

Original Research

Quality of Life in Osteogenesis Imperfecta (OI): A Comparative Study with Insights from Handle with Care by Jodi Picoult

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ABSTRACT:

Osteogenesis Imperfecta (OI) is one of the rare genetic disorder characterized by bone fragility, significantly impacting the lives of affected children and their families. This study delves into the complex dimensions of OI's influence on the quality of life (QoL) for these individuals, a crucial step in crafting a tailored Q o L measure for children with OI. This paper also draws intriguing parallels between the research findings and Jodi Picoult's novel Handle With Care, providing a comprehensive perspective on the real-world implications of OI. By combining medical research with literature, it enriches the understanding of OI, emphasizing the potential benefits of integrating insights from literature into medical practice and patient care. The study highlights the urgent need for further research to create a comprehensive Q o L measure for children with OI, underscoring the importance of capturing the intricate nuances of their experiences. The primary purpose of this paper is to address a critical knowledge gap concerning how OI impacts the lives of children and their families. This exploration lays the foundation for a more patient-centered approach to managing OI, facilitating more effective interventions and support systems. In doing so, the paper underscores the significance of embracing patients' perspectives in developing medical measures, championing a holistic and empathetic approach to managing rare medical conditions such as OI. This research is poised to enhance the overall well-being and Q o L of children with OI and their families.

Key Words: Osteogenesis imperfecta, Handle with Care, COL1A1 & COL1A2, Collagen defect, Vroliks syndrome, Sillence classification.

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INTRODUCTION

Osteogenesis Imperfecta (OI), a rare genetic disorder renowned for its hallmark bone fragility,[1] presents a unique intersection of challenges that extend well beyond the realm of clinical diagnosis. As we embark on this research journey, our exploration aims to unravel the intricate layers of OI, delving into not only the physiological intricacies but also the profound impact it has on the quality of life (QoL) for children grappling with this condition and the families that support them. Recognizing OI as more than a skeletal disorder, our research endeavors to paint a comprehensive picture, understanding the far-reaching implications on the lives of those affected. At its core,

OI stems from defects in collagen production, resulting in heightened bone fragility.

While medical research provides an indispensable foundation, the narrative richness of literature serves as an invaluable supplement. Beyond sterile statistics and clinical observations, literature captures the emotional and social dimensions of OI. This section underscores the profound significance of incorporating literary perspectives, enhancing our understanding and fostering a more empathetic approach to rare genetic disorders.

Within this context, Jodi Picoult's masterfully crafted novel, Handle With Care,[2]emerges as a poignant narrative that mirrors the challenges faced by families

navigating life with a child diagnosed with OI. This fictional portrayal not only offers an emotional journey through the complexities of the disorder but also acts as a reflective lens on societal attitudes towards those grappling with rare medical conditions. By juxtaposing the research findings with the narrative arc of *Handle With Care*, the aim is to uncover parallel realities, bridging the world of empirical knowledge with the emotive power of storytelling. As we delve into this comprehensive exploration, the fusion of medical insights and literary narratives seeks to provide a holistic understanding of OI. By illuminating the intricate tapestry of challenges faced by children and their families, we aspire to contribute to the development of more compassionate and tailored interventions, enriching the lives of those people affected by OI.

The Integration of Literature into Medical Research and Practice:

In the ever-evolving landscape of healthcare, the quest for a more holistic understanding of patients and their experiences has led practitioners and researchers to explore unconventional avenues. One such pathway of getting recognition is the integration of literature into medical research and practice. Beyond the cold, clinical data lies a rich tapestry of human emotions, narratives, and complexities that can be illuminated through the lens of literature. This article delves into the rationale, methods, challenges, and successes of integrating literature into the realm of medicine, highlighting its potential to foster empathy, enhance patient care, and redefine the patient-provider relationship.

Health and illness are not mere physiological states but intricate experiences intertwined with emotions, cultural influences, and societal contexts. To capture the full spectrum of these experiences, researchers and healthcare professionals are turning to literature—a medium that offers a nuanced portrayal of the human condition. Literary works, with their diverse characters and intricate plotlines, serve as a mirror reflecting the myriad ways individuals navigate health challenges, providing invaluable insights into the emotional landscapes that often go unnoticed in clinical settings. Numerous case studies showcase the successful integration of literature into medical practices. From improving patient-centered care to enhancing communication between healthcare providers and patients, these models demonstrate the tangible benefits of incorporating literary insights into medical approaches. Lessons learned from historical integration efforts underscore the enduring impact of intertwining the worlds of literature and medicine. In the quest for a more comprehensive understanding of health and illness, the integration of literature into medical research and practice stands as a promising frontier. By acknowledging the multifaceted nature of human experiences, fostering empathy, and embracing the narratives found in literature, healthcare professionals can elevate patient care to new heights.

As the realms of literature and medicine converge, a more compassionate and holistic approach to healthcare emerges—one that recognizes the profound impact of storytelling on healing.

HISTORY

The four major characteristics of OI: fragility of the bones, frail body, hypermobility with easy dislocation of the joints, and blue sclerae, was first given by Edmond Axman 1831, The term “osteogenesis imperfecta” was introduced by professor Willem Vrolikin in 1849 and hence the name Vrolikin syndrome.[3]

Genetic Basis and Mutations in OI Osteogenesis imperfecta is an autosomal dominant (AD) disorder in which the mutation occurs in the genes which encode the production of type I collagen [COL1A1 & COL1A2].[4] Two mutations in the genes are associated with quantitative or structural abnormality in the production or processing of type I collagen.

The genetic underpinnings of OI are fundamental to its manifestation. Numerous mutations in genes responsible for collagen synthesis have been identified, shedding light on the molecular basis of bone fragility. Advances in genetic research have enabled more precise identification of these mutations, paving the way for improved diagnostics and targeted therapies. Clinical Spectrum and Varying Severity\ OI manifests across a diverse clinical spectrum, presenting varying degrees of severity. The Sillence classification designated OI by clinical severity with four subtypes namely: type I-mild, type II-neonatally lethal, type III-moderately severe and type IV- severe [5] caused by mutations in COL1A1 and COL1A2 genes. Later a X-linked variant in the type I presentation was added (in 2013)due to genetic as well as radiological advances, new types have been introduced recently, and the classification has been extended, into types V-XVIII. Types I-V predominantly are considered as autosomal dominant and the rest from VI to XVIII are considered as autosomal recessive [6] understanding this spectrum is crucial for tailoring medical interventions. Research in this area not only delineates the range of clinical manifestations but also identifies factors contributing to this heterogeneity.

Diagnostic Modalities and Advances in Imaging:

Diagnostic modalities for OI have evolved significantly, with contemporary imaging techniques playing a pivotal role. High-resolution imaging, including bone densitometry and CT scans, allows for more accurate and detailed assessments of bone health. These advancements contribute to early and precise diagnosis, facilitating timely interventions and personalised treatment plans. Interrelationship between osteogenesis imperfecta and dentinogenesis imperfecta

Dentinogenesis imperfecta is also a hereditary disorder which has high penetration and low mutation

rate [7]. Oral examinations with panoramic radiographs showed 40–80% of children with types III and IV OI had DI in their primary dentition. Dental anomalies (opalescent teeth, obliterated pulp cavities, and constricted coronal-radicular junctions) were considered as distinguishing genetic features of OI even before the Silence classification [8] Children with Dentinogenesis Imperfecta experience vertical enamel fractures leading to exposure dentin and subsequent pre-matured severe attrition. Early diagnosis of DI with OI is recommended, Dentinogenesis imperfecta can be repairable unlike OI to improve the function and esthetics' of dentition with the available dental treatments [9]

Therapeutic Approaches and Emerging Treatments:

Historically, OI management centered on supportive measures, including physical therapy and pain management. However optimal management needs a multidisciplinary approach includes a physician, physiotherapist, Orthotics, clinical psychologists, speech and language therapists, dieticians, and social workers. The gold standard treatment of Osteogenesis imperfecta is considered to be the use of bisphosphonates. (10) The Ultimate goal of bisphosphonate therapy is to counteract the high cellular turnover, inspite of the poor-quality collagen, and to increase the bone volume.[11].

The bisphosphonate in children is Pamidronate. The first study with Pamidronate in children with OI,[dose of 1.5–3 mg/kg for 3 consecutive days in 30 children, with repeated administrations at 4–6 months over 5 years] was given by Glorieux et al. in 1998, in 1992–1997. The new mode of treatment, is transplantation of mesenchymal stem cells (MSC) specifically starting treatment before birth or early after birth, to prevent irreversible damage Furthermore ongoing research delves into gene therapy, Bone anabolic agents, Denosumab and other innovative treatments, signaling a paradigm shift in OI management.[12]

Psychosocial Impact and Quality of Life Considerations:

Beyond the physical aspects, research increasingly recognizes the psychosocial impact of OI on individuals and their families. Studies delve into the emotional and social dimensions, exploring the quality of life (QoL) of those living with OI. Understanding these aspects is integral to providing holistic patient care and developing interventions that address the broader impact of the disorder.

Global Perspectives and Epidemiological Insights:

The prevalence of OI ranges from about 1:15,000 to 1:20,000 births[13]OI exhibits regional variations, and global perspectives are essential for a comprehensive understanding of the disorder. Epidemiological insights contribute to identifying genetic and environmental factors influencing OI incidence. This

global perspective aids in tailoring healthcare strategies to diverse populations and understanding the broader impact on healthcare systems.

Ongoing Research and Future Directions:

The landscape of OI research is dynamic, with ongoing efforts to uncover new genetic mutations, refine diagnostic criteria, and explore innovative therapeutic avenues. Collaborative initiatives across international research networks are shaping the future of OI research, emphasizing the need for continued exploration, cross-disciplinary collaboration, and the identification of new avenues for improved patient outcomes. The current understanding of OI in medical research is marked by significant strides in unraveling its genetic basis, characterizing its clinical spectrum, advancing diagnostic modalities, exploring novel therapies, recognizing psychosocial impacts, understanding global perspectives. This evolving knowledge base not only informs the scientific community but also holds the potential to enhance patient care and outcomes for those living with OI. Literature's Role in Shaping Perspectives Literature serves as a powerful medium for conveying the intricacies of the human experience, offering a unique lens through which to explore and understand complex issues such as Osteogenesis Imperfecta (OI). In this section, we delve into the significance of fiction in capturing human experiences, utilizing Jodi Picoult's "Handle With Care" as a narrative lens to shed light on the real-world implications of OI. Fiction, with its emotive storytelling and character-driven narratives, has the ability to transcend the clinical and scientific aspects of medical conditions. It allows readers to vicariously experience the challenges, triumphs, and emotional nuances of individuals living with rare disorders like OI. By delving into the emotional landscapes of characters, fiction provides a more profound understanding of the psychosocial dimensions that often elude conventional medical discourse. This section emphasizes the transformative power of fiction in fostering empathy and enriching perspectives on the human condition. Jodi Picoult's Handle With Care stands as a poignant example of literature's capacity to illuminate the realities of living with a rare medical condition. Through the O'Keefe family's journey, Picoult intricately weaves a narrative that explores the emotional, ethical, and societal complexities surrounding OI. The novel serves as a narrative lens, providing readers with a deeply personal and relatable insight into the challenges faced by families touched by OI. By leveraging the power of storytelling, Handle With Care acts as a bridge between medical facts and the lived experiences of those affected.

The thematic elements within Handle With Care offer a mirror to the real-world implications of OI, transcending the fictional realm to echo the challenges faced by individuals living with the disorder. Themes such as resilience, the impact on familial dynamics,

ethical dilemmas associated with medical decisions, and societal perceptions of disability are delicately woven into the narrative. By analyzing these themes, we gain insights into the emotional and social dimensions of OI, enabling a more holistic understanding that extends beyond clinical parameters. In examining Jodi Picoult's novel, we recognize its role as a powerful narrative tool, providing a textured portrayal of the human experiences associated with OI. Through this exploration, we aim to bridge the gap between medical research and the nuanced realities depicted in literature, fostering a more empathetic and comprehensive approach to understanding and addressing the multifaceted implications of OI.

Conducting a comparative analysis between medical narratives and the literary portrayal in *Handle with Care* allows for a nuanced examination of commonalities and disparities. By juxtaposing clinical findings with fictional narratives, we strive to extract valuable insights that can inform a more comprehensive and empathetic approach to OI. This comparative analysis serves as a bridge between the objective lens of medical research and the subjective, emotive storytelling within literature, enriching our overall understanding of OI. In exploring these parallels, our intention is to foster a holistic perspective on OI, recognizing the interplay between empirical research and the emotive power of literature in shaping our understanding of this rare genetic disorder.

The Role of Storytelling in Medical and Literary Contexts Storytelling serves as a crucial bridge between the realms of medicine and literature, facilitating a nuanced understanding of complex issues such as health, illness, and the human condition. In both medical and literary contexts, the ethical use of storytelling contributes to a more comprehensive, empathetic, and responsible exploration of the challenges faced by individuals, particularly those dealing with medical conditions. This section delves into the significance of ethical storytelling in both domains. The role of Storytelling in both medical and literary contexts is pivotal in shaping a nuanced understanding of human experiences, particularly those associated with medical conditions. By upholding principles of privacy, validity, empathy, and constructive messaging, Storytelling contributes to a richer, more holistic exploration of health-related narratives, fostering a bridge between the scientific and humanistic dimensions of healthcare. **The Holistic Approach: Integrating Literature into Patient Care** The integration of literature into patient care marks a transformative approach that transcends traditional medical practices, acknowledging the significance of narrative and human experience in the healing process. In this section, we explore the value of incorporating literature into patient care, emphasizing the potential benefits for both healthcare providers

and patients. The practice of medicine has traditionally focused on the biological aspects of illness, often overlooking the intricate human stories behind medical conditions. Integrating literature into patient care brings forth the human element, recognizing that each patient is more than a collection of symptoms. Literary narratives provide a platform for patients to express their experiences, fears, and hopes, fostering a deeper understanding that goes beyond clinical data. Literature serves as a powerful tool to enhance communication between healthcare providers and patients. Narratives can serve as a common ground for dialogue, enabling patients to articulate their emotions and healthcare providers to comprehend the holistic impact of a medical condition. By sharing literary resources, healthcare professionals can bridge communication gaps, creating a more empathetic and collaborative care environment. Medical conditions, beyond their physical manifestations, have profound psychosocial implications. Literature provides a medium to explore these dimensions, offering healthcare providers a more holistic understanding of the emotional and social challenges patients face. Integrating this awareness into patient care plans allows for a comprehensive approach that addresses both the physiological and psychological aspects of illness. Integrating literature into medical education enhances the capacity of healthcare professionals to engage with patients on a deeper level. Educational initiatives that expose medical students and practitioners to relevant literary works cultivate a more empathetic and culturally sensitive healthcare workforce. This exposure broadens perspectives and encourages

CONCLUSION

The current state of medical understanding regarding OI reflects an amalgamation of genetic insights, clinical spectrum awareness, diagnostic advancements, and a burgeoning array of therapeutic approaches. However, true progress lies not solely in the accumulation of empirical data but in our ability to empathize with and comprehend the lived experiences of those affected by OI. The incorporation of literature into our understanding of OI becomes paramount, as it acts as a conduit for the emotional and social dimensions often overlooked in traditional medical discourse. In traversing the diverse landscapes of Osteogenesis Imperfecta (OI), from the intricacies of its genetic basis to the emotional complexities depicted in Jodi Picoult's *Handle With Care*, our exploration illuminates the transformative potential that arises from the integration of medical research and literature. This synthesis not only enhances our comprehension of the multifaceted nature of OI but also advocates for a paradigm shift in healthcare – one that embraces the holistic narratives of patients, transcending the confines of clinical data. Jodi Picoult's *Handle With Care* serves as a poignant reminder that behind every medical condition is a

human story, rich with emotion, ethical dilemmas, and societal complexities. The parallels drawn between research findings and the fictional narrative unveil shared threads of struggle, resilience, and familial dynamics, reinforcing the idea that literature serves as a reflective mirror, capturing the essence of real-world implications. In conclusion, the integration of literature into medical research and patient care transcends disciplinary boundaries, weaving a tapestry that recognizes the synergy between empirical knowledge and the emotive power of storytelling. By embracing this holistic perspective, we embark on a collective journey toward a healthcare landscape where compassion, understanding, and the human narrative stand as integral pillars in the pursuit of improved well-being for individuals and families facing rare and complex medical conditions like OI.

overlay panel Heeseog Kang, Smriti Aryal A.C., Joan C. Marini.

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