Health Disparities in Physician-Patient Communication for the Diagnosis of Osteogenesis Imperfecta

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Submitted to: Journal of the Health Disparities Think Tank

Citation Suggestion: Komal, T., Mazumder, A., Oh, A., Kurani, A. (2024). Health Disparities in Physician-Patient Communication for the Diagnosis of Osteogenesis Imperfecta. *Journal of the Health Disparities Think Tank.*

INTRODUCTION AND LITERATURE REVIEW

Health disparities are social and economic factors that result in unequal medical care across different populations, thus creating differences in the quality of medical health across a community ("Race, Ethnicity, and Health").

This is especially serious in populations suffering from rare diseases because high-end resources and technology are needed to detect and treat these diseases early on. Thus, such diseases can be complex to diagnose if the family does not have access to medical care due to low income, lack of education, and language barriers. These factors increase the possibility of delayed medical care, which increases the risks of permanent or untreatable diseases. The most common and adverse health disparities affecting patients with rare diseases include immigrant and minority status, low health literacy, low income, and living in medically under-resourced communities ("Race, Ethnicity, and Health").

Such health disparities can lead to delays in diagnosis, medical treatment, psychological care, genetic counseling, and, most importantly, the progression of the rare disease. These consequences are further exacerbated when considering that timing is crucial with rare diseases timely diagnosis is the most critical factor with rare diseases, second to treatment availability ("How Do Healthcare Providers Diagnose Osteogenesis Imperfecta (OI)?", 2021). However, some of these factors are preventable and changeable. Delays caused by health disparities should be eliminated to improve the patient's medical health significantly.

One such rare disease is osteogenesis imperfecta (OI). OI is classified as a "very rare" genetic and musculoskeletal disease that occurs in every 15,000 to 20,000 people (Deguchi et al., 2021). This disease results when a patient's bones become very brittle and weak, often resulting in several fractures at once from a single low-traumatic event. The pathogenesis of this disease involves mutations in genes that control type I collagen synthesis or in genes responsible for the post-translational modification of type I collagen. Collagen is a substantial component of bones; therefore, deficiencies in this protein can result in severe weakness and fragility. Some other signs of this disease include hearing loss, scoliosis, short stature, reduced respiratory function, cardiac valve regurgitation, blue sclerae, and dentinogenesis imperfecta (Marini & Dang, 2020).

Osteogenesis Imperfecta has several classifications, which are dictated by the structural abnormalities of the type I collagen and thus result in varied phenotypes. The mildest form is OI Type I, which has blue sclerae but no bone deformities. The second mildest form is Type IV, which results in bone deformities but normal sclerae. On the other hand, OI Type III is the most severe and is exhibited in patients who survive the newborn period, resulting in severe bone deformities and short stature (Marini & Dang, 2020).

The diagnosis process for this disease requires various tests from the patient. For example, physicians must take accurate family histories, clinical signs, and genetic tests to diagnose patients ("How Do Healthcare Providers Diagnose Osteogenesis Imperfecta (OI)?", 2021). To obtain these results, physicians often require access to medical resources such as X-rays, bone density tests, and bone biopsy equipment ("How Do Healthcare Providers Diagnose Osteogenesis Imperfecta (OI)?", 2021). Other resources may include gene counseling for family members and genetic therapy for severe cases (Marini & Dang, 2020).

With all these considerations and resources required for diagnosis, it is evident that the patients that are most subject to delays in OI diagnosis are usually racial minorities living in medically under-resourced areas. Moreover, racial minorities often face significant language barriers, poor health literacy, low income, and lower living standards, which are frequently associated with limited medical resources in their communities. Consequently, living in these contexts predisposes these patients to delayed or missed diagnoses, significantly worsening patient health outcomes (Nguyen et al., 2022).

According to the NIH, nearly 10% of the U.S. population has a rare disease, and nearly 2.5% faces delayed diagnosis, such as up to 7 or more years (National Organization for Rare Disorders, 2020). This prevalence raises the question, How can we decrease the effects of health disparities in hopes of increasing diagnosis and early intervention rates amongst racial minorities suffering from Osteogenesis Imperfecta in under-resourced areas across the U.S? This question is crucial to explore, as it continues to impact a substantial portion of our community. The answers could lead to more data, research, and new insights within the orthopedic field. Most importantly, disadvantaged populations will benefit from the increased care and research.

CHARACTERIZING THE STATE OF DISPARITIES

Three main health disparities associated with the misdiagnosis or delayed diagnosis of osteogenesis imperfecta are illiteracy and language barriers, as well as a lack of medical resources within a community.

Illiteracy, the lack of knowledge in a particular area, can come in many forms, including. Health literacy, genetic literacy, and language illiteracy. These illiteracies directly impact the patient experience by preventing the communication of pertinent medical history and symptoms, constituting a major component of medical diagnosis (Nguyen et al., 2022). Medical history, especially, gives clues to a patient’s genetics and, therefore, can provide definite signals about genetic disorders and conditions.

Moreover, health literacy sets the stone for general health inquiries. Without such health education, patients often forgo seeking medical care, which can result in further complications of a rare disease. Likewise, a lack of genetic literacy or understanding of genes and how they can be passed down can result in delayed or missed diagnoses. Being unable to perceive patterns of symptoms among family members can create a series of missed rare disease cases that can become extenuating for the family. Finally, language barriers significantly hinder the communication of symptoms and medical histories, even if the patient has strong genetic knowledge and health literacy. Often, language barriers prevent patients from reaching out due to a lack of confidence and fear of being misunderstood (Nguyen et al., 2022). If that is not the case, a lack of medical interpreters can intensify this disparity. Furthermore, physicians may attempt to compensate for the lack of medical data from patients through increased testing. Consequently, this action can exacerbate patients' financial limits, leading patients to become financially hesitant to come back for returning appointments.

Additionally, a lack of medical resources within the hospital/clinic can substantially increase these disparities. For the diagnosis of rare diseases such as osteogenesis imperfecta, a more comprehensive range of medical equipment, technology, and specialists is needed. More specifically, diagnosing osteogenesis imperfecta requires specialized medical devices such as X-rays, bone density tests/scanning equipment, magnetic resonance imaging (MRI) devices, equipment for bone biopsies, and medical staff, including geneticists, orthopedists, and pathologists ("How Do Healthcare Providers Diagnose Osteogenesis Imperfecta (OI)?", 2021).

In under-resourced areas, which often lack even basic medical equipment and devices, this can lead to referrals to more expensive hospitals (a challenge for those in underserved communities) or result in misdiagnoses, delayed diagnoses, or entirely missed diagnoses. Either alternative prolongs the diagnosis process, resulting in the progression of osteogenesis imperfecta in the individual. A special degree of training is necessary for doctors to support these specific communities with health illiteracy or medical equipment.

ANALYZING THE IMPACT OF THESE DISPARITIES ON THE DIAGNOSIS OF OSTEOGENESIS IMPERFECTA

Impact of Illiteracies (Health, Genetic, Language) on Diagnosis of OI

Health, genetic, and language illiteracies tend to decrease the efficient flow of information, compromising the patient-physician relationship and preventing adequate, timely, and accurate diagnosis of osteogenesis imperfecta. These factors further contribute to the progression of the disease into more advanced stages, which, if prolonged, may be incurable. In other cases, the prolonged diagnosis can complicate further relations as parents make reproductive choices unknowingly.

Family Disadvantages/Social Impact

As seen in the case study of a 3-year-old toddler, language barriers and ineffective information retrieval on the part of the patient resulted in a delayed diagnosis (Nguyen et al., 2022). A stark example is when the patient's parents did not reveal any symptoms until a later appointment. The parents were also not educated about the concept of genetic disorders, and hence they were unable to connect their toddler's disease to their aunt, who had previously shown similar symptoms and was diagnosed with OI. Moreover, the family could not communicate with the physician due to language barriers. A lack of certified medical interpreters made it even more difficult to obtain accurate information. These factors contributed to an unreliable family history, which prolonged the diagnosis of OI.

A shortage of certified medical interpreters leads to using non-certified interpreters like family or friends, which may lead to miscommunications, lack of clarification, and knowledge gaps (Nguyen et al., 2022).

On the other hand, the importance of early diagnosis is exemplified in the case study of a woman with osteogenesis imperfecta. A 33-year-old woman, presented to the clinic with dentinogenesis imperfecta, slightly bluish sclera, and extraskeletal abnormalities and spoke Turkic. Her previous doctor visits likely missed the diagnosis due to unclear communication and a lack of understanding between the patient and the physician. As a result, her physician was unable to provide an early diagnosis, which led to complications down the line. This prolonged diagnosis also led to a delayed diagnosis in her kids of osteogenesis imperfecta (Sezer et al., 2008).

Family Impact

Such prolonged diagnoses can have an impact on family reproductive decisions. For example, adults tend to choose to have fewer children if they know they have genetic conditions that could be passed down. According to a study, 50% of couples with a 1-50% risk of passing down a genetic disease refrained from having more children. Hence, the prolonged diagnosis of OI can impact family relationships and present moral, emotional, and financial challenges for the family (Baird, 2022). Moreover, parents and caregivers tend to struggle with mental health and financial situations. As osteogenesis imperfecta tends to be a life-long struggle, patients and their families have less time for leisure activities and have to commit more time to hospital visits, applying for financial aid, and paying extra attention to their children. Due to these factors, studies show that parents or caregivers of patients suffering from OI suffer from increased stress (Sudore & Schillinger, 2009).

ANALYZING THE ROOTS OF THESE DISPARITIES AND SEARCHING FOR POLICY SOLUTIONS

According to the results of various case studies involving the delayed diagnosis of OI, it becomes clear that the methods for obtaining accurate histories and evidence from the patient vary across physicians and can lead to discrepancies or inadequate history taking. Improved communication techniques are required to develop patient-physician trust, which can be facilitated by training and education programs (Sudore & Schillinger, 2009).

Not all physicians receive the proper training to support health illiterate populations specifically, as this demographic requires extra and focused support. Physicians must be trained adequately to bring up data that the patient otherwise would have brought up. It thus becomes a clear responsibility of the physician to develop advanced communication skills to perform examinations and obtain patient histories accurately regardless of their patient's background and to effectively explain concepts and questions to the patient.

There have been attempts to bridge this gap in knowledge, namely through TACCT, a Tool for Assessing Cultural Competence Training (Jernigan et al., 2016). Other related programs have been implemented in U.S. medical schools; however, there is a degree of variability among the programs. For example, some U.S. medical schools place this skill as a

minor component of communication courses. Only 40% of US medical schools have included health literacy training in their curriculum (Sagi et al., 2022). This data highlights the need for increased health literacy training to accommodate patients from lower literacy levels and enhance patient-physician communication.

Better physician communication training and strengthened referral networks across rural and medically underserved areas may increase early diagnosis rates (Kornelsen et al., 2023). Data indicates that referrals are more common in nonrural areas than in rural areas by approximately 70% (Geissler, 2019). This indicates that fewer medical visits and diagnoses occur for rare diseases such as osteogenesis imperfecta. Hence, it is important to strengthen the referral network amongst rural and medically underserved communities.

Thus, analyzing the causes of delayed diagnosis in rare diseases such as osteogenesis imperfecta illuminates the need for systematic changes in medical school cultural competency training and strengthening physician referral networks in rural and other medically underserved areas.

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