ANNOTATION

of the dissertation submitted for the degree of Doctor of Philosophy (PhD)
in the specialty 8D10102 – "Medicine" **Khasenkyzy Altyngul**

Topic: Improvement of surgical treatment and early rehabilitation of osteogenesis imperfecta in children

Relevance of the problem

Osteogenesis imperfecta (OI) is a hereditary connective tissue disorder characterized by impaired bone structure development, increased fragility, multiple pathological fractures, and various extraskeletal manifestations. The pathogenesis is based on a genetically determined defect in the synthesis of type I collagen, the main structural protein of bone tissue.

According to epidemiological studies, the global prevalence of OI is approximately 1 case per 15,000–20,000 newborns. In the Republic of Kazakhstan, according to the information system "Electronic Register of Patients," in 2025, 119 children aged 0–17 years with a diagnosis of OI were under dynamic observation. At the same time, the actual prevalence may differ from official statistics due to limited knowledge of genetic variants, availability of molecular genetic diagnostics, and difficulties in correlating genotype with clinical phenotype. Structural insufficiency of bone tissue in OI leads to deformities of long bones, impaired limb biomechanics, and frequent fractures. Therefore, priority areas of medical care include fracture prevention, deformity correction, restoration of functional capacity, and the use of effective surgical methods combined with early rehabilitation.

In modern clinical practice, a wide range of surgical methods for stabilizing long tubular bones using intramedullary devices is employed. Telescopic systems are considered the most promising due to their dynamic adaptation to bone growth in children, significantly reducing the frequency of complications and repeated interventions.

Despite the widespread use of modern technologies and implants, some patients remain at risk of postoperative complications and limited functional recovery. Although telescopic systems have a positive impact on clinical outcomes, there is still room for further improvement of surgical techniques and optimization of patient management algorithms.

Special attention should be given to the development and implementation of effective early postoperative rehabilitation programs. The global literature lacks sufficient systematic studies evaluating the impact of early rehabilitation on functional outcomes in children with OI, highlighting the need for comprehensive clinical research and optimized approaches.

Improving surgical strategies and developing effective early rehabilitation algorithms aimed at reducing complications and enhancing functional outcomes remain important and relevant tasks in modern orthopedics. This study contributes to the further development of this field.

Aim of the study

To improve treatment outcomes in children with osteogenesis imperfecta by developing and clinically evaluating an improved system of intramedullary osteosynthesis and an early postoperative rehabilitation algorithm.

Object of the study

Patients with osteogenesis imperfecta during treatment and after its completion.

Subject of the study

Development and clinical evaluation of surgical treatment methods and early rehabilitation in children with osteogenesis imperfecta.

Objectives of the study

1. To conduct a retrospective analysis of the results of conventional surgical treatment methods in children with OI and identify the causes of complications and unsatisfactory outcomes.
2. To develop an improved intramedullary osteosynthesis system for pathological fractures of long bones and evaluate its clinical effectiveness.
3. To assess the outcomes of using an intramedullary locking telescopic implant in patients with OI.
4. To develop and implement early postoperative rehabilitation algorithms and evaluate their impact on functional outcomes.

Scientific novelty of the study

-Key causes of complications in conventional surgical treatment were identified, including insufficient axial and rotational stability and mismatch between implant and bone growth.

-Requirements for intramedullary fixation in children were scientifically substantiated, leading to the development of an improved telescopic implant design that enhances stability (Patent No. 10590, 2025).

-The superiority of telescopic fixation over conventional methods was demonstrated, with a statistically significant reduction in complications and reoperations (40.0% vs 13.3%, $p < 0.05$).

-The dependence of functional outcomes on the timing of rehabilitation initiation was established.

-The effectiveness of a combined approach (stable fixation + early rehabilitation) was proven (GFAQ $p = 0.001$).

Scientific and practical significance

-An improved intramedullary telescopic implant has been introduced into clinical practice, increasing stability and reducing complications.

-An early postoperative rehabilitation algorithm has been developed and implemented, improving functional recovery and reducing rehabilitation time.

-A comprehensive approach improves treatment outcomes, reduces reoperations, and enhances quality of life.

-The results can be applied in pediatric traumatology, orthopedics, and rehabilitation centers.

-The findings are recommended for use in medical education and residency programs.

Main provisions for defense

-A locking telescopic intramedullary implant with full positioning has been developed, eliminating rotational instability and improving axial stability.

-The method reduces complications and improves functional outcomes.

-Early rehabilitation enhances patient functional capacity.

Implementation and approbation

The developed device and methods were implemented in the "Alana Multidisciplinary Clinic" (Astana). The results are used in medical education at Astana Medical University. The study was presented at multiple international and national conferences (2022–2024).

Publications: 1 Scopus-indexed article, 2 national journal articles, 5 conference abstracts, 1 patent, and 1 copyright certificate.

Author's Contribution

The author independently formulated the aim and objectives of the study and developed the research protocol. Primary clinical and diagnostic data were collected, and an electronic database was created for subsequent analysis. The author participated in patient diagnosis, selection of surgical treatment strategies, postoperative management, and analysis of clinical outcomes. Statistical processing of the data was performed, and the obtained results were described, with formulation of the scientific statements, conclusions, and final remarks of the dissertation.

In addition, the author actively contributed to the preparation of scientific publications on the research topic, published both in journals recommended by the Committee for Quality Assurance in Science and Higher Education of the Ministry of Science and Higher Education of the Republic of Kazakhstan and in international peer-reviewed journals. The author also participated in the development of presentation materials for international scientific conferences.

General Characteristics of Materials and Methods

The dissertation study was conducted in accordance with a developed plan for the examination and treatment of patients with osteogenesis imperfecta (OI). Approval for the study was obtained from the local Ethics Committee of the NJSC "Astana Medical University" (Protocol No. 11, November 14, 2023).

The study was divided into five stages:

Stage I – A search and analysis of contemporary literature on epidemiology, classification, etiology, pathogenesis, diagnosis, and treatment methods of OI in children. The search was conducted in international scientific databases, including PubMed, Elsevier, Orphanet, Cochrane Library, TripDatabase, BMJ Best Practice, Google Scholar, as well as national resources such as eLIBRARY and RSCI.

Stage II – A retrospective analysis of medical records to identify the causes of complications following conventional surgical treatment methods. The analysis included records of 12 patients with OI treated between 2021 and 2023. The clinical base of the study was the

Department of Pediatric Orthopedics No. 2 of the Corporate Fund "University Medical Center" (Astana).

Stage III – Development and clinical implementation of an intramedullary locking telescopic implant. A utility model patent was obtained (Patent of the Republic of Kazakhstan No. 10590 dated May 23, 2025) for the device "Intramedullary Locking Telescopic Implant for Tubular Bones". Implementation certificates confirming its clinical use were obtained.

Stage IV – Development, optimization, and implementation of an early postoperative rehabilitation algorithm aimed at improving patients' functional outcomes after surgical treatment. A certificate confirming registration of the algorithm as an intellectual property object was obtained (No.69032 dated March 20, 2026).

Statistical analysis was performed in accordance with accepted standards using the SPSS software package (SPSS Statistics Version 27, USA), with additional analyses conducted using Posit PBC (formerly RStudio, PBC), USA.

Stage V – Comparative evaluation of the effectiveness of the proposed surgical method versus conventional approaches.

Study design: a combined (prospective–retrospective), non-randomized clinical study.

At this stage, 22 patients (30 segments) diagnosed with OI were included. Patients were divided into groups depending on the treatment method: Main group – 10 patients (15 segments) treated using the developed implant, observed at the "Center for Rare Bone Pathology" of the Alanda Multidisciplinary Clinic (Astana). Control group – 12 patients (15 segments) treated using conventional methods, observed at the Department of Orthopedics No. 2 of the "University Medical Center" CF (Astana). Treatment effectiveness was assessed in the postoperative period at 6, 12, 18, and 24 months. Functional outcomes were evaluated using the Gillette Functional Assessment Questionnaire (GFAQ) and the Hoffer-Bullock (H-B) scale.

Conclusions

1. After conventional surgical treatment, complications such as implant migration, the need for reoperation, cortical layer fracture, and local inflammatory complications were identified, and the causes of their occurrence were determined.
2. An intramedullary osteosynthesis system for the treatment of pathological fractures of long tubular bones was developed and implemented into clinical practice.
3. The use of the developed intramedullary osteosynthesis system provides axial stability of the bone, ensures accurate restoration of the biomechanical axis of the limbs, and reduces the risk of implant displacement or migration throughout the entire period of use.
4. The use of an intramedullary locking telescopic implant in combination with an early rehabilitation algorithm contributes to improved functional outcomes in patients.

Practical Recommendations

1. The intramedullary locking telescopic implant is recommended for use in the surgical treatment of osteogenesis imperfecta in children to achieve stable fixation of long tubular bones.
2. Early initiation of rehabilitation measures is recommended, using a systematic комплекс of sequential interventions individually adapted to each patient's condition.