results of this study will allow for suggestions to be made as to how to improve the referral rate from allied-healthcare professionals and how to better genetics education amongst these professionals.

2 May 2018/MP

Awareness of Genetic Counselling Services Amongst Allied-Healthcare Professionals:

Jamey Thom

808293

MSc (Med) Genetic Counselling

Supervisor:

Ms. Tabitha Haw

MSc (Med) Genetic Counselling, genetic Counsellor and Lecturer, Clinical and Counselling Section, Division of Human Genetics, School of Pathology, Faculty of Health Sciences, University of the Witwatersrand

1. Background literature analysis and critique

1. Genetic Counselling

Genetic counselling is defined as "the process of helping people understand and adapt to the medical, psychological and familial implications of the genetic contribution to disease." (Resta *et al.*, 2006).When patients with confirmed genetic diagnoses are referred to a Genetic Clinic, genetic counsellors are tasked with the responsibility of interpreting family and medical histories to determine the possibility of disease occurrence/recurrence and to provide support. In addition, their role is to educate patients about inheritance, testing, management, prevention, resources and research (Resta *et al.*, 2006).

Genetic counselling is based on non-directive, client-centred principles in order to preserve the patient's autonomy and feelings of worth and competence (Kromberg, Wessels and Krause, 2013). Psychosocial counselling is also provided to patients in order to promote the making of informed choices and management of their condition (Resta *et al.*, 2006).

Within the Genetic Clinics, genetic counsellors provide patients with multiple services. These include counselling for prenatal genetic diagnosis during pregnancy, counselling patients with known diagnoses and counselling surrounding carrier, diagnostic and predictive testing for adult onset conditions. The genetic counsellors ensure that the genetic information is distributed in a meaningful way that increases knowledge and provides psychosocial support (Kromberg, Sizer and Christianson, 2013). In order to carry out these genetic services, genetic counsellors must have counselling skills, detailed knowledge of human genetics and testing available, as well as the ability to communicate complex genetic information to patients (Kromberg, Wessels and Krause, 2013).

1.1 Genetic Counselling Services in South Africa

The genetic counselling profession is a developing healthcare profession which requires a MSc (Med) degree and registration with the Health Professions Council of South Africa (HPCSA) (Kromberg, Wessels and Krause, 2013). In Johannesburg, genetic counselling services are available in both the private and public sector (Kromberg, Wessels and Krause, 2013). This study will focus on genetic counselling services that are provided in the public sector at Charlotte Maxeke Johannesburg Academic Hospital, Chris Hani Baragwanath Academic Hospital and Rahima Moosa Mother and Child Hospital (Kromberg, Sizer and Christianson, 2013). Genetic Clinics are held at each of these hospitals on alternate weeks. Services at the Genetic Clinic are provided by medical geneticists and genetic counsellors (Kromberg,

Wessels and Krause, 2013). Medical geneticists see patients who do not yet have a diagnosis and thus their role is both diagnostic and to offer counselling to affected patients and families (Kromberg, Wessels and Krause, 2013). Genetic counsellors usually consult with patients with confirmed diagnoses such as chromosome abnormalities, single gene disorders such as haemophilia and cystic fibrosis as well as counselling pregnant women at risk of having a baby with a birth defect (Skirton *et al.*, 2015). Thus, their role is to counsel patients with these diagnoses and to offer information, psychosocial support and management options.

2. Prevalence of Genetic Conditions and Referrals

In South Africa, it has been reported that a minimum of 6.8% of births are affected by a congenital disorder and that of these 80.5% have a genetic or partially genetic cause (Malherbe, 2015). This is in agreement with the birth prevalence of genetic disorders which was estimated at 53.4 per 1000 live births by Christionson *et al.*, in 2006. However, while there are multiple individuals born with genetic conditions, it is evident that those who would benefit from genetic counselling are not being referred to Genetic Clinics. For example, each year there are around 17 000 babies born at Chris Hani Baragwanath Academic Hospital. (Chris Hani Baragwanath Hospital, 2017). Based on the birth prevalence estimate of the number of babies born with genetic disorders that was provided by Christionson *et al.*'s study, this means that around 923 babies would have been born with genetic disorders in 2017. A review of the patients seen at the Genetic Clinic at Chris Hani Baragwanath Academic Hospital every alternate Thursday revealed that in 2017 only 425 new patients were referred to the Genetic Clinic. Therefore, in 2017 an estimated 498/923 (54%) of individuals with genetic diagnoses did not receive a referral to genetic services. This indicates that it is very likely that the majority of patients with genetic disorders are not receiving referral to genetic counselling at Chris Hani Baragwanath Academic Hospital and emphasises the need to increase the referral rate to genetic services.

2.1. Referral to Genetic Counselling

When a physician diagnoses a patient with a genetic condition it is their responsibility to design treatment and management plans which aim to improve the symptoms of the disorder. Many patients that have genetic disorders have developmental delay, speech delay, feeding problems and low muscle tone and are thus frequently referred to allied-healthcare professionals for treatment (Lapham *et al.*, 2000). It has been reported that of the allied-healthcare professionals, referrals to physiotherapists, occupational therapists and speech therapists occur most frequently (Lapham *et al.*, 2000). Therefore, it is expected that patients with the following

confirmed genetic diagnoses (Table 1) will be frequently referred to the aforementioned allied-healthcare professionals for the management of their symptoms.

Table 1: Common genetic conditions seen by Physiotherapists, Occupational Therapists and Speech Therapists for the treatment of motor, speech and developmental concerns.

Allied-Healthcare Professional	Common Conditions Seen
Physiotherapists	Down Syndrome, Duchene Muscular Dystrophy, Haemophilia, Spinal Muscular Atrophy, Cystic Fibrosis
Occupational Therapists	Down Syndrome, Turner Syndrome, Prader Willi Syndrome
Speech Therapists	Chromosome abnormalities, Huntington's Disease, Fragile X Syndrome

2.2 Referral to Genetic Counselling by Allied-Healthcare Professionals

Amongst the healthcare professionals that should be referred to when a physician makes a genetic diagnosis is a genetic counsellor. A study conducted in South Africa in 2013 indicated that general practitioners make up the second largest referral source to Genetic Clinics (Kromberg, Wessels and Krause, 2013). Ideally, physicians should be the individuals who refer patients to genetic counselling as they are the professionals who first consult with patients. However, as aforementioned, there are a number of patients who are missed. Owing to the fact that allied-healthcare professionals are seeing patients that have been referred to them with confirmed genetic diagnoses, it affords them the opportunity to note individuals who have not yet been seen for genetic counselling and subsequently refer them to the Genetic Clinic.

3. Scope of Practice of Allied-Healthcare Professionals

In the HPCSA guidelines that outline the scope of practice of physiotherapy in South Africa, it states that it is the responsibility of the therapist to "select treatment techniques according to the diagnosis given by the referring medical practitioner and in conjunction with other supplementary health services personnel connected with the treatment and management of the patient" (Health Professions Council of South Africa,1976). In a paper by Van Der Reyden which evaluated the legislation for everyday occupational therapy practice, the author highlights that a registered occupational therapists is responsible and accountable for the

delegation of tasks and referrals (Reyden, 2010). Regulations that define the scope of practice of speech-language therapy states that it is within the clinical services of a speech therapist to provide intervention and support services for patients (Department of Health, 2017). This indicates that there are published guidelines that state that it is within the scope of practice of physiotherapists, occupational therapists and speech therapists to refer their patients to additional services. It is thus important to determine whether these allied-healthcare professionals are aware of genetic counselling as an additional service for the management and support of patients with genetic diagnoses.

4. International Findings Regarding Referral

A study conducted in the United States of America found that allied-healthcare professionals are not adequately informed about the availability and role of genetic counselling and thus appropriate referrals are often not made (Lapham *et al.*, 2000). Lapham *et al.* aimed to determine the genetics education needs of allied-healthcare professionals. They observed that allied-healthcare professionals estimated that 16% of their patients had a genetic disorder but that less than 20% of the individuals that took part in the study had referred their patients for genetic counselling (Lapham *et al.*, 2000). The fact that the only study that addresses this issue is international, indicates that there is both a lack of literature surrounding this topic and a need to determine the rate of referral to Genetic Clinics from allied-healthcare professionals in the current South African context.

5. Barriers to Genetic Counselling Referral

While this study aims to determine the referral rate from allied-healthcare professionals to genetic counselling, it is important to explore previously reported barriers to genetic counselling referral. This is important as it will allow for evaluation of whether or not similar barriers exist in South Africa. The first has been found to be a lack of knowledge of genetics and genetic conditions (Weitz, 2010; Delikurt *et al.*, 2015). Allied-healthcare professionals with genetics knowledge are more likely to refer patients to Genetic Clinics (Guilbert and Cheater, 1990; Hayflick *et al.*, 1998), however, studies have indicated that allied-healthcare professionals are not able to recognise patients with genetic conditions who would benefit from genetic counselling. As a result referrals are not made (Delikurt *et al.*, 2015).

The second barrier that has been found to exist is inadequate awareness of genetic services (Delikurt *et al.*, 2015). A study conducted in 2016 which looked at general practitioners (GPs) based in the private healthcare sector in Johannesburg, South Africa, found that GPs had a

limited knowledge of available genetic counselling services (van Wyk *et al.*, 2016). Although this study evaluated the knowledge of GPs, a lack of awareness of genetic services in allied-healthcare professionals would similarly prevent them from utilising opportunities to refer patients that are seen at their clinics to a genetic counsellor. Closely linked to this is a lack of knowledge on how to refer patients (Delikurt *et al.*, 2015). This situation was highlighted in Lapham *et al.*'s study that concluded that there is a need for guidelines on how to make appropriate referrals (Lapham *et al.*, 2000).

A third barrier which exists is the attitude of the allied-healthcare professional (Delikurt *et al.*, 2015). Research has indicated that individuals who have negative attitudes, misconceptions or who do not believe in the utility of genetic services are less likely to refer their patients for genetic counselling or other genetic services (Hayflick *et al.*, 1998; Delikurt *et al.*, 2015).

6. Scope of research

In light of the suspected high number of patients with genetic conditions that are not being referred for genetic counselling, it is firstly important to determine whether or not allied-healthcare professionals are referring patients with confirmed genetic diagnoses to genetic counselling. Secondly, the fact that allied-healthcare professionals see patients with genetic conditions and it is within their scope of practice to refer, it is important to evaluate these professionals' knowledge of genetic services and the referral procedure in order to determine if there is poor knowledge of genetic services, especially if the referral rate is low. In addition, this study is needed to identify barriers that prevent referrals from taking place. Once all these factors have been examined, the research could potentially be used to make recommendations as to how to decrease barriers and increase knowledge and in addition increase the referral rate from allied-healthcare professionals.

2. Aims and Objectives

2.1 Aim

To determine whether allied-healthcare professionals are aware of genetic counselling services, whether they refer patients for genetic counselling and to identify barriers that may inhibit referral.

2.2 Objectives

- To evaluate whether allied-healthcare professionals are able to determine which diagnosed conditions would benefit from genetic counselling.
- To determine whether allied-healthcare professionals are aware of available genetic services and what genetic counselling entails.
- To determine whether or not allied-healthcare professionals are referring patients with confirmed genetic diagnoses to Genetic Clinics.
- If allied-healthcare professionals are not found to be referring patients to Genetic Clinics, to identify barriers that inhibit referrals to Genetic Clinics.
- To make recommendations as to how to improve the referral rate to Genetic Clinics.

3. Methods

3.1 Study Participants

The study sample will consist of allied-healthcare professionals, namely physiotherapists, occupational therapists and speech therapists. The Division of Human Genetics (NHLS/Wits) has Genetic Clinics at Charlotte Maxeke Johannesburg Academic Hospital, Chris Hani Baragwanath Academic Hospital and Rahima Moosa Mother and Child Hospital and thus the study participants will be recruited from these hospitals. An information document, informed consent form (appendix one) and an adapted three-page questionnaire (appendix two) that will take approximately five to ten minutes to complete will be administered to each of the participants who consent. The expected sample size is 89. This number is based on the number of physiotherapists, occupational therapists and speech therapists based at the three aforementioned teaching hospitals who meet the inclusion criteria and accounts for 20% of individuals who may be absent or choose not to participate (Table 2)

Table 2: The number of Allied-Healthcare Professionals at each hospital per profession.

CMJAH	Physiotherapists	16
	Occupational Therapists	11
	Speech Therapists	8
CHBAH	Physiotherapists	36
	Occupational Therapists	20
	Speech Therapists	13
RM	Physiotherapists	2
	Occupational Therapists	2
	Speech Therapists	3
Total		**111**
Total with 20% non-participation/absence		**89**

The researcher will obtain permission letters from the head of physiotherapy, occupational therapy and speech therapy from each of the hospitals. In addition, permission will be obtained from the CEOs of each of the hospitals to conduct the research.

3.2 Inclusion Criteria

1. Physiotherapists, occupational therapists and speech therapists that are registered with the HPCSA.

2. Physiotherapists, occupational therapists and speech therapists with working experience in the current hospital of one year or more.

3.3 Exclusion Criteria

1. Physiotherapists, occupational therapists and speech therapists that have only completed the demographic information section of the questionnaire.

2. Physiotherapists, occupational therapists and speech therapists that have completed the entire questionnaire, excepting the demographic section.

3.4 Study Design

This is a quantitative, cross-sectional study which will involve the completion of a self-reported questionnaire (appendix two). Owing to the need to conduct this research in a short period of time, the questions that make up the questionnaire are not original and have been adapted from previously published research conducted by Hayflick *et al.* in 1998. In their study, a three-page validated survey was mailed to all members of the American Academy of Paediatrics, The American College of Obstetrics and Gynaecology and the American College of Physicians to determine their awareness and utilisation of genetic services. The questions that have been utilised from this study have been adapted for this research report to evaluate allied-healthcare professional's perception and utilisation of genetic services.

The questions that have been chosen from Hayflick *et al.*'s study were selected based on their ability to provide information that fulfils the five objectives of this research (Table 3). Therefore, questions were selected from Hayflick *et al* 's study based on their ability to provide information on the first objective which is whether or not allied-healthcare professionals are aware of available genetic services and what genetic counselling entails.

The second objective that questions were selected to answer was to evaluate whether allied-healthcare professionals are able to determine which diagnosed conditions would benefit from

genetic counselling. It should be highlighted that this study only aims to determine if allied-healthcare professionals refer individuals with a confirmed genetic diagnosis to genetic counselling. This is to ensure that the number of patients with confirmed genetic diagnoses that are seen increase but that inappropriate referrals of patients with non-genetic causes do not occur.

Thirdly, questions were selected to determine whether or not allied healthcare professionals are referring patients with confirmed genetic diagnoses to Genetic Clinics. Questions were also selected to identify barriers that prevent referrals to the Genetic Clinic and lastly, questions were formulated to evaluate the best means of educating allied-healthcare professionals. This information obtained will also allow for recommendations to be made as to how to improve the referral rate.

Table 3: Research objectives and corresponding questions from study questionnaire

Study Objectives	Questions pertaining to each objective
Knowledge of genetic conditions	Q1; Q1.1; Q4; Q6
Knowledge of genetic counselling	Q3
Knowledge of referral procedure to genetic counselling	Q2; Q5b
Barriers to genetic counselling referral	Q5
Suggestions for genetics education	Q7; Q8; Q9

Questions included in the questionnaire are multiple choice or require the selection of an answer that utilises a likert scale which represents the participants attitude towards the question. There is one open ended question that welcomes additional thoughts and comments. Demographic data that will be collected includes the hospital at which the participant works, their qualifications, profession, age, length of time for which they have been practicing, the average number of patients seen per month and the department in which they work. In order to ensure that the questionnaire is easy to understand it will be shared with genetic counsellors and doctors within the Human Genetics department of the NHLS for feedback in order to eradicate any problems with wording or understanding.

The administration of the questionnaires will be done according to the following flow diagram (Figure 1). The questionnaire will be administered in person to ensure that a larger sample is collected as email communication may be lost or go unnoticed. The researcher will attempt to attend a meeting when all the employees are together but this may not occur as not all

departments have group meetings. In that event, a convenient time for the majority will be set-up.

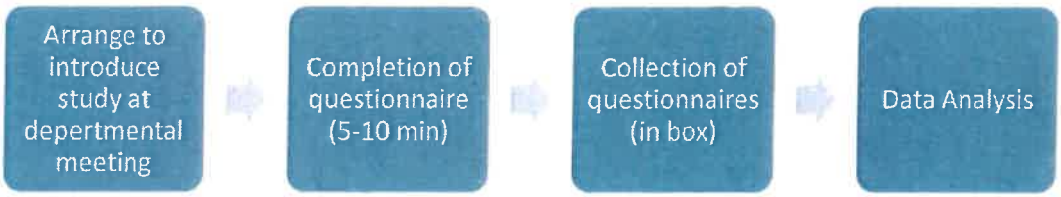

Figure 1: A flow diagram showing the steps that will be followed to administer the questionnaire to the Allied-Healthcare Professionals that will be included in the study.

3.5 Data Analysis

The data collected during this study will be analysed quantitatively and descriptive statistics will be used to summarise the data. Data analysis will occur under the five objectives of this study. Each of the questions will be counted individually according to the frequency of answers. This will allow for percentages to be determined as to how many individuals gave a certain answer for each question. Frequencies will be captured in excel and reported in categories for each profession and for the demographic details. A chi-squared test will be used to note any differences between the demographic details, if the sample size is large enough.

7. Ethics

All participants of the research project will be informed that this project aims to determine the awareness of genetic counselling services amongst allied-healthcare professionals. The reason why more information will not be given is to ensure that the answers of the participants are not biased. The participation will be entirely voluntary and should individuals decline to participate, it will not involve any penalty or loss of benefits to which the participant is otherwise entitled. All participants will sign an informed consent document (appendix one). The data obtained will be kept confidential as only the researcher and supervisor will have access to the original questionnaires. Submission to the Human Research Ethics Committee (Medical) of the University of the Witwatersrand will be made on the 7th February 2019.

8. Timing

	Dec 2018	Jan 2019	Feb 2019	Mar 2019	Apr 2019	May 2019	Jun 2019	Jul 2019
Literature Review								
Protocol Write-up								
Protocol Assessment								
Ethics Application								
Collecting Data								
Data Analysis								
Writing up: Research Report								
Writing up: Paper								

9. Budget

9.1 Predicted Budget

Resource	Cost	Source of Funding
Researcher's travel expenses (minimum 5 trips to CMJAH, CHBAH and RM) = 79.5km at R1.02/km	R405.45	Self-funded
Printing and binding of research report (27 pages) (x2) at 39c per page	R21.06	Self-funded
Photocopying of three-page questionnaires (111) at 39c per page	R43.29	Covered by the Department
Total	**R469.80**	

10. Limitations

One of the limitations of this study is the availability of the allied-healthcare professionals. The completion of the questionnaire will take some time and certain individuals may feel they are too busy to complete it. This may lead to a reduction in the number of individuals who take part in the study and as a result may lead to a decreased sample size. A second limitation is obtaining all the permission letters needed to conduct this research. Owing to the fact that a large number of permission letters need to be obtained and doing so takes time, permission may not be received to conduct the research in all the hospitals. This could lead to a decrease in sample size which will affect data analysis. The last problem is that the questionnaire that will be administered is not validated. Therefore, the results will need to be considered with this in mind.

11. References

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Appendix One

INFORMTION DOCUMENT

Dear Sir/Madam

Introduction: My name is Jamey Thom and I am a MSc (Med) genetic Counselling Masters student in the Division of Human Genetics, National Health Laboratory Services (NHLS) and University of the Witwatersrand (Wits). As part of my studies, I have to undertake a research project, and I am investigating the awareness of genetic counselling services amongst allied-healthcare professionals.

As part of this project I would like to invite you to take part in answering a questionnaire. This activity will involve you answering ten questions once off and will take about five to ten minutes to complete.

You will not receive any direct benefits from participating in this study, and there are no disadvantages or penalties for not participating. You may withdraw at any time or not answer any question if you do not want to. The questionnaire will be completely confidential and anonymous as I will not be asking for your name and the information you give to me will be held securely and will not be disclosed to anyone else but my supervisor.

If you have any questions about this research, feel free to contact me on the details listed below. This study will be written up as a research report. If you wish to receive a summary of this report, I will be happy to send it to you upon request. If you have any queries, concerns or complaints regarding the ethical procedures of this study, you are welcome to contact the University Human Research Ethics Committee, telephone (medical) 011 717 1252, email shaun.schoeman@wits.ac.za.

Yours sincerely,
Jamey Thom

Researcher: Miss Jamey Thom | 011 489 9216 | jamey.thom@nhls.ac.za
Supervisor: Ms. Tabitha Haw | 011 489 9338 | tabitha.haw@nhls.ac.za

NATIONAL HEALTH LABORATORY SERVICE
School of Pathology, University of the Witwatersrand

DIVISION OF HUMAN GENETICS

WITS UNIVERSITY

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[T] +27 11 489 9211 | [F] +27 11 489 9226 | [E] human genetics@nhls.ac.za

Appendix One

Informed Consent

With regards to the research study entitled "**Awareness of Genetic Counselling Services Amongst Allied-Healthcare Professionals**" I (Name), _____consent to answering a self-reported questionnaire and the results being analysed by the researcher and supervisor.

- I consent to the use of the data collected from the questionnaire to be used for the aforementioned project.
- I acknowledge that I have read and understood the information document.
- I understand that findings from this study will not be reported back to me and that results may be published within accredited journals.
- I understand that my information will remain anonymous unless required by law.

Participant Signature_____ Date_____

Witness Signature_____ Date_____

1 Modderfontein Road, Sandringham, Johannesburg, South Africa

Prof Eric Buch Dr Karmani Chetty
Private Bag X8, Sandringham, 2131, South Africa
+27 (0) 11 386 6000/ 0860 00 NHLS(6457)
5200296

Appendix Two

Allied-Healthcare Professionals Genetics Survey

Please answer the following questions about yourself and your practice.

Hospital_____

Profession_____

Qualifications_____

Age_____

How long have you been practicing? _____

How many patients do you see on average per month? _____

In which department do you work? _____

1. Is referral for genetic counselling or consultation available to you? (circle one)

a) Yes
b) No
c) Don't know
d) It's not my role

1.1 If yes, to whom would you refer your patients?

_____

2. If it is available to your patients, approximately how many referrals to genetic services have you made in the past 12 months? (circle one)

a) None
b) 1-2
c) 3-5
d) 6-10
e) >10

3. If you were to refer patients for genetic counselling, which of the following diagnosis would you refer? (circle all that apply)

a) Autism
b) Cystic Fibrosis
c) Cerebral Palsy
d) Down Syndrome
e) Duchene Muscular Dystrophy
f) Attention Deficit Hyperactivity Disorder
g) Charcot Marie Tooth Disorder
h) Cleft lip and/or palate

4. When referral to genetic counselling is made, how important are the following factors in your decision to refer? (select one for each option)

	Very Important	Important	Moderately Important	Of Little Importance	Unimportant
a. Patients interest in genetic counselling					
b. Severity of the disorder					
c. Availability of treatment for the disorder					
d. Desire for management recommendations					
e. Family's desire for recurrence information					

5. When you do not to refer to genetic counselling, how often is each of the following a reason? (select one for each option)

	Very Frequently	Frequently	Occasionally	Rarely	Never
a. Was not aware of genetic services					
b. Do not know how to refer patients					
c. Do not see much benefit for the patient					
d. Do not understand what genetic counsellors do					
e. Unable to get appropriate information from the Genetic Clinic					
f. Do not feel it is my responsibility to refer					

6. Of the following, which do you feel is the responsibility of a genetic counsellor? (circle all that apply)

a) To help couples have a child with desirable characteristics
b) To provide emotional support to patients and families coping with a genetic condition
c) To provide information about how genetics contributes to health problems
d) To provide treatment of genetic conditions
e) To provide information about the chances of a child having a genetic condition
f) To help people understand their options for genetic testing
g) To provide referrals and resources for families with a genetic condition
h) To help prevent the birth of children with genetic conditions

7. Would you be interested in furthering your genetics knowledge? (tick one)

Strongly Agree		Agree		Undecided		Disagree		Strongly Disagree	

8. If you would like to increase your knowledge about genetics, in which areas would you like to do so? (circle all that apply)

a) Overview of genetic disorders commonly seen by allied-healthcare professionals
b) Identifying genetic resources for clients
c) Genetic counselling and what the profession entails
d) Referral procedure to genetic clinics
e) New genetic technologies and advances

9. If you would like to increase your knowledge, which of the following educational resources do you feel would be the most beneficial? (circle all that apply)

a) Online resources
b) Written information
c) Academic lectures
d) Discussion groups

10. We welcome any additional thoughts and comments

Thank you very much for your time!

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG | FACULTY OF HEALTH SCIENCES

Appendix Three

STATEMENT OF PRINCIPLES FOR POSTGRADUATE SUPERVISION

IN A CONTEXT OF ACADEMIC FREEDOM AND WITHIN A FRAMEWORK OF INDIVIDUAL AUTONOMY AND THE PURSUIT OF KNOWLEDGE THIS STATEMENT IS WRITTEN IN THE BELIEF THAT THERE IS A RECIPROCAL RELATIONSHIP AND MUTUAL ACCOUNTABILITY BETWEEN SUPERVISOR AND STUDENT.

THE SUPERVISOR AND THE STUDENT:

1. Will establish agreed roles and clear processes to be maintained by both parties. In the case of join supervision everybody's role needs to be clarified.
2. Will meet regularly and as frequent as is reasonable to ensure steady progress towards the completion of the proposal, research report, or dissertation or thesis. This time varies but the normal minimum requirement for face-to-face contact spread across each year of registration is: 10 contact hours for an Honours project, 15 contact hours for a Masters by a research report and 24 contact hours for a Masters by dissertation and a PhD.
3. Will keep appointments, be punctual and respond timeously to messages.
4. Will keep one another informed of any planned vacations or absences as well as changes in his/her personal circumstances that might impact on the work schedule. Unplanned absences or delays should be discussed as soon as possible and arrangements should be made, to catch up lost time.
5. Will ensure that research on animal or human subjects is concluded according to the procedures and the requirements of the relevant University Ethics committee.
6. Will together complete progress reports on the research project, as requested by each Faculty Graduate Studies Committee.

THE SUPERVISOR

1. Undertakes to provide guidance for the student's research project in relation to the design and scope of the project, the relevant literature and information sources, research methods of data analysis.
2. Has a responsibility to be accessible to the students.
3. Will be prepared for the meeting with student. This includes being up to date on the latest work in his/her area of expertise.
4. Will expect written work as jointly agreed, and will return that work with constructive criticism within a timeframe (a suggestion of 2-4 weeks) jointly agreed at the outset of the research.
5. Will provide advice that can help the student to improve his/her writing. This may include referrals for language training and academic writing. The supervisor will provide guidance on technical aspects of writing such as referencing as well as on the discipline specific requirements. Detailed correction of drafts and instruction in aspects of language and style are not the responsibility of the supervisor.

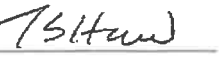
6. Will support the student in the production of a research report, dissertation or thesis. Provision should be allowed for adequate, mutually respectful, discussion around recommendations made.
7. Will assist with the construction of a written time schedule, which outlines the expected completion dates of successive stages of the work.
8. Will ensure the student has the opportunity to present work at postgraduate/staff seminars/national/international conferences as appropriate.
9. Will assist with the publication of research articles appropriate.
10. Will discuss the ownership of research conducted by the student in accordance with the University guidelines and rules on intellectual property, co-authorship and copyright.
11. Will ensure that the research is conducted in accordance with the University's policy on plagiarism.
12. Will ensure that the student is made aware in writing of the inadequacy of progress and/or of any work where the standard is below par. Acceptability will be according to criteria previously supplied to the student.
13. Has a duty to refuse to allow the submission of sub-standard work for examination, regardless of the circumstances. If the student chooses to submit without the consent of the supervisor, then this should be clearly recorded and the appropriate procedures followed.

THE STUDENT

1. Undertakes to work independently under the guidance of the supervisor. This includes reading widely to ensure that the literature pertinent to his/her chosen topic has been identified and consulted.
2. Is obliged to make appointments to see the supervisor and will arrange meeting times well in advance.
3. Will think carefully about how to get maximum benefit from these contact sessions by planning what s/he wants in these sessions.
4. Should submit written work for discussion with the supervisor well in advance of a scheduled meeting. The kind and frequency of written work should be agreed with the supervisor at the outset of the research.
5. Written work that is submitted should be relatively free from basic spelling mistakes, incorrect punctuation and grammatical errors. Responsibility for the accuracy of language, the overall structure and coherence of the final research report, dissertation or thesis rests with the student.
6. Undertakes to heed the advice given by the supervisor and to engage in discussion around suggestions made. Ultimately the student has to take responsibility for the quality and presentation of the work.
7. Should strive, within reasonable bounds, to maintain a focus on his/her research area and to work within the agreed time schedule.
8. Will prepare material for presentations at seminars and conferences.
9. Undertakes to submit papers for publication.
10. Agrees to honour agreements about ownership of the research and in accordance with the University's guidelines and rules in relation to co-authorship, copyright and intellectual property.
11. Will ensure that the work contains no instances of plagiarism and that all citations are properly referenced and that the list of references is accurate, complete and consistent.
12. Agrees to work in accordance with the criteria of acceptability as supplied by the supervisor.
13. Undertakes not to place the supervisor under undue pressure to submit work for examination until the supervisor is satisfied that it has reached an acceptable level of quality. We confirm that we have read and understood this statement and agree to be guided by its principles for as long as we continue to work together.

Please note: The University and Faculty endorse the **Singapore Statement** on research integrity. The principles of the Singapore Statement include honesty, accountability, professionalism and stewardship. Our responsibilities as researchers (i.e. both as students and supervisors) are as pledged in the Singapore Declaration: the assurance of appropriate "data integrity, data sharing, record keeping, authorship, publication, peer review, conflict of interest, reporting misconduct and irresponsible research, communicating with the public, complying with regulations, education, and social responsibilities (World Conference on Research Integrity, 2010)". These principles and responsibilities are important in training our postgraduate students and in promoting global research integrity. Ref: Resnik, DB and Shamoo AE (2011). The Singapore Statement on Research Integrity.Account Res. 18:71-75.

Name of student: Jamey Thom Student's signature: 

Name of Supervisor: Tabitha Haw Supervisor's signature: 

Name of Co-Supervisor: N/A Co-Supervisor's signature: N/A

The broad area of study is: Awareness of Genetic Counselling Services Amongst Allied-healthcare Professionals

Provisional submission date is: 31/07/2019 Degree: MSc (Med) Genetic Counselling

School: Pathology Faculty: Health Sciences

Date: 30/01/2019

Specific agreement pertaining to: ownership and joint publication, funding, may be attached and signed.

GRIEVANCE PROCEDURES: It should be acknowledged that during the course of the research that both students and supervisors can feel aggrieved. In this event, these should be dealt with as swiftly as possible by the parties involved and, if necessary, the Postgraduate Coordinators and Committees. There is, in addition, a University Grievance Policy to help guide deliberations.

Appendix B: Ethic clearance certificate

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG

HUMAN RESEARCH ETHICS COMMITTEE (MEDICAL)

Office of the Deputy Vice-Chancellor (Research & Post Graduate Affairs)

TO: Ms J Thom
School of PathologySchool of Pathology
Department of Human Genetics
National Health Laboratory Service

E-mail: jameythom@gmail.com

CC: Supervisor: Ms T Haw <Tabitha.Haw@nhls.ac.za>
and <HREC-Medical.ResearchOffice@wits.ac.za>

FROM: Iain Burns
Human Research Ethics Committee (Medical)
Tel: 011 717 1252

E-mail: Iain.Burns@wits.ac.za

DATE: 2019/05/21

REF: *R14/49*

PROTOCOL NO: **M190246** *(This is your ethics application study reference number. Please quote this reference number in all correspondence relating to this study)*

PROJECT TITLE: *Awareness of genetic counselling services amongst allied health care professionals*

Please find attached the Clearance Certificate for the above project. I hope it goes well and that an article in a recognized publication comes out of it. This will reflect well on your professional standing and contribute to the Government funding of the University.

MSWorks2000/Iain0007/Clearscan.wps

UNIVERSITY OF THE
WITWATERSRAND
JOHANNESBURG

R14/49 Ms J Thom

HUMAN RESEARCH ETHICS COMMITTEE (MEDICAL)
CLEARANCE CERTIFICATE NO. M190246

NAME: (Principal Investigator)	Ms J Thom
DEPARTMENT:	School of Pathology Department of Human Genetics National Health Laboratory Service
PROJECT TITLE:	Awareness of genetic counselling services amongst allied health care professionals
DATE CONSIDERED:	2019/02/22
DECISION:	Approved unconditionally
CONDITIONS:	
SUPERVISOR:	Ms T Haw
APPROVED BY:	Dr CB Penny, Chairperson, HREC (Medical)
DATE OF APPROVAL:	2019/05/21

This clearance certificate is valid for 5 years from date of approval. Extension may be applied for.

DECLARATION OF INVESTIGATORS

To be completed in duplicate and **ONE COPY** returned to the Research Office Secretary on the 3rd Floor, Phillip Tobias Building, Parktown, University of the Witwatersrand, Johannesburg.
I/we fully understand the conditions under which I am/we are authorized to carry out the above-mentioned research and I/we undertake to ensure compliance with these conditions. Should any departure be contemplated, from the research protocol as approved, I/we undertake to submit details to the Committee. **I agree to submit a yearly progress report**. When a funder requires annual re-certification, the application date will be one year after the date when the study was initially reviewed. In this case, the study was initially reviewed in **February** and will therefore reports and re-certification will be due early in the month of **February** each year. Unreported changes to the application may invalidate the clearance given by the HREC (Medical).

Principal Investigator Signature

Date

PLEASE QUOTE THE CLEARANCE CERTIFICATE NUMBER IN ALL ENQUIRIES

Appendix C: Turn-it-in report

808293:Research_Report_-_Final.docx

ORIGINALITY REPORT

0%	0%	0%	0%
SIMILARITY INDEX	INTERNET SOURCES	PUBLICATIONS	STUDENT PAPERS

MATCH ALL SOURCES (ONLY SELECTED SOURCE PRINTED)

2%

★ Submitted to University of Witwatersrand

Student Paper

Exclude quotes	On	Exclude matches	< 2%
Exclude bibliography	On		

Appendix D: Journal of Genetic Counselling author guidelines

AUTHOR GUIDELINES

SECTIONS

1. AIMS AND SCOPE

The **Journal of Genetic Counseling (JOGC)**, published for the National Society of Genetic Counselors, is a timely, international forum addressing all aspects of the discipline and practice of genetic counseling. The journal focuses on the critical questions and problems that arise at the interface between rapidly advancing technological developments and the concerns of individuals and communities at genetic risk. The publication provides genetic counselors, other clinicians and health educators, laboratory geneticists, bioethicists, legal scholars, social scientists, and other researchers with a premier resource on genetic counseling topics in national, international, and cross-national contexts.

As a crucial resource for genetic counselors and associated professionals, the Journal's primary purpose is to report original research in the following areas:

- **Genetic Counseling Theory, Methods, and Practice**: addresses genetic counseling in clinical or non-clinical settings;
- **Public Health, Public Policy, and Access and Genetics Service Delivery**: addresses public health genomics, health behaviors, legal or policy aspects related to genetic counseling and genetic testing, precision medicine, health disparities, models of genetics services delivery;
- **Education and Genetics Professional Workforce Issues**: addresses educational training, professional development, and workforce topics related to genetic counseling;
- **Ethical, Legal, Psychological, and Social Issues**: addresses ethical, legal, psychological, and/or social issues related to genetic counseling, genetic services, and/or genetic information regarding individuals, communities, and the public
- **Risk Assessment**: addresses algorithms, theoretical models, or empirical data for use in genetic counseling risk assessment.

In addition to research articles, regular features of the Journal of Genetic Counseling include case presentations, editorials, rapid publications, and letters to the editor. Note: The Journal does not publish non-human animal studies.

Return to Guideline Sections

2. SUBMISSION

Once the submission materials have been prepared in accordance with the Author Guidelines, manuscripts should be submitted via the Journal's Editorial Manager

site: https://www.editorialmanager.com/jogc/default.aspx. More details on how to use Editorial Manager are also available at https://www.editorialmanager.com/jogc/default.aspx.

A manuscript is considered for review and possible publication on the condition that it is submitted solely to the journal, and that the manuscript or a substantial portion of it is not under consideration elsewhere. Presentation of the content at meetings prior to submission is acceptable. However, authors should kindly note that submission implies that the content has not been published or submitted for publication elsewhere except as a brief abstract in the proceedings of a scientific meeting or symposium. Note, this journal uses iThenticate's CrossCheck software to detect instances of overlapping and similar text in submitted manuscripts.

The submission system will prompt the author to use an ORCiD ID (a unique author identifier) to help distinguish their work from that of other researchers. Click here to find out more.

For help with submissions, please contact the Editorial Office at JOGC@Wiley.com. When necessary, the Editorial Office staff may refer questions to the Editor-in-Chief.

Return to Guideline Sections

3. MANUSCRIPT CATEGORIES AND GENERAL REQUIREMENTS

MANUSCRIPT CATEGORIES

Original Articles. The Journal of Genetic Counseling seeks papers reporting exciting, timely, original research in the discipline and practice of genetic counseling. The Journal considers papers using a form of systematic study or inquiry to address a question to be original research. Systematic study can be approached using a variety of methods, such as empirical methods, systematic literature review methods, normative or conceptual research methods. Original articles:

- include an abstract and key words;
- are no more than 25 double-spaced pages in length for quantitative studies and no more than 35 double-spaced pages in length for qualitative or non-empirical studies (excluding Supplemental Information);
- have no more than 5 display items (tables + figures), and any additional display items will need to be submitted as Supplemental Information. Large tables should always be published as online only material;
- report relevant information per appropriate methodologic guideline (see Research Reporting Guidelines below).

Case Studies. Case studies are a valuable tool in the presentation of genetic counseling practice. They can serve to demonstrate a counseling model or to stimulate thought about a difficult ethical or counseling situation the author has encountered. In a case study, the paper is focused on the case(s) presented with the intention of alerting the reader to broader issues relevant to practice for the readers' consideration. Note: the Journal of Genetic Counseling does not publish case studies whose sole purpose is to report clinical and molecular information. Case Studies should be concise and focused. They should address observations of patient encounters (usually 1-3) or a single small family that add substantially to the practice and discipline of genetic counseling. Case Studies:

- include an abstract and key words;
- are no more than 15 double spaced pages in length (excluding Supplemental Information);
- have no more than 2 display items (tables + figures), and any additional display items will need to be submitted as Supplemental Information. Large tables should always be published as online only material.

Professional Issues. These article types feature pieces that communicate reflections by the author on the discipline and practice of genetic counseling. Professional Issues:

- include an abstract and key words;
- are no more than 25 double-spaced pages in length (excluding Supplemental Information);
- have no more than 2 display items (tables + figures). and any additional display items will need to be submitted as Supplemental Information. Large tables should always be published as online only material.

Invited Commentary. This type of paper is generally solicited from the Editor but is a submission welcomed from all contributors. It should have a title page and be accompanied by a list of key words for indexing purposes. Commentaries/Editorials often address matters of interest or controversy to the readership.

Brief Reports. These are very brief reports offered in a letter format reporting an observation that adds to the knowledge of the discipline and practice of genetic counseling. They are no more than 9 double spaced manuscript pages in length (excluding Supplemental Information). The manuscripts are not subdivided into sections nor do they include an abstract. Key words are required for indexing purposes.

Correspondence. These are letters to the editor and generally comment on previously published work in the Journal of Genetic Counseling. These are kept brief and to the point; they do not include an abstract, key words, tables, or figures. Like all other material published in the Journal of Genetic Counseling, correspondence is subject to editorial or peer review. The corresponding author of the original manuscript which is the subject of the submitted letter will be offered the opportunity to respond. If a response is provided, every effort will be made to publish these letters together. Only one round of comment is allowed.

Rapid Communications. The Journal of Genetic Counseling features a new section devoted to the rapid communication of full-length, critically reviewed papers reporting new and important advances that are highly likely to have an immediate and critical impact on the discipline and practice of genetic counseling. Our goal is that these manuscripts will be published online approx. 4 weeks after acceptance. In order to have a manuscript considered for Rapid Communication, authors must send a letter of intent along with an abstract to the Editor for consideration prior to submission. The letter of intent should outline the author's rationale for publishing the article as a rapid publication. The Editor or Deputy Editor will respond to the author with a decision. Manuscripts accepted for Rapid publication must adhere to the format of an original research article in the Journal of Genetic Counseling.

Practice Guidelines. These article types address specific areas of genetic counseling and are submitted by the National Society of Genetic Counselors' Practice Guidelines Committee.

Review Articles. The Journal of Genetic Counseling publishes occasional topical reviews. Authors should contact the Editor-in-Chief prior to submission. Note: submissions that describe a systematic process for reviewing the literature to address a research question (e.g., systematic reviews, scoping reviews) are considered original research and are included in the Original Article category.

Book Reviews. Authors may contact the Editor-in-Chief with a proposal to submit a book review. The topic of the reviewed book should be closely aligned with the mission of the Journal. If the proposal is approved for the submission, instruction will be provided by the editor.

Conference Reports. The Journal of Genetic Counseling occasionally publishes an executive summary of an important conference or scientific meeting that involves topics related to the scope of the Journal. The Journal also on occasion publishes the abstracts of an important meeting on a selected basis. Authors should contact the Editor-in-Chief prior to submission.

Corrigenda and Errata. These manuscripts are brief communications to correct errors in previously published work in the Journal of Genetic Counseling. The former is for errors that were responsibility of the author(s), and the latter are for errors that are responsibility of the Journal, including editorial staff and production. These may be written by the corresponding author of the relevant manuscript or they may be composed by an editor.

GENERAL REQUIREMENTS

Format

Manuscripts should be double-spaced with 1 inch margins and 12 point font.

English Language

Manuscripts must be submitted in grammatically correct American English. Manuscripts that do not meet this standard cannot be reviewed. Authors for whom English is a second language may wish to consult an English-speaking colleague or consider having their manuscript professionally edited before submission to improve the English. A list of independent suppliers of editing services can be found at https://wileyeditingservices.com/en/. All services are paid for and arranged by the author, and use of one of these services does not guarantee acceptance or preference for publication.

Revisions

Please submit a marked version (tracked, highlighted, etc) and unmarked version of revised manuscripts.

Ethical Compliance

For all research involving human participants, please include a statement in the Methods section confirming that your study was reviewed by an institutional review board/human investigations committee/ethics committee (include name of committee) and approved or waived as human subjects research.

The Journal of Genetic Counseling does not publish research involving non-human animals.

Informed Consent

The Journal requires that all appropriate steps be taken in obtaining informed consent of all human subjects participating in the research comprising the manuscript submitted for review and possible publication, and statements to this effect must be included under the subheadings, "Human Studies and Informed Consent". For all manuscript categories, identifying information should not be included in the manuscript unless the information is essential for scientific purposes and the study participants or patients (or parents or guardians) give written informed consent for publication. The editors reserve the right to reject manuscripts that do not comply with these requirements. The author will be held responsible for false statements or failure to fulfill these requirements.

Conflict of Interest Statement

The Journal requires that all authors disclose any potential sources of conflict of interest. Any interest or relationship, financial or otherwise, that might be perceived as influencing an author's objectivity is considered a potential source of conflict of interest. These must be disclosed when directly relevant or directly related to the work that the authors describe in their manuscript. Potential sources of conflict of interest include, but are not limited to, patent or stock ownership, membership of a company board of directors, membership of an advisory board or committee for a company, and consultancy for or receipt of speaker's fees from a company. The existence of a conflict of interest does not preclude publication in this journal.

If the authors have no conflict of interest to declare, they must also state this in the manuscript. It is the responsibility of the corresponding author to review this policy with all authors and collectively to list in the manuscript under the subheading "Conflict of Interest" ALL pertinent commercial and other relationships.

The above policies are in accordance with the Uniform Requirements for Manuscripts Submitted to Biomedical Journals produced by the International Committee of Medical Journal Editors (http://www.icmje.org/).

Return to Guideline Sections

4. PREPARING THE SUBMISSION

Parts of the Manuscript

The manuscript should be submitted in separate files: cover letter; main text file; tables; figures; supplementary information files.

Cover Letter

The cover letter should include a statement that the work presented in the manuscript has not been published elsewhere and is not currently under review elsewhere.

If the study includes original data, at least one author must confirm in the cover letter that he or she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.

Main Text File

The main text file should be presented in the following order (as appropriate for article type):

1. Title Page
2. Abstract and keywords
3. Main text
4. Author Contributions
5. Acknowledgements
6. Conflict of Interest
7. Human Studies and Informed Consent
8. Animal Studies
9. References
10. Figure legends

Tables, figures and supplementary information files should be supplied as separate files. Figures must be clearly labeled.

Title Page

The title page should include (in this order) the title of the article, authors' names (no degrees), authors' institutional affiliations where the work was conducted, with a footnote for the author's present address if different from where the work was conducted, and suggested running head. The affiliation should comprise the department, institution (usually university or company), city, and state (or nation) and should be typed as a numbered footnote to the author's name. The suggested running head should be less than 80 characters (including spaces) and should comprise the article title or an abbreviated version thereof. The title page should also include the telephone number and e-mail address of the one author designated to review proofs.

Please denote cases of equal authorship with a footnote: In the case of joint first authorship, a footnote should be added to the author listing, e.g. 'X and Y should be considered joint first author' or 'X and Y should be considered joint senior author.

Authors may benefit from referring to Wiley's best practice tips on Writing for Search Engine Optimization.

Abstract

Please provide an unstructured abstract of no more than 300 words containing the major keywords summarizing the article. The abstract should include a description of the study's objective, methods or methodological approach, sample, measures or main outcome variables, main results, and conclusion.

Keywords

Please provide three to six keywords to be used for indexing the article. Please refer to this list.

Main Body

For Original Research articles, all major sections should carry section headings (such as Introduction, Methods, Results, Discussion, Conclusions, etc.) type centered. Side headings in Methods section should include, as appropriate: Participants, Instrumentation, Procedures, and Data Analysis. The Discussion should begin with a very succinct summary of the major conclusions of the paper and then go on to focus on the interpretation and significance of the findings with concise objective comments that describe their relation to other work in the area. It should not repeat information in the results. Side headings in Discussion should include: Study Limitations, Practice Implications, and Research Recommendations. The journal uses US spelling.

Footnotes should be avoided in the main text. When their use is absolutely necessary, footnotes should be numbered consecutively using Arabic numerals and should be typed at the bottom of the page to which they refer. Place a line above the footnote, so it is set off from the text. Use the appropriate superscript numeral for citation in the text.

Author Contributions

Please include a statement delineating the contributions of each author using the criteria recommended by the International Committee of Medical Journal Editors (ICMJE). The statement should mention each author separately by name. ICMJE criteria are:

- Substantial contributions to the conception or design of the work; or the acquisition, analysis, or interpretation of data for the work; AND
- Drafting the work or revising it critically for important intellectual content; AND
- Final approval of the version to be published; AND
- Agreement to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

If the study includes original data, at least one author must confirm that he or she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis. Please include this statement in the cover letter.

Acknowledgements

Contributions from anyone who does not meet the criteria for authorship should be listed, with permission from the contributor, in an Acknowledgments section. Financial and material support should also be mentioned. Authors should list all funding sources and are responsible for the accuracy of their funder designation. If in doubt, please check the Open Funder Registry for the correct nomenclature: www.crossref.org/services/funder-registry.

If this paper is to be considered for the Journal of Genetic Counseling Best Trainee Paper award, please include a statement indicating that the research presented in the paper was conducted while the first author was in training or to fulfill a degree requirement of the first author. See the Best Trainee Paper Award tab on the journal website for more information about this award. Thanks to anonymous reviewers is not considered appropriate to include in Acknowledgements.

Conflict of Interest Statement

The Conflict of Interest Statement should mention each author separately by name. Recommended wording is as follows:

Author X declares that she has no conflict of interest.

Author Y has received research grants from Drug Company A.

Author Z has received a speaker honorarium from Drug Company B and owns stock in Drug Company C.

If multiple authors declare no conflict, this can be done in one sentence:

Author X, Author Y and Author Z declare that they have no conflict of interest.

Submitting authors should ensure they liaise with all co-authors to confirm agreement with the final statement.

Human Studies and Informed Consent

For manuscripts reporting studies that involve human participants, a statement identifying the ethics committee that approved the study and confirmation that the study conforms to recognized standards is required, for example: Declaration of Helsinki; US Federal Policy for the Protection of Human Subjects; or European Medicines Agency Guidelines for Good Clinical Practice. It should also state clearly in the text that all persons gave their informed consent prior to their inclusion in the study.

The Journal requires that all appropriate steps be taken in obtaining informed consent of any and all human subjects participating in the research comprising the manuscript submitted for review and possible publication, and a statement to this effect must be included in the Human Studies and Informed Consent section of the manuscript. Participant anonymity should be preserved and all identifying information should be excluded in the manuscript.

Photographs need to be cropped sufficiently to prevent human subjects being recognized (an eye bar must not be used because of insufficient de-identification). Images and information from individual participants will only be published where the authors have obtained the individual's free prior informed consent. If any identifying information about participants is included in the article, the following sentence should also be included:

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Authors do not need to provide a copy of the consent form to the publisher; however, in signing the author license to publish, authors are required to confirm that consent has been obtained. Wiley has a standard patient consent form available for use.

Animal Studies

The Journal of Genetic Counseling does not publish non-human animal studies. To affirm that this is the case for your submission, please include the following sentence under this subheading in the manuscript:

'No non-human animal studies were carried out by the authors for this article'

References

The accuracy of references is the responsibility of the authors. Only published papers and those in press may be included in the reference list. The Journal has a strong preference against the inclusion of conference abstracts (published or unpublished) or unpublished data in manuscripts. However, if done, unpublished data and submitted manuscripts must be cited parenthetically within the text. Personal communications should also be cited within the text; permission in writing from the communicator is required.

References should be prepared according to the *Publication Manual of the American Psychological Association* (6th edition). The APA website includes a range of resources for authors learning to write in APA style, including an overview of the manual, free tutorials on APA Style basics, and an APA Style Blog. For more information about APA referencing style, please also refer to the APA FAQ.

EndNote users can download the style here.

According to APA style, in text citations should follow the author-date method whereby the author's last name and the year of publication for the source should appear in the text, for example, (Jones, 1998). Multiple citations should be listed alphabetically by author's last name. The complete reference list should appear alphabetically by name at the end of the paper.

Authors should note that the APA referencing style requires that a Digital Object Identifier (DOI) be provided for all references where available. Also, for journal articles, issue numbers are not included unless each issue in the volume begins with page one.

Reference examples follow:

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Shelton, B. A., John, D., Gibbs, J. T., Huang, L. N., Ruble, D. N., Martin, C. L., … Seltzer, M. M. (1996). The division of household labor. *Annual Review of Sociology, 22*, 299–322. *doi:*10.1146/annurev.soc.22.1.299

Note: for more than seven author names list first six with three dots and then last author name.

Book

Bradley-Johnson, S. (1994). *Psychoeducational assessment of students who are visually impaired or blind: Infancy through high school* (2nd ed.). Austin, TX: Pro-ed.

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Every figure must have a legend. Legends should be concise but comprehensive – the figure and its legend must be understandable without reference to the text. Include definitions of any symbols used and define/explain all abbreviations and units of measurement. Figures should be numbered (with Arabic numerals) and referred to by number in the text.

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Tables should be self-contained and complement, not duplicate, information contained in the text. Tables should be numbered (with Arabic numerals) and referred to by number in the text. They should be supplied as editable files, not pasted as images. The table should have a brief explanatory title, and legends should be concise but comprehensive – the table, legend, and footnotes must be understandable without reference to the text. All abbreviations must be defined in table footnotes. Footnotes should be indicated by superscript lowercase letters and *, **, *** should be reserved for P-values. Statistical measures such as SD or SEM should be identified in the table headings. Each table should be on a separate sheet of paper at the end of the submission.

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Authors are encouraged to send the highest quality figures possible. Line art should be exported at 600 dpi or higher, and halftone images should be exported at 300 dpi or higher.

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Supporting information is information that is not essential to the article, but provides greater depth and background. It is hosted online and appears without editing or typesetting. It may include copies of surveys or interview questions, consent forms, tables, figures, videos, datasets, etc.

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The following points provide general advice on formatting and style.

- **Abbreviations**: In general, terms should not be abbreviated unless they are used repeatedly and the abbreviation is helpful to the reader. Initially, use the word in full, followed by the abbreviation in parentheses. Thereafter use the abbreviation only.
- **Units of measurement**: Measurements should be given in SI or SI-derived units. Visit the Bureau International des Poids et Mesures (BIPM) website for more information about SI units.
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- **Trade Names**: Chemical substances should be referred to by the generic name only. Trade names should not be used. Drugs should be referred to by their generic names. If proprietary drugs have been used in the study, refer to these by their generic name, mentioning the proprietary name and the name and location of the manufacturer in parentheses.
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- **Pedigrees**: Pedigrees should follow the recommendations for standardized nomenclature accepted by the National Society of Genetic Counselors. Authors should consult the following references for these recommendations:
    - Bennett, R. L. , Steinhaus, K. A., Uhrich, S. B., O' Sullivan, C. K., Resta, R. G. , Lochner-Doyle, D., Markel, D. S., Vincent, V., & Hamanishi, J. (1995). Recommendations for Standardized Human Pedigree Nomenclature. Journal of Genetic Counseling, 4, 267-279.

- Bennett, R. L., Steinhaus French, K., Resta, R. G., & Lochner Doyle, D. (2008). Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling, 17, 424-433.

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5. EDITORIAL POLICIES AND ETHICAL CONSIDERATIONS

Peer Review and Acceptance

The acceptance criteria for all papers are the quality and originality of the research and its significance to journal readership and the practice and discipline of genetic counseling. Papers will only be sent to review if the Editors determine that the paper meets the appropriate quality and relevance requirements.

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Although it would be rare for a paper submitted to the Journal of Genetic Counseling to report novel nucleotide sequence data, should that be the case, the novel nucleotide sequence data including genetic mutations must be submitted to a public database prior to publication and a sentence naming the database should be included in the manuscript.

Human Studies and Subjects

For manuscripts reporting studies that involve human participants, a statement identifying the institutional review board/human investigations committee/ethics committee that approved the study and confirmation that the study conforms to recognized standards is required. It should also state clearly in the text that all persons gave their informed consent prior to their inclusion in the study.

Patient anonymity should be preserved. Information from individual patients will only be published where the authors have obtained the individual's free prior informed consent. Authors do not need to provide a copy of the consent form to the publisher; however, in signing the author license to publish, authors are required to confirm that consent has been obtained. Wiley has a standard patient consent form available for use if needed.

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- Qualitative research: COREQ
- Quality improvement studies: SQUIRE

- Study protocols: SPIRIT

Studies reporting on genetic counseling as an intervention should refer to and follow guidelines from:

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7. PUBLICATION PROCESS AFTER ACCEPTANCE

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Once the paper is typeset, the author will receive an email notification with full instructions on how to provide proof corrections.

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Wiley Author Resources

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