Real-World Healthcare Resource Utilization & Associated Drivers Among Patients With Osteogenesis Imperfecta

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Introduction

- OI is a rare genetic disorder of connective tissues caused by an abnormality in the synthesis or processing of type 1 collagen, leading to skeletal fragility and increased fracture risk[1]
- OI currently affects 1 in 15,000 to 20,000 births in the United States[1]
- The clinical spectrum of OI is highly heterogeneous, ranging from milder and nearly asymptomatic phenotypic presentations to severe forms comprised of large deformities, multiple fractures, and necessitating full caregiver support[1,2]
- The management of OI is complex and requires an interprofessional team-based approach. The need for this level of care and differences in phenotypic expression across the disease may potentially influence differences in patient HCRU patterns
- Potential differences in real-world HCRU patterns and the drivers associated among patients with OI are poorly characterized, limiting the ability to plan for care needs for both payers and health systems

Objective

- This study aims to characterize HCRU patterns and the potential reasons that may drive these patterns in patients with OI

Methods

- This retrospective observational study utilized Amplity AnswerY™, which is Amplity's proprietary real-world database and platform built from HIPAA-compliant transcriptions of US prescriber–patient visits. Using artificial intelligence and NLP, it extracts, visualizes, and summarizes treatment discussions and clinical decisions. Since 2017, AnswerY has covered inpatient and outpatient care across more than 70 specialties. Prior to January 2025, AnswerY was known as Amplity Insights
- NLP was used to search and analyze the AnswerY database and platform from 1732 providers for mentioned diagnosis of OI from January 1, 2017, to July 1, 2025
- HCRU was defined as mentions of or related to ED use, hospitalizations, and outpatient visits, and was quantified descriptively
- Drivers behind HCRU in patients with OI were reported qualitatively
- Based on HCRU, 2 subgroups clinically different in severity became apparent. Thus, disease burden scores and treatment patterns were also analyzed among the 2 subgroups

Strengths and Limitations

- Level and diffuse patterns of care for OI in this study mirror and are consistent with current literature, bolstering confidence in the robustness of the results
- As a rare disease, OI may be misdiagnosed or described poorly within the records of AnswerY, potentially influencing the results
- Severity classification is determined retrospectively, and as such, the study does not provide information on many potential factors to look for when proactively determining a new patient's severity status

Patients with OI are primarily younger, and are diagnosed with Type 1 disease

Patient Demographics

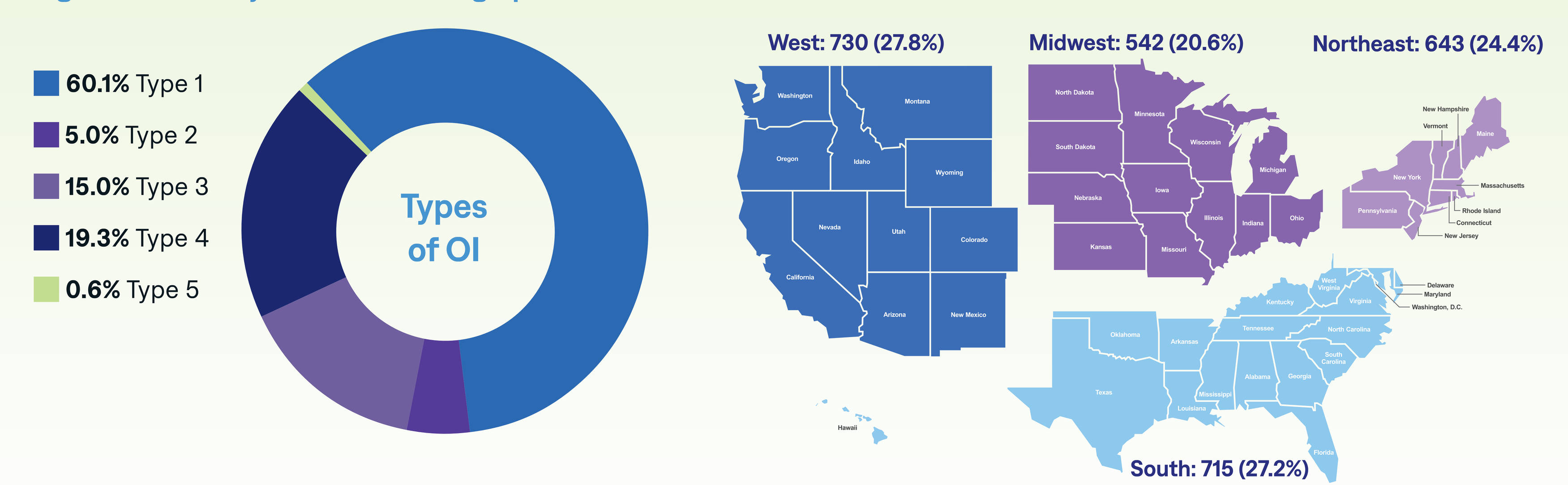
- AnswerY identified 2545 patients with OI from 1732 providers from 2017 to 2025
- **Table 1** and **Figure 1** show demographics and clinical characteristics of patients with OI
- Patients with OI were younger (age, mean ± SD: 38.7 ± 24.5 years), and of patients with a known ethnicity (24.5%), 88.3% of patients were of White ethnicity, 8.8% were of African-American ethnicity, and 2.9% were of Hispanic ethnicity
- Among patients with a known OI type, type 1 disease was the most common, with 60.1% (n=1462) of patients being classified underneath this category
  - Type 3 and type 4 disease were the next most common forms of OI observed. Additional forms were observed minimally
- Geographic distribution of patients were relatively even, with a slight favor towards the Western (27.8%) and Southern (27.2%) regions

Table 1: Demographics for Patients with OI Stratified by Severity[a]

	Overall	High Severity	Low Severity
Total Number of Patients	2545	1817	446
Age			
Patients with record, n (%)	1872 (73.6)	1284 (70.7)	300 (67.3)
Mean (SD)	37.8 (24.5)	38.4 (24.6)	26.1 (18.8)
Gender			
Patients with record, n (%)	1980 (77.8)	1574 (86.6)	406 (91.0)
Male, n (%)	907 (45.8)	737 (46.8)	170 (41.9)
Female, n (%)	1073 (54.2)	837 (53.2)	236 (58.1)
Ethnicity			
Patients with record, n (%)	623 (24.5)	528 (29.1)	49 (11.0)
African-American, n (%)	55 (8.8)	44 (8.3)	8 (16.3)
Hispanic, n (%)	18 (2.9)	14 (2.7)	0 (0)
White, n (%)	550 (88.3)	470 (89.0)	41 (83.7)

[a]When determining severity subgroups, 282 patients had an unknown or unclear disease severity.

Figure 1: Summary of Patient Demographics of All Patients with OI

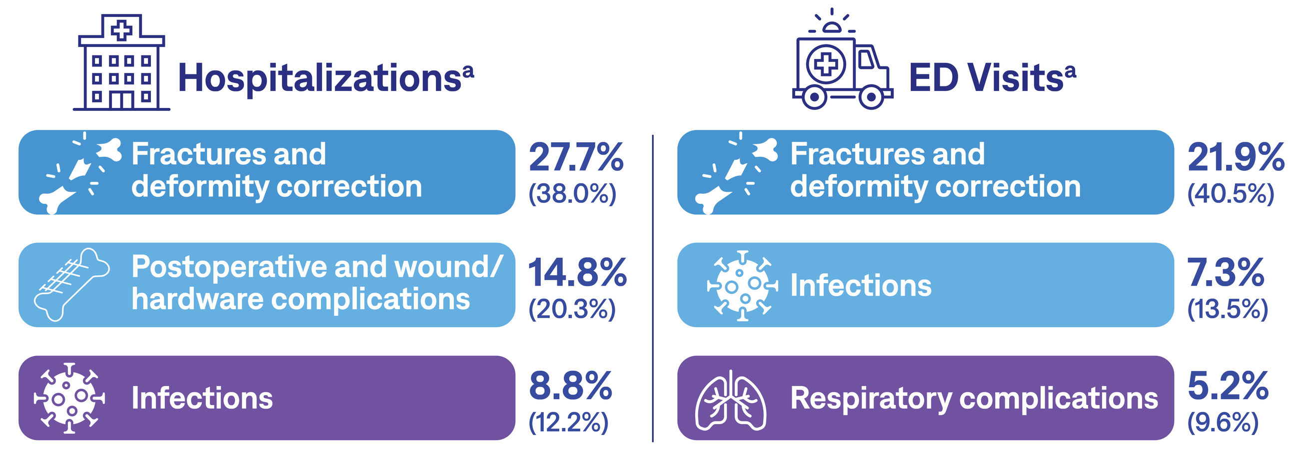


Patients with OI primarily seek medical care due to fractures, deformity correction, and infections

HCRU, Sequencing, and Disease Burden

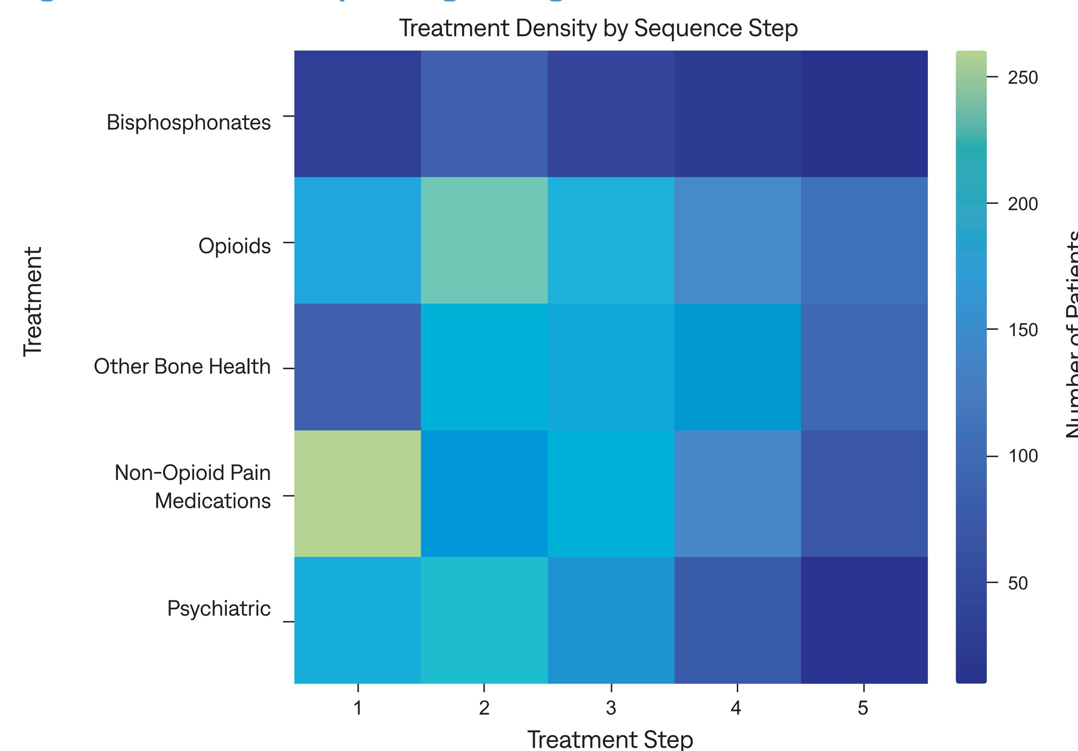
- **Figure 2** shows the drivers of hospitalizations and ED visits among all patients with OI
- Hospitalizations and ED visits among all patients with OI were primarily driven by fractures, deformity corrections, and infections
- In addition to the above, hospitalizations were also driven by postoperative and hardware complications, while ED visits were driven by respiratory complications

Figure 2: Drivers of HCRU Among Patients with OI



[a]Statistics are displayed as follows: percentage of full cohort (percentage of patients with a qualified HCRU visit).

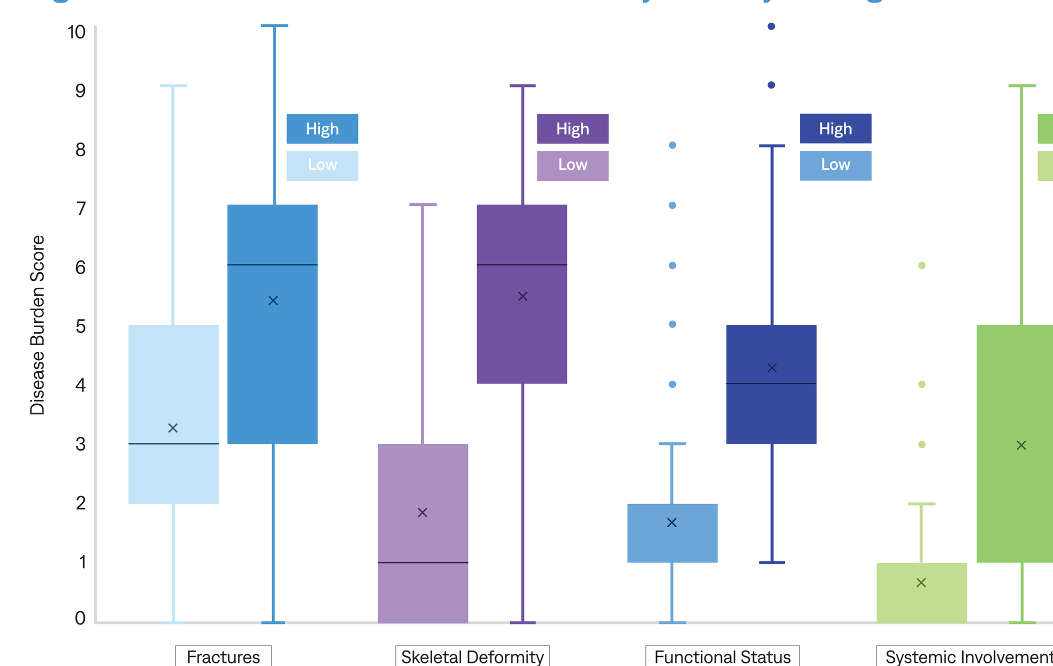
Figure 3: Treatment Sequencing Among Patients with OI



- **Figure 3** shows treatment sequencing among all patients with OI (higher values on the gradient represent greater treatment utilization)
- Management of OI is individualized and based on disease severity
- Consistent with what is known about treating patients with OI, patients in this cohort received symptomatic therapies first, suggesting a focus on symptom control and stabilization
- Bisphosphonates and other bone health agents peak at step 2, and are not commonly used in later lines, suggesting that their use tapers once benefits are maximized
- Psychiatric medications are prevalent in steps 1 and 2, implying some psychosocial impact associated with OI

- Based on disease burden scores, 2 distinct disease severity subgroups became apparent. **Figure 4** shows these scores stratified by disease severity. Horizontal lines and crosses represent the median and mean values, respectively
- Among patients who were quantifiable, patients with lower severity OI had a lower number of fractures compared with those with higher severity (lower severity vs higher severity; mean number of fractures: 2.23 vs 5.73)
- Among patients with OI, patients with lower severity disease also showed lower disease burden score overall in every category

Figure 4: Disease Burden Scores Stratified by Severity Among Patients with OI

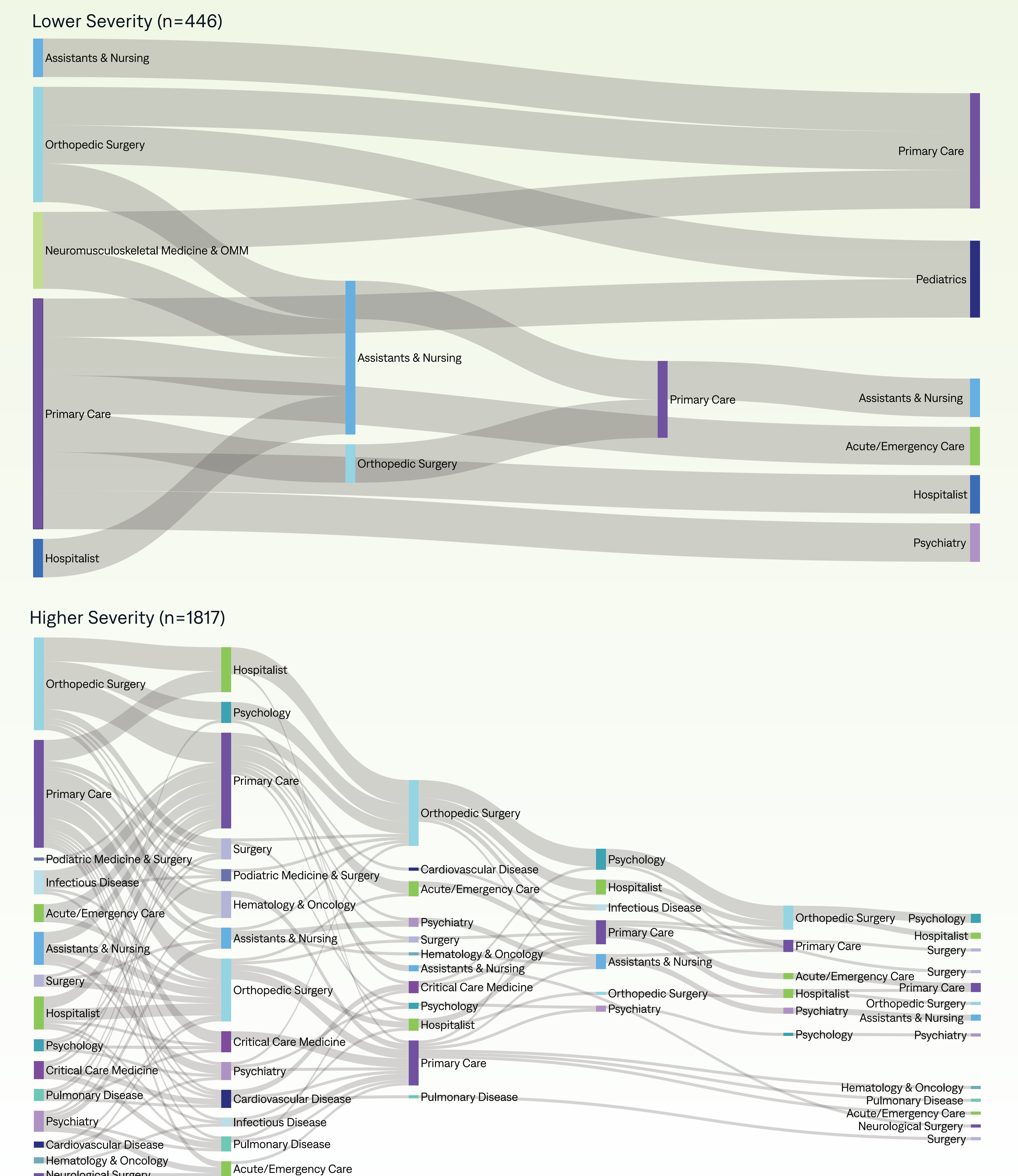


Overall, patients with OI have complex HCRU patterns. However, the complexity in transitions of care is primarily observed in patients with high severity disease

Transitions of Care

- The analysis identified 2 subgroups based on HCRU. **Figure 5** shows the transitions of care pathways among patients with OI stratified by severity
- A lower severity subgroup (n=446) experienced lower HCRU with periodic specialist visits, whereas a higher severity subgroup (n=1817) experienced high rates of recurrent fractures, multiple surgeries, and multidisciplinary outpatient/post-acute care
- Compared with the lower severity subgroup, the higher severity subgroup had more patients with ≥1 documented OI-associated ED visits (33.4% vs 11.9%) and hospitalizations (51.6% vs 6.1%)

Figure 5: Transitions of Care Among Patients with OI



Conclusions

- In this real-world US cohort, all patients with OI have a high HCRU burden driven directly by the disease and indirectly through complications
- Across the entire cohort, observed treatment patterns were consistent with what is known for this patient population, with symptom control preceding disease-modifying treatments
- Two distinct subgroups based on HCRU were identified in this study, with a majority of patients falling into the higher severity subgroup
- High disease burden scores and early maximization of certain treatment-related benefits highlight the lifelong, pervasive impact of OI on patients, while the multidisciplinary care approach outlined in treatment pathways similarly emphasizes the significant strain on healthcare providers and systems
- The findings above highlight the heterogeneity of disease within patients with OI and support a multidisciplinary and tailored approach toward treatment, particularly among high-burden subgroups
- Furthermore, there may be a benefit to mirroring the tailored approach to treatment with a similar effort toward designing care pathways, payer policies, and reimbursement frameworks

DISCLOSURES

DI, RV, EM, and MJ* are employees of Amplity Inc.

*Presenting Author

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ABBREVIATIONS

ED, emergency department; HCRU, healthcare resource utilization; NLP, natural language processing; OI, osteogenesis imperfecta; OMM, osteopathic manipulative medicine