

CITATION REPORT

List of articles citing

**Osteogenesis imperfecta: clinical diagnosis,
nomenclature and severity assessment**

DOI: 10.1002/ajmg.a.36545

**American Journal of Medical Genetics, Part A, 2014,
164A, 1470-81.**

Source: <https://exaly.com/paper-pdf/58817198/citation-report.pdf>

Version: 2024-04-28

This report has been generated based on the citations recorded by exaly.com for the above article. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

#	Paper	IF	Citations
454	Type I procollagen C-propeptide defects: study of genotype-phenotype correlation and predictive role of crystal structure. 2014 , 35, 1330-41		27
453	Bisphosphonate therapy in pediatric patients. 2014 , 13, 109		23
452	The ever-expanding conundrum of primary osteoporosis: aetiopathogenesis, diagnosis, and treatment. 2014 , 40, 55		5
451	Osteogenesis imperfecta: diagnosis and treatment. 2014 , 12, 279-88		44
450	CRTAP mutation in a patient with Cole-Carpenter syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 587-91	2.5	13
449	Prenatal Diagnosis of Skeletal Dysplasias and Connective Tissue Disorders. 2015 , 681-699		
448	New Genetic Forms of Childhood-Onset Primary Osteoporosis. 2015 , 84, 361-9		22
447	Osteogenesis Imperfecta A Tale of 50 Years. 2015 , 52, 1073-4		1
446	Molecular Outcome, Prediction, and Clinical Consequences of Splice Variants in COL1A1, Which Encodes the pro α 1(I) Chains of Type I Procollagen. 2015 , 36, 728-39		16
445	Hindlimb Skeletal Muscle Function and Skeletal Quality and Strength in +/G610C Mice With and Without Weight-Bearing Exercise. 2015 , 30, 1874-86		15
444	Nosology and classification of genetic skeletal disorders: 2015 revision. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2869-92	2.5	380
443	Initial report of the osteogenesis imperfecta adult natural history initiative. <i>Orphanet Journal of Rare Diseases</i> , 2015 , 10, 146	4.2	29
442	Genotype and phenotype analysis of Taiwanese patients with osteogenesis imperfecta. <i>Orphanet Journal of Rare Diseases</i> , 2015 , 10, 152	4.2	21
441	Stem cell transplantation before birth - a realistic option for treatment of osteogenesis imperfecta?. 2015 , 35, 827-32		18
440	Bone structure assessed by HR-pQCT, TBS and DXL in adult patients with different types of osteogenesis imperfecta. 2015 , 26, 2431-40		33
439	Clinical and Molecular Characterization of Osteogenesis Imperfecta Type V. 2015 , 6, 164-72		17
438	Cole-Carpenter syndrome is caused by a heterozygous missense mutation in P4HB. 2015 , 96, 425-31		65

437	Mutations in SEC24D, encoding a component of the COPII machinery, cause a syndromic form of osteogenesis imperfecta. 2015 , 96, 432-9	104
436	Interdisciplinary Care Improves Functional Mobility in an Individual with Type IX Osteogenesis Imperfecta. 2015 , 11, 84-9	
435	Local amino acid sequence patterns dominate the heterogeneous phenotype for the collagen connective tissue disease Osteogenesis Imperfecta resulting from Gly mutations. 2015 , 192, 127-37	13
434	[Diseases revealed by the mouth]. 2015 , 22, 151-2	
433	A look behind the scenes: the risk and pathogenesis of primary osteoporosis. 2015 , 11, 462-74	141
432	The dynamics of adult haematopoiesis in the bone and bone marrow environment. 2015 , 170, 472-86	24
431	Differential diagnosis and diagnostic flow chart of joint hypermobility syndrome/ehlers-danlos syndrome hypermobility type compared to other heritable connective tissue disorders. 2015 , 169C, 6-22	75
430	Pregnancy outcomes in women with osteogenesis imperfecta. 2016 , 29, 2358-62	5
429	Molecular Consequences of the SERPINH1/HSP47 Mutation in the Dachshund Natural Model of Osteogenesis Imperfecta. 2015 , 290, 17679-17689	33
428	Quality of life in caregivers of children and adolescents with Osteogenesis Imperfecta. 2015 , 13, 41	16
427	Classification of osteogenesis imperfecta. 2015 , 165, 264-70	24
426	Altered cytoskeletal organization characterized lethal but not surviving Brtl ^{+/-} mice: insight on phenotypic variability in osteogenesis imperfecta. 2015 , 24, 6118-33	18
425	Perinatal Diagnostic Approach to Fetal Skeletal Dysplasias: Six Years Experience of a Tertiary Center. 2015 , 34, 287-306	6
424	Molecular diagnosis of hypophosphatasia and differential diagnosis by targeted Next Generation Sequencing. 2015 , 116, 215-20	47
423	Identification and in vivo functional characterization of novel compound heterozygous BMP1 variants in osteogenesis imperfecta. 2015 , 36, 191-5	22
422	Vascular Pathobiology. 2016 , 85-124	6
421	Genetic Diagnosis of Skeletal Dysplasias. 2016 , 173-189	
420	Anesthetic Management in a Gravida with Type IV Osteogenesis Imperfecta. 2016 , 2016, 7429251	2

419	Multiparametric Classification of Skin from Osteogenesis Imperfecta Patients and Controls by Quantitative Magnetic Resonance Microimaging. 2016 , 11, e0157891	4
418	Recurrent Proximal Femur Fractures in a Teenager With Osteogenesis Imperfecta on Continuous Bisphosphonate Therapy: Are We Overtreating?. 2016 , 31, 1449-54	27
417	Mutations in SEC24D cause autosomal recessive osteogenesis imperfecta. 2016 , 89, 517-519	15
416	Evidence for a Role for Nanoporosity and Pyridinoline Content in Human Mild Osteogenesis Imperfecta. 2016 , 31, 1050-9	29
415	THE CLINICAL CHARACTERISTICS AND EFFICACY OF BIPHOSPHONATES IN ADULT PATIENTS WITH OSTEOGENESIS IMPERFECTA. 2016 , 22, 1267-1276	4
414	EFFECTS OF LONG-TERM ALENDRONATE TREATMENT ON A LARGE SAMPLE OF PEDIATRIC PATIENTS WITH OSTEOGENESIS IMPERFECTA. 2016 , 22, 1369-1376	6
413	Orthopaedic Considerations for the Adult With Osteogenesis Imperfecta. 2016 , 24, 298-308	26
412	Cardiological assessment of a cohort of Egyptian patients with osteogenesis imperfecta type III. 2016 , 17, 197-200	0
411	Worsening of Callus Hyperplasia after Bisphosphonate Treatment in Type V Osteogenesis Imperfecta. 2016 , 53, 250-2	7
410	Osteopatías con alteraciones de la densidad ósea. 2016 , 49, 1-12	
409	Premature Atherosclerosis and Drug Eluting Stent Restenosis in an Adult with Osteogenesis Imperfecta. 2016 , 25, e166-e168	1
408	Asymptomatic parental mosaicism for osteogenesis imperfecta associated with a new splice site mutation in. 2016 , 4, 972-978	3
407	Cardiovascular disease in patients with osteogenesis imperfecta - a nationwide, register-based cohort study. 2016 , 225, 250-257	15
406	Osteogenesis imperfecta in children and adolescents-new developments in diagnosis and treatment. 2016 , 27, 3427-3437	102
405	Mortality and Causes of Death in Patients With Osteogenesis Imperfecta: A Register-Based Nationwide Cohort Study. 2016 , 31, 2159-2166	55
404	Quality of life in osteogenesis imperfecta: A mixed-methods systematic review. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 62-76	2.5 37
403	Studies of chain substitution caused sub-fibril level differences in stiffness and ultrastructure of wildtype and oim/oim collagen fibers using multifrequency-AFM and molecular modeling. 2016 , 107, 15-22	17
402	Atypical femur fracture in an adolescent boy treated with bisphosphonates for X-linked osteoporosis based on PLS3 mutation. 2016 , 91, 148-51	20

401	DNA sequence analysis in 598 individuals with a clinical diagnosis of osteogenesis imperfecta: diagnostic yield and mutation spectrum. 2016 , 27, 3607-3613		85
400	Diagnostic conundrums in antenatal presentation of a skeletal dysplasia with description of a heterozygous C-propeptide mutation in COL1A1 associated with a severe presentation of osteogenesis imperfecta. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 3303-3307	2.5	2
399	Pharmacological and biological therapeutic strategies for osteogenesis imperfecta. 2016 , 172, 367-383		35
398	Clinical characteristics and the identification of novel mutations of COL1A1 and COL1A2 in 61 Chinese patients with osteogenesis imperfecta. 2016 , 14, 4918-4926		14
397	Mutation analysis of the COL1A1 and COL1A2 genes in Vietnamese patients with osteogenesis imperfecta. 2016 , 10, 27		24
396	Skeletal phenotypes in adult patients with osteogenesis imperfecta-correlations with COL1A1/COL1A2 genotype and collagen structure. 2016 , 27, 3331-3341		21
395	An overlapping phenotype of Osteogenesis imperfecta and Ehlers-Danlos syndrome due to a heterozygous mutation in COL1A1 and biallelic missense variants in TNXB identified by whole exome sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 1080-5	2.5	14
394	Study of the Determinants of Vitamin D Status in Pediatric Patients With Osteogenesis Imperfecta. 2016 , 35, 339-45		4
393	Value of rare low bone mass diseases for osteoporosis genetics. 2016 , 5, 773		7
392	Bisphosphonates: Pharmacokinetics, bioavailability, mechanisms of action, clinical applications in children, and effects on tooth development. 2016 , 42, 212-7		32
391	SPARC/osteonectin in mineralized tissue. 2016 , 52-54, 78-87		117
390	Advances in the Classification and Treatment of Osteogenesis Imperfecta. 2016 , 14, 1-9		55
389	Skeletal Dysplasias. 2016 , 255-409		2
388	Dental panoramic indices and fractal dimension measurements in osteogenesis imperfecta children under pamidronate treatment. 2016 , 45, 20150400		52
387	The psychosocial experience of individuals living with osteogenesis imperfecta: a mixed-methods systematic review. 2016 , 25, 1877-96		33
386	Two novel mutations in TMEM38B result in rare autosomal recessive osteogenesis imperfecta. 2016 , 61, 539-45		18
385	Genetic control of bone mass. 2016 , 432, 3-13		41
384	Myostatin deficiency partially rescues the bone phenotype of osteogenesis imperfecta model mice. 2016 , 27, 161-70		21

383	Developmental charts for children with osteogenesis imperfecta, type I (body height, body weight and BMI). 2017 , 176, 311-316	12
382	Genetic causes and mechanisms of Osteogenesis Imperfecta. 2017 , 102, 40-49	58
381	TGF- β Family Signaling in Connective Tissue and Skeletal Diseases. 2017 , 9,	51
380	Clinical and Molecular Heterogeneity of Osteogenesis Imperfecta. 2017 , 6, 1-63	
379	The Spine in Patients With Osteogenesis Imperfecta. 2017 , 25, 100-109	24
378	Perioperative Management of Pediatric Patients with Osteogenesis Imperfecta Undergoing Orthopedic Procedures. 2017 , 7, 142-149	2
377	Phenotypic Spectrum in Osteogenesis Imperfecta Due to Mutations in TMEM38B: Unraveling a Complex Cellular Defect. 2017 , 102, 2019-2028	14
376	Compound heterozygous mutations in COL1A1 associated with an atypical form of type I osteogenesis imperfecta. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1907-1912	2.5 8
375	Novel missense loss-of-function mutations of WNT1 in an autosomal recessive Osteogenesis imperfecta patient. 2017 , 60, 411-415	9
374	Osteoporosis in childhood. 2017 , 29, 535-546	16
373	Bisphosphonate Treatment and the Characteristics of Femoral Fractures in Children With Osteogenesis Imperfecta. 2017 , 102, 1333-1339	11
372	A novel mutation in a family with osteogenesis imperfecta associated with phenotypic variabilities. 2017 , 4, 17007	3
371	MECHANISMS IN ENDOCRINOLOGY: Genetics of human bone formation. 2017 , 177, R69-R83	21
370	Novel mutations in the SEC24D gene in Chinese families with autosomal recessive osteogenesis imperfecta. 2017 , 28, 1473-1480	14
369	An ENU-induced splice site mutation of mouse Col1a1 causing recessive osteogenesis imperfecta and revealing a novel splicing rescue. 2017 , 7, 11717	2
368	Osteogenesis imperfecta: diagnosis and treatment. 2017 , 24, 381-388	42
367	Gene mutation spectrum and genotype-phenotype correlation in a cohort of Chinese osteogenesis imperfecta patients revealed by targeted next generation sequencing. 2017 , 28, 2985-2995	34
366	PLS3 sequencing in childhood-onset primary osteoporosis identifies two novel disease-causing variants. 2017 , 28, 3023-3032	24

365	PLS3 Deletions Lead to Severe Spinal Osteoporosis and Disturbed Bone Matrix Mineralization. 2017 , 32, 2394-2404		23
364	Novel COL1A1 Mutation c.3290G>T Associated With Severe Form of Osteogenesis Imperfecta in a Fetus. 2017 , 20, 455-459		3
363	Pamidronate "zebra lines": A treatment timeline. 2017 , 12, 850-853		4
362	Health-Related Quality of Life in Adults with Osteogenesis Imperfecta. <i>Calcified Tissue International</i> , 2017 , 101, 473-478	3.9	17
361	The clinical features of osteogenesis imperfecta in Vietnam. 2017 , 41, 21-29		11
360	Delineation of Ehlers-Danlos syndrome phenotype due to the c.934C>T, p.(Arg312Cys) mutation in COL1A1: Report on a three-generation family without cardiovascular events, and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 524-530	2.5	29
359	Using GWAS to identify novel therapeutic targets for osteoporosis. 2017 , 181, 15-26		28
358	Tooth agenesis in osteogenesis imperfecta related to mutations in the collagen type I genes. 2017 , 23, 42-49		23
357	Fracture Rates and Fracture Sites in Patients With Osteogenesis Imperfecta: A Nationwide Register-Based Cohort Study. 2017 , 32, 125-134		60
356	Craniofacial manifestations in osteogenesis imperfecta type III in South Africa. 2017 , 3, 17021		4
355	Cole-Carpenter syndrome-1 with a de novo heterozygous deletion in the P4HB gene in a Chinese girl: A case report. 2017 , 96, e9504		7
354	WHOLE-BODY VIBRATION EXERCISE IMPROVES FUNCTIONAL PARAMETERS IN PATIENTS WITH A SYSTEMATIC REVIEW WITH A SUITABLE APPROACH. 2017 , 14, 199-208		10
353	Efficacy of Denosumab for Osteoporosis in Three Female Patients with Osteogenesis Imperfecta. 2017 , 242, 115-120		14
352	CLINICAL FEATURES AND PATTERN OF FRACTURES AT THE TIME OF DIAGNOSIS OF OSTEOGENESIS IMPERFECTA IN CHILDREN. 2017 , 35, 171-177		6
351	Mutational analysis of COL1A1 and COL1A2 genes among Estonian osteogenesis imperfecta patients. 2017 , 11, 19		24
350	Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. 2018 , 110, 368-377		21
349	Osteogenesis Imperfecta Type I: Recognition in Primary Care. 2018 , 14, 470-476		
348	Pain and quality of life of children and adolescents with osteogenesis imperfecta over a bisphosphonate treatment cycle. 2018 , 177, 891-902		11

347	Fracture Patterns Differ Between Osteogenesis Imperfecta and Routine Pediatric Fractures. 2018 , 38, e207-e212		12
346	Difference between Methods for Estimation of Basal Metabolic Rate and Body Composition in Pediatric Patients with Osteogenesis Imperfecta. 2018 , 72, 21-29		5
345	Health-related quality of life of children and adolescents with osteogenesis imperfecta: a cross-sectional study using PedsQL 2018 , 18, 95		8
344	Altered corneal biomechanical properties in children with osteogenesis imperfecta. 2018 , 22, 183-187.e1		5
343	Pain experiences of adults with osteogenesis imperfecta: An integrative review.. 2018 , 2, 9-20		4
342	Autosomal Recessive Osteogenesis Imperfecta Caused by a Novel Homozygous COL1A2 Mutation. <i>Calcified Tissue International</i> , 2018 , 103, 353-358	3.9	8
341	Expanding the Clinical Spectrum of Phenotypes Caused by Pathogenic Variants in PLOD2. 2018 , 33, 753-760		12
340	Genetic analysis of osteogenesis imperfecta in the Palestinian population: molecular screening of 49 affected families. <i>Molecular Genetics & Genomic Medicine</i> , 2018 , 6, 15-26	2.3	20
339	MicroRNAs in Bone Diseases: Progress and Prospects. 2018 ,		
338	Implant therapy for a patient with osteogenesis imperfecta type I: review of literature with a case report. 2018 , 4, 36		2
337	Analyses of LMNA-negative juvenile progeroid cases confirms biallelic POLR3A mutations in Wiedemann-Rautenstrauch-like syndrome and expands the phenotypic spectrum of PYCR1 mutations. 2018 , 137, 921-939		15
336	Efficacy and Safety of Denosumab Therapy for Osteogenesis Imperfecta Patients with Osteoporosis-Case Series. 2018 , 7,		13
335	Clinical, Genetics, and Bioinformatic Characterization of Mutations Affecting an Essential Region of in Patients with BMND18. 2018 , 2018, 8953217		5
334	Anesthesia in children with osteogenesis imperfecta: Retrospective chart review of 83 patients and 205 anesthetics over 7 years. 2018 , 28, 1050-1058		9
333	Cyclic bisphosphonate therapy reduces pain and improves physical functioning in children with osteogenesis imperfecta. <i>BMC Musculoskeletal Disorders</i> , 2018 , 19, 344	2.8	14
332	A moderate form of osteogenesis imperfecta caused by compound heterozygous mutations. 2018 , 9, 132-135		5
331	Case 3: A 10-year-old Boy with Saber Shins. 2018 , 39, 523-526		
330	Use of flexible intramedullary nailing in combination with an external fixator for a postoperative defect and pseudarthrosis of femur in a girl with osteogenesis imperfecta type VIII: a case report. 2018 , 13, 191-197		1

329	The dental perspective on osteogenesis imperfecta in a Danish adult population. 2018 , 18, 175	5
328	Brief Review on Metabolic Bone Disease. 2018 , 8, 611-640	8
327	Pauwels Osteotomy for Femoral Neck Nonunion in Two Adult Siblings with Osteogenesis Imperfecta. 2018 , 30, 53-59	2
326	Ribcage deformity and the altered breathing pattern in children with osteogenesis imperfecta. 2018 , 53, 964-972	14
325	A comparative study of quality of life, functional and bone outcomes in osteogenesis imperfecta with bisphosphonate therapy initiated in childhood or adulthood. 2018 , 113, 137-143	10
324	Skeletal Dysplasias. 2018 , 175-196	
323	Mechanical elasticity of proline-rich and hydroxyproline-rich collagen-like triple-helices studied using steered molecular dynamics. 2018 , 86, 105-112	16
322	Adults with osteogenesis imperfecta: Clinical characteristics of 151 patients with a focus on bisphosphonate use and bone density measurements. 2018 , 8, 168-172	8
321	Cumulative radiation exposure from medical imaging and associated lifetime cancer risk in children with osteogenesis imperfecta. 2018 , 114, 252-256	2
320	Splicing defect in FKBP10 gene causes autosomal recessive osteogenesis imperfecta disease: a case report. 2018 , 19, 86	3
319	Effects of a telescopic intramedullary rod for treating patients with osteogenesis imperfecta of the femur. 2018 , 12, 97-103	12
318	Uncommon IFITM5 mutation associated with severe skeletal deformity in osteogenesis imperfecta. 2018 , 82, 477-481	7
317	Rare Copy Number Variants in Array-Based Comparative Genomic Hybridization in Early-Onset Skeletal Fragility. <i>Frontiers in Endocrinology</i> , 2018 , 9, 380	5-7 11
316	Multifaced Roles of the $\alpha 1$ Integrin in Ehlers-Danlos and Arterial Tortuosity Syndromes' Dermal Fibroblasts. 2018 , 19,	14
315	From Structure to Phenotype: Impact of Collagen Alterations on Human Health. 2018 , 19,	59
314	Osteogenesis imperfecta and the teeth, eyes, and ears-a study of non-skeletal phenotypes in adults. 2018 , 29, 2781-2789	20
313	Dentinogenesis imperfecta type II in Swedish children and adolescents. <i>Orphanet Journal of Rare Diseases</i> , 2018 , 13, 145	4-2 7
312	Identification of a de novo fetal variant in osteogenesis imperfecta by targeted sequencing-based noninvasive prenatal testing. 2018 , 63, 1129-1137	12

311	Genotype-phenotype correlation among Malaysian patients with osteogenesis imperfecta. 2018 , 484, 141-147	5
310	A review of syndromes associated with blue sclera, with inclusion of malformations of the head and neck. 2018 , 126, 252-263	9
309	Osteogenesis Imperfecta: Muscle-Bone Interactions when Bi-directionally Compromised. 2018 , 16, 478-489	8
308	Fracturas patológicas del adulto y del niño. 2018 , 51, 1-16	
307	Longitudinal analysis of the audiological phenotype in osteogenesis imperfecta: a follow-up study. 2018 , 132, 703-710	3
306	The clinical application of single-sperm-based SNP haplotyping for PGD of osteogenesis imperfecta. 2019 , 65, 75-80	13
305	Dental alterations on panoramic radiographs of patients with osteogenesis imperfecta in relation to clinical diagnosis, severity, and bisphosphonate regimen aspects: a STROBE-compliant case-control study. 2019 , 128, 621-630	5
304	Bleeding and bruising in Osteogenesis Imperfecta: International Society on Thrombosis and Haemostasis bleeding assessment tool and haemostasis laboratory assessment in 22 individuals. 2019 , 187, 509-517	6
303	Hearing impairment and osteogenesis imperfecta: Literature review. 2019 , 136, 379-383	7
302	Pathogenic Variants and Phenotype Characteristics in Ukrainian Osteogenesis Imperfecta Patients. 2019 , 10, 722	15
301	Use of denosumab in osteogenesis imperfecta: A case report. 2019 , 26, 68-73	
300	Hybrid minigene splicing assay verifies the pathogenicity of a novel splice site variant in the COL1A1 gene of a chinese patient with osteogenesis imperfecta type I. 2019 , 50, 2215-2219	1
299	Genotype-Phenotype Association Analysis Reveals New Pathogenic Factors for Osteogenesis Imperfecta Disease. 2019 , 10, 1200	4
298	Nosology and classification of genetic skeletal disorders: 2019 revision. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 2393-2419	2.5 216
297	Radiotherapy Late Effects and Osteogenesis Imperfecta: Dos and Don'ts in Clinical Practice. 2019 , 12, 322-331	
296	Osteogenesis imperfecta caused by COL1A1, CRTAP and LEPRE1 mutations. Report of 2 cases. 2019 , 153, 336-337	
295	L'ostéogénèse imparfaite : quelle prise en charge chez l'adulte ? 2019 , 86, 14-19	
294	De novo and inherited pathogenic variants in collagen-related osteogenesis imperfecta. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e559	2.3 5

293	The orthopaedic management of long bone deformities in genetically and acquired generalized bone weakening conditions. 2019 , 13, 12-21	9
292	Morphology of Osteogenesis Imperfecta Collagen Mimetic Peptide Assemblies Correlates with the Identity of Glycine-Substituting Residue. 2019 , 20, 3013-3019	
291	IFITM5 pathogenic variant causes osteogenesis imperfecta V with various phenotype severity in Ukrainian and Vietnamese patients. 2019 , 13, 25	7
290	In vitro bone-like nodules generated from patient-derived iPSCs recapitulate pathological bone phenotypes. 2019 , 3, 558-570	32
289	Historical contingency shapes adaptive radiation in Antarctic fishes. 2019 , 3, 1102-1109	22
288	Interpreting Osteogenesis Imperfecta Variants of Uncertain Significance in the Context of Physical Abuse: A Case Series. 2019 , 8, 63-68	1
287	Cyclic pamidronate treatment for osteogenesis imperfecta: Report from a Brazilian reference center. 2019 , 42, 252-260	2
286	Accumulation of microdamage and low bone mass in the femoral head as a cause of subchondral insufficiency fracture in a patient with osteogenesis imperfecta. 2019 , 37, 768-772	1
285	Calcium intake improvement after nutritional intervention in paediatric patients with osteogenesis imperfecta. 2019 , 32, 619-624	1
284	Sclerostin antibody reduces long bone fractures in the oim/oim model of osteogenesis imperfecta. 2019 , 124, 137-147	15
283	Dentinogenesis imperfecta in Osteogenesis imperfecta type XI in South Africa: a genotype-phenotype correlation. 2019 , 5, 4	3
282	Genotype-phenotype correlation study in 364 osteogenesis imperfecta Italian patients. 2019 , 27, 1090-1100	27
281	Osteogenesis Imperfecta: A Pediatric Orthopedic Perspective. 2019 , 50, 193-209	16
280	Challenges of delivery of dental care and dental pathologies in children and young people with osteogenesis imperfecta. 2019 , 20, 473-480	1
279	Stress, Depression, and Quality of Life Among Caregivers of Children With Osteogenesis Imperfecta. 2019 , 33, 437-445	5
278	Current concepts in osteogenesis imperfecta: bone structure, biomechanics and medical management. 2019 , 13, 1-11	26
277	Mobility in osteogenesis imperfecta: a multicenter North American study. 2019 , 21, 2311-2318	6
276	The management of osteogenesis imperfecta in adults: state of the art. 2019 , 86, 589-593	7

275	A self-assembling collagen mimetic peptide system to simultaneously characterize the effects of osteogenesis imperfecta mutations on conformation, assembly and activity. 2019 , 7, 3201-3209		3
274	Dental implants in individuals with osteogenesis imperfecta: a 6-year follow-up study. 2019 , 12, 272-277		0
273	A Case of Osteogenesis Imperfecta Type II With Additional Balanced Translocation t(1;20)(p13;p11.2). 2019 , 38, 263-271		
272	New Insights Into Monogenic Causes of Osteoporosis. <i>Frontiers in Endocrinology</i> , 2019 , 10, 70	5.7	33
271	Assessing disease experience across the life span for individuals with osteogenesis imperfecta: challenges and opportunities for patient-reported outcomes (PROs) measurement: a pilot study. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 23	4.2	8
270	Buschke-Ollendorff Syndrome, Marfan Syndrome and Osteogenesis Imperfecta. 2019 , 1139-1150		
269	The Parathyroid and Disorders of Calcium and Bone Metabolism. 2019 , 409-479		0
268	Scoliosis and Cardiopulmonary Outcomes in Osteogenesis Imperfecta Patients. 2019 , 44, 1057-1063		9
267	Genotype-phenotype relationship in a large cohort of osteogenesis imperfecta patients with COL1A1 mutations revealed by a new scoring system. 2019 , 132, 145-153		7
266	Personalized surgery approach in severe form of osteogenesis imperfecta type III: point of view. 2019 , 28, 505-508		1
265	Olecranon Fractures in Pediatric Patients With Osteogenesis Imperfecta. 2019 , 39, e558-e562		1
264	The evolving therapeutic landscape of genetic skeletal disorders. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 300	4.2	10
263	A novel Ser40Trp variant in IFITM5 in a family with osteogenesis imperfecta and review of the literature. 2019 , 28, 120-125		10
262	Déficit auditif et ostéogénèse imparfaite : revue de la littérature. 2019 , 136, 371-375		
261	Osteogenesis imperfecta caused by COL1A1, CRTAP and LEPRE1 mutations. Report of 2 cases. 2019 , 153, 336-337		2
260	Rickets and Osteomalacia. 2019 , 339-354		2
259	Novel mutations in BMP1 induce a rare type of osteogenesis imperfecta. 2019 , 489, 21-28		11
258	Use of denosumab in osteogenesis imperfecta: A case report. 2019 , 26, 68-73		

257	Congenital disorders of bone and blood. 2019 , 119, 71-81		5
256	SURGICAL MANAGEMENT OF RETINAL DETACHMENT IN OSTEOGENESIS IMPERFECTA: CASE REPORT AND REVIEW OF THE LITERATURE. 2019 , 13, 43-46		5
255	Incidence and treatment of femur fractures in adults with osteogenesis imperfecta: an analysis of an expert clinic of 216 patients. 2020 , 46, 165-171		4
254	Total hip arthroplasty for Protrusio Acetabuli in a young adult Osteogenesis Imperfecta features and Marfanoid features: A case report. 2020 , 11, 96-98		1
253	Osteogenesis imperfecta. Report of 15 Cases. 2020 , 16, 165-168		1
252	Osteogenesis imperfecta. Report of 15 cases. 2020 , 16, 165-168		
251	Osteonectin regulates the extracellular matrix mineralization of osteoblasts through P38 signaling pathway. 2020 , 235, 2220-2231		20
250	Mutation spectrum of COL1A1/COL1A2 screening by high-resolution melting analysis of Chinese patients with osteogenesis imperfecta. 2020 , 38, 188-197		2
249	Impact of fracture characteristics and disease-specific complications on health-related quality of life in osteogenesis imperfecta. 2020 , 38, 109-116		4
248	Juvenile osteoporosis with calvarial doughnuts: Progressive high-turnover bone loss responsive to bisphosphonate therapy. 2020 , 87, 271-272		2
247	A xenograft model to evaluate the bone forming effects of sclerostin antibody in human bone derived from pediatric osteogenesis imperfecta patients. 2020 , 130, 115118		4
246	Association between ribs shape and pulmonary function in patients with Osteogenesis Imperfecta. 2020 , 21, 177-185		7
245	Two novel mutations of COL1A1 in fetal genetic skeletal dysplasia of Chinese. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1105	2.3	6
244	Fatigue in adults with Osteogenesis Imperfecta. <i>BMC Musculoskeletal Disorders</i> , 2020 , 21, 6	2.8	4
243	Hearing loss in individuals with osteogenesis imperfecta in North America: Results from a multicenter study. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 697-704	2.5	9
242	Ostéoporose juvénile associée à des « Calvarial Doughnuts » : une ostéoporose résolutive, à haut remodelage osseux, évoluant favorablement sous bisphosphonates. 2020 , 87, 67-68		
241	Healthcare trajectory of children with rare bone disease attending pediatric emergency departments. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 2	4.2	
240	COL1-related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlers-Danlos syndrome overlap. 2020 , 97, 396-406		16

239	Osteogenesis imperfecta type 1 with an incidental finding of bilateral radioulnar synostosis. 2020 , 29, 155-157		
238	Interaction between KDELR2 and HSP47 as a Key Determinant in Osteogenesis Imperfecta Caused by Bi-allelic Variants in KDELR2. 2020 , 107, 989-999		11
237	A Multicenter Study of Intramedullary Rodding in Osteogenesis Imperfecta. 2020 , 5,		0
236	Prenatal diagnosis of fetal skeletal dysplasia using 3-dimensional computed tomography: a prospective study. <i>BMC Musculoskeletal Disorders</i> , 2020 , 21, 662	2.8	1
235	Respiratory impairment impacts QOL in osteogenesis imperfecta independent of skeletal abnormalities. 2020 , 15, 153		2
234	Rare genetic diseases affecting skeletal development and oral health disparities among children and adolescents: a pathway analysis. 2020 , 70, 469-476		1
233	Gene Expression Profile and Acute Gene Expression Response to Sclerostin Inhibition in Osteogenesis Imperfecta Bone. 2020 , 4, e10377		1
232	Osteogenesis imperfecta type I: The role of deep phenotyping in a patient with a ruptured uterus. 2020 , 63, 104095		
231	Malocclusion in individuals with osteogenesis imperfecta: A systematic review and meta-analysis. 2020 ,		0
230	RNA sequencing analysis reveals increased expression of interferon signaling genes and dysregulation of bone metabolism affecting pathways in the whole blood of patients with osteogenesis imperfecta. 2020 , 13, 177		2
229	Genetische Ursachen und Therapie der Osteogenesis imperfecta. 2020 , 29,		0
228	Explanation for mild and severe osteogenesis imperfecta phenotypes due to splice variants at c.2029-1 in COL1A1. 2020 , 21, 100803		
227	Exome Sequencing Reveals a Phenotype Modifying Variant in ZNF528 in Primary Osteoporosis With a COL1A2 Deletion. 2020 , 35, 2381-2392		2
226	Novel Compound Heterozygous Mutations in Cause Rare Autosomal Recessive Osteogenesis Imperfecta. 2020 , 11, 897		1
225	Echocardiographic study in children with osteogenesis imperfecta. 2020 , 30, 1490-1495		0
224	Osteogenesis imperfecta: Review of 40 patients. 2020 , 154, 512-518		
223	High Bone Mineral Density Osteogenesis Imperfecta in a Family with a Novel Pathogenic Variant in COL1A2. 2020 , 93, 263-271		1
222	Growing Rod Surgery for Early-Onset Scoliosis in an Osteogenesis Imperfecta Patient. 2020 , 144, 178-183		1

221	New 3D Cone Beam CT Imaging Parameters to Assist the Dentist in Treating Patients with Osteogenesis Imperfecta. 2020 , 8,		1
220	Biology and Biomechanics of the Heart Valve Extracellular Matrix. 2020 , 7,		14
219	Cervical kyphosis: A predominant feature of patients with osteogenesis imperfecta type 5. 2020 , 13, 100735		2
218	A Baseline Measurement of Quality of Life in 322 Adults With Osteogenesis Imperfecta. 2020 , 4, e10416		2
217	Osteoporose im Kindes- und Jugendalter. 2020 , 13, 163-171		0
216	Whole Exome Sequencing with Comprehensive Gene Set Analysis Identified a Biparental-Origin Homozygous c.509G>A Mutation in Gene Clustered in Two Taiwanese Families Exhibiting Fetal Skeletal Dysplasia during Prenatal Ultrasound. 2020 , 10,		3
215	Reproductive options for families at risk of Osteogenesis Imperfecta: a review. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 128	4.2	4
214	Inter- and Intrafamilial Phenotypic Variability in Individuals with Collagen-Related Osteogenesis Imperfecta. 2020 , 13, 960-971		5
213	Rare splicing mutation in COL1A1 gene identified by whole exomes sequencing in a patient with osteogenesis imperfecta type I followed by prenatal diagnosis: A case report and review of the literature. 2020 , 741, 144565		5
212	Bisphosphonate-related osteonecrosis of the jaws and dental surgery procedures in children and young people with osteogenesis imperfecta: A systematic review. 2020 , 121, 556-562		10
211	Long bone fracture characteristics in children with medical conditions linked to bone health. 2020 , 103, 104396		2
210	A Clinical Perspective on Advanced Developments in Bone Biopsy Assessment in Rare Bone Disorders. <i>Frontiers in Endocrinology</i> , 2020 , 11, 399	5.7	3
209	Combined technique of titanium telescopic rods and external fixation in osteogenesis imperfecta patients: First 12 consecutive cases. 2020 , 22, 316-325		2
208	Osteogenesis imperfecta: Review of 40 patients. 2020 , 154, 512-518		
207	Tibial-tubercle avulsion and patellar-tendon rupture in pre-pubertal child with osteogenesis imperfecta(OI): Case report and review of current treatment in OI. 2020 , 11, 339-343		1
206	Sclerostin-Antibody Treatment Decreases Fracture Rates in Axial Skeleton and Improves the Skeletal Phenotype in Growing oim/oim Mice. <i>Calcified Tissue International</i> , 2020 , 106, 494-508	3.9	9
205	A molecular dynamics approach on the Y393C variant of protein disulfide isomerase A1. 2020 , 96, 1341-1347		0
204	Mutations in COL1A1/A2 and CREB3L1 are associated with oligodontia in osteogenesis imperfecta. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 80	4.2	4

203	Comparative analysis of the viscoelastic properties of collagen-like proteins by virtual creep test. 2021 , 39, 2744-2753	0
202	Osteogenesis imperfecta: Novel genetic variants and clinical observations from a clinical exome study of 54 Indian patients. 2021 , 85, 37-46	2
201	Reduced mesiodistal tooth dimension in individuals with osteogenesis imperfecta: a cross-sectional study. 2021 , 79, 262-267	0
200	The evolution of the nosology of osteogenesis imperfecta. 2021 , 99, 42-52	6
199	Hospital admissions of patients with osteogenesis imperfecta in the English NHS. 2021 , 32, 1207-1216	2
198	Comparable Effects of Strontium Ranelate and Alendronate Treatment on Fracture Reduction in a Mouse Model of Osteogenesis Imperfecta. 2021 , 2021, 4243105	0
197	Bone and growth: basic principles behind rare disorders. 2021 , 171, 86-93	
196	Bone Health in Children. 2021 , 201-222	
195	Pathophysiology of respiratory failure in patients with osteogenesis imperfecta: a systematic review. 2021 , 53, 1676-1687	4
194	Bone properties in osteogenesis imperfecta: what can we learn from a 'bone biopsy beyond histology?'. 2021 , 171, 111-119	1
193	Metabolic Bone Diseases in the Pediatric Population. 2021 , 25, 94-104	0
192	Genetic Disorders of Bone or Osteodystrophies of Jaws-A Review. 2021 , 8, 41-50	1
191	Neonatal Bone Disorders. 2021 , 9, 602552	1
190	Cytoplasmic polyadenylation by TENT5A is required for proper bone formation. 2021 , 35, 109015	5
189	Targeting cellular stress in vitro improves osteoblast homeostasis, matrix collagen content and mineralization in two murine models of osteogenesis imperfecta. 2021 , 98, 1-20	3
188	Increased Osteocyte Lacunae Density in the Hypermineralized Bone Matrix of Children with Osteogenesis Imperfecta Type I. 2021 , 22,	2
187	Prenatal Diagnosis of Skeletal Dysplasias and Connective Tissue Disorders. 2021 , 783-802	
186	Case Report: Hyperplastic Callus of the Femur Mimicking Osteosarcoma in Osteogenesis Imperfecta Type V. <i>Frontiers in Endocrinology</i> , 2021 , 12, 622674	5.7 0

185	Two novel bi-allelic KDELR2 missense variants cause osteogenesis imperfecta with neurodevelopmental features. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 2241-2249	2.5	2
184	Pain Phenotypes in Rare Musculoskeletal and Neuromuscular Diseases. 2021 , 124, 267-290		10
183	COL1-Related Disorders: Case Report and Review of Overlapping Syndromes. 2021 , 12, 640558		2
182	Current Overview of Osteogenesis Imperfecta. 2021 , 57,		1
181	The Polygenic and Monogenic Basis of Paediatric Fractures. 2021 , 19, 481-493		0
180	Plastin 3 in health and disease: a matter of balance. 2021 , 78, 5275-5301		3
179	Genetic analysis in Japanese patients with osteogenesis imperfecta: Genotype and phenotype spectra in 96 probands. <i>Molecular Genetics & Genomic Medicine</i> , 2021 , 9, e1675	2.3	2
178	OSTEOGNESIS IMPERFECTA. 2021 , 32, 311-318		
177	Ocular characteristics and complications in patients with osteogenesis imperfecta: a systematic review. 2021 ,		3
176	Collagen transport and related pathways in Osteogenesis Imperfecta. 2021 , 140, 1121-1141		5
175	Analysis of bone architecture using fractal-based TX-Analyzer in adult patients with osteogenesis imperfecta. 2021 , 147, 115915		
174	Histopathology of osteogenesis imperfecta bone. Supramolecular assessment of cells and matrices in the context of woven and lamellar bone formation using light, polarization and ultrastructural microscopy. 2021 , 14, 100734		5
173	Clinical, genetic characteristics and treatment outcomes of children and adolescents with osteogenesis imperfecta: a two-center experience. 2021 , 1-10		0
172	A roadmap to surgery in osteogenesis imperfecta: results of an international collaboration of patient organizations and interdisciplinary care teams. 2021 , 92, 608-614		1
171	Lower Extremity Deformity in Osteogenesis Imperfecta: Overview and Surgical Approach. 2021 , 31, 100882		
170	Localized chondro-ossification underlies joint dysfunction and motor deficits in the mouse model of osteogenesis imperfecta. 2021 , 118,		0
169	Pregnancy in women with osteogenesis imperfecta: pregnancy characteristics, maternal, and neonatal outcomes. 2021 , 3, 100362		2
168	Advancing human disease research with fish evolutionary mutant models. 2021 ,		4

167	Osteogenesis imperfecta in children. 2021 , 148, 115914	2
166	From late fatherhood to prenatal screening of monogenic disorders: evidence and ethical concerns. 2021 , 27, 1056-1085	1
165	Acetabular Protrusion in a Cohort of Patients with Osteogenesis Imperfecta Evaluated in a Pediatric Hospital.	
164	Novel Mutations Within Collagen Alpha1(I) and Alpha2(I) Ligand-Binding Sites, Broadening the Spectrum of Osteogenesis Imperfecta - Current Insights Into Collagen Type I Lethal Regions. 2021 , 12, 692978	0
163	Efficacy of Romosozumab for Osteoporosis in a Patient With Osteogenesis Imperfecta: A Case Report. 2021 ,	0
162	Rib Fracture in a Term Newborn with Respiratory Distress. 2021 , 22, e559-e563	1
161	Role of rs193922155 in the etiopathogenesis of osteogenesis imperfecta with description of the phenotype: A case report. 2021 , 100, e27021	
160	Gender-Related Impact of Sclerostin Antibody on Bone in the Osteogenesis Imperfecta Mouse. 2021 , 12, 705505	0
159	Osteoarthritis in osteogenesis imperfecta: A nationwide register-based cohort study. 2022 , 154, 116222	0
158	Knowledge, attitude and practices regarding health self-management among patients with osteogenesis imperfecta in China: an online cross-sectional survey. 2021 , 11, e046286	0
157	Fractures following pregnancy in Osteogenesis imperfecta - A self-controlled case series using Danish Health Registers. 2022 , 154, 116177	
156	Multiscale characterization of pathological bone tissue. 2021 ,	1
155	Glaucoma Syndromes: Insights into Glaucoma Genetics and Pathogenesis from Monogenic Syndromic Disorders. 2021 , 12,	2
154	A systematic review and meta-analysis on the efficacy of stem cell therapy on bone brittleness in mouse models of osteogenesis imperfecta. 2021 , 15, 101108	0
153	High bone mass phenotype in a cohort of patients with Osteogenesis Imperfecta caused due to and C-propeptide cleavage variants in. 2021 , 15, 101102	
152	Osteogenesis Imperfecta and Child Abuse From a Forensic Point of View. 2021 , 13, e12790	0
151	Temporomandibular Joint and Cervical Spine Mobility Assessment in the Prevention of Temporomandibular Disorders in Children with Osteogenesis Imperfecta: A Pilot Study. 2021 , 18,	1
150	Disorders of calcium metabolism. 2021 , 309-388	0

149	Plethora of Traumatic Lesions of Bilateral Knee Extensor Mechanism in Osteogenesis Imperfecta. <i>Frontiers in Endocrinology</i> , 2020 , 11, 603638	5.7	2
148	Incidence and treatment of adult femoral fractures with osteogenesis imperfecta: An analysis of a center of 72 patients in Taiwan. 2021 , 18, 1240-1246		2
147	Comparative characteristics of the morphological structure of teeth in children with I and III types of osteogenesis imperfecta (in vitro). 2021 , 20, 271-274		
146	Mother's sense of coherence and dental characteristics in children and adolescents with osteogenesis imperfecta: A paired study. 2021 , 41, 170-177		
145	Regenerative Approaches in Oral Medicine. 2021 , 197-264		
144	Knochenverletzungen. 2016 , 89-106		1
143	Osteogenesis imperfecta mutations in plastin 3 lead to impaired calcium regulation of actin bundling. 2020 , 8, 21		7
142	Cardiopulmonary Status in Adults with Osteogenesis Imperfecta: Intrinsic Lung Disease May Contribute More Than Scoliosis. 2020 , 478, 2833-2843		7
141	Economic burden of smoking attributed illnesses in Pakistan.		1
140	Cytoplasmic polyadenylation by TENT5A is required for proper bone formation.		1
139	Historical contingency shapes adaptive radiation in Antarctic fishes.		1
138	A rare combination of amniotic constriction band with osteogenesis imperfecta. 2015 , 2015,		0
137	Management of atypical femoral fracture in a patient with osteogenesis imperfecta. 2017 , 2017,		4
136	Osteogenesis imperfecta-pathophysiology and therapeutic options. 2020 , 7, 9		9
135	Polymorphisms associated with low bone mass and high risk of atraumatic fracture. 2015 , 64, 621-31		5
134	Pamidronate Therapy Increases Trabecular Bone Complexity of Mandibular Condyles in Individuals with Osteogenesis Imperfecta. <i>Calcified Tissue International</i> , 2021 , 1	3.9	0
133	Encyclopedia of Ophthalmology. 2014 , 1-2		
132	Bulbous epiphysis and popcorn calcification as related to growth plate differentiation in osteogenesis imperfecta. 2015 , 12, 202-6		1

- 131 Clinical application of antenatal genetic diagnosis of osteogenesis imperfecta type IV. **2015**, 21, 964-9
- 130 STAPEDOTOMY TO TREAT MIXED HEARING LOSS IN OSTEOGENESIS IMPERFECTA: A CASE STUDY. **2015**, 5, 43-48
- 129 Evaluation and Management of Pathologic Femur Fractures in Children. **2016**, 195-213
- 128 Osteogenesis Imperfecta. **2016**, 718-721
- 127 Rib Cage Anomalies in Severe Osteogenesis Imperfecta [Functional Investigations]. **2017**, 297-306
- 126 Creatine Kinase as Biomarker in Osteogenesis Imperfecta. **2017**, 177-199
- 125 Hereditäre Bindegewebserkrankungen. **2017**, 1-17
- 124 Leczenie niedosłuchu w osteogenesis imperfecta [przegląd piśmiennictwa]. **2017**, 6, 9-15
- 123 Management of a patient with osteogenesis imperfecta and trisomy 18. **2017**, 23, 146-151
- 122 Osteogenesis Imperfecta. **2018**, 433-435
- 121 Osteogenesis imperfecta. **2018**, 8, e2018040
- 120 Hereditäre Bindegewebserkrankungen. **2018**, 883-899
- 119 Encyclopedia of Ophthalmology. **2018**, 1299-1300
- 118 OSSICULOPLASTY IN HEARING LOSS TREATMENT OF PATIENTS WITH OSTEOGENESIS IMPERFECTA. **2018**, 8, 25-30 1
- 117 Mobility in Osteogenesis Imperfecta: A Multicenter North American Study.
- 116 Osteogenesis Imperfecta: Diagnostic Feature. **2018**, 15, 224-232 3
- 115 Assessing Disease Experience across the Life Span for Individuals with Osteogenesis Imperfecta: Challenges and Opportunities for Patient-Reported Outcomes (PROs) Measurement.
- 114 The Hip in Osteogenesis Imperfecta. **2019**, 715-734

- 113 HEARING LOSS DUE TO OSTEOGENESIS IMPERFECTA IN TWO CHILDREN. **2019**, 9, 35-39
- 112 Clinic Case of Rare Type VI Osteogenesis Imperfecta. **2019**, 16, 30-35
- 111 Surgical treatment of comminuted intraarticular distal femur fracture in patient with osteogenesis imperfecta type I. **2019**, 7, 87-96 1
- 110 PHENOTYPIC CHARACTERISTICS IN OSTEOGENESIS IMPERFECTA PATIENTS. **2019**, 21, 266-271
- 109 Experience of the application of pamidronic acid in the therapy in children with osteogenesis imperfecta. **2019**, 19, 282-287
- 108 Current approach to diagnosis and treatment of children with osteogenesis imperfecta. **2019**, 7, 87-102 1
- 107 Hereditary Disorders of Collagen and Elastic Fibers. **2020**, 1-16
- 106 Osteogenesis Imperfecta in the Spine. **2020**, 221-230 2
- 105 Chirurgia toracică en pacient cu osteogeneză imperfectă. **2020**, 79, 94-96
- 104 Skelett. **2020**, 349-379
- 103 Osteogenesis imperfecta: a literature review and a clinical case of a perinatal-lethal type of disease. **2021**, 226-234
- 102 Introduction to Osteogenesis Imperfecta. **2020**, 3-9
- 101 Pediatric Rehabilitation. **2020**, 285-317
- 100 Osteogenesis imperfecta: treatment and surgical management. **2021**, 33, 74-78 1
- 99 Dental management of a child with a rare bone disorder: a case report with a six-year follow up. 68,
- 98 Knochen und Kalziumstoffwechsel. **2020**, 105-133 1
- 97 Bilateral Anterior Inferior Iliac Spine Avulsion Fractures in an Adolescent with Type I Osteogenesis Imperfecta: A Case Report. **2020**, 10, e2000271
- 96 Morphologic Features and Treatment of the Hip and Proximal Femur in OI. **2020**, 129-146

- 95 Osteoblasts in Human Skeletal Diseases. **2020**, 499-512
- 94 Changes in Biomaterial Properties of Bone With Disease and Treatment. **2020**, 456-480
- 93 Non-collagen pathogenic variants resulting in the osteogenesis imperfecta phenotype in children: a single-country observational cohort study. **2021**,
- 92 Osteogenesis Imperfecta and Split Foot Malformation due to 7q21.2q21.3 Deletion Including COL1A2, DLX5/6 Genes: Review of the Literature.
- 91 'In-Out-In' K-wires sliding in severe tibial deformities of osteogenesis imperfecta: a technical note. **2021**, 30, 257-263 1
- 90 OSTEOGENESIS IMPERFECTA. **2021**, 683-705
- 89 The effect of bisphosphonate medication on orthodontics and orthognathic surgery in patients with osteogenesis imperfecta. **2019**, 8, Doc06 2
- 88 Osteogenesis imperfecta due to a possible new COL1A2 mutation; the importance of phenotyping and diagnostic challenges. **2016**, 16, 168-71 1
- 87 Early Motor Delay: An Outstanding, Initial Sign of Osteogenesis Imperfecta Type 1. **2017**, 7, 63-66 2
- 86 Revision Rates for Osteogenesis Imperfecta Patients Treated with Telescopic Nails. A follow-up Study After a 7-year Experience. **2020**, 13, 543-547
- 85 Three Patient Kindred with a Novel Phenotype of Osteogenesis Imperfecta due to a Variant. **2021**, 13, 218-224
- 84 Infrequent association of two rare diseases: amniotic band syndrome and osteogenesis imperfecta. **2021**, 10, 0
- 83 Early documentation of inherited disorders through family studies. **2022**, 17-26
- 82 Oral health-related quality of life in children with osteogenesis imperfecta. **2021**, 1 0
- 81 Perspective of the GEMSTONE Consortium on Current and Future Approaches to Functional Validation for Skeletal Genetic Disease Using Cellular, Molecular and Animal-Modeling Techniques.. *Frontiers in Endocrinology*, **2021**, 12, 731217 5.7 1
- 80 An Unusual Presentation of Osteogenesis Imperfecta: A Case Report. **2021**, 11, 1
- 79 Osteogenesis Imperfecta: Search for Mutations in Patients from the Republic of Bashkortostan (Russia).. **2022**, 13, 0
- 78 Osteogenesis imperfecta in 140 Turkish families: Molecular spectrum and, comparison of long-term clinical outcome of those with COL1A1/A2 and biallelic variants.. **2021**, 155, 116293 0

- 77 Bone hydration: How we can evaluate it, what can it tell us, and is it an effective therapeutic target?. **2022**, 16, 101161 1
- 76 Three Patient Kindred with a Novel Phenotype of Osteogenesis Imperfecta due to a COL1A1 Variant. **2021**, 13, 218-224
- 75 Outcomes of Operative Treatment of Forearm Deformity in Children with Osteogenesis Imperfecta: 18 Cases. **2022**, 2, 21-30 0
- 74 Identification and development of the novel 7-genes diagnostic signature by integrating multi cohorts based on osteoarthritis.. **2022**, 159, 10
- 73 Phenotypic Spectrum and Molecular Basis in a Chinese Cohort of Osteogenesis Imperfecta With Mutations in Type I Collagen.. **2022**, 13, 816078 0
- 72 Metaphyseal and posterior rib fractures in osteogenesis imperfecta: Case report and review of the literature.. **2022**, 16, 101171
- 71 Spine Pathologies in Osteogenesis Imperfecta: A Review. **2022**, 28, 118-127
- 70 Does the Skeletal Phenotype of Osteogenesis Imperfecta Differ for Patients With Non-COL1A1/2 Mutations? A Retrospective Study in 113 Patients.. **2022**, 42,
- 69 Fractures in Osteogenesis Imperfecta: Pathogenesis, Treatment, Rehabilitation and Prevention.. **2022**, 9, 2
- 68 Osteogenesis Imperfecta/Ehlers-Danlos Overlap Syndrome and Neuroblastoma-Case Report and Review of Literature.. **2022**, 13,
- 67 Comprehensive Risk Assessments and Anesthetic Management for Children with Osteogenesis Imperfecta: A Retrospective Review of 252 Orthopedic Procedures Over 5 Years.. **2022**, 0
- 66 Modern approaches to surgical treatment of children with osteogenesis imperfecta. **2021**, 265-270
- 65 Comprehensive pain management strategy for infants with moderate to severe osteogenesis imperfecta in the perinatal period.. **2021**, 3, 156-162
- 64 High Heterogeneity of Temporal Bone CT Aspects in Osteogenesis Imperfecta Is Not Linked to Hearing Loss.. **2022**, 11,
- 63 Dental phenotype in an adolescent with osteogenesis imperfecta type XII.. **2022**, 15, 0
- 62 Table_1.DOCX. **2020**,
- 61 Table_1.XLSX. **2020**,
- 60 Data_Sheet_1.XLSX. **2018**,

59	Image_1.TIF. 2018 ,		
58	Image_2.TIF. 2018 ,		
57	DataSheet_1.pdf. 2019 ,		
56	Table_1.xlsx. 2019 ,		
55	Revision Rates for Osteogenesis Imperfecta Patients Treated with Telescopic Nails. A follow-up Study After a 7-year Experience. 2020 , 13, 543-547		0
54	Hereditary Disorders of Collagen and Elastic Fibers. 2022 , 871-886		
53	Educational Case: Osteogenesis imperfecta. <i>Academic Pathology</i> , 2022 , 9, 100025	1.3	
52	Prevalence and Hospital Admissions in Patients With Osteogenesis Imperfecta in The Netherlands: A Nationwide Registry Study.. <i>Frontiers in Endocrinology</i> , 2022 , 13, 869604	5.7	1
51	Nuss Procedure for pectus excavatum repair in a patient with osteogenesis imperfecta. <i>Journal of Pediatric Surgery Case Reports</i> , 2022 , 82, 102311	0.3	
50	Hereditary Disorders of the Dermis. 2016 , 119-135.e6		0
49	Dissecting the phenotypic variability of osteogenesis imperfecta.. <i>DMM Disease Models and Mechanisms</i> , 2022 , 15,	4.1	1
48	Prenatal trio-based whole exome sequencing in fetuses with abnormalities of the skeletal system.. <i>Molecular Genetics and Genomics</i> , 2022 ,	3.1	1
47	Osteogenesis imperfecta.		
46	Prevalence of scoliosis and impaired pulmonary function in patients with type III osteogenesis imperfecta. <i>European Spine Journal</i> ,	2.7	
45	Poetics of brittle bone disease: using found poetry to explore childhood bioethics. <i>Journal of Poetry Therapy</i> , 1-18	0.7	
44	The prevalence of musculoskeletal pain and therapy needs in adults with Osteogenesis Imperfecta (OI) a cross-sectional analysis. <i>BMC Musculoskeletal Disorders</i> , 2022 , 23,	2.8	
43	Knochenverletzungen. 2022 , 107-128		
42	Patient-reported outcomes in a Chinese cohort of osteogenesis imperfecta unveil psycho-physical stratifications associated with clinical manifestations. <i>Orphanet Journal of Rare Diseases</i> , 2022 , 17,	4.2	

- 41 An Update on Animal Models of Osteogenesis Imperfecta. *Calcified Tissue International*, 3.9
- 40 Analysis of the clinical and genetic characteristics of a Chinese family with osteogenesis imperfecta type I. *Molecular Genetics & Genomic Medicine*, 2.3
- 39 Quantitative gait analysis in children with osteogenesis imperfecta: relationship between gait deviations and clinical features. **2022**, 88, 255-262
- 38 Osteogenesis Imperfecta Diagnosed in an Active Duty Female Due to CREB3L1 Heterozygosity.
- 37 Morphological Study of Dental Structure in Dentinogenesis Imperfecta Type I with Scanning Electron Microscopy. **2022**, 10, 1453 ○
- 36 Early-onset osteoporosis: Rare monogenic forms elucidate the complexity of disease pathogenesis beyond type I collagen. ○
- 35 Descemet Membrane Endothelial Keratoplasty in a Patient With Descemet Membrane Detachment and Rieger-Like Anomaly Associated With Osteogenesis Imperfecta and Mutation in COL1A1. **2022**, Publish Ahead of Print,
- 34 Evaluation of the Severity of Malocclusion in Children with Osteogenesis Imperfecta. **2022**, 11, 4862
- 33 Functional status of individuals with osteogenesis imperfecta: data from a reference center. **2022**,
- 32 Craniofacial morphology in adults with osteogenesis imperfecta ▯ cross-sectional study.
- 31 Chemically-induced osteogenic cells for bone tissue engineering and disease modeling. **2022**, 289, 121792 ○
- 30 Biomechanical, Microstructural and Material Properties of Tendon and Bone in the Young Oim Mice Model of Osteogenesis Imperfecta. **2022**, 23, 9928 ○
- 29 Role of Extracellular Matrix and Inflammation in Abdominal Aortic Aneurysm. **2022**, 23, 11078 1
- 28 Does the c.-14C>T Mutation in the IFITM5 Gene Provide Identical Phenotypes for Osteogenesis Imperfecta Type V? Data from Russia and a Literature Review. **2022**, 10, 2363 ○
- 27 Rosemary Extract-Induced Autophagy and Decrease in Accumulation of Collagen Type I in Osteogenesis Imperfecta Skin Fibroblasts. **2022**, 23, 10341 ○
- 26 Anthropometrics of Polish children with osteogenesis imperfecta: a single-centre retrospective cohort study. **2022**, 22, ○
- 25 Skin chronological aging drives age-related bone loss via secretion of cystatin-A. ○
- 24 Chronic pain in adults with osteogenesis imperfecta and its relationship to appraisal, coping, and quality of life: A cross-sectional study. **2022**, 101, e30256 ○

- 23 A multicenter study to evaluate pain characteristics in osteogenesis imperfecta. ○
- 22 COL1A2 (p.Gly322Ser) Mutation Causes Late-Onset Osteogenesis Imperfecta: A Case Report. **2022**, ○
- 21 Brittle Bone Disease: A Case Report. **2022**, ○
- 20 Mutant MESD links cellular stress to type I collagen aggregation in osteogenesis imperfecta type XX. **2023**, 115, 81-106 ○
- 19 Incidence and nonunion rates of tibial fractures in adults with osteogenesis imperfecta: a retrospective cohort study of 402 patients with 42 fractures at an expert clinic. **2022**, 23, ○
- 18 Osteogenesis Imperfecta: A Case Series and Literature Review. **2023**, ○
- 17 Comparison of the use of intramedullary telescopic system and titanium elastic rods in children with osteogenesis imperfecta I and III types. **2023**, 373-379 ○
- 16 Corneal Biomechanical Characteristics in Osteogenesis Imperfecta With Collagen Defect. **2023**, 12, 14 ○
- 15 Key4OI Recommendations for Lung Function Guidance in Osteogenesis Imperfecta Based on an Internationally Performed Comprehensive ICHOM Procedure. **2023**, ○
- 14 Assessment of the Retinal Nerve Fibre Layer, Retina, and Choroid in Osteogenesis Imperfecta. ○
- 13 Postawy rodziców wobec dzieci z wrodzonymi wadami kości. **2022**, 51-71 ○
- 12 LAMELLAR THICKNESS MEASUREMENTS IN CONTROL AND OSTEOGENESIS IMPERFECTA HUMAN BONE, with development of a method of automated thickness averaging to simplify quantitation.. **2022**, ○
- 11 From Genetics to Clinical Implications: A Study of 675 Dutch Osteogenesis Imperfecta Patients. **2023**, 13, 281 ○
- 10 Mesenchymal stem cells in the treatment of osteogenesis imperfecta. **2023**, 12, ○
- 9 Landscape of Well-Coordinated Fracture Healing in a Mouse Model Using Molecular and Cellular Analysis. **2023**, 24, 3569 1
- 8 Nosology of genetic skeletal disorders: 2023 revision. **2023**, 191, 1164-1209 ○
- 7 Successful helmet therapy in an infant with positional brachycephaly associated with perinatal severe osteogenesis imperfecta. ○
- 6 Systematic review of health related-quality of life in adults with osteogenesis imperfecta. **2023**, 18, ○

- 5 Osteogenesis imperfecta. **2023**, 23, 182-188 ○
- 4 Bi-allelic mutation in SEC16B alters collagen trafficking and increases ER stress. **2023**, 15, ○
- 3 Lrp5 p. Val667Met Variant Compromises Bone Mineral Density and Matrix Properties in Osteoporosis. ○
- 2 Transition of young adults with metabolic bone diseases to adult care. 14, ○
- 1 Prevalence of Monogenic Bone Disorders in a Dutch Cohort of Atypical Femur Fracture Patients. ○