

# Bridging Genetics and Ophthalmology: The Emerging Role of Ophthalmic Genetic Counseling in the Philippines

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A significant number of eye conditions have genetic etiology, such as, inherited retinal degenerations (IRD), retinoblastoma, corneal dystrophies – and including the more common ones – cataracts, glaucomas, errors of refraction, age-related macular degenerations, etc. The advancement of technology has improved genetic testing, and therefore promotes a better understanding of the connection between ophthalmology and genetics. Genetic information can be overwhelming for many individuals, and even clinicians may find it challenging to explain genetic concepts to patients. Genetic counseling (GC) addresses this gap. This profession helps patients understand how genetics affects health, the course of the disease, and family risk. GC is both an art and a science. Genetic information can have significant negative consequences for the patients if interpreted wrongly, and may, at times, pose serious risks. As an ophthalmologist myself, genetics is an underrecognized aspect in ophthalmology – especially in developing countries like the Philippines.

Ophthalmic genetic counseling (OGC) is a field of genetic counseling intertwining with the fields of genetics and ophthalmology. Although rare, it

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stands on an important ground of healthcare service where it aims to discuss and provide education of genetic eye disorders to the patients and family members. While OGC could be new to the Philippines, it has its roots from the mid-20<sup>th</sup> century when genetic counseling started to rapidly expand<sup>1</sup>. Globally, this field has already started helping individuals with genetic eye diseases and catering genetic services to provide support to affected patients. While there are only a few published literatures regarding OGC, it is considered already as an emerging field more prominently in first-world countries where genetic testing is available<sup>1</sup>. This paper aims to provide an introduction to OGC and its relevance not only in a global context, but as well as in the Philippine setting.

## **Roles, Responsibilities of the Ophthalmic Genetic Counselor**

The significance of counseling patients is beginning to be more superficial as more eye disorders with genetic predisposition become unraveled. The role generally of a genetic counsellor is similar to other subspecialties: (1) provide information and educate a layperson about basic

biology, inheritance patterns, and options for genetic testing<sup>1</sup>, (2) calculate risks of inheritance, (3) conduct a pre- and post-genetic testing education and counseling, and (4) provide psychosocial support on the genetic information learned by affected individuals and their families. Sutherland et. al.<sup>1</sup> described OGC as counseling of vision-related issues requiring (1) basic knowledge of eye terminology, anatomy, and universal definitions and standards for visual function such as legal versus complete blindness, (2) eye test descriptions (e.g., visual acuity, visual fields, electroretinogram, visual evoked potentials, etc.), (3) awareness of vision aids, (4) knowledge of level of visual function such as vision affecting employment and driving abilities; (5) discussion on mobilities and low vision options, (6) identification of local support services and research initiatives<sup>1</sup>. These described tasks pose challenges to the genetic counselor, often requiring to allot more time and effort to read on ophthalmology prior to counseling patients.

### Challenges of Ophthalmic Genetic Counseling in Asia

In Asia<sup>2</sup>, it was emphasized by Hui et al. that genetic counseling in the region is a growing clinical trend as of 2022. Recently, a Professional Society of Genetic Counselors in Asia was established in 2015 introducing genetic counseling to the underserved countries including the Philippines<sup>2</sup>. Furthermore, the challenges of genetic counseling in Asia are elaborated by Hui et al<sup>2</sup> to as follows: (1) inadequate knowledge of genetics across different physicians, (2) shortage and absence of genetic counsellors, (3) limited access to genetic services, and (4) absence of legal policies and regulations on genetic discrimination<sup>2</sup>. What even adds more to these difficulties are the lack acknowledgment, limited monetary compensation and remuneration, and ineffective regulatory body execution for genetic counselors<sup>2</sup>. The recent establishment of the Philippine Society of Genetic Counselors (PSGC) in 2021 further reflects the growing recognition of genetic counseling as an essential component of healthcare in the country. The formation of the society reflects that the profession is continuously evolving hoping for more collaborations between genetic counselors and subspecialties like ophthalmology which may help expand access to

culturally sensitive genomic services for Filipino patients and families.

### Ophthalmic Genetic Counseling in the Philippines: Factors and Challenges

OGC assumes an important role and faces many challenges because of many inherent factors relating to the country's healthcare system. One of the main challenges in the Philippines points to language barrier system. The Philippines is known for its language diversity with over 167 Filipino ethnolinguistic groups and 150 languages spoken across the archipelago<sup>3</sup>. Words like "genetics," "DNA," "retinoblastoma" or "retina" sometimes do not exist in a vernacular language counterpart, often requiring counselors to take more steps ahead to expound on these concepts<sup>2,3</sup>. Additionally, even the disease concepts in itself, such as retinoblastoma (RB) and IRD which are rarely discussed diseases especially in the rural areas, adds extra effort for the genetic counselor in further translating the counseling itself.

Another important consideration is the cultural context through which patients and families understand disease causation. Abad et al. described several Filipino explanatory models of illness, including *namamana* (inheritance), *libi*, *sumpa* or *gaba* (curse), *namaligno* (supernatural causes), *pasma*, and *kaloob ng Diyos* (God's will), which may coexist with biomedical explanations of disease<sup>4</sup>. In the context of inherited ocular disorders, these beliefs largely influence how families interpret diagnoses, communicate risk within families, and make decisions regarding genetic testing and surveillance. With this in consideration, the art of genetic counseling in the Philippines does not only look at providing accurate genomic information but at the same time, carefully incorporating culturally sensitive information with acknowledging patients' beliefs and values. This approach may improve understanding, trust, and acceptance of genetic services while ensuring that counseling remains patient-centered and culturally sensitive.

Relating to the existing genetic eye disorders in the country, another foreseen challenge is the relatively high prevalence of heritable eye disorders, namely, retinoblastoma, glaucoma, retinal degenerations, corneal and optic nerve disorders. All

of which are relatively prevalent, and more often, these eye conditions go undetected until its later stages, leading to significant morbidities and challenges in the treatment and management. In essence, the knowledge of ophthalmic genetic counseling becomes a vital tool in the early and timely identification and management which can potentially mitigate the impact on affected patients and families.

Another point where genetic counseling assumes a significant position in the field of healthcare is the fact that genetic counseling is part of the aspect of preventive healthcare. The healthcare system of the Philippines is increasingly recognizing the importance of preventive measures approach to local diseases. Counseling aligns with this approach by providing patients and families the information and knowledge to adopt proactive measures in managing eye health. In my personal view, this approach does not only reduce the burden to the system, but promotes a culture of wellness and prevention. However, the ethics of predictive testing for genetic eye diseases is an issue<sup>5</sup>. A review conducted by Mezer et al elaborated various attitudes towards predictive testing of various ocular conditions, such as different approaches of acceptance of untreatable conditions such as retinitis pigmentosa<sup>5</sup>. Furthermore, predictive testing for one disease differs considerably from another condition. For example, in glaucoma, appropriate treatment may cure or slow down the progression; therefore, it is beneficial to diagnose patients as early as possible, while predictive testing for retinitis pigmentosa, being an untreatable condition, brings more damage to the psychosocial wellbeing of affected individuals and their families<sup>5</sup>.

Additionally, access to specialized medical care, including genetic counseling services, is limited in the Philippines, notably in the rural, underserved areas. In such cases of limited access, OGC can improve resource allocation and facilitate more targeted interventions, ensuring all Filipinos have access to genetic information and care. A good example are families affected with RB. RB is a childhood cancer of the eye affecting 1:17,000 live births<sup>6,7,8</sup> where about 40% of the cases are hereditary<sup>6</sup>. In such cases, little is known about the reproductive behavior of couples who are at an increased risk for having a child with RB. A study

based in Netherlands<sup>6</sup> showed that RB had not been a factor in the reproductive decisions of thirty-eight out of 81 respondents (47%) because of limited access to genetic information. Given that RB carries numerous enduring consequences, not only for the individual but also for the family, extending beyond genetic and physical impacts, the family's quality of life and financial stability are jeopardized if genetic counseling is not addressed to prevent long-term repercussions<sup>8,9</sup>. Similarly, one of the challenges for lack of access to genetic information is that most of the time, OGC is done by the ophthalmologist. In a study by Cohen et al.<sup>10</sup>, genetic information is learned from the ophthalmologist mainly (69%), where the rest is composed of geneticist, oncologist and genetic counselors<sup>10</sup>. It is not surprising that because of the shortage or even absence of genetic counselors, ophthalmologists tend to play the role of a counselor despite the inadequate training for disclosure of genetic information.

Moreover, genetic counseling in the Philippines faces challenges in religion, culture and language. The mixture of various ethnic and cultural groups influences significantly the decision-making process of families and how they cope to genetic information<sup>2,3</sup>. In contrast to this existing knowledge of the challenges in terms of cultural beliefs and practices of the Filipino people<sup>3</sup>, OGC aligns with the strong family ties and family-centered approach rich in the Filipino culture. By considering the familial context of the Filipinos, genetic counselors can foster open communication and in effect, promote more tailored guidance without cultural nuances. This, in my personal view, is the advantage of OGC towards its introduction to the Philippine setting. This approach incorporates psychosocial elements into genetic counseling to ensure that the patients are well-informed and that their autonomy is respected<sup>3</sup>.

Finally, genetic counseling offers enormous economic implications. The preventive and more targeted approach, as discussed earlier, in OGC promotes a more cost-effective approach by enabling early intervention and potentially reducing short- and long-term economic burden associated with advanced eye conditions. A study by Soliman et al<sup>11</sup> compared the overall outcomes of prenatal versus postnatal screening of familial RB where the burden of economic cost is significantly reduced for

appropriate and well-planned prenatal and postnatal screening of RB<sup>11</sup>. In this similar study, a prenatal molecular diagnosis of RB on the parents increased the likelihood of infants born without detectable tumors which equated to better vision outcomes, and lesser invasive therapy. The role of prenatal molecular diagnosis facilitates anticipatory planning mentally, physically and emotionally for both the affected child and family<sup>11</sup>.

### Introduction of a New Field to the Philippine Setting

In the field of developing healthcare in the Philippines, OGC serves as a field envisioning the path towards personalized and precision medicine. This field hopes for a promising future in revolutionizing the understanding of patients, families and healthcare workers in the etiology, diagnosis and treatment of heritable eye conditions present in the country. Even with the diverse cultural background and traditions (e.g., strong family ties) among Filipinos, OGC complements as a hope in providing individuals with a unique opportunity to understand their genetic conditions. Soon, the future holds hope and promises for better life outcomes for those affected, as more diagnostic examinations and management become available, such as the introduction of preimplantation genetic diagnosis for RB predisposition<sup>12</sup>. The advent of newer technologies, in fact, has allowed the emergence of more novel options to treat, and this, in effect, could significantly reduce the incidence of inherited ocular disorders.<sup>12,13</sup> This, in general, ultimately results and reflects to a lesser burden to the economy of the Philippine government. As the Philippines embraces a new era of genetics, the ophthalmologists and geneticists are hopeful that precision medicine unfolds in the country, offering a glimpse into the future of treating genetic eye disorders that were once thought incurable.

In summary, OGC emerges not only as an additional complement to the field of genetic counseling, but also serves to shed light in the future of treatment of genetic eye disorders. OGC has in its wings its enormous potentials to make a lasting impact not only in the early and timely detection, promotion of more expanded preventive measures, and fostering culture of autonomy and informed decision-making, but also, of utmost importance, to

the overall wellbeing of the affected individuals and their families. Finally, genetic counselors must be able to adapt to the growing specialized field of genetics across different subspecialties together with the rapid advent of emerging precision medicine across the world.

### Ethics Compliance Statement

The author affirms that this perspective manuscript complies with the ethical standards of the *Philippine Journal of Ophthalmology* and adheres to the principles of responsible authorship and scholarly integrity. This manuscript does not involve research on human participants, patient data, or identifiable clinical images; therefore, institutional review board (IRB) approval and informed consent were not required. Any clinical scenarios referenced are either hypothetical, anonymized, or drawn from publicly available information without compromising patient privacy. The author certifies that all opinions expressed are their own, based on current evidence, professional experience, and critical analysis. All sources of information have been properly cited, and no confidential or proprietary information has been disclosed.

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