

Fractures in Children with Osteogenesis Imperfecta: Analysis Over a 10-Year Follow-Up Period

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INTRODUCTION: Osteogenesis Imperfecta (OI) is a rare genetic collagen disorder that results in a myriad of clinical manifestations in pediatric patients. From an orthopaedic standpoint, weak bone density and an increased risk of multiple bone fractures pose an elevated risk of mortality in these children. This large study investigates the occurrences of various fracture manifestations in OI patients.

METHODS:

A comprehensive multinational database was queried for deidentified patient records between 2003-2022. Children with a diagnosis of OI with a minimum of 10-year follow up were gathered and compared to children without an OI diagnosis that experienced fractures. Statistical analysis was performed and statistical significance was determined at 0.05.

RESULTS: A total of 1,216,357 fractures were identified, among which 1,967 (0.16%) patients with a diagnosis of OI are compared to 1,214,390 (99.84%) patients without an OI. The age at index fracture was 0.2 and 0.3 years, respectively. A higher proportion of patients in the OI cohort are White and female (P <0.01). Longitudinally, OI patients experienced higher rates (P <0.0001) of skull and facial bone (HR = 8.52, 95% CI [6.04-12.01]), cervical vertebra and other parts of the neck (HR = 10.34, 95% CI [5.92-18.04]), ribcage and thoracic spine (HR = 28.83, 95% CI [17.32-47.98]), lumbar spine and pelvis (HR = 19.08, 95% CI [11.32-32.16]), shoulder and upper arm (HR = 11.12, 95% CI [8.76-14.11]), forearm (HR = 8.69, 95% CI [6.98-10.81]), wrist and hand (HR = 6.43, 95% CI [4.89-8.45]), femur (HR = 66.12, 95% CI [38.82-112.62]), lower leg to ankle (HR = 22.83, 95% CI [17.07-30.53]), and foot and toe fractures (HR = 10.83, 95% CI [7.78-15.06]).

DISCUSSION AND CONCLUSION: This is one of the largest studies to-date investigating fracture patterns in children with Osteogenesis Imperfecta. Compared to healthy patients, children with OI are highly susceptible to fractures in various locations in the body including the appendicular and axial skeleton.

Measure	Risk Ratio	95% CI	LB	UB	P-Value
skull and facial bones	5.225	(3.747, 7.286)	3.747	7.286	0.000
cervical vertebra and other parts of neck	5.733	(3.325, 9.886)	3.325	9.886	0.000
rib(s), sternum and thoracic spine	16.063	(9.731, 26.513)	9.731	26.513	0.000
lumbar spine and pelvis	9.188	(5.505, 15.333)	5.505	15.333	0.000
shoulder and upper arm	5.566	(4.444, 6.972)	4.444	6.972	0.000
forearm	3.565	(2.907, 4.372)	2.907	4.372	0.000
wrist and hand level	2.395	(1.846, 3.107)	1.846	3.107	0.000
femur	38.143	(22.519, 64.606)	22.519	64.606	0.000
lower leg, including ankle	10.902	(8.241, 14.423)	8.241	14.423	0.000
foot and toe, except ankle	4.886	(3.555, 6.717)	3.555	6.717	0.000