A study based on the French Nationwide Hospitalization Database PMSI to characterize the Osteogenesis Imperfecta population between 2019 and 2023

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BACKGROUND

Osteogenesis imperfecta (OI) is a hereditary connective tissue disorder typically caused by mutations in genes involved in the biosynthesis and folding of type 1 collagen. A hallmark of the disease is frequent fractures resulting from minimal trauma[1].

National epidemiological studies on OI remain scarce. At the end of 2024, there were 3,610 patients diagnosed with OI in the French national rare diseases database, La Banque Nationale de Données Maladies Rares (BNDMR)[2].

Accordingly, the prevalence is estimated between 1:10,000 and 1:20,000 births[3].

OBJECTIVES

The objective of this study was to provide quantitative data about the clinical characteristics and healthcare use of this population over a 5-year period, using the French national database, Programme de Medicalisation des Systèmes d'Information (PMSI)[4].

METHODS

PMSI Database

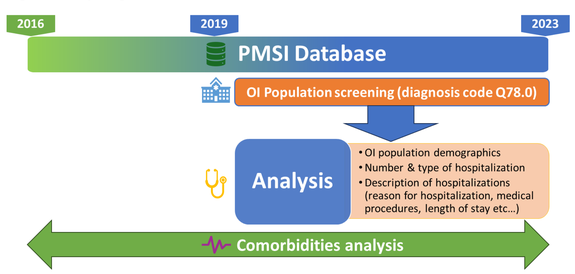
PMSI is the French Hospitalization Database of hospital stays. The database contains a record for each acute inpatient stay and represents about 25 million records per year in both public and private care facilities. Its use has been mandatory since 1996.

Clinical information, such as patient characteristics, diagnosis and medical procedures, is collected alongside the duration in hospital stay. Anonymous patient identifier enables record linkage.

Methodology

Data were extracted as OI using the specific ICD-10 diagnosis code Q78.0 over a 5-year period (2019–2023). The analysis included data on hospitalizations, medical procedures, and length of stay. Comorbidities data was also collected from an extended period (2016-2023), shown in **Figure 1**.

Figure 1. Study design



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1. Lafage-Proust MH et al. Joint Bone Spine. 2019 Oct;86(5):589-593.
2. Banque Nationale de Données Maladies Rares. Number of cases per rare disease registered in the BNDMR as of 3rd December 2024. https://www.bndmr.fr/publications/nombre-de-cas-par-mr/
3. ORPHANET https://www.orpha.net/en/disease/detail/216804
4. Boudemaghe T et al. Int J Epidemiol. 2017 Apr 1;46(2):392-392d.

RESULTS

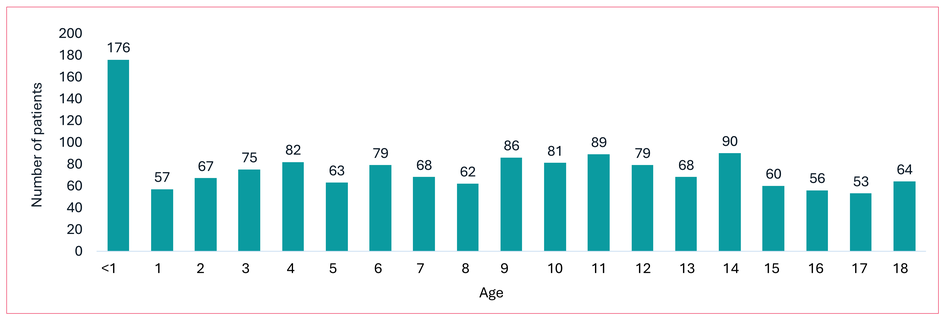
Patient Demographics

We identified a total of 2,971 patients with a median age of 19 years across all age groups (Min-Max: 0-96). 48.9% were aged ≤18 years, **Figure 2**.

Figure 2 Patient demographics

	Male	Female	Number of hospitalizations	Average length of stay
1,455 Patients ≤18 years	802 (55%)	653 (45%)	11,771	1.9 days
1,516 Patients > 18 years	802 (40%)	913 (60%)	11,555	2.7 days

Figure 3. Number of OI children by age at first hospitalisation between 2019 and 2023 (N = 1,455)

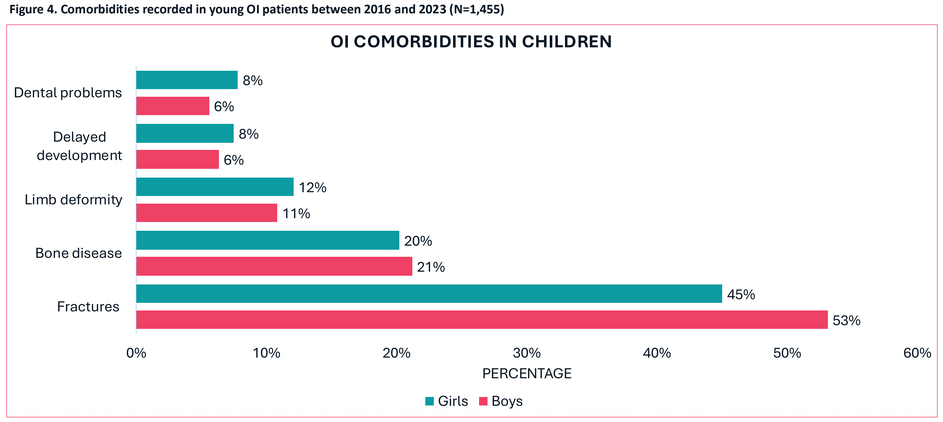


Comorbidities In Children And Adolescents

Between 2016 and 2023, **49%** of OI children were hospitalized with fractures, with **35%** of them having femur fractures.

Main comorbidities recorded were fractures, other bone diseases (e.g., anomalies of bone continuity, scoliosis, spondylopathies), limb deformities, delayed development and dental problems, **Figure 4**.

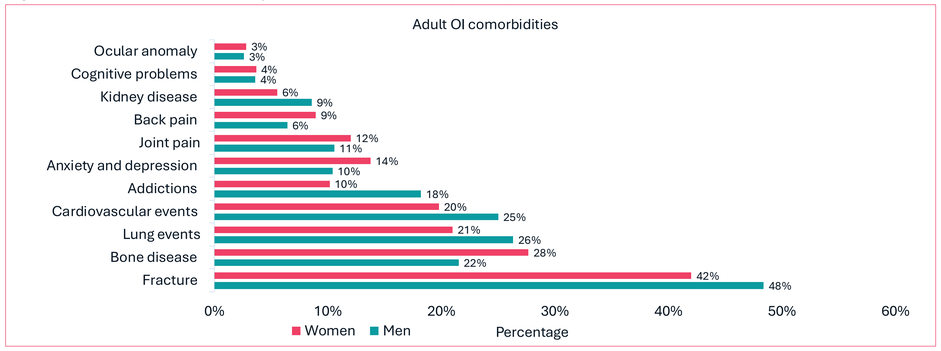
Figure 4. Comorbidities recorded in young OI patients between 2016 and 2023 (N=1,455)



Comorbidities in adults

In OI adult patients, the most frequent comorbidities recorded were fractures, bone disease (e.g. Osteoporosis, abnormalities of gait and mobility, scoliosis, arthropathy…), pain, respiratory and cardiovascular events, **Figure 5**.

Figure 5. Comorbidities recorded in Adult OI patients between 2016 and 2023 (N = 1,516)



Pregnancy

Between 2019 and 2023, 30% of women of childbearing age gave birth, half of them by caesarian section. Complications were recorded in 38% of childbirths such as premature labor (13%) and fetal distress (16%), **Figure 6** and **7**.

Figures 6. Pregnancy in OI women

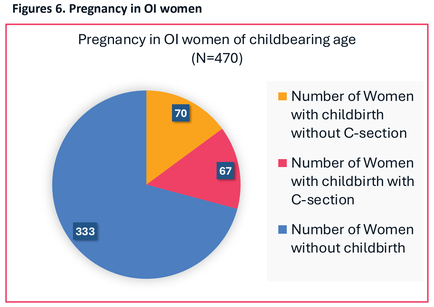
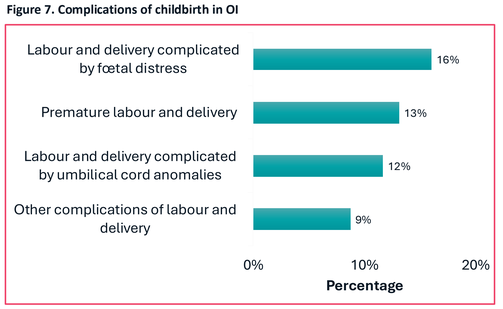


Figure 7. Complications of childbirth in OI



LIMITATIONS

The study was not designed to estimate OI prevalence even though the number of patients identified (2,971) is close to the prevalence of the BNDMR (3,610).

Using hospital discharge data (PMSI) likely underestimates the true number of certain OI related issues such as dental problems and other outpatient treated comorbidities.

CONCLUSIONS

The PMSI database provides valuable insights into the healthcare landscape in OI. Fractures remain a major burden across all age groups and both sexes, underscoring the fragility that characterises this condition. In children, complications include limb deformities and growth delays, while in adults, extra-skeletal manifestations and psychological challenges are more prevalent. These findings highlight the need for continued efforts to develop and implement more effective strategies to reduce fracture incidence and improve the quality of life for people with OI.

DISCLOSURES AND ACKNOWLEDGMENTS

MCS: received consultancy fees from Mereo Biopharma. JLG, HB: Employees Tekkare. OS, JS, AMD: Employees, Shareholders Mereo Biopharma