Scholars Academic Journal of Biosciences
Abbreviated Key Title: Sch Acad J Biosci
ISSN 2347-9515 (Print) | ISSN 2321-6883 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Medicine

Exploring the Roles of Genetic Counseling in Pediatric Medicine

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DOI: 10.36347/sajb.2024.v12i01.004 | **Received:** 15.12.2023 | **Accepted:** 21.01.2024 | **Published:** 27.01.2024

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Abstract **Review Article**

In the field of pediatric treatment, genetic counseling is an essential instrument for lowering the prevalence of oncological disorders and uncommon diseases. The primary objective of this inquiry was to conduct a thorough literature analysis with the purpose of acquiring the most recent information on genetic counseling in the field of pediatrics. According to the findings of the investigation, the increased significance of genetic counseling can be attributed to the fact that technology improvements that can be implemented at each stage of the counseling process have contributed to the process. The identification of rare illnesses and oncology situations, such as retinoblastoma, can be accomplished with the astounding assistance of genetic counseling. Genetic counseling can be done to provide premarital information on genetic problems in order to reduce the likelihood of developing future illnesses such as thalassemia. This can be accomplished by utilizing genetic counseling. Genetic counseling is a vital practice that gives families and healthcare practitioners the ability to make well-informed decisions regarding an individual's health.

Keywords: Genetic counseling, rare diseases, oncology, family, healthcare providers.

INTRODUCTION

The present study explores genetic counseling in pediatrics from various points of view. It starts with the definition of the concept of genetic counseling and covers the roles of genetic counseling in oncology and rare diseases.

1- An Overview of Genetic Counseling

Hereditary counseling, which is also referred to as genomic counseling, encompasses the provision of support to individuals in understanding and adapting to the physical, psychological, and familial ramifications associated with hereditary disease factors (Ormond, 2013).

As previously said, genetic counseling is one of the strategies used to avoid hereditary illnesses (Rahmat *et al.*, 2022). Genetic counseling, as opposed to standard care or routine interventions such as health education or psychoeducation, provides a more complete approach (Mwangi and Mbwayo, 2020). Genetic counseling includes a recurrence calculating procedure, a carrier screening test, and decision-making that can affect the patient's psychosocial condition. Alternative therapies, on the other hand, largely provide broad disease information (Setiawan *et al.*, 2018). According to multiple studies, genetic counseling is used because it can help an individual understand and adjust to medical, psychological, familial, and genetic variables associated with a condition that is not addressed by alternative interventions (Setiawan *et al.*, 2018).

Genetic counseling, as defined by the National Society of Genetic Counselors (Rujito, 2018), is a communication process that addresses health issues about the inheritance of diseases within a family. Genetic counseling refers to the systematic presentation of genetic disease issues to a family in a suitable manner (Setiawan *et al.*, 2020), Multiple stakeholders participate in this procedure, including one or more competent individuals who assist the family or individual in comprehending the medical information associated with the diagnosis, prognosis, and treatment of genetic disorders.

Genetic counselors evaluate the probability of patients developing a particular disease by analyzing their medical and familial backgrounds. They provide instruction on current research, inheritance patterns, genetic testing, management, and preventative measures (Mochiki *et al.*, 2023). Furthermore, they offer direction to empower patients and their families to make well-informed choices and adapt to the potential hazard or state linked to the ailment (Shickh *et al.*, 2023).

Genetic counseling provides a customized approach that is modified to accommodate the unique requirements of each patient and family, empowering them to understand their ailment, familial history, and available treatment options (Morales *et al.,* 2023). Genetic counselors fulfill an essential function by proficiently conveying complex genetic principles, possessing expertise on a wide range of genetic tests, and aiding families in managing potential apprehensions (including uncertainty and risk) (Lowe *et al.*, 2023).

In addition to handling familial and psychosocial concerns, they guide families as they navigate the decision-making process concerning genetic testing (Dean *et al.,* 2023).

2- Genetic Counseling for Families

While it is recommended that patients and their families or surrogate decision-makers who undergo genetic testing, such as genome-wide sequencing (GWS), receive genetic counseling, little is known about the specific gaps in research regarding genetic counseling practices for genomic or genetic testing in pediatric and neonatal intensive care units (NICUs and PICUs) (Kim *et al.,* 2024). NICUs have utilized genetic testing to identify or validate clinical diagnoses in patients with genetic disorders for many years. In NICUs, recent research has highlighted the efficacy of rapid sequencing in the diagnosis of neonates that are critically ill (Gubbels *et al.,* 2020; Wojcik *et al.,* 2023). NICU admissions are characterized by a multitude of complications, including but not limited to preterm birth, low birth weight, infection, asphyxia, surgically required congenital abnormalities, and a substantial prevalence of genetic disorders (Chow *et al.,* 2015). Genetic disorders, which include chromosomal abnormalities, congenital deformities, and deformations, are the leading cause of infant mortality, accounting for approximately 20% of all child fatalities (Banu *et al.,* 2023).

Individuals who are admitted to Pediatric Intensive Care Units (PICUs) are generally younger than 18 years old and may exhibit a wide range of symptoms, including severe and chronic rare disorders, incidental injuries, and acute illnesses (Demers *et al.*, 2022).

Similarly, families of children admitted to the Pediatric Intensive Care Unit (PICU) endure a multitude of stressors; this emphasizes the importance of delivering care that addresses the needs of the entire family within such environments (Baird, 2014).

3- Genetic Counseling in Pediatric Rare Diseases

A rare disease in Europe affects less than one in every 2000 people, or less than 0.05% of the population, according to the 2005 definition provided by the European Organization for Rare Diseases. According to the Rare Diseases Act (H.R.4013—107th Congress 2002), an illness is considered rare in the US if it affects fewer than one person out of every 200,000. According to some authorities, a disease is considered "ultrarare" if its occurrence rate is less than one in two million (Hennekam, 2011). When taken together, unusual diseases, however rare in and of themselves, are projected to afflict 4-8% of the general population (Boycott *et al.,* 2017). Approximately 7000 uncommon disorders are thought to have an 80% hereditary propensity (Amberger *et al.,* 2019). Most instances mostly impact young children; 30% of affected individuals are thought to die before turning five (European Organization for Rare Diseases, 2005). The intricate character of uncommon medical disorders and their involvement in several physiological systems set them apart. Because these conditions are uncommon, most professionals are often unaware of them. They thus pose serious difficulties for the diagnosing procedure (Elliott, 2020).

Early and precise diagnosis of uncommon genetic abnormalities in infants may be possible with the assistance of exome and genome sequencing (E/GS) (Wright *et al.,* 2018; Crellin *et al.,* 2023). Healthcare funders are increasingly acknowledging the financial and therapeutic advantages that early diagnostic investigations employing E/GS can provide (Crellin *et al.,* 2023). Therefore, the test's accessibility in clinical environments has been enhanced (Sachdev *et al.,* 2021). Genetic services are facing challenges in meeting the increasing demand (Fennell *et al.,* 2020). To enhance patient access, test ordering must be extended beyond genetic services and into standard clinical practice (Burton *et al.,* 2017). The challenge and intricacy of implementing practice change in the healthcare industry are widely recognized (Bauer *et al.,* 2015). To promote transformation and improve the delivery of healthcare services, interventions are necessary (Skivington *et al.,* 2021). It is critical to comprehend the viewpoints and requirements of diverse stakeholders, such as families and patients, to identify and formulate efficacious treatments (Crellin *et al.,* 2023).

There is an increasing acknowledgment within the healthcare sector of the importance of collaborating with patients, and caregivers specifically in the context of pediatrics, to enhance the quality of services delivered and ensure that their needs are adequately addressed (Bombard *et al.,* 2018). This is supported by the fact that patient experience is now incorporated into quality-of-care benchmarks (ACSQHC, 2011).

4- Pediatric Genetic Counseling in Oncology

The initial stage in the early identification and prevention of hereditary cancer with onset in infancy involves the careful selection of appropriate patients and families for cancer genetic counseling (Schneider and Jasperson, 2015). Recently, guidelines have been established and are constantly being developed to identify specific types of tumors and other warning signs that indicate the need for a hereditary cancer risk assessment (Hampel, 2015). However, these guidelines

are insufficient in addressing the various forms of tumors that occur in youth and may necessitate a referral. Specifically, although certain types of tumors like neuroblastoma or leukemia may not be worrisome on their own, the presence of other indicators such as multiple tumors or a family history of cancer in specific tumor types can warrant a referral in the context of pediatric medicine. Moreover, our understanding of the specific tumor/cancer types that are prone to arise from a germline mutation is constantly evolving. Within approximately one year, it was reported that patients diagnosed with hypodiploid acute lymphocytic leukemia (ALL), sonic hedgehog medulloblastoma, or anaplastic rhabdomyosarcoma have a notably greater likelihood of having a germline TP53 mutation compared to patients with different subtypes of ALL, medulloblastoma, or rhabdomyosarcoma (Schneider and Jasperson, 2015).

An additional concern with hereditary cancer syndromes that appear in youngsters is the potential absence of any familial medical record of the condition. The child's close relatives may have been in their youth when they went through this, which could clarify why the family's cancer history is not readily evident until the medical records of more distant relatives are examined. Occasionally, the absence of any family history can be attributed to recessive inheritance or de novo inheritance of a condition (Hettmer *et al.,* 2014; Schneider and Jasperson, 2015).

It is recommended that patients, their families, or surrogate decision-makers who are undergoing genetic testing, particularly for genome-wide sequencing (GWS), seek genetic counseling (Kim *et al.,* 2024). However, research on the specific gaps in genetic counseling considerations regarding genomic or genetic testing in pediatric and neonatal intensive care units (NICUs and PICUs, respectively) is lacking (Elliott and Friedman, 2018). Genetic testing has been utilized by neonatal intensive care units (NICUs) to identify or validate clinical diagnoses in patients with genetic disorders for many years (Wojcik *et al.,* 2022; Kim *et al.,* 2024). In NICUs, recent studies have highlighted the efficacy of rapid sequencing for the diagnosis of critically ill infants (Meng *et al.,* 2017; Kingsmore and Cole, 2022; Rodriguez *et al.,* 2022).

Infants are admitted to Neonatal Intensive Care Units (NICUs), where they present with a range of complications including preterm birth, low birth weight, infection, asphyxia, surgically correctable congenital deformities, and a notable prevalence of genetic disorders (Heidari *et al.,* 2013). Genetic disorders, which include chromosomal abnormalities, congenital deformities, and deformations, are the leading cause of infant mortality, accounting for approximately 20% of all child fatalities (Murphy *et al.,* 2017).

Limited research has been conducted on the application and practicality of genetic and genomic testing in pediatric intensive care units (PICUs). Pediatric Intensive Care Units (PICUs) frequently see patients who are below the age of 18. These individuals present with a wide range of conditions, including but not limited to acute illnesses, incidental injuries, and chronic and severe rare disorders (Ibiebele *et al.,* 2018). Previous studies have established a correlation between the Neonatal Intensive Care Unit (NICU) and a variety of difficulties that have a substantial impact on the experiences of caregivers upon the admission of their child. It is not uncommon for caregivers working in the NICU to encounter heightened levels of anxiety and despondency (Del Fabbro and Cain, 2016; Williams *et al.,* 2018).

Similarly, families of children admitted to the Pediatric Intensive Care Unit (PICU) endure a multitude of stressors; this emphasizes the importance of delivering care that addresses the requirements of the entire family within such environments (Debelić *et al.,* 2022).

Reduced MHC-I expression, a low tumor mutational burden (TMB), and a proportionally low amount of neoantigens are characteristics of pediatric malignancies (Bao *et al.,* 2021; Jardim *et al.,* 2021). For childhood-onset malignancies, alternative therapeutic approaches are required to increase MHC-I expression on tumor cells. Major Histocompatibility Complex class I (MHC-I) expression has a large potential to be upregulated in many types of cancer as a means of immunotherapy, specifically to restore immunological control of malignancies (Guillaume *et al.,* 2024). It can increase immunotherapy's response rates and increase the treatment's efficacy for malignancies that weren't responding before (Wang *et al.,* 2021).

5- Genetic Counseling in Pediatric Retinoblastoma

The most prevalent type of cancer in young eyes, retinoblastoma, can run in families or be passed down through generations. Everyone working with patients must be aware of the seriousness of this diagnosis because of the presence of another cancer risk factor. Patients with retinoblastoma, regardless of whether the disease runs in their family or is unilateral, must have genetic testing and counseling as part of their treatment plan. In addition to influencing primary tumor care, a patient's genetic status in retinoblastoma dictates the length and frequency of follow-up exams to detect any further eye tumors or the onset of secondary cancer. Genetic counseling and follow-up measures are discussed in this chapter, which also compares hereditary and non-hereditary retinoblastomas. It also includes two clinical instances that show how different types of retinoblastoma manifest and how different treatments work (Soliman *et al.,* 2017; Im *et al.,* 2023).

6- The Role of Genetic Counselors

Genetic counselors engage in various research activities, including securing grants, participating in studies, designing studies, and applying study findings

(Wainstein *et al.*, 2023). At the trainee level, there is an additional level of research integration. As to the 2019 guidelines from the Accreditation Council for Genetic Counseling (ACGC, 2019), all accredited genetic counseling training programs are required to provide research findings to maintain their recognition. Hence, it is logical to anticipate that genetic counselors will inevitably come across research methods in some capacity (Wainstein *et al.*, 2023). According to a recent content survey of the Journal of Genetic Counseling, spanning the years 2011-2017, the term "research methodology" was mentioned in only nine pieces (1.9%), making it the least common topic (Wallgren *et al.*, 2021). Unlike the thirty-six percent of studies that employed quantitative approaches, forty-four percent of the assessed publications utilized qualitative methods for data analysis. The proportion of qualitative articles published in each journal issue shown a statistically significant increase, suggesting a rising recognition of the importance of qualitative research methodologies. It is unsurprising that genetic counselors are inclined towards qualitative research methods. During interviews or focus groups, professionals can utilize their training and clinical expertise to effectively address sensitive topics, employ advanced communication techniques, synthesize information, and evaluate outcomes (Biesecker *et al.*, 2019). Nevertheless, possessing these talents in isolation does not ensure the creation of top-notch, therapeutically advantageous qualitative research; a solid theoretical basis and meticulousness are also needed (Wainstein *et al.*, 2023).

7- CONCLUSIONS

The practice of genetic counseling is an essential component of pediatric medicine. This type of counseling educates healthcare workers, who are accountable for the treatment of children and the advice they provide to their families regarding health issues, about the possibility of genetic disorders. More than only malignancies and unusual diseases, genetic counseling can be helpful in a variety of situations. To prevent diseases like thalassemia, which is a prominent example, it is essential to have a genetic counseling approach that is more all-encompassing.

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