Genetic Counselling In Kerala Context: An Exploratory Study

Dissertation submitted to Kerala University In partial fulfilment of the requirements for the award of the Degree of

MSc Counselling Psychology

By

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CERTIFICATE

This is to certify that the Dissertation entitled "Genetic Counselling In Kerala Context : An Exploratory Study" is an authentic work carried out by Dilsha K, Reg. No. 60422115010 under the guidance of DR Pramod SK during the fourth semester of M.Sc. Counselling Psychology programme in the academic year 2022- 2024.

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DECLARATION

I, Dilsha K, do hereby declare that the dissertation titled ‘Genetic Counselling In Kerala Context : An Exploratory Study’ submitted to the Department of Counselling Psychology, Loyola College of Social Sciences, Sreekariyam, under the supervision of Dr Pramod SK , Assistant professor of the Department of Counselling Psychology, for the award of the degree of Master’s in Science of Counselling Psychology, is a bonafide work carried out by me and no part thereof has been submitted for the award of any other degree in any University

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DILSHA K

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ABSTRACT

Genetic counselling is a crucial service that provides essential guidance and support to individuals and families dealing with genetic conditions. This field educates about genetic disorders, including their causes, implications, and management options, while offering emotional and professional support to help families make informed decisions and address mental and physical health challenges proactively. Despite its importance, genetic counselling faces significant challenges, including widespread lack of awareness and underutilization of services. This study aims to explore the significance of genetic counselling, identify barriers to its practice, and propose strategies to overcome these barriers, particularly in the context of Kerala, where research on this topic is limited. The study employs a qualitative approach with an interpretivist research design, using thematic analysis to examine data collected from eight mental and health professionals who work with clients with genetic conditions. Data were gathered through purposively selected participants and semi-structured interviews. The findings reveal a noticeable scarcity of genetic counselling services in Kerala, exacerbated by inconsistent protocols and a shortage of qualified counsellors. This service gap negatively impacts the quality of care and reinforces social stigma surrounding genetic disorders. To address these issues, the study recommends several measures: establishing dedicated genetic counselling centers to improve service delivery; integrating genetic counselling into medical and academic curricula to better prepare future healthcare professionals; launching public education campaigns to raise awareness and combat stigma through various media platforms; making genetic counselling and testing more affordable and accessible via government policies and financial support; and developing standardized protocols to ensure consistent, high quality care while safeguarding patient privacy. Supporting research in genetics and the professional development of genetic counsellors is also essential to building a robust workforce. By addressing barriers such as lack of awareness, inadequate qualifications, and social stigma, and integrating counselling with other support systems, these strategies aim to improve the quality of life for individuals and families affected by genetic conditions, fostering a more inclusive and supportive environment.

CHAPTER I

INTRODUCTION

According to Indian Genetic Disease Database, in India, the genetic disease database manages data for a colossal 1.18 billion people, documenting 5,760 patients and carriers. It covers an astounding 6,647 diseases and has recorded 780 unique mutations, showcasing an exceptional range of genetic variation. Data concluded that Genetic diseases can be controlled best through an integrative approach of community education, population screening, genetic counselling, carrier identification and neonatal screening (Nucleic Acid Research, 2011). Then what is Genetic Counselling? How it is performed? What are the benefits? To address the questions there are no extensive studies done in Kerala. So this research paper will try to explore these questions in light of increasing genetic mutations and disorders so as to assist the individuals and families to ensure their mental wellbeing.

Genetic counselling is defined as "the process of helping people understand and adapt to the medical, psychological and familial implications of genetic conditions to disease" (ScienceDirect, 2009). Many health conditions are known to run in families, referred to by doctors as "genetic" or "hereditary" conditions. Individuals with a family history of serious health problems may seek to understand their own risk for these diseases. Similarly, prospective parents might want to know if they or their partner could pass a hereditary condition to their child. To address these concerns, genetic counselling can be a valuable resource. Genetic counsellors provide more than just an assessment of the likelihood of hereditary conditions being passed from one generation to the next. They also offer support in managing the emotional impact that genetic conditions can have on families (WebMD, 2021).

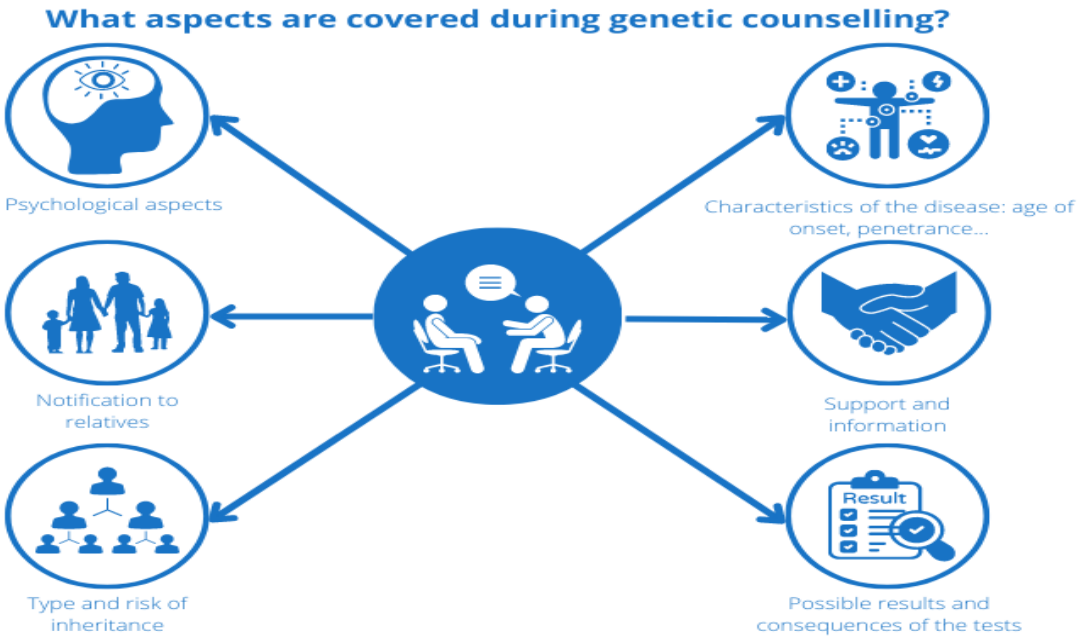
Minesh Khathri explained that people might seek genetic counselling for various reasons. For example, those planning to have a baby might want to learn about screening for genetic conditions. Women who have had difficulties with conception or recurrent miscarriages may also seek guidance through genetic counselling. Mothers who have had a child with a genetic birth defect and are concerned about the possibility of it happening again could benefit from this counselling as well. Additionally, individuals interested in understanding their family's history of genetic conditions or seeking information about genetic conditions prevalent in specific ethnic groups—such as sickle-cell anemia among African-Americans—might find genetic counselling useful.

Those curious about the process of genetic testing can also seek this expertise. Genetic counsellors are trained not only in genetics but also in psychological counselling, providing valuable perspective and support (Khathri, 2021).

The profession of genetic counselling began nearly 50 years ago in the United States, and has grown internationally in the past 30 years (National Library of Medicine, 2019). Sheldon Clark Reed was an American biologist and geneticist who coined the term genetic counselling and advocated for the wider use of genetic counselling as a means to educate the public. The history of genetic counselling as a clinical practice has its origins in eugenics, a now-discredited movement aimed at improving human populations through controlled breeding. Although medical geneticists distanced themselves from eugenic beliefs after World War II, the separation was sometimes more superficial than substantial. Despite this, the genetic counselling profession itself does not have direct links to eugenics. Since its inception, genetic counselling has been closely tied to medical genetics and geneticists. Key milestones in the development of genetic counselling as a distinct profession include the establishment of master's-level training programs at Sarah Lawrence College in 1969, the creation of the National Society of Genetic Counsellors, the development of a certification process managed by genetic counsellors independently of medical geneticists, the launch of a professional journal, and collaborations with a broad range of clinicians beyond just medical geneticists, such as obstetricians, oncologists, surgeons, cardiologists, and neurologists (OXFORD ACADEMIC, 2019).

So in essence, Genetic counselling is a structured communication process designed to empower patients and their families to make informed and autonomous decisions where there is a presence of genetic condition. It aims to provide a comprehensive understanding of genetic information and its implications, facilitating the effective utilization of this information in clinical and personal contexts. This process involves the interpretation of genetic testing results, assessment of genetic risk, and provision of psychosocial support to help individuals navigate the complexities of genetic data and its impact on health and family planning.

Figure1: different aspects covered during Genetic Counselling



Resource:Genosalut,https://www.genosalut.com/en/genetic-testing-and counselling/geneticcounselling/

The gene is considered the basic unit of inheritance. Genes are passed from parents to offspring and contain the information needed to specify physical and biological traits. Most genes code for specific proteins, or segments of proteins, which have differing functions within the body. Humans have approximately 20,000 protein-coding genes (National Human Genome Research Institute, 2024).

Genes, composed of DNA, are fundamental to determining an organism's characteristics, including appearance, survival traits, and behavior. All living beings possess genes, which significantly influence health throughout life. DNA, or deoxyribonucleic acid, contains the instructions essential for the development, growth, and reproduction of life. Chromosomes, located in each cell's nucleus, house genes, with each gene containing specific DNA sequences that encode instructions for making proteins. These proteins affect physical traits like hair color, height, and eye color, and can also influence susceptibility to genetic disorders. DNA is inherited from parents, with humans having 46 chromosomes in total 23 from each parent. A gene is made up of sequences of four nucleotide bases: adenine (A), cytosine (C), guanine (G), and thymine (T). The order of these bases determines the genetic instructions encoded in DNA strands, such as those influencing eye color.

Human chromosomes vary in size, ranging from 50 to 300 million base pairs, with the entire human genome comprising about 3 billion bases and around 20,000 genes. Genes impact numerous internal and external factors, including disease susceptibility. Mutations in genes, which can be inherited or caused by environmental factors like toxins, can lead to genetic disorders such as sickle-cell anemia and Huntington's disease. The Human Genome Project, initiated in 1990, aimed to map the entire human genome to enhance understanding of genetic factors in diseases and develop new strategies for diagnosis, treatment, and prevention (MedicalNewsToday, 2024).

As cited in US Centers for Disease Control and Prevention (2024) following are the key terms and concepts that reveals the significance of genetics:

1. *DNA*

DNA, or deoxyribonucleic acid, contains the instructions necessary for the functioning of the body. It is composed of two strands that twist around each other, forming a structure known as a double helix. Each DNA strand includes nitrogen bases, which constitute the DNA code. There are four types of bases: thymine (T), adenine (A), cytosine (C), and guanine (G). Bases on one strand of DNA pair with complementary bases on the other strand, creating the "rungs" of the DNA ladder. The sequence of these bases varies across different segments of DNA. The DNA sequence, or the order of the bases (Ts, Cs, Gs, and as), is crucial for reading genetic information. Although the fundamental components of DNA are consistent across almost all living organisms, the sequence of these components differs *(US Centers for Disease Control and Prevention, 2024).*

2. *Inheritance*

Genetic inheritance is the process through which DNA is passed from parents to offspring. *(US Centers for Disease Control and Prevention, 2024).*

3. *Genome*

The genome refers to the complete set of DNA within an organism's body. *(US Centers for Disease Control and Prevention, 2024).*

4. *Chromosomes*

DNA is organized into structures known as chromosomes. Each chromosome consists of a single, long DNA molecule with numerous genes. Chromosomes are inherited from one's parents and

come in pairs. Humans have 46 chromosomes, arranged in 23 pairs. Each child inherits one chromosome from each pair from the mother and one from the father. There are 22 pairs of autosomes, and the 23rd pair consists of sex chromosomes, which determine an individual's sex. Females have two X chromosomes, while males have one X and one Y chromosome. Daughters receive an X chromosome from each parent, while sons receive an X from their mother and a Y from their father. *(US Centers for Disease Control and Prevention, 2024).*

5. *Genes and Proteins*

Each chromosome contains multiple genes. Genes are segments of DNA that provide instructions for producing proteins. Proteins are essential for the structure and function of the body. Individuals possess two copies of every gene, inherited one from each parent. The combination of genes inherited from parents can influence various traits, such as physical appearance and susceptibility to certain diseases. *(US Centers for Disease Control and Prevention, 2024).*

6. *Alleles*

Alleles are different forms of the same gene, differing slightly in their DNA sequence. These variations contribute to an individual's unique characteristics. Each person inherits two alleles for each gene one from each parent. If both alleles are identical, the individual is homozygous for that gene; if they differ, the individual is heterozygous. While most allele differences do not significantly affect protein function, some variations can influence traits like blood type or be associated with health conditions. Certain alleles may impact the body's ability to produce specific proteins, potentially leading to genetic disorders. *(US Centers for Disease Control and Prevention, 2024).*

7. *Cells*

Cells are the fundamental units of life, with trillions present in the human body. Various cell types constitute different tissues and organs, each performing specialized functions. For instance, skin cells, blood cells, heart cells, brain cells, and kidney cells are among the diverse types necessary for bodily functions. A cell's basic structure includes a jelly-like substance called cytoplasm, enclosed by a membrane. The cytoplasm contains specialized structures,

including the nucleus, which houses DNA organized into chromosomes. *(US Centers for Disease Control and Prevention, 2024).*

8. *Gene Expression*

Gene expression is the process by which proteins are produced according to genetic instructions. While every cell in the body contains the same DNA, different genes are activated or deactivated depending on the cell type, leading to variations in function and appearance. For example, muscle cells and nerve cells have distinct gene expression profiles. Gene expression can also be influenced by age, behavior, and environmental factors, such as smoking or physical activity *(US Centers for Disease Control and Prevention, 2024).*

9. *DNA Methylation*

DNA methylation involves adding a chemical group (methyl group) to DNA, a process that can also be reversed through demethylation. Generally, methylation suppresses gene activity, while demethylation activates genes. This mechanism helps regulate gene expression without altering the DNA sequence *(US Centers for Disease Control and Prevention, 2024).*

10. *Genetic Change (Mutation, Gene Variant, Genetic Variant)*

    Genetic changes, also known as mutations or variants, refer to alterations in the DNA sequence. Not all genetic changes cause problems, but some can affect protein function, potentially leading to diseases. Such changes can be inherited if they occur in germ cells (sperm or eggs), while mutations in somatic cells (body cells) are not passed on to offspring. Genetic changes can occur during DNA replication or due to environmental factors like high radiation. Despite the body's repair mechanisms, some changes may still occur. *(US Centers for Disease Control and Prevention, 2024).*

11. *Copy Number Variation (CNV)*

    Copy number variation refers to differences in the number of DNA repeats across individuals. While CNVs contribute to genetic diversity, some variations are associated with diseases. *(US Centers for Disease Control and Prevention, 2024).*

12. Environmental Factors

Environmental factors encompass exposures related to one's living conditions, such as air pollution, lifestyle behaviors, and dietary choices. *(US Centers for Disease Control and Prevention, 2024).*

13. Epigenetics

Epigenetics involves changes in gene activity influenced by environmental factors and behaviors. These changes can activate or deactivate genes without altering the DNA sequence. Epigenetic modifications, such as DNA methylation, can impact protein production and are subject to change with age, environmental exposures, and lifestyle factors (Centers for Disease Control and Prevention. (n.d.). *Genomics and health, 2024).*

Classification of Genetic Disorders

As outlined in "Parks' Textbook of Preventive and Social Medicine (23rd Edition)" classification of genetic disorders encompasses chromosomal disorders, single-gene disorders, mitochondrial disorders, multifactorial disorders, and complex genetic disorders. Each category includes specific types of disorders with distinct inheritance patterns and manifestations, illustrating the broad spectrum of genetic conditions and their underlying genetic mechanisms.

1. Chromosomal Disorders

Numerical Chromosome Abnormalities: These disorders arise from an abnormal number of chromosomes. Examples include:

a. Down syndrome (Trisomy 21): Characterized by an extra copy of chromosome 21.

b. Turner Syndrome: Resulting from a missing X chromosome in females.

c. Klinefelter Syndrome: Occurs in males with an extra X chromosome (XXY).

Structural Chromosome Abnormalities: These involve changes in the structure of chromosomes, such as:

a. Deletion: Loss of a chromosome segment (e.g., Cri du Chat Syndrome).

b. Duplication: Extra copies of a chromosome segment.

c. Translocation: A segment of one chromosome is transferred to another chromosome (e.g., Philadelphia Chromosome in Chronic Myeloid Leukemia).

d. Inversion: A chromosome segment is reversed within the chromosome.

2. Single-Gene Disorders

Autosomal Dominant Disorders: A single copy of the mutant gene is sufficient to cause the disorder. Examples include:

a.Huntington's disease: Neurodegenerative disorder with onset in adulthood.

b.Marfan syndrome: Connective tissue disorder affecting the heart, eyes, and skeleton.

Autosomal Recessive Disorders: Two copies of the mutant gene are required for the disorder to manifest. Examples include:

a.Cystic Fibrosis: Affects the respiratory and digestive systems.

b.Sickle Cell Disease: Affects red blood cell shape and function.

X-Linked Disorders: The gene causing the disorder is located on the X chromosome. Examples include:

a.Hemophilia: A bleeding disorder due to defective blood clotting factors.

b.Duchenne Muscular Dystrophy: A severe form of muscle degeneration.

3. Mitochondrial Disorders

Involve mutations in mitochondrial DNA: These disorders affect energy production and are inherited maternally. Examples include:

a.Leigh Syndrome: A severe neurological disorder.

b.Mitochondrial Myopathy: Muscle weakness and pain.

4. Multifactorial Disorders

Multifactorial disorders result from the interaction of multiple genes and environmental factors: Examples include:

a.Type 2 Diabetes: Influenced by genetic susceptibility and lifestyle factors.

b.Heart Disease: Associated with both genetic predisposition and lifestyle factors.

5. Complex Genetic Disorders

Involve interactions between genetic and environmental factors, often with a combination of genetic and non-genetic components: Examples include:

a.Cancer: Various types of cancer can be influenced by genetic mutations and environmental exposures.

b.Neurodevelopmental Disorders: Such as autism spectrum disorders, which involve a complex interplay of genetic and environmental factors (Park, 2015)

Early Diagnosis and Treatment

Parks in the same textbook "Parks' Textbook of Preventive and Social Medicine (23rd Edition)" gave the diagnosis methods and available treatment for Genetic disorders as follows:

Detection of genetic carriers: It is now possible to identify the healthy carriers of a number of genetic disorders, especially the inborn errors of metabolism. The female carriers of Duchenne type of muscular dystrophy, an X-linked disorder, can now be detected by elevated levels of serum creatine kinase in 80 per cent of carriers. In some conditions, carriers can be recognized with a high degree of certainty (e.g., acatalasia); in some only a proportion of carriers can be detected (e.g., haemophilia, PKU, galactosaemia); in other conditions, no method has yet been found which will distinguish carriers (e.g., alkaptonuria) (Park, 2015)

Prenatal diagnosis: Amniocentesis in early pregnancy (about 14-16 weeks) has now made it possible for prenatal diagnosis of conditions associated with chromosomal anomalies (e.g., Down's Syndrome); many inborn errors of metabolism (e.g., Tay-Sach's disease, galactosaemia, Maple syrup urine disease, Alpha-thalassaemia and neural tube defects) .Amniocentesis Examination of

a sample of amniotic fluid makes possible the prenatal diagnosis of chromosomal anomalies and certain metabolic defects. The procedure can be used as early as 14th week of pregnancy when abortion of the affected fetus is still feasible. The diagnosis of chromosomal anomalies is made by culture and Kryotyping of fetal cells from the amniotic fluid, and of metabolic defects by biochemical analysis of the fluid. (Park, 2015)

Screening of newborn infants: today there are long list of screening tests for the early diagnosis of genetic abnormalities - sex chromosome abnormalities, congenital dislocation of hip, PKU, congenital hypothyroidism, sickle cell disease, cystic fibrosis, Duchenne muscular dystrophy, congenital adrenal hyperplasia, G6PD deficiency etc. (Park, 2015)

Neonates should be routinely examined for congenital abnormalities, particularly dislocation of the hip, which can be simply corrected at this stage. Biochemical screening of newborn infants was first used for PKU in 1966. Heel-prick blood samples are usually collected at 5-10 days after birth. Several drops of blood are collected on filter paper (the Guthrie card), which is sent to screening laboratory. Screening of newborns for congenital hypothyroidism is carried out in most developed countries. Sickle-cell disease can be detected cheaply and reliably by hemoglobin electrophoresis using Guthrie blood spots. Neonatal screening for cystic fibrosis is based on the measurement of immunoreactive trypsin in Guthrie blood spots. (Park, 2015)

Recognizing pre-clinical cases: today there is pretty long list of screening tests for the early diagnosis of hereditary diseases. For example, heterozygotes for phenylketonuria can be detected by a phenylalanine tolerance test. A simple urine examination for sugar after morning breakfast is good enough to detect diabetics. Examination of sibs and close relatives of diabetics by a glucose tolerance test will often reveal preclinical cases of acholuric jaundice. A raised serum uric acid should arouse suspicion of gout. Sickle cell trait can be uncovered by subjecting the red cells to reduced oxygen tension. Thalassemia minor can be detected by studying the blood picture. (Park, 2015)

Rehabilitation: with many genetic or partially genetic conditions causing physical or mental disability, much can be done for the patient and for his family in helping him to lead a better and more useful life. (Park, 2015)

Genetic counselling: Genetic counselling can have the greatest impact when individuals or couples at genetic risk are identified prospectively, i.e., before they have developed symptoms themselves or produced their first affected child. Prospective counselling is technically possible only when carriers can be accurately identified. To some extent, the established genetic population-screening services may serve as models for the development of future genetic screening programmers. The most Immediate and practical service that genetics can render in medicine and surgery is genetic counselling. Genetic counselling may be prospective or retrospective. (Park, 2015)

(1) Prospective genetic counselling: This allows for the true prevention of disease. This approach requires identifying heterozygous individuals for any particular defect by screening procedures and explaining to them the risk of their having affected children if they marry another heterozygote for the same gene. In other words, if heterozygous marriage can be prevented or reduced, the prospects of giving birth to affected children will diminish. The application in this field, for example, are sickle cell anemia and thalassemia. It is possible that this kind of prevention may find wider application to cover a number of other recessive defects. (Park, 2015)

(2) Retrospective genetic counselling: Most genetic counselling is at present retrospective, i.e., the hereditary disorder has already occurred within the family. A survey carried out by the WHO showed that genetic advice was chiefly sought in connection with congenital abnormalities, mental retardation, psychiatric illness and inborn errors of metabolism and only a few sought premarital advice. The WHO recommends the establishment of genetic counselling centers in sufficient numbers in regions where infectious disease and nutritional disorders have been brought under control and in areas where genetic disorders have always constituted a serious public health problem (e.g., sickle cell anemia and thalassemia) (Park, 2015).

The methods which could be suggested under retrospective genetic counselling are: (I) contraception (ii) pregnancy termination and (iii) sterilization depending upon the attitudes and cultural environment of the couples involved (Bhanot, 2015).

Anna and Christine proposed that Genetic counselling plays a crucial role in supporting individuals undergoing genetic evaluations and those at risk for genetic disorders. It is a communication process designed to assist patients in making informed decisions by considering their personal preferences, values, family dynamics, educational background, and cultural norms. This approach

helps shape the patient's understanding of and response to their genetic diagnosis or risk, ensuring that decisions align with their personal circumstances (PubMed Central, 2018).

As cited in textbook named 'Understanding Genetics: A District of Columbia Guide for Patients and Health Professionals' (2010), through genetic counselling, patients receive essential education about the role of genetic and non-genetic risk factors for diseases. They learn about fundamental genetic concepts such as inheritance patterns, penetrance, and variable expressivity. Counsellors also provide information on potential treatment options, preventive measures, and risk factor modifications tailored to the individual's genetic profile. This personalized education is crucial for enabling patients to manage their genetic risks effectively (National Library of Medicine, 2008).

The impact of genetic risk factor information on behavior can be complex and varies based on the specific condition, the genetic factor's contribution to disease risk or severity, and individual beliefs about genetics. For example, while a positive correlation between a family history of cardiovascular disease and perceived risk has been observed, the effect on health related behavior change is not always straightforward. Studies have shown that knowing one's family history of cardiovascular disease does not consistently lead to behavioral changes or altered risk perception (ScienceDirect, 2017).

Genetic counselling also ensures that patients provide informed consent by discussing the benefits, risks, and limitations of genetic testing. This process includes exploring the option of not undergoing testing. Knowing one's genetic predisposition can enhance diagnostic accuracy, management strategies, and preventive measures. Moreover, there can be psychological and cognitive benefits associated with understanding one's genetic risks (American College of Obstetricians and Gynecologists, 2017).

Family dynamics and privacy concerns are significant aspects of genetic counselling. Family members may face challenges such as loss of privacy when sharing medical histories and potential stigma from being labeled with a genetic predisposition. Changes in family relationships may also occur, such as guilt or survivor's guilt among relatives. Concerns about the misuse of genetic information by employers or insurers can also deter individuals from pursuing genetic services. Legal protections like the Genetic Information Nondiscrimination Act (GINA) of 2008 address some of these concerns by preventing discrimination based on genetic information (Genetic Discrimination, National Human Genome Research Institute).

As genetic testing evolves, traditional pretest and posttest counselling methods may need adaptation, particularly when germ line genetic testing informs therapeutic options, such as targeted cancer therapies. In these cases, patient preferences regarding the receipt of genetic findings should be honored, and the relevance and potential benefits of this information must be discussed. The limitations and potential harms of receiving genetic information also need to be addressed (ScienceDirect, 2017).

Population based genetic screening programs, such as newborn screening or ethnicity based screening, offer examples of how to manage patient education and informed consent on a larger scale. Studies of population based screening for BRCA1 and BRCA2 gene variants have shown high levels of satisfaction and minimal distress among participants, although these outcomes may differ in clinical settings compared to self referred individuals (Centers for Disease Control and Prevention, 2023).

In summary, genetic counselling is essential for providing tailored support to individuals navigating genetic risks. It ensures that patients are informed, their decisions are aligned with their personal circumstances, and they are prepared for the psychological and practical implications of genetic information (ScienceDirect, 2017).

Counselling, as defined by the National Society of Genetic Counsellors (NSGC), is a process aimed at helping individuals understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. This involves interpreting family and medical histories to evaluate the likelihood of a disease occurring or recurring within a family. It also includes providing education about genetic concepts such as inheritance patterns, testing options, management strategies, prevention methods, available resources, and ongoing research. Additionally, counselling promotes informed decision-making and helps individuals adjust to their genetic risks or conditions (PubMed Central, 2022).

Genetic counselling can be relevant at various stages of life. Before conception, it may be needed when individuals are carriers of specific genetic traits or chromosomal abnormalities. During pregnancy, it is offered if there are concerns related to maternal age or abnormalities detected in prenatal tests. After birth, it addresses congenital anomalies that may be identified. In childhood, it is provided for developmental delays or other issues, and in adulthood, it is relevant for

conditions with later onset, such as hereditary cancer syndromes (Centers for Disease Control and Prevention, 2024).

A significant component of genetic counselling involves creating a pedigree, or family tree, to illustrate the inheritance of traits within a family. Standard symbols are used to depict this inheritance. The person who presents with the genetic condition and brings the family to seek counselling is referred to as the index patient or proband (proposita if female and propositus if male). The individual who seeks the counselling services is known as the consultant, and often, the index patient and consultant are different individuals. This distinction underscores the importance of considering various perspectives in the counselling process (National Library of Medicine, 2008).

Individuals often seek genetic counselling primarily to assess the risk of heritable diseases in their offspring. Advances in genetic testing now allow for the direct detection of many disease-causing mutations in carriers and affected individuals. However, for some genetic disorders, prenatal diagnosis may not be feasible, or the specific gene responsible for the disease remains unknown. In such cases, genetic counsellors must be knowledgeable about available options and provide information on choices like adoption, artificial insemination, or the use of donated eggs. When the genotype of an affected individual is identified through DNA analysis and the inheritance pattern of the disease is understood, genetic counsellors can offer more precise predictions regarding the risk of recurrence and provide prenatal diagnosis (National Library of Medicine, 2008).

Estimating recurrence risks involves understanding the inheritance pattern of a genetic condition and thoroughly analyzing family history. For disorders following Mendelian inheritance patterns, recurrence risks for specific family members can be calculated using Mendelian principles. For instance, a child of a heterozygous individual with an autosomal dominant (AD) disorder with complete penetrance has a 50% chance of being affected, while the recurrence risk for siblings of an individual with an autosomal recessive (AR) disorder is 25%. Conversely, the inheritance mechanisms for most chromosomal and multifactorial disorders are not fully understood, so recurrence risk estimates rely on empirical data. For example, the risk of recurrence for a sibling of a child with isolated cleft lip/palate is about 3–5%, and if a child has Down syndrome due to trisomy 21, the empiric recurrence risk is approximately 1% (National Library of Medicine, 2013).

For consanguineous couples, such as first cousins, the risk of having an abnormal offspring is estimated to be around 4.5–5%, compared to up to 3% for unrelated parents. In some communities, premarital genetic testing has become common practice. This is particularly true among West Africans where sickle cell disease is prevalent, Asians from regions with high thalassemia rates, and Eastern European Ashkenazi Jews with a higher carrier rate for Tay–Sachs disease. The range of conditions for which preconceptual carrier screening is offered continues to expand, influenced partly by the parents' ancestry (National Library of Medicine, 2013).

Genetic Counselling and Future

Genetic counselling plays a critical role in understanding genetic risks, making informed reproductive decisions, and ultimately preventing genetic disorders in future generations. By providing individuals and families with the necessary knowledge and support, genetic counselling enables proactive steps to minimize the occurrence of hereditary conditions. (National Society of Genetic Counsellors, 2022).

Understanding Genetic Risks

Genetic counselling empowers individuals to understand their genetic risks based on family history and inheritance patterns. Counsellors analyze medical histories and identify potential genetic disorders, allowing families to make informed decisions about their reproductive options. This understanding is crucial for couples planning pregnancies, especially if they have a family history of genetic conditions. (National Society of Genetic Counsellors, 2022).

Informed Family Planning Decisions

The role of genetic counselling in family planning is significant. It helps couples assess the risk of passing on genetic conditions to their children, enabling them to take preventive measures if necessary. Counselling can inform decisions about genetic testing, prenatal screening, and the option of assisted reproductive technologies, reducing anxiety and uncertainty in the family planning process. (National Society of Genetic Counsellors, 2022).

Proactive Prevention

By identifying and discussing the implications of genetic risks, genetic counsellors assist families in developing strategies to mitigate the likelihood of genetic disorders in future generations. This proactive approach can include discussions about lifestyle changes or medical interventions that can influence health outcomes for offspring. (National Society of Genetic Counsellors, 2022).

Emotional and Psychological Support

Genetic counselling is not just about information; it encompasses emotional support during the often challenging decision-making process surrounding genetic risks and family planning. Counsellors aid families in processing the emotional impact of genetic information and navigating concerns related to hereditary conditions. This emotional support is essential for helping families cope with complex genetic issues and aids in facilitating informed choices. (National Society of Genetic Counsellors, 2022).

Integration of Genetic Testing

As advancements in genetic testing continue to evolve, genetic counsellors help families understand how these developments impact their health and reproductive choices. With improved testing techniques, potential risks can be assessed more accurately, enabling better management and preventive strategies. Genetic counselling thus aligns education about genetic risks with the availability of testing options to provide comprehensive guidance.

Overall, genetic counselling is vital for families looking to understand and navigate the complexities of genetic information, ensuring that future generations can experience healthier outcomes through informed decisions and proactive management of genetic risks (National Society of Genetic Counsellors, 2022).

Despite its growing importance, there remains a notable gap in qualitative research exploring the lived experiences and perceptions of both counsellors and clients. This research paper aims to address this gap by investigating the significance of genetic counselling from a qualitative perspective. By examining the responses of participants, study seeks to uncover insights in to the need and effectiveness of genetic counselling, the challenges faced in that area and strategies to

promote the filed. Employing a qualitative research approach, this study will utilize in-depth interviews and thematic analysis to explore these dimensions. The findings are expected to provide valuable contributions to the field, offering practical recommendations for improving genetic counselling practices and informing policy development. This introduction will be followed by a detailed review of the existing literature, methodology, and analysis, ultimately culminating in a discussion of the study's implications for practice and future research.

Need and Significance of the Study

India's vast population, high birth rate, and prevalence of consanguineous marriages contribute to a significant prevalence of genetic disorders nationwide (Hindustan Times, 2023). Cancer, a critical and life-threatening condition, affects numerous individuals, with Kerala alone detecting approximately 35,000 new cases annually and treating nearly 100,000 patients. Despite the presence of treatment facilities in government hospitals and specialized centers, such as Medical Colleges, the Regional Cancer Centre, Malabar Cancer Centre, and Cochin Cancer Centre, challenges including delayed detection, high treatment costs, limited centers, and low awareness contribute to high mortality rates (Economic Review, State Planning Board, Kerala, 2016).Genetic disorders not only result in personal crises but also strain familial relationships, highlighting the need for understanding emotional reactions and communication both individually and as couples. These conditions often lead to psychological distress and depression, underscoring the importance of genetic counseling to manage risks, reduce stress, and prevent future issues while providing necessary support (Batnariu et al., 2012).

This study aims to explore the significance of genetic counseling, identify barriers to its practice, and examine the sensitivity required in delivering genetic information. It will also seek to develop strategies to promote genetic counseling and support individuals and families in navigating their awareness, cultural beliefs, and practices that influence their perceptions of genetic disorders and healthcare decisions. By facilitating discussions on family dynamics and promoting understanding among family members with genetic conditions or susceptible, the study can address the stigma surrounding genetic disorders and advocate for informed decision-making. Furthermore, with limited research on the Kerala population, where cultural norms may differ, the study has the potential to drive policy changes and healthcare improvements, thereby promoting better access to genetic services and support.

Statement of the Problem

Genetic counseling is a vital aspect of healthcare, particularly in regions such as Kerala, where the prevalence of consanguineous marriages may contribute to a higher incidence of genetic disorders. However, there is a notable lack of comprehensive infrastructure, awareness, and accessibility to genetic counseling services throughout the state. Genetic conditions can cause significant disruptions in the lives of individuals and their families, often leading to psychological disturbances such as anxiety and depression. Consequently, the issue is framed as "Genetic Counseling in the Kerala Context: An Exploratory Study," highlighting the need to investigate and address these challenges within the region.

CHAPTER II
REVIEW 0F LITERATURE

The objective of this chapter is to provide a comprehensive overview of existing studies, focusing on their methodologies, findings, and contributions to the field. By critically examining these works, the chapter will highlight the strengths and limitations of current research, thus identifying gaps that could inform and enhance the current study. An integrative review approach is employed organizing the relevant studies chronologically to illustrate the evolution of the field over time (Whittemore & Knafl (2005).The integrative review: Updated methodology. *Journal of Advanced Nursing*).This also include theoretical elements that highlights what is genetic counselling and its utility. Overall, the reviews facilitated a deeper understanding of the progress made in genetic counselling, underscored methodological advancements, and elucidated areas requiring further investigation, thereby building a robust foundation for subsequent research by synthesizing past findings and addressing existing knowledge gaps.

1. The Hindu Bureau (2024 April 07) Consanguineous marriages increase the risk of Hereditary Eye Diseases, say doctors The Hindu published study conducted by LV Prasad Eye Institute revealed a strong correlation between such marriages, particularly uncle-niece unions, and the incidence of eye disorders. Early detection and genetic counselling are crucial for managing HEDS and preventing their transmission to future generations. However, cultural beliefs and limited awareness often hinder the adoption of preventive measures. There is a pressing need for increased awareness about the risks associated with consanguinity and the importance of genetic counselling in affected populations.

2. Arumugam, S., & et al. (2023). Conducted a study titled ‘Knowledge, Attitude, and Practice about the Process of Genetic Counselling among Clinicians’. Using the pre validated questionnaires the study found that approximately 90% of clinicians recognize the importance of collecting a multi-generational family history, providing advice on inheritance patterns, recurrence risk, and genetic tests for disorders with genetic causes. However, the study suggests that clinicians may face challenges, such as lack of awareness or time constraints, in addressing genetic aspects thoroughly. It emphasizes the need for comprehensive genetic counselling in multiple sessions to alleviate the burden of genetic disorders in society. The recruitment method for the participants is

not mentioned, raising concerns about potential bias if they came from specific departments or had pre-existing beliefs about genetic counselling.

3. The study by Agouti, J.M et al in 2023 titled the importance of on-site genetic counselling for prospective assisted reproductive technology patients aimed to evaluate the benefits of integrating genetic counselling into an assisted reproductive technology (ART) center. By analyzing data from 1340 couples undergoing ART treatment, the researchers found that 11.2% were referred to genetic counselling. The majority of referrals were due to known genetic risks, while others had potential genetic concerns. The integration of genetic counselling facilitated informed decision-making, identified couples requiring further testing, and optimized ART outcomes. The authors concluded that an on-site genetic counselling service significantly enhances the ART process, improving patient care and reducing the workload of ART staff.

4.In a recent study titled 'Familiarity, Knowledge, Attitude and Willingness towards Genetic Counselling among the South Indian Population' a cross sectional quantitative research ,conducted by Prabha, L.J., & Revathy, K.S. (2022), the objective was to gauge awareness and explore individual factors like knowledge, attitude, and willingness towards genetic counselling (GC) in the South Indian population. The findings demonstrated a positive attitude and willingness toward GC, with distinctive knowledge patterns compared to similar studies. The study suggests the need for more qualified professionals in the field and advocates for integrating genetic counselling and testing as primary preventive measures within government healthcare sectors. These measures are seen as essential in alleviating the burden of genetic diseases in both families and society. The study can incorporate how cultural beliefs, practices, and stigma might influence attitudes and access to genetic counselling.

5. Kumar et al. (2021) investigated the knowledge, attitude, and practice of prenatal diagnosis among obstetricians and gynecologists (OBGYNs) in Kerala, India, using a cross-sectional survey design. Their findings, while highlighting a generally positive attitude towards prenatal diagnosis, revealed knowledge gaps regarding specific tests and protocols. This underscores the potential significance of genetic counselling. By providing in-depth information about prenatal testing options, interpreting results, and addressing patient concerns, genetic counsellors can bridge these

knowledge gaps and ensure informed decision-making by pregnant women and their partners. The study also identified variations in OBGYN practices, suggesting the need for standardized guidelines. However, the cross-sectional design and regional focus limit the generalizability of these findings (Kumar et al., 2021).

6. Khatak, S & Wadhva, N studied Public perception of genetic counselling in India: Opening mind Eyes in October 2020. During the lockdown period from June 30th to July 6th, 2020, a survey aimed at raising awareness about genetic counselling was conducted using a structured questionnaire. The survey successfully engaged nearly 400 respondents, predominantly (69%) from non-medical backgrounds. Gender distribution was balanced, with 53% male and 47% female participants contributing to the study. Among the respondents, 61% were unmarried individuals, highlighting the growing recognition of genetic counselling as a more dependable alternative to traditional methods such as horoscope matching when considering marriage and future family planning.

7. El-Deen et al (2020) cross-sectional study examined the burden experienced by primary caregivers of children with Down syndrome in Egypt. Using the Zarit Burden Interview, the researchers found that over 40% of caregivers reported moderate to severe burden, with factors such as caregiver gender, family structure, child's age, and medical conditions influencing this outcome. These findings highlight the significant emotional and psychological toll on caregivers of individuals with Down syndrome, emphasizing the need for comprehensive support services to address their well-being.

8. Cassis F et al in 2020 studied Psychological interventions for people with hemophilia. Managing hemophilia presents significant challenges, both physically and psychologically. Individuals with hemophilia often face difficulties in social interactions, physical activities, and emotional well-being. This review aimed to assess the effectiveness of psychological therapies in helping people with hemophilia cope with their condition. While the review identified several studies on various psychological interventions, the quality of evidence was generally low. The available research suggests potential benefits of psycho-education and computerized learning programs in improving coping strategies and quality of life, but more rigorous studies are needed to establish the effectiveness of these interventions. Overall, there is a clear need for further research on

psychological interventions for people with hemophilia to provide more robust evidence for clinical practice.

9. Sheethal Sharda (2020) in a newspaper article named *All in the genes* highlights how advancements in genetics and genomics have led to personalized medicine, where treatments are tailored to individual patients based on their genetic makeup. While these advancements offer potential benefits, they also introduce complexities that both doctors and patients struggle to understand. Genetic counsellors bridge the gap between complex genetic information and patient comprehension, empowering individuals to make informed healthcare decisions aligned with their personal values and family goals. Essentially, genetic counselling is becoming an essential component of quality healthcare.

10.Kumar, A., et al (2019) studied Genetic counselling in India: Current status and future directions. His article uses a review methodology. The authors analyze existing literature on genetic counselling in India to provide an overview of the current state and future directions for the field. Genetic counselling services in India are limited and unevenly distributed. Their findings were most services are concentrated in major cities and tertiary care hospitals. There is a shortage of qualified genetic counsellors. Awareness about genetic counselling among healthcare professionals and the public low. Lack of standardized protocols and reimbursement policies for genetic counselling services. The article highlights the potential benefits of genetic counselling, including: Improved decision-making for families at risk of genetic disorders Reduced psychological distress for patients and families. Increased utilization of preventive measures and early detection strategies. The limitation of study are this is a review article and does not present original research findings. The authors do not provide a systematic analysis of the literature. The limitations of existing research on genetic counselling in India are not explicitly discussed.

11. Family Shares Their Experience with Fragile X (20 May 2019) Attitude (Video: https://www.youtube.com/watch?v=CUUOF9NaqPo)The genetic condition Fragile X poses significant challenges for those affected and their families. The three sisters, who discovered their family's connection to Fragile X only recently, detail their difficulties:

The family's journey to understanding Fragile X was marked by delays and confusion. The diagnosis of their children came at different times, often with mistaken diagnoses like autism, before Fragile X was identified. This delay in diagnosis led to periods of uncertainty and frustration for the parents, as they struggled to connect symptoms with the correct condition. Discovering that multiple children were affected by Fragile X had a profound emotional impact. Parents felt guilt for passing on the condition, despite it being an unknown factor. The realization that their children had this condition brought both relief and distress, as it answered long-standing questions but also introduced new challenges. Children with Fragile X face various educational and social hurdles. They often experience delayed milestones, learning difficulties, and significant challenges with social interactions. Their behaviors, such as heightened sensory sensitivities and extreme reactions to new environments, complicate their ability to engage in typical activities and schooling. Managing Fragile X impacts family dynamics and daily life. The need for constant vigilance and adaptation to the children's needs—such as preparing visual aids for routine activities—can be overwhelming. Additionally, the strain of coordinating care and navigating the education system adds to the family's burden. The family highlights the importance of early intervention and tailored support. They emphasize that appropriate strategies, like using social stories and sensory diets, significantly improve their children's well-being. However, accessing such support and educating others about Fragile X remains an ongoing challenge. Overall, Fragile X presents a complex set of difficulties that affect every aspect of life for those impacted and their families, from emotional and practical challenges to the need for specialized support and understanding.

12. Prajnya, R et al (2017) studied prenatal screening for genetic disorders Suggested guidelines for the Indian Scenario. Prenatal screening is a vital tool for preventing genetic disorders and congenital disabilities. While the article emphasizes the importance of prenatal testing for conditions like Down syndrome, beta-thalassemia, and neural tube defects, it also underscores the critical need for genetic counselling. The lack of standardized guidelines, coupled with insufficient physician knowledge about screening tests and timing, highlights the gap in prenatal care. This emphasizes the crucial role of genetic counsellors in providing accurate information, explaining test results, and supporting patients in making informed decisions. To maximize the benefits of prenatal screening, the article calls for the development of comprehensive guidelines and improved

access to genetic counselling services. In essence, genetic counselling is essential to ensure that prenatal screening is effective and beneficial for expectant parents.

13.Haven, J. (2017 February 09) What Is Genetic Counselling? (Videohttps://www.youtube.com/watch?v=7yIW0L9dLCQ)

Genetic counselling is a concept that remains unfamiliar to many individuals, with an even smaller number understanding the specifics of what genetic counsellors do. Genetic counsellors possess advanced training in human genetics and counselling and are employed in various settings. At Shodair Hospital in Helena, within the Medical Genetics department, a team of skilled professionals includes geneticists, a nurse navigator, other genetic counsellors, a nutritionist, a psychologist, and lab staff. This team collaborates to provide answers for families. Shodair employs a unique model by offering services at outreach sites across the state, including on reservations, thus reaching underserved patients with cutting-edge services. Genetic counsellors often face emotionally charged situations, such as delivering life-altering news about conditions like Down syndrome, cystic fibrosis, autism, or Huntington's disease. The genetic counsellor has a strong interest in cancer genetics, exemplified by a case involving a patient named Mary. At age 47, Mary was referred for genetic counselling due to a family history of breast and ovarian cancer. The counsellor reviewed Mary's family and personal history to assess her lifetime risk of breast cancer, which was over 20%, qualifying her for additional breast MRI screenings. They discussed the general genetics of cancer, the role of genetic predisposition, and various testing options. Mary chose to pursue genetic testing for BRCA1 and BRCA2, the two most common cancer-related genes. Testing for BRCA genes has increased significantly, facilitating important family discussions. For patient Mary, a holiday conversation led her to seek genetic counselling. She tested positive for a BRCA1 mutation, indicating an 85% risk of breast cancer and a 40% risk of ovarian cancer. The counsellor discussed options including increased screening, surgery, and medication, and informed Mary that her daughters had a 50% chance of inheriting the mutation. Mary decided on follow-up screenings but was later diagnosed with stage 4 ovarian cancer and passed away within a month. Her daughters have different approaches to managing their potential risk. This case highlights the complex, emotional nature of genetic counselling and underscores the growing importance of genetic testing.

14. Barton et al (2016) review highlights the significant psychological burden experienced by individuals with cystic fibrosis (CF) and their caregivers. Through a literature review, they found elevated rates of depression and anxiety, emphasizing the need for regular psychological assessments using standardized tools. The study also revealed a correlation between psychological distress and decreased adherence to treatment regimens, leading to poorer health outcomes. These findings underscore the importance of integrating psychological support into CF care, including the role of genetic counselling in addressing the emotional challenges associated with a chronic genetic condition.

15. In their review titled "The economic burden of inherited genetic diseases: A challenge for health systems worldwide" (Modell & Drury, 2016), the authors analyze existing literature to demonstrate the significant economic strain these conditions place on healthcare systems globally. The review methodology highlights direct medical costs (treatment, hospitalization) as well as indirect costs (loss of productivity, caregiver time), emphasizing the substantial financial burden, particularly in low- and middle-income countries. This underscores the importance of genetic counselling, as it can potentially prevent the occurrence of genetic conditions, thereby mitigating the economic burden on individuals and healthcare systems as a whole. However, it's important to acknowledge that this is a review and doesn't present original research, and specific cost data may vary depending on location and healthcare systems. (Modell & Drury, 2016)

16. Ghorpade, N., et al (September, 2016) studied impact of genetic counselling named ‘Impact of a Genetic Counselling Replica on Parents of Children with Down Syndrome: A South Indian Cohort Study’ and summarized that, in situations where parents have children with Down syndrome and lack awareness of the condition, genetic counselling becomes crucial. Two phases of genetic counselling sessions were conducted with eight parents in this study. Following the sessions, parents gained a better understanding of the potential reasons for their child's condition. The researchers concluded that genetic counselling serves as a valuable tool to assist parents of children with Down syndrome. The study could include more about the interventions and it’s before and after effects.

17. Das, K., & Mohanty, D (October, 2011) organized a study entitled ‘Genetic counselling in tribes in India’ during Neonatal Screening Programmed undertaken for sickle cell disease in Kalahandi district of Orissa and Community Screening Programmed in primitive tribes of India in

four States viz. Orissa, Gujarat, Tamil Nadu and Maharashtra. Genetic counselling in these areas has to be continuous to achieve success and therefore the need for setting up of permanent centers in the tribal areas in India.

18. Coad et al administered a study on title ‘Parents’ and children’s communication about genetic risk: a qualitative study, learning from families’ experiences, in 2011. The research utilized semi-structured interviews to explore various aspects related to genetic risk information within these families, focusing on roles of family members, communication dynamics, and psychological outcomes. Grounded theory was employed to analyze findings from 33 participating families, encompassing a total of 79 individuals, including children aged 8-11 years and young people aged 12 and older. Parents highlighted multiple challenges in communicating effectively with their children about genetic risks, emphasizing the need for more guidance from healthcare professionals on how to provide developmentally appropriate information. The study concluded by advocating for enhanced support for families in discussing genetic risk information across a child's development. It underscored the benefits of ongoing, open communication about genetic risks throughout childhood, suggesting that such approaches could improve coping mechanisms and facilitate acceptance of the implications associated with genetic conditions.

19. Rolnick et al. (2011) surveyed genetic service providers to understand their perspectives on barriers to referral for genetic counselling on title Barriers to Identification and Referral for Genetic Counselling. Genetic counselling plays a crucial role in this process by providing information, risk assessment, and emotional support. However, significant gaps exist between identification of at-risk individuals and referral for genetic counselling. The study found limited use of referral guidelines (46%), suggesting a need for improved education and standardized protocols for identifying high-risk patients. barriers, including: Lack of awareness or knowledge about genetic counselling (36%), Fear of the implications of a positive test result (20%), Concerns about insurance coverage and potential discrimination (52%), Distance to appointments and lack of access to services (48%), Prioritization of other health concerns over cancer risk assessment (72%), Discouragement from family members (28%). Additionally, concerns about the psychological impact of genetic testing, potential insurance implications, and limited access to services are commonly reported. While Rolnick et al. (2011) provides valuable insights, the study focuses

solely on the perspectives of genetic service providers. Future research should explore patient perspectives on these barriers to gain a more comprehensive understanding. Additionally, research investigating the effectiveness of interventions aimed at addressing these barriers is crucial.

20. Aitken et al (2007) carried out a study on topic ‘Communicating genetic information in families – a review of guidelines and position papers’ using data sources from the websites of national human genetics societies, bioethics committees, research institutes, legal recommendations committees, and regional and international health organizations adopting method meta-analysis. The study focuses on the communication of genetic information within families, reviewing existing guidelines and position papers to understand best practices and recommendations. This review is crucial for informing genetic counselling practices, as effective communication of genetic information can significantly impact family dynamics and patient outcomes. Forrest systematically identified and analyzed guidelines and position papers related to the communication of genetic information. This involved a comprehensive search of relevant databases and professional organizations. The study involved a thematic analysis of the selected documents to distill key recommendations and practices. The study concluded that Effective communication requires clarity in conveying complex genetic information and sensitivity to the emotional impact on family members and Guidelines advocate for ensuring that family members understand the implications of genetic information to support informed decision-making. The study’s findings offer practical recommendations for genetic counsellors, including the need for tailored communication strategies and supportive resources for families. The review underscores the importance of developing and implementing standardized guidelines to ensure consistency in genetic counselling practices. The identified gaps suggest a need for further research on the effectiveness of different communication strategies and the impact of genetic information on family dynamics.

21. Family system characteristics and psychological adjustment to cancer susceptibility genetic testing: a prospective study by Asperen et al in 2007 .This study examined prospectively the contribution of family functioning, differentiation to parents, family communication and support from relatives to psychological distress in individuals undergoing genetic susceptibility testing for a known familial pathogenic BRCA1/2 or Hereditary nonpolyposis colorectal cancer-related mutation. Family functioning, differentiation to parents, hereditary cancer-related family

communication and perceived support from relatives were assessed in 271 participants for genetic testing before test result disclosure. Hereditary cancer distress (assessed by the Impact of Event Scale) and cancer worry (assessed by the Cancer Worry Scale) were assessed before, 1 week after, and 6 months after test result disclosure. Participants reporting more cancer-related distress over the study period more frequently perceived the communication about hereditary cancer with relatives as inhibited, the nuclear family functioning as disengaged-rigid or enmeshed-chaotic, the support from partner as less than adequate and the relationship to mother as less differentiated. Especially, open communication regarding hereditary cancer and partner support may be important buffers against hereditary cancer distress. Identifying individuals with insufficient sources of support and addressing the family communication concerning hereditary cancer in genetic counselling may help the counselee to adjust better to genetic testing.

22. McDaniel's (2005) article emphasizes the crucial role of psychotherapy in supporting individuals and families coping with genetic disorders. The author underscores the complex emotional and interpersonal challenges posed by these conditions, including anger, guilt, ambivalence, and communication difficulties. The article highlights the importance of understanding family dynamics and developmental issues in addressing these challenges. It advocates for the inclusion of family therapists within genetic healthcare teams to provide comprehensive support through individual, couple, and family therapy, as well as psych educational groups. Essentially, the article positions psychotherapy as a vital component in managing the psychological impact of genetic disorders.

23. Mahay, S.B. (2002) conducted a study titled 'The Burden of Genetic Disorders in India and a Framework for Community Control.' The study found a high prevalence of genetic disorders in India, attributed to its large population, high birth rate, and the common practice of consanguineous marriages in many communities. The conclusion emphasized the integration of genetic services into existing primary healthcare and medical services. The recommendation included training physicians in district and medical school hospitals to provide genetic counselling. The study suggested the establishment of additional medical genetics departments in medical schools for continued progress. The study can consider including other relevant areas of medical genetics beyond congenital malformations and specific disorders.

24. Broadstock & Marteau in 2000 conducted study titled Psychological consequences of predictive genetic testing: a systematic review. This systematic literature review aimed to examine the psychological consequences of predictive genetic testing. A comprehensive search across five databases yielded 15 studies involving individuals undergoing testing for Huntington's disease, hereditary breast and ovarian cancer, familial adenomatous polyposis, and spin cerebellar ataxia. The findings indicate that while both carriers and non-carriers experienced decreased distress following testing, pre-test emotional state was a stronger predictor of post-test psychological outcomes than the test result itself. Contrary to expectations, no consistent evidence of increased distress was found in either group. The included studies were primarily focused on Huntington's disease and had short follow-up periods. Additionally, the self-selected nature of participants and the lack of standardized counselling interventions restrict the generalizability of findings and limit conclusions about the relationship between counselling and emotional outcomes. This emphasizes the need for further research with longer follow-up periods, diverse participant populations, and experimental manipulation of counselling interventions to better understand the psychological impact of predictive genetic testing and to inform optimal counselling practices.

25. The Rotterdam/Leiden Genetics Workgroup (1999) conducted a study examining psychological distress in individuals facing predictive DNA testing for autosomal dominant late-onset disorders. The study compared individuals at risk for Huntington's disease, hereditary cerebral hemorrhage with amyloidosis Dutch-type, familial adenomatous polyposis coli, and hereditary breast and ovarian cancer. Participants completed questionnaires assessing intrusion, avoidance, anxiety, depression, hopelessness, and psychological complaints. Additionally, in-depth interviews explored participants' emotional responses to genetic testing. Results indicated higher levels of anxiety, depression, and psychological complaints among individuals at risk for neurodegenerative disorders compared to those at risk for cancer syndromes. Those with high intrusion and avoidance scores exhibited greater psychological distress but also demonstrated more reflective coping mechanisms during interviews. The study highlighted the importance of identifying coping strategies to provide appropriate counselling and support. However, the long-term consequences of denial coping strategies remain unclear. This study contributes to the understanding of the psychological impact of predictive genetic testing by demonstrating the

complex relationship between psychological distress, coping mechanisms, and the type of genetic disorder.

2.1 Conclusion

The literature review reveals that genetic counselling is a critical component in managing genetic disorders and advancing personalized medicine. Research highlights the essential role of genetic counselling in various contexts, including hereditary disease management, prenatal care, and assisted reproductive technologies. Studies indicate that genetic counselling improves decision-making, enhances patient care, and supports families in navigating complex genetic information. However, the studies reviewed also expose several gaps and challenges within the field. While there is a general recognition of the benefits of genetic counselling, issues such as limited access, cultural barriers, and knowledge gaps among healthcare providers persist. Extensive studies done in Kerala are scares. There is a lack of comprehensive data on the availability and accessibility of genetic counselling services in different regions of Kerala. Research can focus on mapping out these services, especially in rural and underserved areas. Addressing these gaps will help enhance the effectiveness of genetic counselling services and improve their integration into healthcare systems, ultimately benefiting patients and families affected by genetic disorders.

CHAPTER III

METHOD

The aim of this chapter is to clearly outline the methodology employed in this study, detailing the methods utilized and the rationale behind their selection. It addresses key components such as the research design, providing an overview of the overall approach; the participants, including criteria for selection; the data collection tools used to gather data; and the data analysis, which was conducted using thematic analysis. Additionally, careful consideration of ethical issues was undertaken during the research planning phase. This structured approach ensures a comprehensive understanding of the methodology and its implementation.

3.1 Research Design

Sunaina Singh (2023) wrote that a research design is the plan or framework used to conduct a research study. It involves outlining the overall approach and methods that will be used to collect and analyze data in order to answer research questions or test hypotheses. A well-designed research study should have a clear and well-defined research question, a detailed plan for collecting data, and a method for analyzing and interpreting the results. A well-thought-out research design addresses all these features (Researcher Life, 2023). This current study use the research design ‘Interpretivism’.

According to *Interpretivism – Research Methods Handbook. (2020),* Interpretivism, often contrasted with Positivism, is a research approach that asserts knowledge in the human and social sciences cannot be understood using the same methods as natural sciences. Unlike Positivism, which seeks objective truths, Interpretivism recognizes that aspects of human experience—such as emotions, values, and cultural influences—cannot be fully known or quantified in an objective manner. Instead, Interpretivism aims to generate understanding and often adopts a relativist perspective, accepting that multiple, subjective viewpoints can coexist.

In Interpretivist research, qualitative methods are preferred to explore complex human phenomena. This research may involve unstructured or "messy" data, and various techniques are employed to handle this type of data. Interpretivism acknowledges that researchers' own cultural and personal biases inevitably influence their studies and often embraces this subjectivity as part of the research process (*Interpretivism – Research Methods Handbook. 2020).*

A significant challenge in Interpretivism is ensuring the validity of qualitative research. Since it accepts that objective knowledge is unattainable, the approach focuses on subjective interpretations and the researcher's role in constructing knowledge. Interpretivism often incorporates a relativist epistemology, integrating different perspectives to form a comprehensive understanding or narrative (*Interpretivism – Research Methods Handbook. 2020).*

Key features of Interpretivism include recognizing that the social world cannot be fully understood from a single individual perspective, as realities are multiple and socially constructed. This approach acknowledges that the interaction between researcher and participants is an inherent aspect of research and emphasizes the importance of context in shaping knowledge. Interpretivism also understands that knowledge can be value-laden, and these values should be made explicit. It focuses on understanding individual experiences rather than seeking universal laws, views causes and effects as interdependent, and considers contextual factors essential to the research process (*Interpretivism – Research Methods Handbook. 2020).*

Interpretivism is often associated with Constructivism, which serves as its ontological and epistemological foundation. Constructivism emphasizes that knowledge is constructed through human interaction and is often used in educational research. Common methods in Interpretivist research include case studies, interviews, focus groups, document analysis, and various forms of qualitative analysis such as thematic analysis and phenomenology (*Interpretivism – Research Methods Handbook. 2020).*

3.2 Participants:

Participants are the individuals who take part in a study, either directly or indirectly via a representative, and who have given their informed consent (Ethical Research Involving Children, 2024).Data was collected from eight mental and health professionals who work with clients with genetic conditions, susceptible to genetic disorders or those who come from family with history of genetic disorders. They were selected through purposive sampling. Each participant was labeled accordingly, and their professions are outlined below:

P1- Infertility Specialist

P2- Gynecologist

P3- Psychiatrist

P4- Counselling Psychologist

P5- Gynecologist

P6 -Medical Geneticist

P7- Counselling Psychologist

P8- Behavior Analyst

3.3 Tools

Research tools encompass a broad array of resources, methods, instruments, software, and techniques used by researchers to gather, analyze, interpret, and communicate data throughout the research process. These tools facilitate various aspects of research, including data collection, organization, analysis, visualization, collaboration, and documentation. They can be physical, such as laboratory equipment and survey instruments, or digital, like software and online databases. Essential for conducting research effectively and rigorously, these tools vary according to the specific objectives, methods, and requirements of each study. Examples include laboratory apparatus, survey questionnaires, statistical software, data visualization tools, literature databases, and collaboration platforms (Research Tools – EvalCommunity, 2024).

The present qualitative study deployed a Semi Structured Interview to gather data from the participants. A semi-structured interview is a data collection method that involves asking questions within a predefined thematic framework, but without a fixed order or specific wording for the questions. This approach is commonly used in qualitative research across various fields, such as marketing, social science, and survey methodology. Semi-structured interviews serve as an exploratory tool, enabling researchers to gather in depth insights while maintaining flexibility. In field research with multiple interviewers, this method provides a consistent theoretical framework, allowing each interviewer to explore different aspects of the research question (Scribbr, 2022).

3.4 Research Questions

1. What is your understanding of ‘Genetic Counselling’?

2. What are the concerns of family with a genetic condition?

3. Importance of conducting Genetic Counselling'?

4. Does genetic counselling helps to improve the gene pool? (If someone has a genetic condition, by providing suggestions and counselling does it reduce the expression of that gene)

5. How Genetic Counselling is being done in Kerala?

6. Is there any barriers for practicing it?

7. What strategies can be implement to promote and expand the Genetic Counselling?

3.5 Informed Consent Form

An informed consent form which includes the terms of confidentiality and the purpose of the study was given to the participants to ensure their voluntary participation in the study. Which is included in Appendices.

3.6 Interview Schedule

Interview schedule prepared to collect the data through online and offline, schedule is given in Appendices.

3.7 Data Analysis

Qualitative research produces unstructured, text-based data such as interview transcripts, observation notes, and multimedia materials like audio and video recordings. Unlike quantitative research, which uses statistical methods, qualitative analysis involves a dynamic process of inductive reasoning to explore values, meanings, and experiences. This analysis includes systematically organizing and coding data into categories, identifying patterns, and drawing insights. While software tools can aid in organizing and managing data, researchers must still interpret and synthesize the findings themselves, as the analysis process remains inherently complex and intuitive (National Library of Medicine, 2008). In this study researcher used ' Thematic Analysis' to analyze and gather information from the verbatim of participants .As cited in Journal of Nelta , 2020 ,Thematic analysis is a qualitative research method that researchers use to systematically organize and analyze complex data sets. It is a search for themes that can capture the narratives available in the account of data sets. It involves the identification of themes through careful reading and re-reading of the transcribed data (King, 2004; Rice & Ezzy, 1999). A rigorous

thematic analysis approach can produce insightful and trustworthy findings (Nowell, Norris, White & Moules, 2017).

CHAPTER IV

RESULT & DISCUSSION

Chapter outlines the results and discussions based on the analysis of the collected data. For each research question (RQ), the results are presented and followed by a discussion that incorporates the researcher's observations, theoretical frameworks, and pertinent literature according to identified themes and subthemes. Major themes and subthemes identified which is given below:

Table 1: Identified major themes and subthemes

Major Themes	Sub Themes
Knowledge	Perception
Concerns of Families	Emotional and psychological impacts, Practical concerns (financial & social), Decision-making processes within families
Role of Genetic counselling - enhancing gene pool	Perceived Benefits, Long-term Implications and Future Directions
Educational and Informative Role	Genetic Risks, Genetic Literacy, Health Management
Practices and Implementation	Processes and Protocols
Access Barriers	Affordability , Awareness, Stigma, Emotional Distress and Lack of Integrated Services
Strategies	Legislative support and Public Education

1. What is your understanding of ‘Genetic Counselling’?

The collected responses are as follows:

- “I work in an infertility settings, according to my knowledge in this field we get cases like Intellectual disability, parents who are diagnosed with Learning Disability and majority of cases with high maternal age and recurrent pregnancy loss at that time we give them information’s regarding their genetic condition and available remedies concentrating on available tests, it is not proper genetic counselling but we don’t have such departments here.” – **P1**
- "I don't have much knowledge on this, only limited. From what I understand, when dealing with patients, particularly those with neurodevelopmental conditions, we ask about their genetic background and family history during the medical history recording. This helps us understand why the disorder occurred and how family history might have played a role. This process is crucial in fields like neurology, pediatrics, prenatal medicine, and even conditions related to metabolism or cancer. Typically, psychiatric social workers handle this to help prevent the transmission of these conditions. It's what I know as genetic counselling." **P7**
- "In my understanding, genetic counselling is provided to families or individuals who may be at risk of developing genetic conditions. Its purpose is to assist them in taking necessary precautions, preparing them mentally to cope with potential situations, and aiding in decision-making processes. However, it's not widely practiced here, so my knowledge on this subject is somewhat limited." **P3**
- "To understand genetic counselling, you really need a strong grasp of genetics and psychological counselling. This includes understanding genetic disorders and counselling techniques, which are crucial for conducting both pre and post genetic counselling sessions. In pre-genetic counselling, it's about understanding how conditions develop and the probability or susceptibility to those conditions. In post-genetic counselling, the focus shifts to educating clients about their conditions and prevention methods. That's why a master's degree in both genetics and counselling is essential to practice genetic counselling." **P6**

- “I don’t have much knowledge regarding this, so am not able to provide you informations for your purpose, I will refer someone else” **P2**
- “I am working as a gynecologist , what I do is prescribe for genetic tests if there is such doubts , I don’t know what this is actually in a practical scenario, we have learnt it during our education, am sorry” **P5**
- "From what I understand, genetic counselling involves evaluating the genetic factors related to a person's concern and providing both emotional support and assistance to the individual and their family." **P4**
- "Genetic counselling is a form of guidance that provides psychoeducation and raises awareness for individuals who are either predisposed to or already have a genetic condition. It covers potential issues, how the condition may develop, its causes, possible solutions, and ways to manage or prevent it. These components are typically included in genetic counselling in my knowledge” **P8**

Through the “RQ1- What is your understanding of ‘Genetic Counselling’?” the researcher aimed to explore professionals' comprehension of genetic counselling, focusing on key aspects like what genetic counselling fundamentally entails, who is responsible for performing genetic counselling and how it can assist the patients. For the above research question the identified major theme is Knowledge and sub theme is Perception.

Theme 1 – Knowledge

The upcoming responses will focus on the theme of knowledge:

“I don’t have much knowledge regarding this, so am not able to provide you information for your purpose” (P2).

"I don't have much knowledge on this, only limited” (P7).

"In my understanding, genetic counselling is provided to families or individuals who may be at risk of developing genetic conditions” (P3).

"From what I understand, genetic counselling involves evaluating the genetic factors related to a person's concern and providing both emotional support and assistance to the individual and their family" (P4).

According to FC Fraser as cited in article ‘What is ideal genetic counselling? A survey of current international guidelines’ (Hietala M et al,2008), the core of genetic counselling is to present medical and genetic facts to the counselees, and to help them to understand their meaning and choose the course of action most appropriate to them in relation to the genetic problem present in the family. Assessing the knowledge of genetic counselling yielded varied responses. Some participants (n=2) indicated limited knowledge in this area, while others'(n=6) responses aligned with the views expressed by FC Fraser. Based on the participants' limited knowledge, genetic counselling is generally provided to individuals with genetic conditions, families with a member diagnosed with a genetic condition, or those at risk of such conditions. This counselling involves discussing the causes and available treatment options, helping individuals or families prepare mentally and take necessary precautions. The limited understanding observed can be attributed to insufficient exposure and experience in this field. A thorough understanding of genetic counselling requires a deeper knowledge of both genetics and counselling. For families affected by or predisposed to genetic conditions, genetic counselling plays a crucial role in guiding them to take appropriate actions. It helps clarify genetic concerns, providing both emotional and professional support. This process is particularly important in specialized fields such as neurology, pediatrics, prenatal medicine, and conditions related to metabolism or cancer.

Genetic counselling is a specialized field within counselling that provides comprehensive guidance and psychoeducation related to genetic conditions. This process involves educating individuals and families about the nature of genetic disorders, including how these conditions arise, their potential implications, and the available options for management and treatment. Genetic counselling is typically conducted by professionals who possess expertise in both counselling techniques and genetic sciences. These professionals help clients understand the genetic factors influencing their health and offer support in interpreting genetic testing results. It provides a framework for understanding genetic risks, planning for potential health challenges, and accessing appropriate medical interventions.

Sub theme 1- Perception

The upcoming responses will address the theme of perception:

"Cases like Intellectual disability, parents who are diagnosed with Learning Disability and majority of cases with high maternal age and recurrent pregnancy loss at that time we give them information's regarding their genetic condition and available remedies concentrating on available tests" (P1).

"This includes understanding genetic disorders and counselling techniques, which are crucial for conducting both pre and post genetic counselling sessions" (P6).

"Providing both emotional support and assistance to the individual and their family" (P4).

The participants have perceived genetic counselling as a service that provides information about genetic conditions, with particular emphasis on intellectual disabilities, neurodevelopmental disorders, learning disabilities in parents, advanced maternal age, and recurrent pregnancy loss. This service aims to help families or individuals take preventative measures, mentally prepare for potential outcomes, and make informed decisions. Participants describe pre-genetic counselling as involving the evaluation of the development and probability of genetic conditions, while post-genetic counselling focuses on educating clients about their specific conditions and strategies for prevention. This perspective illustrates their understanding of genetic counselling as a holistic process designed to address and manage genetic conditions comprehensively. These perceptions are aligned with the study conclusion of research titled 'Familiarity, Knowledge, Attitude and Willingness towards Genetic Counselling among the South Indian Population', concluded that Genetic Counselling aims to people understand and adapt to medical, psychological and familial implications of genetic conditions (Prabha & Revathy,2022). Conversely, one participant (P5) viewed genetic counselling primarily as the prescription of genetic tests, while another participant (P7) perceived it more as a component of medical history taking and prevention rather than a specialized, comprehensive service.

So the genetic counselling can be aimed to individuals and families those who are susceptible or those who live with such condition to take informed decisions regarding family planning and living arrangements.it can give the instrumental and emotional support in those times. This service can safeguard the physical and psychological wellbeing of population. Metcalfe S A (Genetic counselling, patient education, and informed decision-making in the genomic era, 2017) stated appropriate pre-test genetic counselling is recommended to clients who seek information regarding genetics should provide psychosocial support so that couples can consider the pros and cons of being tested and can make informed decisions.

2. What are the concerns of family with a genetic condition?

Collected responses are as follows:

- "Most of the families especially in consanguineous marriages they may already know the probability of that condition so embryo screening after IVF is recommended. Those who don't expect such conditions are hard to handle the spouse blame opposite partner like he/ she is impotent so they are moving to divorce some says they don't have problem but family has so they will move to divorce. Couples with exceeded age also expect the probability but they get upset if the results confirm.so we give awareness like it is not done consciously but it is an unconscious occurrence and remedies are provided." **P1**
- "Families facing genetic conditions often have numerous concerns, primarily centered on health due to either a family history of such conditions or the likelihood of inheriting them. They worry about life expectancy and whether they can maintain good health. These worries extend to emotional and psychological concerns, including stress about passing the condition to the next generation and its potential impact on their quality of life and future. This stress can lead to mood disorders in some cases. Financial challenges are also significant, alongside social stigma, particularly concerning mental illness. This stigma can isolate individuals from social gatherings and make them reluctant to seek treatment or share their condition with others. Caregivers also face burdens as their roles are often impaired by these conditions, turning care into a significant responsibility for others. Discrimination

> can further exacerbate these challenges, affecting job opportunities and potentially leading to underpayment due to perceived inefficiencies compared to others."**P3**

Through the "RQ2- What are the concerns of family with a genetic condition? The researcher wants to explore the effect of genetic conditions on family, how they perceive and handle it. And what all are the psychological turmoil a family goes through in such situations and how the professionals deal such family concerns. The major theme identified is Concerns of family and sub themes are Emotional and psychological impacts, Practical concerns (financial & social) and Decision-making processes within families.

Theme 1- Concerns of Family

The upcoming responses will cover the theme of family-related concerns:

"Most of the families especially in consanguineous marriages they may already know the probability of that condition" (P1).

"Couples with exceeded age also expect the probability but they get upset if the results confirm" (P1).

"Families facing genetic conditions often have numerous concerns, primarily centered on health due to either a family history of such conditions or the likelihood of inheriting them" (P3).

"Affecting job opportunities and potentially leading to underpayment due to perceived inefficiencies compared to others" (P3).

A genetic disorder is a disease caused in whole or in part by a change in the DNA sequence away from the normal sequence and some diseases are caused by mutations that are inherited from the parents and are present in an individual at birth, like sickle cell disease (National Human Genome Research Institute). Families facing genetic conditions often have a range of concerns primarily focused on health due to either a family history of such conditions or the likelihood of inheriting them. These concerns extend to the potential risk of passing the condition to future generations, implications for quality of life, and potential challenges in job opportunities or perceived underemployment due to perceived inefficiencies. Additionally, families formed through

consanguineous marriages or among older couples may anticipate these risks, but the confirmation of a genetic condition can still result in significant distress and emotional upheaval.

Since the conditions are inherited or if someone is a carrier of such mutations the family where that individual can have some concerns like chances of passing to next generations which can be really tiring emotionally. The dominancy and recessiveness of genes can make a tug of war inside each individual. The condition can effect an individual in many ways like career, job, and family and so on. There is a chance but don't know whether it represent or not which can impair the decision making. Janzing and colleagues (2021) advocated interventions to address the concerns of family in study entitled 'The Impact of Rare Genetic Disorders on Family Functioning' , concluded that greater attention needs to be focused on the stress levels experienced by parents raising a child with a rare genetic syndrome, the average stress levels among these parents are notably concerning. Various factors will facilitate such conditions posing a threat to the well-being not only of the affected but also other family members.

Subtheme 1 - Emotional and psychological impacts

The upcoming responses will cover the theme Emotional and Psychological Concerns:

"Those who don't expect such conditions are hard to handle the spouse blame opposite partner like he/ she is impotent so they are moving to divorce some says they don't have problem but family has so they will move to divorce" (P1).

"Couples with exceeded age also expect the probability but they get upset if the results confirm" (P1).

"They worry about life expectancy and whether they can maintain good health. These worries extend to emotional and psychological concerns, including stress about passing the condition to the next generation and its potential impact on their quality of life and future. This stress can lead to mood disorders in some cases" (P3).

"Caregivers also face burdens as their roles are often impaired by these conditions, turning care into a significant responsibility for others" (P3).

The data confirms that this genetic disorders can impact an individual or a family emotionally and psychologically and those who don't expect such conditions are hard to handle such confirmations.

One of the participant (P1) recalled an experience were the spouse blamed the opposite partner like he/ she is impotent so they are moving to divorce when a genetic test showed positive. Participants reported that even though couples with exceeded age and consanguine relation also expect the probability but they also get upset if the results confirm and the clients worry about life expectancy and it impose a question that whether they can maintain good health and life, these worries extend to emotional and psychological concerns, including stress about passing the condition to the next generation and its potential impact on their offspring's quality of life and future. This stresses are leading to mood disorders in some cases (P3). Not only the patients the caregivers also face burdens as their roles are often impaired by these conditions, turning care into a significant responsibility since some conditions can limit the functioning of an individual. Regarding the limitations Gilchrist D M (PubMed Central, 2002) stated that just because a genetic disease has been diagnosed does not necessarily translate into a cure or even a surgical or pharmaceutical treatment that may delay disease progression or ameliorate symptoms.

The impact of a genetic condition on an individual and their family is both profound and multifaceted, affecting emotional and psychological well-being. These conditions can disrupt the harmony of family life, impacting both the spousal relationship and the broader family system. The lack of awareness about the nature and expression of genetic disorders highlights the critical need for education and support. In this context, genetic counselling plays a crucial role. Proper counselling and support are essential in mitigating distress and providing assistance, especially when dealing with life-changing news. Even in cases where genetic risks are anticipated—such as with carrier parents, consanguineous marriages, or advanced-aged couples—the confirmation of a genetic condition can still be deeply distressing. Therefore, addressing the emotional and psychological concerns of the family through genetic counselling is vital. These findings are in line with study conclusion of Hadley D W et al, that is, the psychological impact of genetic conditions often includes feelings of maternal guilt and paternal blame, which can significantly affect families. To address these emotional challenges, genetic counsellors and health professionals should actively monitor for signs of these feelings and offer opportunities for families to discuss and manage them. Providing anticipatory guidance early after a diagnosis can help normalize these common reactions and facilitate open discussions. Support groups and referrals to mental health resources can further assist in alleviating persistent guilt and preventing potential depression. By addressing these psychological impacts proactively, health care providers

can better support families through the emotional complexities of genetic conditions (Genetics in Medicine, 2006).

Subtheme 2 - Practical concerns (financial & social)

The upcoming responses will support the theme Emotional and Psychological Concerns:

"hard to handle the spouse blame opposite partner like he or she is impotent" (P1).

"Alongside social stigma, particularly concerning mental illness. This stigma can isolate individuals from social gatherings and make them reluctant to seek treatment or share their condition with others" (P3).

"Discrimination can further exacerbate these challenges" (P3).

"Due to perceived inefficiencies compared to others." (P3).

In a study by Belmont et al titled 'Estimating the Burden and Economic Impact of Pediatric Genetic Disease' found, This study used a big database of all types of health insurance claims for kids in the U.S. to figure out how much it costs to care for children who might have a genetic disease. The study found that these kids use a lot more health care services, often needing extra procedures and staying in the hospital for much longer. This extra care can cost between $12,000 and $77,000 more per hospital visit compared to kids who don't have a suspected genetic disease. From this it is clear that there is significant financial burden resulting from genetic disorders, which is applicable to our society too, the presenting data confirms that findings. The cost of genetic tests are huge, all families couldn't afford it. This financial strain often leaves families struggling or feeling helpless when dealing with rare genetic conditions, highlighting a broader issue of accessibility and support for those affected.

Genetic disorders are often mistakenly linked to impotence, which can undermine relationship stability (P1). This misattribution is frequently due to a lack of knowledge and awareness about genetics. As a result, individuals may unjustly blame their spouses for the disorders, leading to relationship breakdowns (P1).The unconscious nature of these issues often means that they are not openly discussed in such situations. Additionally, there is a social stigma associated with genetic disorders similar to that of mental illness, where individuals feel compelled to hide these conditions and are reluctant to talk about them in social settings (P3).Moreover, there is a tendency for

discrimination based on perceived inefficiency compared to others, further exacerbating the challenges faced by those with genetic disorders (P3).

Familial concerns related to genetic disorders are evident in both financial and social aspects. The high cost and economic burden associated with treatment can significantly impact families, potentially affecting their willingness and ability to seek and adhere to treatment. Therefore, cost-effective innovations in this field are urgently needed. Similar to the stigma surrounding mental health, genetic disorders also carry a stigma. People often fear revealing such information due to potential discrimination and various negative labels. This stigma, coupled with the psychological strain, can further exacerbate social difficulties for affected individuals. There is a prevalent myth that genetic conditions are the result of conscious choices, reflecting a lack of understanding. To address this, it is essential to integrate genetic counselling and testing into government healthcare services at affordable prices. Additionally, raising awareness through grassroots campaigns about the nature, causes, and manifestations of genetic disorders, along with forming peer support groups for individuals at risk, will be beneficial in mitigating stigma and improving overall support for affected families.

Subtheme 3 – Decision making processes within families.

The following responses will support the theme Decision making processes within families:

"In consanguineous marriages they may already know the probability of that condition so embryo screening after IVF is recommended" (P1).

"They are moving to divorce some says they don't have problem but family has so they will move to divorce" (P1).

Participant P1 noted that in marriages involving consanguinity, couples are often aware of the increased genetic risks associated with having children. To assess these risks and determine the necessary precautions, genetic testing is recommended through genetic counselling. This process aids in informed decision-making for the couple. Couples who are at risk of transmitting a genetic disease to their offspring may face difficult challenges regarding reproductive decision-making. Deciding if, and how, to purse their child wish can be a demanding process (Gultzow T et al , 2021). Participant also noted that Couples are deciding to divorce or break the relation because

they are not aware of the unconscious nature of this genetic disorders. It indicate the lack of proper orientation to couples which is actually impairing their decision making ability.

Gultzow et al. (2021) investigated hereditary diseases and childbearing decisions in their study, "Hereditary Diseases and Child Wish: Exploring Motives, Considerations, and the (Joint) Decision-Making Process of Genetically At-Risk Couples." Their research highlights the need for further exploration into how couples make decisions collaboratively and the types of support they need. They noted that existing research on prenatal and reproductive choices often emphasizes the role of expectant mothers. Their study found that involving both partners in decision-making tends to result in a broader exploration of options, as shared perspectives can enhance the decision-making process and potentially improve reproductive health outcomes. To facilitate this, Gultzow et al. introduced a communication exercise in their decision aid. This exercise is designed to help partners discuss their personal values and reproductive options together, fostering a better understanding of each other's priorities (National Library of Medicine, 2021).

Decision-making is a critical aspect of daily life, as it often involves ensuring our well-being and minimizing harm to our surroundings. In a family setting, decisions can have profound and life-altering effects, often leading to significant ambiguity and anxiety. Therefore, guiding families towards more productive and informed decisions is essential. Genetic counselling can play a pivotal role in this process.

Assessing an individual's background and providing appropriate guidance is crucial. As indicated by previous responses, genetic disorders can sometimes lead individuals to consider ending relationships. Educating them about the nature of these disorders and offering support for a respectful and informed decision about continuing or ending the relationship is important.

3. Does genetic counselling helps to improve the gene pool?

Collected responses are as follows:

- “Am not aware to give much information on that but what I do is prior checkups and testing.” **P1**
- “Yeah, it is very helpful because we can identify the present abnormalities in genes much earlier. This allows us to prevent transmission or reduce risk factors in the

> next generation, particularly in conditions like diabetes, through interventions such as promoting a healthy diet and regular exercise. The degree of benefit perceived by patient will depend on the level of awareness and understanding of available prevention techniques, underscoring the importance of providing insights and support in this regard."**P3**

Through the "RQ3- Does genetic counselling helps to improve the gene pool? The researcher wants to investigate how genetic counselling can help in preventing or reducing the occurrences of genetic disorders and examine the precautions and lifestyle changes that can be adopted to improve genetic patterns and reduce the risk of genetic disorders. The major theme identified is Role of Genetic counselling- enhancing gene pool and sub themes are Perceived Benefits, Long-term Implications and Future Directions.

Theme 3 - Role of Genetic counselling- enhancing gene pool

The following responses shows the theme Role of Genetic counselling- enhancing gene pool:

"Am not aware to give much information on that but what I do is prior checkups and testing." (P1).

"Yeah, it is very helpful because we can identify the present abnormalities in genes much earlier. This allows us to prevent transmission or reduce risk factors in the next generation, particularly in conditions like diabetes, through interventions such as promoting a healthy diet and regular exercise" (P3).

"The degree of benefit perceived by patient will depend on the level of awareness and understanding of available prevention techniques, underscoring the importance of providing insights and support in this regard." (P3)

Investigating the role of genetic counselling in improving the gene pool received a mixed response. One participant (P1) was unsure about this possibility and preferred to conduct genetic tests early, while another participant found it beneficial because identifying genetic abnormalities early helps prevent transmission or reduce risk factors for future generations, particularly in conditions like diabetes. Interventions such as promoting a healthy diet and regular exercise can be implemented.

The perceived benefit to patients depends on their awareness and understanding of these prevention techniques, highlighting the need for comprehensive education and support. Similar effects of genetic counselling are noted in the study by Aspinwall LG et al., titled "CDKN2A Testing and Genetic Counselling Promote Reductions in Objectively Measured Sun Exposure One Year Later," which concluded that genetic counselling about high melanoma risk, with or without test results, led to lower UVR exposure. Test reporting specifically helped carriers reduce their UVR exposure and showed lighter skin pigmentation over time. Non-carriers did not increase UVR exposure after negative results (Genetics in Medicine, 2020).

The role of genetic counselling in directly altering one's gene pool is not entirely clear, but its value lies primarily in prevention and informed decision-making. Genetic counselling provides individuals with a comprehensive understanding of their genetic makeup, including potential abnormalities and predispositions to various conditions. This knowledge is crucial for understanding one's risk for inherited conditions and can guide subsequent health decisions. With insights gained from genetic counselling, individuals can make more informed decisions about their health. For example, they might choose to undergo preventive measures, adopt healthier lifestyle changes, or pursue early interventions based on their genetic risk factors. While genetic counselling itself doesn't change the genetic code, it helps individuals understand how lifestyle and environmental factors can influence gene expression. By making informed choices, such as improving diet, increasing physical activity, or avoiding known risk factors, individuals can potentially control or mitigate the impact of genetic predispositions. By enabling proactive management and preventive strategies, genetic counselling can reduce the risk of developing certain conditions or manage them more effectively if they arise. This can lead to a reduction in health-related suffering and improve quality of life.

Sub theme 1 - Perceived Benefits

Following responses will address the Perceived benefits:

"Can identify the present abnormalities in genes much earlier. This allows us to prevent transmission or reduce risk factors in the next generation, particularly in conditions like diabetes, through interventions such as promoting a healthy diet and regular exercise" (P3).

From the participant's experience (P3), genetic counselling provides several benefits, including early detection and prevention of genetic abnormalities in individuals or families and this early identification allows individuals to take preventive measures, thereby reducing the risk of passing genetic conditions to future generations. Additionally, Participant (P3) reported that genetic counselling encourages lifestyle changes, such as adopting a healthy diet and engaging in regular physical activity, which can further support overall health and well-being.

Similar result found in a study done by Austin J et al titled 'BEHAVIOURAL CHANGES AFTER PSYCHIATRIC GENETIC COUNSELLING: AN EXPLORATORY STUDY', Participants reported that after participating in genetic counselling, they adopted more protective behaviors like exercising, improving sleep, following medication instructions, seeking professional help, and practicing self-care. They also felt more in control, confident, and accepting of their psychiatric condition. This change happened because genetic counselling helped them address feelings of guilt, shame, fear, and confusion about their condition and its causes. The counselling clarified which factors they could control and which they couldn't, and reassured them that mental illness symptoms are not fixed or predetermined for everyone (Science Direct, 2019).

Subtheme 2 - Long-term Implications

Following response highlight theme Long-term Implications:

"Prevent transmission or reduce risk factors in the next generation" (P3).

From the collected data, participants reported that genetic counselling can reduce the transmission of genetic conditions to the next generation and bring long-term changes to an individual's life. It benefits both the current and future generations, demonstrating the long-term implications of genetic counselling.

Ausems M et al conducted study using Breast cancer patients, Seventy-seven first-time counselling attendees (44 of whom were affected) completed questionnaires on their risk understanding, knowledge, perceived control, and distress levels before, immediately after, and six months after counselling. Analysis revealed that affected individuals had stable cognition but experienced reduced anxiety and increased stress reactions immediately after counselling. Unaffected

individuals showed improved cognitions over time, except in knowledge, and similar distress patterns as affected individuals. Overall, counselling helps educate women about breast cancer risk and reduces anxiety, with these benefits appearing to be sustained over time (Science Direct, 2011).

Genetic counselling has long-term effects on both affected and unaffected individuals. For those who are affected, it can significantly reduce anxiety and psychological distress. For those who are unaffected, genetic counselling helps lower anticipated stress and motivates them to take proactive steps to prevent future diagnoses or distress. The benefits of counselling are long-lasting, although further research is needed to determine the exact duration of these effects. And also the effect of environment, social and psychological factors over the duration of its effect can be studied, and how such situations can be managed and addressed through Genetic Counselling, in short building resilience in patients with genetic disorders.

Subtheme 3 – Future Directions

Following response focus theme Future Directions:

"Benefit perceived by patient will depend on the level of awareness and understanding of available prevention techniques, underscoring the importance of providing insights and support in this regard" (P3).

When examining the future of genetic counselling in improving genetic outcomes, participants noted that increasing awareness about genetic counselling and available treatment options is crucial. This underscores the need to provide comprehensive insights not only to the public but also to professionals in the field. The lack of engagement from participants highlights the importance of addressing these gaps in knowledge and communication.

Blair Stevens, Assistant Professor and Director of Prenatal Genetic Counselling Services, University of Texas Health Center said that "Genomic medicine is becoming a key part of standard healthcare, including genetic screening during pregnancy, newborn screening, and cancer screening in adults. To effectively integrate genomic medicine into routine care, expertise from genetic specialists is crucial. This requires collaboration among stakeholders to make complex

genetic screenings accessible and actionable. Genetic counsellors play a vital role in educating policymakers, training future healthcare providers, researching diverse patient needs, and working with advocacy groups to ensure successful implementation"(National Society of Genetic Counsellors,2022).

4. Importance of conducting Genetic Counselling'?

Collected responses are as follows:

- "Like sex, sexual health genetic counselling is also an important area. People are not aware of such area even professionals too. Nowadays the cases are high so such departments and bodies need to be generated and sufficient help and guidelines should be given. Also need to convey what is marriage and importance of compliance between the partners people learn all these things from videos and films which is confabulated materials especially sex" **P1**
- "The importance of genetic counselling lies in raising awareness and providing psycho education. Without it, there's a risk of genetic information being misunderstood or misused, which is why we offer both pre and post genetic counselling sessions." **P6**
- My understanding is that due to genetic conditions, many issues related to physical and psychological well-being are becoming more prevalent today. Therefore, providing counselling before encountering these issues is crucial. For example, conditions range from cancer to neuropsychological disorders; if parents have mood disorders, there is a higher chance of autism in the next generation. Recently, I consulted a couple whose first child was diagnosed with an intellectual disability. When they planned for a second child, they consulted multiple doctors about potential genetic issues. The doctors reassured them it was okay to conceive, but the second child was later diagnosed with the same condition, highlighting the need for thorough genetic counselling. Such counselling can help manage lifestyle, conceive frequency, and necessary checks, making it essential for informed planning.

  P8

Through the "RQ4- Importance of conducting Genetic Counselling'? The researcher aims to investigate the role of genetic counselling in education and awareness-building for clients. Specifically, the study will focus on how genetic counselling contributes to reducing defective gene expression and its familial implications. Additionally, the research will explore the benefits of genetic counselling for society and the broader population for their health management. The major theme identified is Educational and Informative Role and sub themes are Genetic Risks, Genetic Literacy and Health Management.

Major Theme 1 - Educational and Informative Role

Following responses denote the theme Educational and Informative Role:

"Like sex, sexual health genetic counselling is also an important area. People are not aware of such area even professionals too" (P1).

"Convey what is marriage and importance of compliance between the partner's people learn all these things from videos and films which is confabulated materials especially sex" (P1).

"Recently, I consulted a couple whose first child was diagnosed with an intellectual disability. When they planned for a second child, they consulted multiple doctors about potential genetic issues. The doctors reassured them it was okay to conceive, but the second child was later diagnosed with the same condition, highlighting the need for thorough genetic counselling" (P8).

Professionals emphasize the need for increased awareness and education about genetic health, as many people are not well-informed in this area like in sexual health. Genetic counselling can help clarify the implications of marriage and partner compliance, especially when genetic issues arise within a family. Due to the current lack of education, there is a risk of misunderstanding genetic information. Genetic counselling can address these issues through proper psychoeducation. This support is valuable for families planning for children or conception. For instance, one respondent (P8) shared an experience where a family, after consulting a professional, was assured that their

next child would not inherit a genetic condition. Despite this reassurance, the child was later diagnosed with the same disorder, highlighting both the informative role of genetic counselling and the need for improved functioning in the field. The importance of genetic counselling also found in an article written by Aliouche H, Genetic counselling is vital throughout various life stages, including pregnancy planning, where it helps understand inherited conditions and assess risks related to infertility or miscarriage. During pregnancy, it identifies potential fetal issues through tests and evaluates risks from maternal health or environmental factors. For children showing signs of genetic disorders, such as developmental or sensory issues, genetic counselling provides crucial support. Additionally, it aids individuals with a family history of genetic diseases, like cancer or neuromuscular disorders, in managing their health risks effectively (News Medical Life Sciences, 2022).

As genomic medicine advances, there is an increasing need for genetic counselling and genetic counsellors (News Medical Life Sciences, 2022). Similar to mental and sexual health, Genetic health need to be studied. Genetic conditions and gene mutations can affect family stability and relationships, often due to stigma and lack of knowledge. Genetic counselling can provide crucial information about the implications of genetic results, support for partners, and strategies for building resilience within families. Rather than dismissing relationships in light of genetic findings, counselling can offer guidance on managing these situations constructively. As genomic medicine becomes more mainstream, the demand for genetic counselling is expected to rise, underscoring its essential role in improving health outcomes and family planning. As cited in Genetic Testing and Family Planning, Wagner A F wrote that Carrier screening is useful for individuals who know of genetic conditions in their family or want to understand their genetic background before starting a family. It helps prepare for the arrival of a child by identifying potential genetic risks. Notes that genetic counselling often provides reassurance, with the primary outcome being "peace of mind" for patients. The screenings and tests discussed during these counselling sessions are designed to aid in planning, offer reassurance, and support informed decision-making (Northwestern Medicine, 2019).

Subtheme 1 – Genetic risks

The following are the responses related to the subtheme Genetic Risks:

"For example, conditions range from cancer to neuropsychological disorders; if parents have mood disorders, there is a higher chance of autism in the next generation" (P8).

"Recently, I consulted a couple whose first child was diagnosed with an intellectual disability. When they planned for a second child, they consulted multiple doctors about potential genetic issues" (P8).

Identified genetic risks include neuropsychological disorders, cancer, mood disorders in parents, and neurodevelopmental conditions such as intellectual disability in children. These risks are relevant for genetic counselling, which can be used to address and manage these concerns.

Risk assessment is a crucial component of genetic testing and counselling, involving the accurate calculation of an individual's likelihood of carrying a specific genetic mutation or being affected by a genetic disorder. This assessment should use all available data, including genetic test results, family history, the ethnic background of parents, and mutation frequencies within the population. Risk evaluation is an ongoing process, requiring continual analysis and updates as new information becomes available (National Library of Medicine, 2005).

Genetic risk refers to the likelihood that an individual will be affected by genetic abnormalities. This risk can be influenced by inherited genetic variants, combinations of different genes, and environmental factors. Genetic mutations or environmental exposures that promote the development of a disorder can increase this risk. Therefore, awareness of these risk factors and making informed decisions are essential for managing genetic health effectively.

Hull S C stated that risk assessments can be complex and often lack clear-cut answers. Consulting with genetics professionals can provide individuals with a better understanding of what statistical genetic risk means for their personal situation (National Human Genome Research Institute, 2024).

Sub theme 2 – Genetic Literacy

The following responses pertain to the subtheme of genetic literacy:

"People learn all these things from videos and films which is confabulated materials" (P1)

"There's a risk of genetic information being misunderstood or misused" (P6)

"They consulted multiple doctors about potential genetic issues. The doctors reassured them it was okay to conceive" (P8)

Based on the responses, participants noted that people often gather information about genetic disorders and genetic health from videos, where information may be misleading or sensationalized to attract viewers, similar to trends seen in sexual health content which can lead to misunderstandings and misuse of genetic information due to a lack of genetic literacy. One participant (P8) recounted an experience where a client sought the help of a genetic counsellor due to stress related to planning for a child. This highlights the need for improving genetic literacy among the public through genetic counselling.

Gunter C et al defined Genetic Literacy as 'Genetic literacy refers to having enough knowledge of genetic principles to make informed decisions about personal well-being and participate effectively in genetic issues. It involves understanding basic genetic concepts and applying this knowledge to health decisions. Higher genetic literacy enhances communication between patients and providers, aids in making informed health choices, and fosters positive attitudes toward genetic testing and its benefits for family members' (Science Direct, 2022).

Genetic literacy can be crucial because it empowers individuals with the knowledge needed to understand genetic disorders, their development, and prevention strategies and with adequate genetic literacy, people can gather accurate information about genetic health, which helps reduce stigma and discrimination associated with genetic conditions. Genetic counselling can play a key role in this process by providing clear, comprehensive information about genetic health. Genetic Counsellors can assist help individuals and families make informed decisions, which can alleviate psychological and emotional burdens and contribute to overall well-being. By enhancing genetic

literacy, genetic counselling supports better management of genetic conditions and promotes a harmonious and informed approach to health.

Sub theme 3 - Health Management

The following responses relate to the subtheme of health management:

"Nowadays the cases are high so such departments and bodies need to be generated and sufficient help and guidelines should be given" (P1).

"The importance of genetic counselling lies in raising awareness and providing psycho education" (P6).

"Such counselling can help manage lifestyle, conceive frequency, and necessary checks, making it essential for informed planning" (P8).

Genetic counselling plays a significant role in health management, as evidenced by the responses provided. By offering advice, raising awareness, and delivering psychoeducation, genetic counselling supports effective health management. It assists in managing lifestyle choices, making informed decisions about conception, and planning families, ultimately contributing to better health outcomes. Similar findings can be found in an article titled 'Understanding Genetics: A District of Columbia Guide for Patients and Health Professionals', Genetic counsellors are integral members of the healthcare team, offering information and support to families affected by or at risk of genetic disorders. They identify at-risk families, analyze family histories and inheritance patterns, calculate recurrence risks, and provide guidance on genetic testing. Additionally, genetic counsellors help families understand genetic disorders within their cultural, personal, and familial contexts. They offer supportive counselling, advocate for patients, and refer individuals to other health professionals and support services. They also act as a key resource for information on genetic disorders for healthcare professionals, patients, and the public (National Library of Medicine, 2010).

Genetic counselling plays a multifaceted role in health management. It provides psychoeducation to help individuals and families understand genetic conditions, their implications, and the available

management options. This education helps mitigate the psychological and emotional burdens that often accompany genetic disorders by offering support and guidance tailored to the needs of the individual and their family. In addition, genetic counsellors assess risk factors by analyzing family histories and genetic information to determine the likelihood of developing or passing on genetic conditions. They recommend preventive measures, such as lifestyle changes or proactive health screenings, to manage or reduce these risks. Genetic counselling also takes into account the sociocultural contexts of individuals, acknowledging that cultural, personal, and familial factors influence how genetic information is perceived and managed. By integrating these aspects into the counselling process, genetic counsellors help individuals navigate their genetic conditions in a way that aligns with their unique circumstances. Overall, genetic counselling contributes to comprehensive health management by addressing not only the biological aspects of genetic disorders but also the psychological and sociocultural dimensions, ensuring a holistic approach to well-being.

5. How Genetic Counselling is being done in Kerala?

Following responses collected:

- “As per my knowledge I don’t know such practitioners, if it is there it would be minimal. In our settings the guidelines and remedies are given by infertility specialists and concerned doctors.” **P1**
- "When you inquire about the situation in Kerala, there isn't a proper system in place for genetic counsellor practice in hospitals. Generally, doctors provide brief counselling after diagnosis and treatment initiation, rather than beforehand. This delays early identification and prevention efforts. Moreover, public awareness about these issues is quite low. It's crucial to start working on prevention as early as possible. Currently, this field is evolving and might see significant development in the future." **P3**
- "In Kerala, there are very few genetic counsellors available. Those who have completed medical genetics or have a background in genetics and counselling often take on the role.

Besides them, it's quite rare. Many of our clients gather information from centers in Pune, and we provide counselling based on that." **P6**

- "Practicing in such a label like Genetic counsellor in my knowledge is rare, I don t know such professionals personally" **P4**
- Genetic counselling, unlike other areas such as family or marital counselling, is less practiced and not as vibrant, particularly in Kerala. It is not commonly explored. In hospital settings, however, it is conducted in a formal manner. In my view, understanding genetic risk factors and taking appropriate precautions is essential and beneficial.**P8**

Through the "RQ5- How Genetic Counselling is being done in Kerala? The researcher aims to investigate several key aspects of genetic counselling in Kerala. The focus is on understanding how genetic counselling is practiced within the state, including the methodologies and procedures used, the qualifications and backgrounds of genetic counsellors, the researcher will assess the guidelines and regulations governing the practice of genetic counselling in Kerala. This includes reviewing official standards, protocols, and best practices established by local health authorities and professional organizations. Understanding these guidelines will provide insight into the regulatory framework and quality control measures in place for genetic counselling services. And finally the acceptance and perception of genetic counselling among the population of Kerala. The major theme identified Practices and Implementation and sub themes are Processes, Protocols, and Cultural Sensitivity.

Major theme 1 - Practices and Implementation

The following responses address the subtheme of practices and implementation:

"As per my knowledge I don't know such practitioners, if it is there it would be minimal" (P1)

"Generally, doctors provide brief counselling after diagnosis and treatment initiation" (P3)

"In Kerala, there are very few genetic counsellors available" (P6)

"Practicing in such a label like Genetic counsellor in my knowledge is rare, I don t know such professionals personally" (P4)

"Unlike other areas such as family or marital counselling, is less practiced and not as vibrant, particularly in Kerala. It is not commonly explored" (P8)

Analyzing the participants' responses regarding the practices and implementation of genetic counselling in Kerala reveals a significant scarcity of genetic counselling services in the state. Two participants (P1 and P4) reported that they are unaware of any practicing genetic counsellors or organizations specializing in genetic counselling. Another participant (P8) noted that genetic counselling is both underutilized and insufficiently explored in Kerala. Additionally, P1 highlighted the rarity of practitioners and the minimal presence of genetic counselling services. These responses clearly indicate a shortage of genetic counsellors and a lack of widespread practice of genetic counselling in Kerala.

The Hindu Bureau (2023, November 26) Rajiv Gandhi Centre for Biotechnology to support identification of rare genetic disorders in kids. The Hindu, cited that The Rajiv Gandhi Centre for Biotechnology (RGCB) in Kerala is actively seeking participation from various segments of society through clinical support groups, family support groups, or volunteer groups to address the unique needs of children with rare genetic conditions. The RGCB will collaborate with district and taluk hospitals as well as family groups across Kerala to identify rare pediatric genetic diseases and provide free genetic testing, support, and counselling to affected families. On World Children's Day, November 20, RGCB will host an awareness program focused on pediatric rare genetic disorders. RGCB Director Chandrabhas Narayana emphasized that the center aims to uphold the rights of children to life, health, education, and family life, as outlined by World Children's Day. According to the Ministry of Health and Family Welfare, between 72 and 96 million people in India suffer from some form of rare condition. While there are approximately 7,000 known rare conditions, the ICMR's National Registry for Rare Diseases has reported only 4,000. It is noted that 80% of rare diseases are genetic, with 70% of these conditions beginning in childhood. (The Hindu, 2023). Though there is significant presence of genetic conditions in Kerala, the availability is questionable which can impact the quality of life of Kerala population.

Sub theme 1 – Processes

The responses below explore the subtheme of processes:

"In our settings the guidelines and remedies are given by infertility specialists and concerned doctors." P1

"Generally, doctors provide brief counselling after diagnosis and treatment initiation, rather than beforehand. This delays early identification and prevention efforts" (P3).

"In hospital settings, however, it is conducted in a formal manner" (P8).

When analyzing the responses regarding the practice of genetic counselling in Kerala, it is apparent that participants were unable to specify any particular criteria or standardized procedures for conducting genetic counselling. This observation suggests that there may be a significant gap in the established practices and systematic exploration of genetic counselling within the region. The absence of clearly defined guidelines or processes could be a result of limited implementation and development of genetic counselling services in Kerala. Consequently, this gap underscores the need for more comprehensive research and the establishment of standardized practices to enhance the effectiveness and consistency of genetic counselling in the area.

The actual process of Genetic Counselling is found in an article titled 'Process of Genetic Counselling and Testing', Before genetic testing, individuals will have a pre-test consultation with a genetic counsellor or medical oncologist to review their personal and family cancer history, assess the likelihood of a hereditary cause, discuss the benefits and limitations of genetic testing, and address implications for insurance, legal, and privacy issues. Consent for genetic testing is required and is a voluntary decision. A sample, usually blood but sometimes skin or saliva, will be collected for analysis, with results typically available in 2 to 6 weeks. A follow-up appointment will then discuss the test results, their implications for the individual and family, recommendations for managing cancer risk, and possible referrals to other specialists for further risk management. (National Cancer Centre Singapore, 2021)

The absence of standardized practices and procedures indicates a lack of thorough exploration in the field of genetic counselling. In many general hospital settings, genetic counselling is often reduced to merely conducting genetic tests and interpreting the results, which does not fully benefit the individuals involved. The process outlined by the National Cancer Centre Singapore

demonstrates that there are numerous other factors and considerations that must be addressed. Without a definitive model to guide practitioners, they may approach genetic disorders in a disorganized manner. This lack of a clear process and model can mislead both society and practitioners, potentially reinforcing social stigma and discrimination. Therefore, it is crucial to explore genetic counselling more deeply, develop a standardized practice model, and integrate it into the health system to improve the overall health of the population in Kerala.

Subtheme 2 - Protocols

The following responses examine the subtheme of protocols:

"Those who have completed medical genetics or have a background in genetics and counselling often take on the role" (P6).

"Generally, doctors provide brief counselling after diagnosis and treatment initiation" (P3)

Regarding the protocols for practicing genetic counselling, one participant (P6) reported that individuals with backgrounds in counselling and genetics typically assume this role. In contrast, another participant (P3) indicated that, generally, doctors provide only brief counselling after diagnosis and the initiation of treatment. This variation in practice across different settings highlights the lack of clear protocols and guidelines for genetic counselling.

Here in addition to the lack of processes of practicing genetic counselling, there is lack of protocols of practicing and training of professionals. The lack of such protocols can make adverse issues where unauthorized practicing and practitioners will expand. Since it is genetic information, which a sensitive and confidential matter, the lack of protocols can affect the privacy and protection of individuals and families. The testing can misused like prenatal sex determination and foeticides. Absence of protocols and rules can pave way to malfunctions and malpractices where the quality of service to the population is compromised.so there is need of proper protocols and training and guidelines to practice this genetic counselling.

National Society of Genetic Counsellors (NSGC) introduced a code of ethics that attempts to clarify and guide the conduct of a professional so that the goals and values of the profession are best served. The NSGC Code of Ethics is based upon the distinct relationships genetic counsellors have with 1) themselves, 2) their clients, 3) their

colleagues, and 4) society. The section 1 implies Genetic counsellors prioritize professionalism, competence, and integrity. They strive to obtain accurate information, stay updated with guidelines and standards, work within their expertise, and represent their credentials truthfully. They follow conflict of interest guidelines, disclose any potential biases, and avoid using their position for personal gain. Additionally, they are responsible for maintaining their physical and emotional health to ensure effective professional performance. Section 2 implies, the counsellor-client relationship in genetic counselling is founded on care and respect for the client's autonomy and individuality. Genetic counsellors are committed to providing unbiased services, referring clients to other professionals when necessary, and ensuring clients make informed decisions without coercion. They respect clients' diverse backgrounds and beliefs, maintain privacy and security of confidential information, and avoid exploiting clients for personal or professional gain. Section 3 implies, Genetic counsellors maintain professional relationships with respect, collaboration, and support. They mentor and guide colleagues and trainees, value others' contributions, encourage ethical behavior, and ensure that responsibilities match individuals' expertise. They uphold appropriate boundaries to prevent exploitation, take responsibility only for their own work, and properly acknowledge others' contributions. Additionally, they inform employers about ethical obligations according to the NSGC Code of Ethics. Section 4 implies, Genetic counsellors engage with society to enhance well-being and access to genetic services, guided by principles of honesty, objectivity, and integrity. They advocate against genetic discrimination, provide reliable information to key stakeholders, and educate the public on genetic advancements and their societal impacts. They also promote ethical research practices and work towards legal reforms that align with professional principles for the public good (NSGC Code of ethics, National Society of Genetic Counsellors).

6. Is there any barriers for practicing it?

Following responses collected as below:

- “Lack of knowledge and awareness is a barrier I think, and I don’t know the costs and charges of such services. Initially the lack of awareness need to be alleviated then only something can be done.”**P1**
- “It is quite expensive, since we need more results to confirm the results and inheritance .So the money and time is a major barrier. And the people’s tendency to hide information which reduce the reliability of the information.” **P3**
- "The main issue is the lack of awareness, which creates a significant gap between practitioners and the general public. There's a shortage of genetic counsellors, and those practicing often lack sufficient training and face payment challenges. Financial barriers exist for people too, as there are no insurance schemes to cover these services. Additionally, there's a lack of technical equipment and persistent superstitions surrounding genetic counselling.” **P7**
- "I see a barrier in whether the practitioner is qualified to provide genetic counselling services. In India, there's a licensing board called the 'Board of Genetic Counselling,' but many practitioners lack this licensing. Currently, those who do provide counselling often have a background in counselling and a master's degree in genetics. Even though I have a master's in medical genetics and psychology, which allows me to provide these services, this discrepancy is a significant drawback." **P6**
- “In our psychology curriculum, we cover physiology, but genetics is not extensively studied. Updating or enhancing the curriculum to include more genetics could be beneficial. Limited knowledge in this area might be a barrier.” **P4**
- A lack of professionalism and knowledge among providers can be a significant barrier. To offer accurate information about the likelihood of developing various disorders, a thorough understanding of these conditions is essential. Even when information is provided, it may be perceived in different ways; for example, cancer-related information might elicit fearful responses, so this area must be handled diplomatically. Additionally, before proceeding with tests, one must consider

expenses and the availability of technology and equipment. If a diagnosis is made, it is crucial to provide grievance counselling to both the patient and their caregivers. **P8**

Through the "RQ6- 6. Is there any barriers for practicing it? The researcher wants to explore the barriers of practicing the genetic counselling, the barriers of accessing the service and providing the services and related factors. The major theme identified is Access Barriers and sub themes are Affordability, Awareness, Stigma, Emotional Distress and Lack of Integrated Services.

Major theme 1 - Access Barriers

The responses below delve into the central theme of access barriers:

"Lack of knowledge and awareness is a barrier I think" (P1)

"It is quite expensive, since we need more results to confirm the results and inheritance .So the money and time is a major barrier" (P3)

"There's a shortage of genetic counsellors, and those practicing often lack sufficient training" (P7)

"Whether the practitioner is qualified to provide genetic counselling services. In India, there's a licensing board called the 'Board of Genetic Counselling,' but many practitioners lack this licensing" (P6)

"In our psychology curriculum, we cover physiology, but genetics is not extensively studied" (P4)

"Information might elicit fearful responses" (P8)

In evaluating the barriers to accessing genetic counselling services, two participants reported concerns about the qualifications and proficiency of practitioners in delivering genetic counselling and understanding genetic information. One participant (P6) noted that while there is a "Board of Genetic Counselling" meant to authorize practice, many practitioners lack proper accreditation. Additionally, limited awareness of available genetic counselling services and inadequate education

in genetics further restrict the effectiveness and reach of these services. And the fear and anxiety resulting the process of genetic tests will hinder people from seeking help.

Similar result found in study conducted by Bulsara et al in 2018, titled 'Investigating barriers to genetic counselling and germline mutation testing in women with suspected hereditary breast and ovarian cancer syndrome and Lynch syndrome', identified the major barrier themes as Lack of importance; Level of information received; Timing of referral processes; Fear and anxiety; Resistance to and perceptions of counselling (Science Direct, 2018).

The practice of genetic counselling faces several significant barriers. Firstly, the limited availability of genetic counsellors is a major concern, which is often linked to a lack of awareness and exploration in the field. This shortage can deter potential professionals from entering the field, perpetuating its underdevelopment and limiting the benefits to individuals who need these services. Additionally, the cost and time involved in genetic counselling can be prohibitive, further restricting access to these essential services. Another substantial barrier is the fear associated with genetic testing and potential diagnoses, which can be influenced by social stigma, family pressure, and concerns about potential disruptions to one's life. This fear often leads individuals to avoid seeking counselling or undergoing testing. To address these challenges, efforts must focus on increasing awareness of genetic counselling, reducing associated costs and time commitments, and providing support to alleviate fear and stigma surrounding genetic conditions.

Bradbury A R et al (2017) conducted a study to identify the barriers in practicing genetic counselling titled 'Possible barriers for genetic counsellors returning actionable genetic research results across state lines' the study identified barriers as there is an ethical obligation to return actionable genetic results to participants, but there is currently no legal requirement to do so. This uncertainty complicates the informed consent process and the implementation of these ethical obligations, shortage of genetic counsellors, and their services can be costly, State Licensure Variability and Costs and Resource Limitation (Genetics in Medicine, 2017).

Subtheme 1 – Affordability

The following responses explore the theme of affordability:

"It is quite expensive, since we need more results to confirm the results and inheritance .So the money and time is a major barrier" (P3).

"Financial barriers exist for people too, as there are no insurance schemes to cover these services" (P7).

"Additionally, before proceeding with tests, one must consider expenses" (P8)

The affordability of genetic counselling is a major concern due to its high costs. The process often requires additional tests to confirm results and inheritance patterns, which can be expensive and time-consuming. Additionally, the lack of insurance coverage for genetic counselling services exacerbates the financial burden on individuals. This situation necessitates careful consideration of expenses before proceeding with tests, making the overall cost a significant barrier to accessing these essential services. This finding is consistent with the study outcome of research done by Jackson and colleagues (2011) titled 'Barriers in Identification and Referral to Genetic Counselling for Familial Cancer Risk: The Perspective of Genetic Service Providers' ,concluded that barriers to seeking genetic counselling after referral included risk evaluation viewed as a non-priority, concerns about impact, distance to appointments , lack of insurance , lack of patient/provider knowledge about the value of genetic counselling , discouragement by family members and fear (ResearchGate,2011).

The affordability of genetic testing and counselling presents a significant barrier. Genetic tests are both time-consuming and costly; for example, at DNA Labs India, the cost of a genetic counselling test is 1,000 INR (DNA Labs India). Additionally, participants have reported that such services are rarely available in government healthcare sectors. The absence of health insurance coverage for genetic counselling and tests further deters individuals from seeking these services, as the lack of financial support increases the overall expense.

Subtheme 2 – Awareness

The following responses investigate the theme of awareness:

"Lack of knowledge and awareness is a barrier I think" (P1)

"The main issue is the lack of awareness, which creates a significant gap between practitioners and the general public" (P7)

A significant barrier to the practice of genetic counselling is the lack of knowledge and awareness surrounding the field. This deficiency in awareness creates a notable gap between practitioners

and the general public, making it challenging for individuals to understand the benefits and availability of genetic counselling services. The limited understanding of genetic counselling among both potential clients and the broader community contributes to underutilization of these services and hinders the effective practice of genetic counselling.

A study titled 'Knowledge, Attitude, and Practice About the Process of Genetic Counselling Among Clinicians' conducted by Arumugham S et al (2023) concluded that, As genetic testing technology advances, the demand for genetic counselling in healthcare is growing. It found that while most clinicians recognize the importance of gathering a detailed family history and referring patients for genetic testing, there are notable gaps in awareness and practice. Many clinicians agree on the need to educate patients about genetic conditions and the potential psychological impact on families. However, some still lack awareness of genetic testing's benefits and ethical considerations, leading to inconsistent practices. The study also highlighted that awareness and attitudes towards genetic counselling vary among clinicians based on their specialty and experience. Overall, improving knowledge about genetic testing and counselling is crucial for better integration into healthcare practices and for addressing the rising burden of genetic disorders (National Library of Medicine, 2023).

The current level of awareness among practitioners and the general population regarding genetic counselling is insufficient. Without increased awareness, this field will remain underexplored. To advance genetic health awareness, similar to mental health and sexual health initiatives, grassroots-level efforts are essential. Implementing awareness campaigns through both virtual and traditional media can effectively educate the public. Additionally, establishing genetic counselling services within healthcare settings is crucial for improving public health protection, especially in the era of genomic mutations. Enhanced awareness and accessible counselling services are vital for addressing genetic health challenges effectively.

Subtheme 3 – Stigma

The following responses address the theme of stigma:

"And the people's tendency to hide information which reduce the reliability of the information." (P3)

"Persistent superstitions surrounding genetic counselling." (P7)

Stigma related to genetic counselling often stems from individuals' reluctance to share sensitive information, which undermines the reliability of genetic data. Additionally, persistent superstitions and cultural beliefs surrounding genetic counselling contribute to this stigma, further discouraging people from seeking or disclosing relevant genetic information. These factors collectively hinder the effectiveness and acceptance of genetic counselling services. Stigma is characterized as a deeply discrediting attribute or mark that leads to social devaluation. This stigma can arise from the mark itself or from social interactions where the mark is perceived as indicative of the individual's flawed characteristics (Earnshaw and Chaudoir, 2009). Perceived stigma involves a person's understanding of how others might react to or view them based on a particular trait or identity (Zelaya et al., 2012). Anticipated stigma refers to the expectation of future stigmatizing experiences (Earnshaw et al., 2013). Internalized stigma describes the process by which individuals become aware of, accept, and apply stigma to themselves (Munoz et al., 2011). Experienced or enacted stigma involves actual discriminatory actions or behaviors (Catona et al., 2016). The presence of stigma can negatively affect health outcomes by discouraging individuals from seeking help or a definitive diagnosis, impacting patients across various diseases (Kane et al., 2019).

Baynam G et al (2024) conducted study titled 'Stigma associated with genetic testing for rare diseases—causes and recommendations, and found stigma related to genetic counselling manifests in various forms, significantly affecting individuals with rare and genetic disorders. People with genetic conditions often face stigma through behaviors such as labeling, stereotyping, and status loss, which leads to a diminished social identity (Williams et al., 2010). This stigma influences all stages of the diagnostic and treatment journey, from pre-diagnosis to accessing care and adhering to treatment, negatively impacting the quality of life for patients and their families. Stigmatization can extend to genetic carriers and discourage practices like cascade screening (Kruse et al., 2021). Cultural and societal factors further exacerbate stigma, contributing to gender-based discrimination, reproductive restrictions, and fears of social exclusion (Kruse et al., 2021; Boardman et al., 2020). These issues can also cause distress among patients and their families, as they may experience isolation, shame, and a lack of support, affecting their overall well-being (Ayres et al., 2019; Bogart et al., 2022). Socioeconomic and cultural beliefs often lead to reluctance in seeking diagnoses or treatments, perpetuating stigma and limiting access to genetic counselling and support (Chediak et al., 2022).

Subtheme 4 – Emotional Distress

The upcoming responses explore the theme of emotional distress:

"Cancer-related information might elicit fearful responses, so this area must be handled diplomatically" (P8)

One respondent (P8) noted that genetic information, such as details about cancer risk, can evoke fear in clients and thus needs to be handled with care. This fear and the associated emotional distress can act as a barrier to effective communication and decision-making.

Davies L and colleagues identified eight emotional effects of genetic disorders in their study titled 'the emotional effects of genetic diseases: implications for clinical genetics', the emotions are anxiety, worry about risks to children, guilt, anger, uncertainty, sadness and grief, depression, and redemptive adjustment. Two factors were identified that could modify the emotional effects; these were variability of genetic diseases, and lack of diagnosis/inappropriate care (National Library of Medicine, 2007).The established study findings are congruent with the current study findings.
The nature and impact of genetic disorders can cause significant emotional distress for individuals and families. Concerns about hereditary conditions or susceptibility to genetic anomalies often lead to fear and anxiety regarding genetic testing. Stigma surrounding genetic abnormalities can further exacerbate distress, as can discrimination and inefficiencies in managing the condition. These factors contribute to uncertainty about the future and may disrupt an individual's working, mental, and physical capacities. Genetic disorders can also strain family relationships and lead to conflicts over care decisions. Families may experience grief, especially if the condition affects a child or close relative, and may also feel guilt or shame about passing the condition to future generations. Additionally, the presence of a genetic condition can complicate decision-making processes, requiring careful consideration of various factors.

Subtheme 5 - Lack of Integrated Services

The forthcoming responses address the theme of insufficiently integrated services:

"Additionally, there's a lack of technical equipment" (P7).

"Lack of awareness, which creates a significant gap between practitioners and the general public" (P7).

"If a diagnosis is made, it is crucial to provide grievance counselling to both the patient and their caregivers" (P8).

A significant barrier in genetic counselling is the lack of integrated services. According to respondent P7, there is a notable deficiency in technical equipment, which hampers the effectiveness of genetic counselling. Furthermore, P7 highlights a critical gap in awareness that separates practitioners from the general public, complicating the delivery of care. Additionally, P8 emphasizes the importance of providing grief counselling to both patients and their caregivers following a diagnosis, underscoring that the absence of such support services can further exacerbate the challenges faced by those affected by genetic conditions.

As cited in Permanente Medicine, Kaiser Permanente (2023) shares their experience and the need for an integrative approach to genetic counselling. When patients face potential genetic diagnoses for serious conditions such as breast or ovarian cancer, it is crucial to have accurate information, clear communication, and strong support. Kaiser Permanente's integrated care model allows genetic counsellors and physician geneticists to collaborate closely, streamlining care by reducing delays caused by administrative hurdles like pre-authorizations and outside referrals. This teamwork contributes to the greater efficiency of genetic counselling at Kaiser Permanente compared to other health care organizations.

Dr. Pim Suwannarat, a clinical geneticist and regional medical director for genetics with the Mid-Atlantic Permanente Medical Group, highlights the essential role of genetic counsellors. They not only explain what genetic testing entails and identify the appropriate tests but also interpret and communicate the results—whether negative, positive, or uncertain—in a meaningful way for both patients and their families. Physicians and genetic counsellors also exchange evidence-based information and best practices with colleagues outside their immediate teams. Dr. Suwannarat notes that through community practice groups, they brainstorm new strategies and maintain consistent approaches to testing and treatment. This collaborative network allows for efficient communication across different regions, facilitating patient care even when patients and their families are geographically separated.

Genetic counsellors at Kaiser Permanente are also involved in clinical research, academic publishing, and presenting at national and international conferences. Some work within the organization's molecular genetics laboratories, guiding other health care providers on genetic tests

and laboratory choices. The expertise and compassionate care provided by genetic counsellors significantly impact the quality of patient care, as evidenced by their high satisfaction ratings. Dr. Suwannarat emphasizes that the goal is to ensure patients receive not only testing information but also guidance on how to use their results, share them with their families, and understand their implications for personal care. The annual Genetic Counsellor Awareness Day acknowledges their valuable contributions to Permanente Medicine (Permanente Medicine, 2023).

Inconsistent and fragmented services can create significant barriers in genetic counselling, which deals with sensitive and life-changing issues that require careful management. The lack of integration often results in a burdensome process for patients or clients, potentially diminishing the benefits of genetic counselling. Addressing client needs through integrated services could improve outcomes; for instance, integrating vocational training for individuals with genetic conditions like dwarfism (WebMD, 2021) could be beneficial. Similarly, providing family therapy services for families dealing with genetic disorders can enhance the overall process. Integrating various services under a unified approach can also reduce expenses by avoiding redundant tests and treatments, ultimately ensuring more effective and comprehensive care for patients.

7. What strategies can be implement to promote and expand the Genetic Counselling?

   Responses collected as follows:

   - “First awareness need to be bring, people should know this filed and related studies and research can also help to expand it. This can bring acceleration in hospitals and medical health care systems to begin such services.” **P1**
   - "Having clear criteria for credentials would be beneficial, and it would also be helpful to know if there's already a specific approach in place for practitioners. I don’t know if it is already present. If we can empower it then this field can be bring to prominence" **P7**
   - “In the future, we need grassroots-level interventions similar to raising awareness about mental health stigma. This could include establishing specialized units and clinics dedicated to genetic counselling services. It's crucial to address these issues and advocate for policy changes at the government level." **P3**

- "In Kerala, genetic counselling services are extremely rare. There are no government positions available for this. To expand these services, creating such positions in hospitals and clinics would be beneficial. We need to integrate genetic counselling into society more broadly. Currently, unauthorized practitioners and even doctors and radiologists handle it, which dilutes the impact of therapy. Proper legalization and creating opportunities will help in expanding this field." **P6**
- To advance this field, we need to establish a dedicated specialization and invest in its study. Encouraging students to pursue education and research in this area is crucial. By fostering more research, the field will become more prominent and gain wider recognition. **P4**
- Based on my understanding, we are in an era where numerous mutations are occurring, even with common conditions like fever. As a result, the prevalence of genetic conditions is likely to increase in the coming years. It is essential for individuals to be aware of these conditions from the prenatal period through adulthood, particularly concerning cancer. Providing psycho education across various settings and recognizing genetic counselling as a distinct specialization, similar to other therapies and counselling, will be beneficial. Integrating this focus into academic programs would also be advantageous. **P8**

Through the "RQ7- What strategies can be implement to promote and expand the Genetic Counselling?" The researcher aims to identify and evaluate strategies for enhancing the promotion and expansion of genetic counselling services so as to improve accessibility, integration, and effectiveness of genetic counselling, ensuring that more individuals benefit from these services. The major theme identified is Strategies and sub themes are Legislative Support and Public Education.

Major theme 1 – Strategies

The upcoming responses will focus on the theme of strategies:

"Related studies and research can also help to expand it" (P1).

"We need grassroots-level interventions similar to raising awareness about mental health stigma" (P3).

"Establishing specialized units and clinics dedicated to genetic counselling services" (P3).

"Encouraging students to pursue education and research in this area is crucial. By fostering more research, the field will become more prominent and gain wider recognition" (P4).

"Integrating this focus into academic programs would also be advantageous" (P8).

Participants advocated that strategies to enhance the practicing of genetic counselling can include expanding genetic counselling services can benefit from insights gained from related studies and research. Grassroots-level interventions, akin to those used to address mental health stigma, are needed to raise awareness about genetic counselling. Establishing specialized units and clinics dedicated to genetic counselling would improve service delivery. Encouraging students to pursue education and research in genetics is crucial for advancing the field and increasing its recognition. Additionally, incorporating genetic counselling into academic programs would further support its development and integration.

Kaariainen H (2010) studied 'How to Improve Quality of Genetic Counselling?' and found that to enhance genetic counselling, several key strategies must be adopted. First, it is crucial to safeguard ample resources for genetic centers, ensuring they have the necessary funding, facilities, and staff to deliver comprehensive services. Additionally, developing tools to evaluate the quality of genetic services is essential for maintaining high standards and improving effectiveness. At the same time, all healthcare professionals should receive appropriate training in genetics and genetic testing. This training will enable them to effectively inform patients about genetic issues and make well-informed referrals to specialized genetic counselling when needed. By implementing these strategies, the accessibility and quality of genetic counselling can be significantly improved (ResearchGate, 2010).

To effectively promote the practice of genetic counselling, several strategies should be employed. First, increasing public awareness through targeted campaigns can educate individuals about the benefits and availability of genetic counselling, similar to successful mental health initiatives (Guttmacher & Collins, 2002). Expanding services by establishing specialized clinics and integrating genetic counselling into broader healthcare settings can enhance accessibility and

service delivery (Miller et al., 2018). Additionally, incorporating genetic counselling into medical and academic curricula ensures that future healthcare professionals are well-prepared in genetics (Reilly et al., 2017). Supporting research in the field is crucial for advancing practices and addressing emerging needs (Burke et al., 2015). Finally, promoting career development and providing support for students and professionals in genetics can help build a robust workforce to meet growing demands (Knebel et al., 2021). Implementing these strategies collectively can significantly enhance the reach and effectiveness of genetic counselling services.

Subtheme 1 - Legislative Support

The upcoming responses will concentrate on the theme of legislative support:

"Having clear criteria for credentials would be beneficial" (P7)

"Advocate for policy changes at the government level" (P3)

"Proper legalization and creating opportunities will help in expanding this field" (P6)

From the above responses, it is evident that the field of genetic counselling suffers from a lack of clear legislation. This includes deficiencies in credentialing standards, established policies, and opportunities for growth and development.

The study by Suter S (2020) titled 'Legal Challenges in Genetics, Including Duty to Warn and Genetic Discrimination', concluded that Legal issues in genetic counselling remain largely unresolved and can vary depending on the jurisdiction, largely because the law often lags behind rapid technological advancements. This delay creates uncertainty in how courts will handle emerging dilemmas related to genetic counselling. Additionally, the norms and practices within the genetic counselling community can influence legal perspectives on these issues. Consequently, it is crucial for genetic counsellors to stay actively involved in ongoing discussions and debates about the ethical and legal complexities associated with genetics and genomics to help shape and guide legal approaches (National Library of Medicine, 2020).

To enhance the field of genetic counselling, proper legal support is crucial. Providing high-quality education and training to practitioners can improve service standards and address fragmentation. Promoting research and creating government job opportunities can attract students to the field, benefiting individuals and society. Addressing unauthorized practices due to inadequate

credentials is essential to prevent exploitation, which can lead to emotional distress, worsening of conditions, and financial loss. Accreditation for current genetic counsellors and government support can improve their status and effectiveness. Additionally, establishing new policies and schemes to protect individuals with genetic disorders, along with programs that support career and personal development, can positively impact their daily lives and decision-making.

Subtheme 2 - Public Education

The upcoming responses will center on the theme of public education:

"First awareness need to be bring, people should know this field" (P1).

"In the future, we need grassroots-level interventions similar to raising awareness about mental health stigma" (P3).

"Providing psycho education across various settings and recognizing genetic counselling as a distinct specialization" (P8).

To effectively improve public education about genetic counselling, it is essential to first raise awareness so that people understand the field. This involves grassroots-level interventions, akin to those used to address mental health stigma, to educate the public about the significance and benefits of genetic counselling. Providing psychoeducation in diverse settings and acknowledging genetic counselling as a distinct and specialized field will further enhance public understanding and acceptance. Khatak S and colleagues (2020) conducted a study titled 'Public perception of genetic counselling in India: Opening mind Eyes', the survey conducted through a simple online platform served both as a survey and an awareness program. It highlights how India is evolving by turning challenges into opportunities and moving towards greater self-reliance and self-sufficiency. The demonetization event on November 8, 2016, transformed India's cash-based transactions into a digital system, leading to improved citizenship and adaptation to new constraints. The survey reached 400 respondents, demonstrating that the concept of genetic counselling has begun to gain traction. Notably, 57% of respondents learned about the necessity of genetic counselling through the survey itself. Additionally, 55% of respondents identified ethical issues, lack of education, and ignorance as significant barriers to advancing medical approaches in India. Furthermore, 57% strongly supported the creation of a social online platform to facilitate information sharing and access to help. Overall, the survey successfully raised

awareness about genetic counselling and emphasized the importance of embracing technological advancements for a better future.

Increasing awareness and public education about genetic counselling could be beneficial. Instead of enduring fear, anxiety, shame, and guilt, individuals can consider make informed decisions and seek appropriate treatment. Similar to recent mental health awareness efforts, grassroots-level interventions are needed to advance the field of genetic counselling and maximize its impact. Government initiatives such as campaigns in schools, hospitals, and health centers, as well as offering free treatments and services, can engage the public. Additionally, leveraging virtual media and implementing innovative schemes and programs dedicated to public education can further enhance outreach and effectiveness.

CHAPTER V

SUMMARY & CONCLUSIONS

5.1 Findings

Research Question 1: Understanding of the Genetic Counselling

- Genetic counselling is a specialized field within counselling that provides comprehensive guidance and psychoeducation related to genetic conditions. This process involves educating individuals and families about the nature of genetic disorders, including how these conditions arise, their potential implications, and the available options for management and treatment.
- Genetic counselling is typically conducted by professionals who possess expertise in both counselling techniques and genetic sciences.
- Genetic counselling is generally provided to individuals with genetic conditions, families with a member diagnosed with a genetic condition, or those at risk of such conditions.
- For families affected by or predisposed to genetic conditions, genetic counselling plays a crucial role in guiding them to take appropriate actions.
- It helps clarify genetic concerns, providing both emotional and professional support.
- This service aims to help families or individuals take preventative measures, mentally prepare for potential outcomes, and make informed decisions.
- Genetic counselling involves discussing the causes and available treatment options, helping individuals or families prepare mentally and take necessary precautions.
- Pre-genetic counselling involves the evaluation of the development and probability of genetic conditions, while post-genetic counselling focuses on educating clients about their specific conditions and strategies for prevention.
- A thorough understanding of genetic counselling requires a deeper knowledge of both genetics and counselling.
- Some participants (n=2) indicated limited knowledge in this area.

Research Question 2: Concerns of Family with Genetic Condition

- Families facing genetic conditions often have a range of concerns primarily focused on health due to either a family history of such conditions or the likelihood of inheriting them.
- Concerns extend to the potential risk of passing the condition to future generations, implications for quality of life, and potential challenges in job opportunities or perceived underemployment due to perceived inefficiencies.
- Genetic disorders can impact an individual or a family emotionally and psychologically, and those who don't expect such conditions find it hard to handle such confirmations.
- Families formed through consanguineous marriages or among older couples may anticipate these risks, but the confirmation of a genetic condition can still result in significant distress and emotional upheaval.
- The condition can affect an individual in many ways like career, job, and family and so on.
- Not only the patients but also the caregivers face burdens, as their roles are often impaired by these conditions, turning care into a significant responsibility since some conditions can limit the functioning of an individual.
- Spouses often blame each other, attributing the genetic disorders to one's impotency. These conditions can disrupt the harmony of family life, impacting both the spousal relationship and the broader family system.
- There is a social stigma associated with genetic disorders similar to that of mental illness, where individuals feel compelled to hide these conditions and are reluctant to talk about them in social settings.
- People often fear revealing such information due to potential discrimination and various negative labels. This stigma, coupled with the psychological strain, can further exacerbate social difficulties for affected individuals.
- There is a tendency for discrimination based on perceived inefficiency compared to others, further exacerbating the challenges faced by those with genetic disorders.
- The cost of genetic tests is huge; all families couldn't afford it. This financial strain often leaves families struggling or feeling helpless when dealing with rare genetic conditions, highlighting a broader issue of accessibility and support for those affected.

- The high cost and economic burden associated with treatment can significantly impact families, potentially affecting their willingness and ability to seek and adhere to treatment.
- There is a prevalent myth that genetic conditions are the result of conscious choices, reflecting a lack of understanding. To address this, it is essential to integrate genetic counselling and testing into government healthcare services at affordable prices.
- Decision-making is a critical aspect of daily life, as it often involves ensuring our well-being and minimizing harm to our surroundings. In a family setting, decisions can have profound and life-altering effects, often leading to significant ambiguity and anxiety. Therefore, guiding families towards more productive and informed decisions is essential. Genetic counselling can play a pivotal role in this process.

Research Question 3 – Role of genetic counselling to improve the gene pool.

- The role of genetic counselling in directly altering one's gene pool is not entirely clear, but its value lies primarily in prevention and informed decision-making.
- While genetic counselling itself doesn't change the genetic code, it helps individuals understand how lifestyle and environmental factors can influence gene expression.
- With insights gained from genetic counselling, individuals can make more informed decisions about their health.
- By making informed choices, such as improving diet, increasing physical activity, or avoiding known risk factors, individuals can potentially control or mitigate the impact of genetic predispositions.
- By enabling proactive management and preventive strategies, genetic counselling can reduce the risk of developing certain conditions or manage them more effectively if they arise. This can lead to a reduction in health-related suffering and improve quality of life.
- Genetic counselling can reduce the transmission of genetic conditions to the next generation and bring long-term changes to an individual's life. It benefits both the current and future generations.
- Genetic counselling has long-term effects on both affected and unaffected individuals. For those who are affected, it can significantly reduce anxiety and psychological distress. For

those who are unaffected, genetic counselling helps lower anticipated stress and motivates them to take proactive steps to prevent future diagnoses or distress.

- The benefits of counselling are long-lasting, although further research is needed to determine the exact duration of these effects.
- The future of genetic counselling in improving genetic outcomes depends on increasing awareness about genetic counselling and available treatment options.

Research Question 4: Importance of conducting Genetic Counselling

- Genetic counselling can provide crucial information about the implications of genetic results, support for partners, and strategies for building resilience within families.
- Rather than dismissing relationships in light of genetic findings, counselling can offer guidance on managing these situations constructively.
- Genetic Counsellors can assist individuals and families make informed decisions, which can alleviate psychological and emotional burdens and contribute to overall well-being.
- By offering advice, raising awareness, and delivering psychoeducation, genetic counselling supports effective health management. It assists in managing lifestyle choices, making informed decisions about conception, and planning families, ultimately contributing to better health outcomes.
- Genetic counselling also takes into account the sociocultural contexts of individuals, acknowledging that cultural, personal, and familial factors influence how genetic information is perceived and managed. By integrating these aspects into the counselling process, genetic counsellors help individuals navigate their genetic conditions in a way that aligns with their unique circumstances.
- Risk assessment is a crucial component of genetic testing and counselling, involving the accurate calculation of an individual's likelihood of carrying a specific genetic mutation or being affected by a genetic disorder.
- Awareness of these risk factors and making informed decisions are essential for managing genetic health effectively.
- Proper psychoeducation. This support is valuable for families planning for children or conception.

- Gather information about genetic disorders and genetic health from videos, where information may be misleading or sensationalized to attract viewers, similar to trends seen in sexual health content which can lead to misunderstandings and misuse of genetic information due to a lack of genetic literacy.

Research Question 5: Genetic Counselling in Kerala

- There is significant scarcity of genetic counselling services in the state.
- There is a significant gap in the established practices and systematic exploration of genetic counselling within the region.
- The absence of clearly defined guidelines or processes could be a result of limited implementation and development of genetic counselling services in Kerala.
- In many general hospital settings, genetic counselling is often reduced to merely conducting genetic tests and interpreting the results, which does not fully benefit the individuals involved.
- This variation in practice across different settings highlights the lack of clear protocols and guidelines for genetic counselling.
- Since it is genetic information, a sensitive and confidential matter, the lack of protocols can affect the privacy and protection of individuals and families.
- This lack of a clear process and model can mislead both society and practitioners, potentially reinforcing social stigma and discrimination.
- The National Society of Genetic Counsellors (NSGC) introduced a code of ethics that attempts to clarify and guide the conduct of a professional so that the goals and values of the profession are best served.
- The absence of protocols and rules can pave the way for malfunctions and malpractices, compromising the quality of service to the population.
- Therefore, it is crucial to explore genetic counselling more deeply, develop a standardized practice model, and integrate it into the health system to improve the overall health of the population in Kerala.
- There is a need for proper protocols, training, and guidelines to practice genetic counselling effectively.

Research Question 6: Barriers for Practicing Genetic Counselling

- There is a significant barrier to the practice of genetic counselling due to the lack of knowledge and awareness surrounding the field. This deficiency creates a notable gap between practitioners and the general public, making it challenging for individuals to understand the benefits and availability of genetic counselling services. This lack of awareness contributes to the underutilization of these services and hinders their effective practice.

- The qualifications and proficiency of practitioners in delivering genetic counselling and understanding genetic information are often inadequate. Although there is a "Board of Genetic Counselling" meant to authorize practice, many practitioners lack proper accreditation, affecting the quality of care provided.

- The limited availability of genetic counsellors is a major concern. This shortage can deter potential professionals from entering the field, perpetuating its underdevelopment and limiting the benefits to individuals who need these services. Additionally, the cost and time involved in genetic counselling can be prohibitive, further restricting access to these essential services. The lack of insurance coverage for genetic counselling services exacerbates the financial burden on individuals.

- Fear and anxiety resulting from the genetic testing process can hinder people from seeking help. Concerns about hereditary conditions or susceptibility to genetic anomalies often lead to fear and anxiety, influenced by social stigma, family pressure, and worries about potential disruptions to one’s life. Stigma related to genetic counselling, stemming from reluctance to share sensitive information and persistent cultural beliefs, further discourages individuals from seeking or disclosing relevant genetic information. This fear and associated emotional distress can act as barriers to effective communication and decision-making.

- Inconsistent and fragmented services create significant barriers in genetic counselling, which deals with sensitive and life-changing issues requiring careful management. The process often necessitates additional tests to confirm results and inheritance patterns, which can be expensive and time-consuming. Addressing client needs through integrated services could improve outcomes; for example, integrating vocational training for individuals with genetic conditions and providing family therapy services for families dealing with genetic disorders could enhance the overall process. Such integration could also reduce expenses by avoiding redundant tests and treatments, ensuring more effective and comprehensive care for patients.

- Families may experience grief, especially if the condition affects a child or close relative, and may also feel guilt or shame about passing the condition to future generations. Genetic disorders can strain family relationships and lead to conflicts over care decisions. The presence of a genetic condition can complicate decision-making processes, requiring careful consideration of various factors.

Research Question 7: Strategies to improve genetic counselling practice

- To enhance genetic counselling services, several strategies should be implemented. Establishing specialized units and clinics dedicated to genetic counselling would significantly improve service delivery. Expanding these services can benefit from insights gained from related studies and research.

- Encouraging students to pursue education and research in genetics is crucial for advancing the field and increasing its recognition. Incorporating genetic counselling into medical and academic curricula ensures that future healthcare professionals are well-prepared in genetics.

- Promoting career development and providing support for students and professionals in genetics can help build a robust workforce to meet growing demands.

- Increasing public awareness through targeted campaigns can educate individuals about the benefits and availability of genetic counselling, similar to successful mental health initiatives.

- Government initiatives, such as campaigns in schools, hospitals, and health centers, as well as offering free treatments and services, can further engage the public.

- Leveraging virtual media and implementing innovative schemes dedicated to public education can enhance outreach and effectiveness.

- Proper legal support is crucial for protecting individuals with genetic disorders. Accreditation for current genetic counsellors and government support can improve their status and effectiveness.

- Establishing new policies and schemes to protect individuals with genetic disorders, along with programs supporting career and personal development, can positively impact their daily lives and decision-making.

- Promoting research in genetics and creating government job opportunities can attract students to the field, benefiting both individuals and society.

- Providing psychoeducation in diverse settings and acknowledging genetic counselling as a distinct and specialized field will enhance public understanding and acceptance of genetic counselling services.

5.2 Suggestions

- Establishing specialized units and clinics dedicated to genetic counselling can significantly improve service delivery. These centers should be equipped to offer comprehensive guidance and support for individuals and families affected by or at risk of genetic conditions.

- Encouraging students to pursue education and research in genetics is crucial. Incorporating genetic counselling into medical and academic curricula will ensure that future healthcare professionals are well-prepared in genetics, advancing the field and increasing its recognition.

- Promoting career development and providing support for students and professionals in genetics can help build a robust workforce. This includes offering training opportunities, career guidance, and support systems for those entering the field.

- Implementing targeted campaigns to increase public awareness about the benefits and availability of genetic counselling is essential. Similar to successful mental health initiatives, these campaigns should focus on educating the public and reducing stigma associated with genetic disorders.

- Government initiatives, such as campaigns in schools, hospitals, and health centers, as well as providing free treatments and services, can engage the public more effectively. Leveraging virtual media and innovative public education schemes can further enhance outreach and effectiveness.

- Proper legal support is crucial for protecting individuals with genetic disorders. Accreditation for genetic counsellors and government support can improve their status and effectiveness. New policies and schemes should be established to protect individuals with genetic disorders and support their career and personal development.

- Promoting research in genetics and creating government job opportunities can attract students to the field and benefit society. Support for research initiatives will further enhance the development and integration of genetic counselling.

- Offering psychoeducation in diverse settings and acknowledging genetic counselling as a specialized field will improve public understanding and acceptance. Specialized support programs should be developed to address the specific needs of families and individuals affected by genetic conditions.

- There is a need to develop and implement standardized practice models for genetic counselling. Establishing clear protocols and guidelines will ensure consistent and effective practice, reduce variation, and protect the privacy and rights of individuals.

- Addressing barriers such as the high cost of genetic tests and lack of insurance coverage is essential. Strategies should be developed to make genetic counselling more affordable and accessible, ensuring that financial constraints do not prevent individuals from seeking necessary care.

5.3 Conclusion

Genetic counselling is an essential service that offers valuable guidance and support to individuals and families navigating the complexities of genetic conditions. This specialized field plays a crucial role in educating people about genetic disorders, including their causes, implications, and available management options. It provides both emotional and professional support, helping families make informed decisions and take proactive steps to address potential health challenges.

Despite its critical importance, genetic counselling faces several significant challenges. Many individuals lack awareness about the benefits and availability of genetic counselling, leading to underutilization of these services. Additionally, the high costs associated with genetic testing

and counselling create a substantial barrier for many families, often preventing them from accessing necessary support.

In regions like Kerala, there is a noticeable scarcity of genetic counselling services, exacerbated by a lack of standardized protocols and guidelines. This inconsistency in service delivery affects the quality of care and contributes to the reinforcement of social stigma surrounding genetic disorders. The shortage of qualified genetic counsellors further limits access and impacts the effectiveness of the services provided.

To address these issues, several actions are recommended. Establishing dedicated genetic counselling centers can improve service delivery and ensure comprehensive care. Integrating genetic counselling into medical and academic curricula will prepare future healthcare professionals to effectively manage genetic issues. Public education campaigns should be launched to raise awareness about genetic counselling, utilizing various media platforms to reach a broader audience and combat stigma.

Making genetic counselling and testing more affordable and accessible through government policies and financial support is essential. Developing and implementing standardized protocols will ensure consistent, high-quality care and protect patient privacy. Supporting research in genetics and the professional development of genetic counsellors will help build a robust workforce capable of meeting growing demands.

Addressing barriers such as lack of awareness, inadequate qualifications, and social stigma is crucial. Integrating genetic counselling with other support systems, like vocational training and family therapy, will provide more comprehensive care and reduce costs. By implementing these strategies, we can enhance the impact of genetic counselling, improve the quality of life for individuals and families affected by genetic conditions, and foster a more inclusive and supportive environment. Genetic counselling has its own significance so addressing the barriers through effective strategies can benefit in Kerala population.

5.4 Limitations of the study

The study was specifically focused on the districts of Malappuram and Thiruvananthapuram in Kerala, which may limit the generalizability of the findings to other regions. Additionally, the participation was constrained by the limited number of practitioners with expertise in genetics. Despite these factors, the study provides valuable insights within its defined scope and highlights areas for further research and exploration.

5.5 Suggestions for further study

Further research in the field of genetics could significantly advance our understanding by delving into several crucial areas. One potential avenue is a more in-depth investigation into the causes and implications of genetic counselling specifically related to rapid gene mutations. This type of research would aim to uncover the underlying mechanisms behind these mutations, their health consequences, and how genetic counselling can effectively address and manage these challenges.

Another important area for future study is the integration of genetics education into school curricula from an early age. By exploring how introducing genetics concepts early in education impacts students' understanding and attitudes toward genetic issues, researchers could provide valuable insights into the long-term benefits of such educational initiatives. This could help in shaping curricula that better prepare students for making informed decisions about their genetic health in the future.

Research into methods for improving gene pools also presents a valuable opportunity. Investigating strategies to enhance genetic diversity and reduce the prevalence of genetic disorders could lead to more effective public health interventions. This could include evaluating the success of genetic screening programs and identifying approaches that promote healthier gene pools across populations.

Additionally, studying the experiences and suggestions of genetic counsellors and practitioners could offer practical recommendations for improving genetic counselling practices. Understanding the day-to-day challenges faced by these professionals and gathering their insights could lead to better training, resource allocation, and overall effectiveness in delivering genetic counselling services.

Finally, research into how individuals perceive genetic risks and make decisions about genetic testing, treatment, and family planning could provide a deeper understanding of personal decision-making processes. Exploring how people interpret genetic information and the factors that influence their choices can offer valuable insights into the psychological and practical aspects of managing genetic risks, thereby improving support systems and decision-making frameworks for individuals and families navigating genetic information.

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APPENDICES

INFORMED CONSENT FORM

Name of the respondent:

Age:

Sex:

Place:

Educational Qualification:

Job:

I am Dilsha K, pursuing my master's degree in Counselling Psychology at Loyola College of Social Sciences, Thiruvananthapuram. I am doing this research behalf of my curriculum on the topic 'significance of Genetic Counselling'. I am going to give you information and invite you to be part of this research. You do not have to decide today whether or not you will participate in the research. Before you decide, you can talk to anyone you feel comfortable with about the research. This consent form may contain words that you do not understand. Please ask me to stop as we go through the information and I will take time to explain. If you have questions later, you can ask them of me or of another researcher.

Purpose of the study

Human health and Genetics are important so this study tries to explore the significance of counselling services in the genetic areas. We believe that you can help us by telling us what you know both about genetic counselling and about local health practices in general. I want to know the significance of this genetic counselling, how the genetic disorders effect the families and what a counselling psychologist can do in that area so as to improve the health and living of humans.

Type of Research Intervention

This research will involve your participation in a semi structured interview that will take about one and a half hour.

Voluntary Participation

The choice that you make will have no bearing on your job or on any work-related evaluations or reports. You may change your mind later and stop participating even if you agreed earlier.

Risks

There are no major risks involved like breaching of confidentiality of your clients, you may need to allocate time of 1 hour for this participation which may interfere your work process.

Benefits

There will be no direct benefit to you, but your participation is likely to help to find out more about genetic counselling which can contribute to our community.

Confidentiality

The information that collect from this current study will be kept private. Any information about you will have a number on it instead of your name. Only the researchers will know what your number is.

Right to Refuse or Withdraw

You do not have to take part in this research if you do not wish to do so, and choosing to participate will not affect your job or job-related evaluations in any way. You may stop participating in the interview at any time that you wish without your job being affected. I will give you an opportunity at the end of the interview to review your remarks, and you can ask to modify or remove portions of those, if you do not agree with my notes or if I did not understand you correctly.

Certificate of Consent

I have read the foregoing information, or it has been read to me. I have had the opportunity to ask questions about it and any questions I have been asked have been answered to my satisfaction. I consent voluntarily to be a participant in this study

Signature of Participant _____

Date _____

SEMI STRUCTURED INTERVIEW SCHEDULE

Introduction:

- Thank you for participating in this interview. The aim of this study is to explore the significance of genetic counselling from the perspective of mental and health professionals. Your insights will help in understanding how genetic counselling is integrated into mental health practice and its impact on families with genetic conditions.

Background Information:

- Could you briefly describe your professional background and your experience with genetic counselling?

Core Questions:

1. **Understanding of Genetic Counselling:**

   What is your understanding of 'Genetic Counselling'?

   Can you describe how you perceive its role within mental health settings?

   How do you think genetic counselling integrates with other mental health services?

2. **Concerns of Families with Genetic Conditions:**

   What are the common concerns or challenges faced by families dealing with a genetic condition?

   How do these concerns impact their mental health and family dynamics?

3. **Importance of Genetic Counselling:**

   In your opinion, what is the importance of conducting genetic counselling?

   Can you provide examples of how genetic counselling has made a difference in your practice or in the lives of families you work with?

4. **Impact on Gene Pool:**

   Do you believe that genetic counselling helps to improve the gene pool? Specifically, does providing suggestions and counselling reduce the expression of genetic conditions?

   [Follow-up] What are your thoughts on the effectiveness of genetic counselling in preventing or managing genetic disorders?

5. **Current Practice in Kerala:**

   How is genetic counselling being implemented in Kerala?

   Are there specific practices or approaches in Kerala that you think are particularly effective or challenging?

6. **Barriers to Practice:**

   Are there any barriers or challenges to practicing genetic counselling in your setting or in Kerala more broadly?

   [Follow-up] How do these barriers affect the delivery and effectiveness of genetic counselling?

7. **Strategies for Promotion and Expansion:**

   What strategies could be implemented to promote and expand the practice of genetic counselling?

Are there particular resources, training programs, or policy changes that you think could support these efforts?

Conclusion:

- Is there anything else you would like to add about the significance of genetic counselling or your experiences with it?
- Do you have any recommendations for further research or improvements in genetic counselling practices?

Closing:

- Thank you for your time and valuable insights. Your input will contribute significantly to understanding and enhancing the role of genetic counselling in mental health settings.