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Nosology and classification of genetic skeletal disorders: 2019 revision

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345	New Targets and Emergent Therapies for Osteoporosis. 2020 , 262, 451-473		7
344	Two novel mutations of COL1A1 in fetal genetic skeletal dysplasia of Chinese. 2020 , 8, e1105		6
343	Current status in therapeutic interventions of neonatal bone mineral metabolic disorders. 2020 , 25, 101075		4
342	Extremity anomalies associated with Robinow syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3584-3592	2.5	3
341	Fourteen-year follow-up of a child with acroscyphodysplasia with emphasis on the need for multidisciplinary management: a case report. 2020 , 21, 189		2
340	Skeletal Phenotypes Due to Abnormalities in Mitochondrial Protein Homeostasis and Import. 2020 , 21,		1
339	Biallelic mutations in LAMA5 disrupts a skeletal noncanonical focal adhesion pathway and produces a distinct bent bone dysplasia. 2020 , 62, 103075		3
338	Schmid metaphyseal chondrodysplasia: an example of radiology guidance to molecular diagnosis. 2020 , 15, 2554-2556		1
337	Hydrops in first trimester as unreported prenatal finding of dyssegmental dysplasia confirmed by exome sequencing. 2021 , 58, 318-320		1
336	Evaluation of FGFR inhibitor ASP5878 as a drug candidate for achondroplasia. 2020 , 10, 20915		2
335	Expanding the phenotypic spectrum of RPL13-related skeletal dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 2776-2781	2.5	2
334	Exploring human genetic skeletal disorders provides important insights into skeletogenesis and elucidates basic developmental signaling pathways. 2020 , 62, 103091		
333	Molecular testing strategies in the evaluation of fetal skeletal dysplasia. 2020 , 1-7		1
332	Insights into dental mineralization from three heritable mineralization disorders. 2020 , 212, 107597		5
331	Multisystemic manifestations in a cohort of 75 classical Ehlers-Danlos syndrome patients: natural history and nosological perspectives. 2020 , 15, 197		9
330	Current Understanding of the Genetics of Intervertebral Disc Degeneration. 2020 , 7, 431		9
329	Chondrodysplasias and Aneurysmal Thoracic Aortopathy: An Emerging Tale of Molecular Intersection. 2020 , 26, 783-795		0

328	MicroRNAs in cartilage development and dysplasia. 2020 , 140, 115564		6
327	Assessment of longitudinal bone growth in osteogenesis imperfecta using metacarpophalangeal pattern profiles. 2020 , 140, 115547		2
326	The Musculoskeletal Knowledge Portal: Making Omics Data Useful to the Broader Scientific Community. 2020 , 35, 1626-1633		8
325	Novel Compound Heterozygous Mutations in Cause Rare Autosomal Recessive Osteogenesis Imperfecta. 2020 , 11, 897		1
324	Expanding the phenotypic spectrum of IFT81: Associated ciliopathy syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 2403-2408	2.5	0
323	Pediatric radiology in the diagnosis and management of skeletal dysplasias - welcome to the era of genomic medicine and modern drug pipelines. 2020 , 50, 1648-1649		1
322	Biallelic cGMP-dependent type II protein kinase gene () variants cause a novel acromesomelic dysplasia. 2020 ,		3
321	Spondyloepiphyseal dysplasia type Stanescu: Expanding the clinical and molecular spectrum of a very rare type II collagenopathy. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 2715-2721	2.5	0
320	Cloning, expression and enzyme activity delineation of two novel CANT1 mutations: the disappearance of dimerization may indicate the change of protein conformation and even function. 2020 , 15, 240		1
319	Cartografía genética de las enfermedades raras constitucionales. 2020 , 53, 1-6		
318	Functioning and equality according to International Classification of Functioning, Disability and Health (ICF) in people with skeletal dysplasia compared to matched control subjects - a cross-sectional survey study. 2020 , 21, 808		2
317	The Genetic Architecture of High Bone Mass. 2020 , 11, 595653		5
316	Whole-exome sequencing identified two novel mutations of DYNC2L1 in fetal skeletal ciliopathy. 2020 , 8, e1524		2
315	The radiologic diagnosis of skeletal dysplasias: past, present and future. 2020 , 50, 1650-1657		6
314	Molecular Genetics and Pathogenesis of Ehlers-Danlos Syndrome and Related Connective Tissue Disorders. 2020 , 11,		10
313	Spondyloepimetaphyseal dysplasia with elevated plasma lysosomal enzymes caused by homozygous variant in MBTPS1. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 1796-1800	2.5	4
312	An apparent new syndrome of extreme short stature, microcephaly, dysmorphic faces, intellectual disability, and a bone dysplasia of unknown etiology. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 1562-1571	2.5	
311	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

310	Biochemical characteristics of the chondrocyte-enriched SNORC protein and its transcriptional regulation by SOX9. 2020 , 10, 7790		0
309	Pseudodiastrophic dysplasia expands the known phenotypic spectrum of defects in proteoglycan biosynthesis. 2020 , 57, 454-460		4
308	A novel mutation within intron 17 of the CUL7 gene results in appearance of premature termination codon. 2020 , 507, 23-30		1
307	A novel homozygous missense variant in MATN3 causes spondylo-epimetaphyseal dysplasia Matrilin 3 type in a consanguineous family. 2020 , 63, 103958		0
306	Health Supervision for People With Achondroplasia. 2020 , 145,		15
305	Sitting Height to Standing Height Ratio Reference Charts for Children in the United States. 2020 , 226, 221-227.e15		8
304	New perspectives on the treatment of skeletal dysplasia. 2020 , 11, 2042018820904016		13
303	Homozygous Loss-of-Function Mutations in CCDC134 Are Responsible for a Severe Form of Osteogenesis Imperfecta. 2020 , 35, 1470-1480		14
302	Genetics of Skeletal Disorders. 2020 , 262, 325-351		0
301	Congenital posterior cervical spine malformation due to biallelic c.240-4T>G RIPPLY2 variant: A discrete entity. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 1466-1472	2.5	2
300	High Fidelity of Mouse Models Mimicking Human Genetic Skeletal Disorders. 2019 , 10, 934		5
299	The changing world of skeletal dysplasia. 2020 , 4, 253-254		4
298	Best practice guidelines for management of spinal disorders in skeletal dysplasia. 2020 , 15, 161		5
297	Osteogenesis imperfecta in a male holstein calf associated with a possible oligogenic origin. 2020 , 40, 58-67		2
296	Finite element analysis of bone strength in osteogenesis imperfecta. 2020 , 133, 115250		5
295	Joint Replacements in Individuals With Skeletal Dysplasias: One Institution's Experience and Response to Operative Complications. 2020 , 35, 1993-2001		3
294	The Connective Tissue Disorder Associated with Recessive Variants in the Zinc Transporter Gene (Spondylo-Dysplastic Ehlers-Danlos Syndrome Type 3): Insights from Four Novel Patients and Follow-Up on Two Original Cases. 2020 , 11,		4
293	Skeletal Dysplasias Caused by Sulfation Defects. 2020 , 21,		8

292	WNT Signaling and Bone: Lessons From Skeletal Dysplasias and Disorders. 2020 , 11, 165		19
291	Multiple epiphyseal dysplasia and related disorders: Molecular genetics, disease mechanisms, and therapeutic avenues. 2021 , 250, 345-359		4
290	Predicting ambulatory function at skeletal maturity in children with moderate to severe osteogenesis imperfecta. 2021 , 180, 233-239		2
289	The third case of TNFRSF11A-associated dysosteosclerosis with a mutation producing elongating proteins. 2021 , 66, 371-377		2
288	Biallelic TMEM251 variants in patients with severe skeletal dysplasia and extreme short stature. 2021 , 42, 89-101		3
287	Radiologic Features of Type II and Type XI Collagenopathies. 2021 , 41, 192-209		1
286	Delineation of the clinical and radiological features of Stuve-Wiedemann syndrome childhood survivors, four new cases and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 856-865	2.5	1
285	[Rehabilitation of orphan diseases in adulthood: osteogenesis imperfecta]. 2021 , 80, 29-42		2
284	Further defining the clinical and molecular spectrum of acromesomelic dysplasia type maroteaux: a Turkish tertiary center experience. 2021 , 66, 585-596		3
283	Human dentin characteristics of patients with osteogenesis imperfecta: insights into collagen-based biomaterials. 2021 , 119, 259-267		3
282	Novel RPL13 Variants and Variable Clinical Expressivity in a Human Ribosomopathy With Spondyloepimetaphyseal Dysplasia. 2021 , 36, 283-297		5
281	The evolution of the nosology of osteogenesis imperfecta. 2021 , 99, 42-52		6
280	General Skeletal Disorders. 2021 , 447-468		
279	Clinical and genetic characteristics and orthopedic manifestations of the Saul/Wilson syndrome in two Russian patients. 2020 , 8, 451-460		3
278	AIFM1-associated X-linked spondylometaphyseal dysplasia with cerebral hypomyelination. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1228-1235	2.5	0
277	Best practice guidelines in managing the craniofacial aspects of skeletal dysplasia. 2021 , 16, 31		5
276	Development of the Screening Tool for Everyday Mobility and Symptoms (STEMS) for skeletal dysplasia. 2021 , 16, 40		3
275	Bone and growth: basic principles behind rare disorders. 2021 , 171, 86-93		

274	Procollagen Trafficking and its Implications in Osteogenesis Imperfecta. 2021 , 23-53		
273	Dysproportionierung und Kleinwuchs. 2021 , 707-721		
272	Imaging of Congenital Skeletal Disorders. 2021 , 25, 22-38		2
271	Opportunities and Challenges in Functional Genomics Research in Osteoporosis: Report From a Workshop Held by the Causes Working Group of the Osteoporosis and Bone Research Academy of the Royal Osteoporosis Society on October 5th 2020. 2020 , 11, 630875		2
270	Massively Parallel Sequencing for Rare Genetic Disorders: Potential and Pitfalls. 2020 , 11, 628946		4
269	De novo Leu619Pro variant causes a new channelopathy characterised by giant cell lesions of the jaws and skull, skeletal abnormalities and polyneuropathy. 2021 ,		1
268	Rare skeletal disorders: a multidisciplinary postnatal approach to diagnosis and management. 2021 , 171, 94-101		
267	Further expanding the mutational spectrum of brain abnormalities, neurodegeneration, and dysosteosclerosis: A rare disorder with neurologic regression and skeletal features. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1888-1896	2.5	5
266	Osteoclasts recycle via osteomorphs during RANKL-stimulated bone resorption. 2021 , 184, 1330-1347.e13		49
265	Control of osteocyte dendrite formation by Sp7 and its target gene osteocrin.		
264	[Rare bone disorders and respective treatments]. 2021 , 62, 486-495		1
263	Mild metatropic dysplasia: emphasis on the magnetic resonance imaging of articular cartilage thickening. 2021 , 7, 20200155		
262	Compound Heterozygous Frameshift Mutations in MESD Cause a Lethal Syndrome Suggestive of Osteogenesis Imperfecta Type XX. 2021 , 36, 1077-1087		6
261	Growth Topics in FGFR3-Related Skeletal Dysplasias. 2021 , 7, 82-98		0
260	The role of IFT140 in early bone healing of tooth extraction sockets. 2021 ,		1
259	Total Knee Arthroplasty in Spondyloepiphyseal Dysplasia with Irreducible Congenital Dislocation of the Patella: Case Report and Literature Review. 2021 , 17, 275-283		
258	A case of brachymetacarpia in a skeleton from a Mudejar cemetery from Spain (13th-14th century AD). 2021 , 31, 621-627		
257	Abnormalities in Tooth Formation after Early Bisphosphonate Treatment in Children with Osteogenesis Imperfecta. 2021 , 109, 121-131		3

256	Utility of genetic testing for prenatal presentations of hypophosphatasia. 2021 , 132, 198-203		1
255	Differentiation of Hypertrophic Chondrocytes from Human iPSCs for the In Vitro Modeling of Chondrodysplasias. 2021 , 16, 610-625		3
254	Deficiency of TMEM53 causes a previously unknown sclerosing bone disorder by dysregulation of BMP-SMAD signaling. 2021 , 12, 2046		1
253	Outcomes of Temporary Hemiepiphyseal Stapling for Correcting Genu Valgum in Children with Multiple Osteochondromas: A Single Institution Study. 2021 , 8,		
252	Signaling Pathways in Bone Development and Their Related Skeletal Dysplasia. 2021 , 22,		6
251	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. 2021 , 66, 995-1008		6
250	Sleep-disordered breathing and its management in children with rare skeletal dysplasias. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 2108-2118	2.5	1
249	Prenatal Diagnosis of Skeletal Dysplasias and Connective Tissue Disorders. 2021 , 783-802		
248	Compressive Strength of Iliac Bone ECM Is Not Reduced in Osteogenesis Imperfecta and Increases With Mineralization. 2021 , 36, 1364-1375		4
247	Novel mutations in BMP1 result in a patient with autosomal recessive osteogenesis imperfecta. 2021 , 9, e1676		0
246	Description of four patients with TRIP11 variants expand the clinical spectrum of odontochondroplasia (ODCD) and demonstrate the existence of common variants. 2021 , 64, 104198		1
245	Skeletal dysplasias in art and antiquities: A cultural journey through genes, environment, and chance. 2021 , 187, 199-212		1
244	New gene discoveries in skeletal diseases with short stature. 2021 , 10, R160-R174		1
243	Current Overview of Osteogenesis Imperfecta. 2021 , 57,		1
242	The role of biomineralization in disorders of skeletal development and tooth formation. 2021 , 17, 336-349		8
241	-Associated Diastrophic Dysplasia and rMED-Clinical Features in Affected Finnish Children and Review of the Literature. 2021 , 12,		0
240	Towards an ICF-based self-report questionnaire for people with skeletal dysplasia to study health, functioning, disability and accessibility. 2021 , 16, 236		
239	Osteocyte transcriptome mapping identifies a molecular landscape controlling skeletal homeostasis and susceptibility to skeletal disease. 2021 , 12, 2444		12

238	KIAA0753-related skeletal ciliopathy: a ninth case, extending the phenotype and reporting a novel variant. 2021 , 30, 142-146		1
237	Monoallelic mutations in <i>SLCO2A1</i> cause autosomal dominant primary hypertrophic osteoarthropathy. 2021 , 36, 1459-1468		0
236	The Polygenic and Monogenic Basis of Paediatric Fractures. 2021 , 19, 481-493		0
235	Impact of Intrinsic Muscle Weakness on Muscle-Bone Crosstalk in Osteogenesis Imperfecta. 2021 , 22,		0
234	Natural history of TRPV4-Related disorders: From skeletal dysplasia to neuromuscular phenotype. 2021 , 32, 46-55		3
233	Identification of clinical and radiographic predictors of central nervous system injury in genetic skeletal disorders. 2021 , 11, 11402		
232	Genetic Analysis Using a Next Generation Sequencing-Based Gene Panel in Patients With Skeletal Dysplasia: A Single-Center Experience. 2021 , 12, 670608		0
231	Preoperative Microbiological Screening in Pediatric Assessment of Skeletal Dysplasias Cases Before Planned Orthopedic Surgery. 2020 , 9, 281-285		
230	Genetic analysis in Japanese patients with osteogenesis imperfecta: Genotype and phenotype spectra in 96 probands. 2021 , 9, e1675		2
229	Oligogenic Inheritance of Monoallelic , , , and Variants Leading to a Phenotype Similar to Odontochondrodysplasia. 2021 , 12, 680838		2
228	Enfermedades raras constitucionales. 2021 , 56, 1-13		
227	A Novel <i>IFITM5</i> Variant Associated with Phenotype of Osteoporosis with Calvarial Doughnut Lesions: A Case Report. 2021 , 109, 626-632		1
226	Osteogenesis Imperfecta in Two Finnish Lapphund Puppies. 2021 , 12, 177-185		
225	Spondyloepimetaphyseal dysplasia <i>EXTL3</i> -deficient type: Long-term follow-up and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3104-3110	2.5	1
224	Combined exome sequencing and deep phenotyping in highly selected fetuses with skeletal dysplasia during the first and second trimesters improves diagnostic yield. 2021 , 41, 1401-1413		2
223	Broadening the phenotypic spectrum of <i>Beta3GalT6</i> -associated phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3153-3160	2.5	0
222	Rehabilitation einer 47-jährigen Patientin mit Osteogenesis imperfecta. 2021 , 60, 172-176		
221	Pediatric rib pathologies: clinicoimaging scenarios and approach to diagnosis. 2021 , 51, 1783-1797		0

220	Destructive juvenile idiopathic arthritis: do not overlook rare genetic skeletal disorders. 2021 , 3, e404	
219	Sox9 Determines Translational Capacity During Early Chondrogenic Differentiation of ATDC5 Cells by Regulating Expression of Ribosome Biogenesis Factors and Ribosomal Proteins. 2021 , 9, 686096	3
218	A Heterozygous Missense Variant in in a Stillborn Romagnola Calf with Skeletal-Cardio-Enteric Dysplasia. 2021 , 11,	
217	An variant links aberrant Rac1 function to early-onset skeletal fragility. 2021 , 5, e10509	
216	A novel mutation in the gene: Diagnostic approach from relatively common skeletal dysplasias to an extremely rare Odontochondrodysplasia. 2021 ,	
215	Diagnostic yield of rare skeletal dysplasia conditions in the radiogenomics era. 2021 , 14, 148	2
214	Molecular diagnosis for 55 fetuses with skeletal dysplasias by whole-exome sequencing: A retrospective cohort study. 2021 , 100, 219-226	3
213	Chondrodysplasias With Multiple Dislocations Caused by Defects in Glycosaminoglycan Synthesis. 2021 , 12, 642097	1
212	Thundering hoofbeats and dazzling zebras: A model integrating current rare disease perspectives in paleopathology. 2021 , 33, 196-208	1
211	Phenotypic and genotypic spectrum of CTSK variants in a cohort of twenty-five Indian patients with pycnodysostosis. 2021 , 64, 104235	0
210	FAM20C Overview: Classic and Novel Targets, Pathogenic Variants and Raine Syndrome Phenotypes. 2021 , 22,	1
209	Frequency of carriers for rare recessive Mendelian diseases in a Brazilian cohort of 320 patients. 2021 , 187, 364-372	4
208	A Novel Heterozygous Variant in a Short Patient Born Small for Gestational Age with Recurrent Patellar Dislocation: A Case Report. 2021 ,	
207	Genetic causes of the skeletal system abnormalities diagnosed by prenatal sonography with the use of exome sequencing: single institution experience. 2021 ,	4
206	Osteogenesis imperfecta in children. 2021 , 148, 115914	2
205	A cross-sectional nationwide survey of osteosclerotic skeletal dysplasias in Japan. 2021 ,	0
204	Genotypic and Phenotypic Characteristics of 29 Patients With Rare Types of Osteogenesis Imperfecta: Average 5 Years of Follow-Up. 2021 , 12, 622078	
203	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

202	Muscle transcriptome in mouse models of osteogenesis imperfecta. 2021 , 148, 115940		2
201	Phenotypic Characterization of Immortalized Chondrocytes from a Desbuquois Dysplasia Type 1 Mouse Model: A Tool for Studying Defects in Glycosaminoglycan Biosynthesis. 2021 , 22,		
200	Zebrafish Models for Human Skeletal Disorders. 2021 , 12, 675331		0
199	The phenotypic spectrum of AMER1-related osteopathia striata with cranial sclerosis: The first Canadian cohort. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3793-3803	2.5	1
198	Role of rs193922155 in the etiopathogenesis of osteogenesis imperfecta with description of the phenotype: A case report. 2021 , 100, e27021		
197	A Roadmap to Gene Discoveries and Novel Therapies in Monogenic Low and High Bone Mass Disorders. 2021 , 12, 709711		3
196	Induction and expansion of human PRRX1 limb-bud-like mesenchymal cells from pluripotent stem cells. 2021 , 5, 926-940		3
195	Current state of the art in treatment of Mendelian disease: Skeletal dysplasias. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3359-3368	2.5	1
194	Cilia kinases in skeletal development and homeostasis. 2021 ,		
193	Whole Exome Sequencing Analysis in Fetal Skeletal Dysplasia Detected by Ultrasonography: An Analysis of 38 Cases. 2021 , 12, 728544		2
192	Splice Site Variant in Dogo Argentino Dogs with Disproportionate Dwarfism. 2021 , 12,		0
191	Congenital Disorders of Deficiency in Glycosaminoglycan Biosynthesis. 2021 , 12, 717535		7
190	Differential diagnosis of a diffuse sclerosis in an identified male skull (early 20th century Coimbra, Portugal): A multimethodological approach for the identification of osteosclerotic dysplasias in skeletonized individuals. 2021 , 34, 134-141		1
189	Splice Defect in Cane Corso Dogs with Dental-Skeletal-Retinal Anomaly (DSRA). 2021 , 12,		
188	The Role of Sonic Hedgehog in Human Holoprosencephaly and Short-Rib Polydactyly Syndromes. 2021 , 22,		3
187	Molecular diagnosis in a cohort of 114 patients with rare skeletal dysplasias. 2021 , 187, 396-408		0
186	Deciphering osteoarthritis genetics across 826,690 individuals from 9 populations. 2021 , 184, 4784-4818.e17		24
185	Neonatal hydrocephalus: an atypical presentation of malignant infantile osteopetrosis. 2021 , 37, 3695-3703		0

184	Foetal thoracic hypoplasia: concomitant anomalies and neonatal outcomes. 2021 , 1-6		
183	Congenital Metabolic Bone Disorders as a Cause of Bone Fragility. 2021 , 22,		0
182	Reply to "Genetics of Osteogenesis Imperfecta in Suspected Child Abuse". 2021 , 217, 1019-1020		1
181	Acrocapitofemoral dysplasia: Novel mutation in IHH in two adult patients from the third family in the literature and progression of the disease. 2021 , 64, 104343		
180	A novel intronic variant in PIGB in Acrofrontofacionasal dysostosis type 1 patients expands the spectrum of phenotypes associated with GPI biosynthesis defects. 2021 , 153, 116152		
179	De novo mutation in COL2A1 leads to lethal foetal skeletal dysplasia. 2021 , 153, 116169		1
178	B3GAT3-related linkeropathy and an in-frame homozygous deletion in an adult patient. 2021 , 64, 104342		2
177	Resveratrol Reduces COMPopathy in Mice Through Activation of Autophagy. 2021 , 5, e10456		4
176	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 517-527	2.5	1
175	First Report of Two Egyptian Patients with Desbuquois Dysplasia due to Homozygous Mutations. 2021 , 12, 279-288		0
174	A CRISPR-engineered swine model of COL2A1 deficiency recapitulates altered early skeletal developmental defects in humans. 2020 , 137, 115450		8
173	Prenatal diagnosis of skeletal dysplasias using whole exome sequencing in China. 2020 , 507, 187-193		9
172	Osteocyte Transcriptome Mapping Identifies a Molecular Landscape Controlling Skeletal Homeostasis and Susceptibility to Skeletal Disease.		3
171	The transcriptome of regenerating zebrafish scales identifies genes involved in human bone disease.		1
170	Gene Testing in Everyday Clinical Use: Lessons from the Bone Clinic. 2021 , 5, bvaa200		1
169	New developments in chondrocyte ER stress and related diseases. 2020 , 9,		11
168	Osteogenesis imperfecta: an update on clinical features and therapies. 2020 , 183, R95-R106		24
167	A trans-eQTL network regulates osteoclast multinucleation and bone mass. 2020 , 9,		10

- 166 Clinical and genetic characteristics of rare variants of acromelic skeletal dysplasias caused by mutations in the FBN1 gene. **2021**, 9, 327-337
- 165 Osteogenesis Imperfecta: Current and Prospective Therapies. **2021**, 11, 3
- 164 Diagnostic utility of next-generation sequencing-based panel testing in 543 patients with suspected skeletal dysplasia. **2021**, 16, 412 1
- 163 Mouse Dspp frameshift model of human dentinogenesis imperfecta. **2021**, 11, 20653 2
- 162 Expanding the Phenotype of the -Related Ciliopathy and Identification of Three Neurogenetic Disorders in a Single Family. **2021**, 12, 0
- 161 Pamidronate Therapy Increases Trabecular Bone Complexity of Mandibular Condyles in Individuals with Osteogenesis Imperfecta. **2021**, 1, 0
- 160 A trans-eQTL network regulates osteoclast multinucleation and bone mass.
- 159 Human iPSC-derived hypertrophic chondrocytes reveal a mutation-specific unfolded protein response in chondrodysplasias.
- 158 Clinical, Genetic and Orthopedic Characteristics of Desbuquois Dysplasia. **2021**, 27, 71-83
- 157 Skeletal disorders. **2020**, 369-379
- 156 Further delineation of a recognizable type of syndromic short stature caused by biallelic SEMA3A loss-of-function variants. *American Journal of Medical Genetics, Part A*, **2021**, 185, 889-893 2.5 3
- 155 Skeletal Dysplasia. **2021**, 121-135
- 154 Collagen Type 1 and Osteogenesis Imperfecta. **2020**, 125-129
- 153 Management of Osteogenesis Imperfecta: A Multidisciplinary Comprehensive Approach. **2020**, 12, 417-429 0
- 152 [Sclerosing hyperostotic bone disorders]. **2021**, 61, 1096-1106
- 151 Control of osteocyte dendrite formation by Sp7 and its target gene osteocrin. **2021**, 12, 6271 6
- 150 Resveratrol reduces COMPopathy in mice through activation of autophagy.
- 149 Spine radiograph in dysplasias: A pictorial essay. **2020**, 30, 436-447

- 148 Fetal Skeletal Dysplasias that Involve the Face: Binder Syndrome and Nager Syndrome. **2021**, 16, 140-144
- 147 Frameshift Variant in British Shorthair Cats with Complex Skeletal Dysplasia.. **2021**, 12,
- 146 Variable skeletal phenotypes associated with biallelic variants in. **2021**, 0
- 145 Orthopedic concerns of a child with short stature. **2021**, 34,
- 144 Development and validation of an expanded targeted sequencing panel for non-invasive prenatal diagnosis of sporadic skeletal dysplasia. **2021**, 14, 212 0
- 143 Perspective of the GEMSTONE Consortium on Current and Future Approaches to Functional Validation for Skeletal Genetic Disease Using Cellular, Molecular and Animal-Modeling Techniques.. **2021**, 12, 731217 1
- 142 Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability.. **2022**, 3, 100074 3
- 141 Prenatal Diagnosis of a Lethal Skeletal Dysplasia. **2021**, 22, e859-e865
- 140 How to select a key finding for a syndrome searching: A systemic approach. **2021**, 2, 280
- 139 Health-related Quality of Life in Adult Patients with Multiple Epiphyseal Dysplasia and Spondyloepiphyseal Dysplasia.. **2021**, 6, 20210048
- 138 Combination of osteogenesis imperfecta and type 1 diabetes mellitus. **2022**, 24, 470-476
- 137 Drug Treatment of Low Bone Mass and Other Bone Conditions in Pediatric Patients.. **2022**, 1
- 136 Fabric-elasticity relationships of tibial trabecular bone are similar in osteogenesis imperfecta and healthy individuals.. **2021**, 155, 116282
- 135 Regenerating zebrafish scales express a subset of evolutionary conserved genes involved in human skeletal disease.. **2022**, 20, 21 3
- 134 The Skeletal System. **2022**, 805-827
- 133 Osteogenesis Imperfecta: characterization of fractures during pregnancy and post-partum.. **2022**, 17, 22
- 132 Clinical and Genetic Characteristics of COL2A1-Associated Skeletal Dysplasias in 60 Russian Patients: Part I.. **2022**, 13, 0
- 131 SLC10A7, an orphan member of the SLC10 family involved in congenital disorders of glycosylation.. **2022**, 1 0

130	Risk Factors for Rebound After Correction of Genu Valgum in Skeletal Dysplasia Patients Treated by Tension Band Plates.. 2022 ,		
129	Functional Validation of Osteoporosis Genetic Findings Using Small Fish Models.. 2022 , 13,		0
128	Patient-reported prevalence of gastrointestinal issues in the adult skeletal dysplasia population with a concentration on osteogenesis imperfecta.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	1
127	An ultra-rare case of immunoskeletal dysplasia with neurodevelopmental abnormalities in an Indian patient with homozygous c.953C > T variant in EXTL3 gene: a case report.. 2022 , 22, 78		0
126	Oral bone biology.. 2022 ,		1
125	Changes in skeletal dysplasia nosology.. 2021 , 62, 689-696		
124	A de novo heterozygous HOXA11 variant in a patient with mesomelic dysplasia with urogenital abnormalities.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	0
123	Novel missense ACAN gene variants linked to familial osteochondritis dissecans cluster in the C-terminal globular domain of aggrecan.. 2022 , 12, 5215		0
122	A homozygous hypomorphic BNIP1 variant causes an increase in autophagosomes and reduced autophagic flux and results in a spondylo-epiphyseal dysplasia.. 2022 ,		0
121	A Novel Osteochondrodysplasia With Empty Sella Associates With a Variant.. 2022 , 13, 845889		0
120	Delineation of dual molecular diagnosis in patients with skeletal deformity.. 2022 , 17, 139		
119	Identification of variants in ACAN and PAPSS2 leading to spondyloepi(meta)physeal dysplasias in four Chinese families.. 2022 , e1916		0
118	Novel Loss-of-Function Mutations in Cause Acromesomelic Dysplasia, Maroteaux Type.. 2022 , 13, 823861		0
117	Osteogenesis Imperfecta/Ehlers-Danlos Overlap Syndrome and Neuroblastoma-Case Report and Review of Literature.. 2022 , 13,		
116	Clinical and genetic characterization of three Russian patients with pycnodysostosis due to pathogenic variants in the CTSK gene.. 2022 , e1904		
115	Things come in threes: A new complex allele and a novel deletion within the CFTR gene complicate an accurate diagnosis of cystic fibrosis.. 2022 , e1926		
114	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia.. 2022 ,		0
113	Further evidence for attenuated phenotype with variants in the BMPER gene causing DSD: Case report and literature review.. 2022 , 65, 104470		

112	Is Plasma C-Type Natriuretic Peptide Level Suitable for Diagnosing and Typing Skeletal Dysplasia?. 2022 , 3, 18-23	
111	DDRKG1 is required for the proper development and maintenance of the growth plate cartilage.. 2022 ,	0
110	The Treatment of Subtrochanteric Fracture with Reversed Contralateral Distal Femoral Locking Compression Plate (DF-LCP) Using a Progressive and Intermittent Drilling Procedure in Three Osteopetrosis Patients.. 2021 ,	0
109	Bones and Teeth. 2021 , 520-540	
108	Genetics of monogenic disorders of calcium and bone metabolism.. 2021 ,	0
107	Novel subtype of mucopolysaccharidosis caused by arylsulfatase K (ARSK) deficiency.. 2021 ,	6
106	Schmerzen, Bewegungseinschränkungen und entzündliche Veränderungen am Gelenk –immer eine Arthritis?. 2021 , 41, 393-400	
105	Physical Activity, Exercise, and Sports in Individuals with Skeletal Dysplasia: What Is Known about Their Benefits?. 2022 , 14, 4487	
104	The fibrillinopathies: new insights with focus on the paradigm of opposing phenotypes for both FBN1 and FBN2.. 2022 ,	1
103	Fetal Flat-Facies on Prenatal Ultrasound: Is it Chondrodysplasia Punctata? A Retrospective Chart Review of 62 Fetuses. 1	
102	A second individual with rhizomelic spondyloepimetaphyseal dysplasia and homozygous variant in GNPAT1.. 2022 , 104495	
101	Table_1.xlsx. 2020 ,	
100	Table_2.xlsx. 2020 ,	
99	Table_3.docx. 2020 ,	
98	Table_1.DOCX. 2020 ,	
97	A novel nonsense mutation of TGFBR1 in a fetus with atypical Loays-Dietz syndrome 1.. 2022 , 61, 127-128	
96	Clinical, radiological and molecular studies in 24 individuals with Dyggve-Melchior-Clausen dysplasia and Smith-McCort dysplasia from India.. 2022 ,	
95	The Modified Shields Classification and 12 Families with Defined DSPP Mutations. 2022 , 13, 858	0

94	Diagnostic distribution and postnatal evaluation of prenatally detected short femur: A single center experience.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	
93	Dissecting the phenotypic variability of osteogenesis imperfecta.. 2022 , 15,		1
92	Prenatal trio-based whole exome sequencing in fetuses with abnormalities of the skeletal system.. 2022 ,		1
91	Genetic spectrum of prenatally diagnosed skeletal dysplasias in a Finnish patient cohort.		
90	Computational Biology Insights into Genotype-Clinical Phenotype-Protein Phenotype Relationships between Novel SLC26A2 Variants Identified in Inherited Skeletal Dysplasias.		
89	Cartilage Homeostasis and Osteoarthritis. 2022 , 23, 6316		2
88	Pubertal growth in osteogenesis imperfecta caused by pathogenic variants in COL1A1/COL1A2. 2022 ,		0
87	Clinical Approach to Inborn Errors of Metabolism in Paediatrics. 2022 , 3-123		
86	Mosaic deletions of known genes explain skeletal dysplasias with high and low bone mass.		0
85	A missense mutation in DDRGK1 gene associated to Shohat-type spondyloepimetaphyseal dysplasia: Two case reports and a review of literature. <i>American Journal of Medical Genetics, Part A</i> ,	2.5	0
84	Biallelic KIF24 variants are responsible for a spectrum of skeletal disorders ranging from lethal skeletal ciliopathy to severe acromesomelic dysplasia.		
83	The MAP3K7 gene: further delineation of clinical characteristics and genotype/phenotype correlations.		
82	An Update on Animal Models of Osteogenesis Imperfecta.		
81	Spondyloepimetaphyseal dysplasia-Maroteaux type due to dominant TRPV4 mutation: expanding the phenotype with a case report.		
80	Fetal autopsy for the diagnosis of skeletal dysplasia and comparison with prenatal ultrasound findings over a 16-year period. 2022 ,		
79	Prenatal ultrasonographic features in Blomstrand osteochondrodysplasia: Antenatal case series confirmed by postmortem radiology and molecular diagnosis.		0
78	Molecular Basis for Hypochondroplasia in Japan. 2022 , 3, 428-432		
77	Prenatal Diagnosis of Jeune Syndrome Caused by Compound Heterozygous Variants in DYNC2H1 Gene: Case Report with Rapid WES Procedure and Differential Diagnosis of Lethal Skeletal Dysplasias. 2022 , 13, 1339		

- 76 Refining nosology by modelling variation among facial phenotypes: the RASopathies. *jmedgenet-2021-108366* ○
- 75 Functional Independence of Taiwanese Children with Osteogenesis Imperfecta. **2022**, 12, 1205 ○
- 74 Screening of Mineralogenic and Osteogenic Compounds in Zebrafish Tools to Improve Assay Throughput and Data Accuracy. **2022**, 15, 983 1
- 73 A novel variant in GNPAT1 gene causing a spondylo-epi-metaphyseal dysplasia resembling PGM3 Desbuquois like dysplasia. ○
- 72 Quality of Life in Children and Adolescents with Stickler Syndrome in Spain. **2022**, 9, 1255
- 71 Molecular identification of T-box transcription factor 6 and prognostic assessment in patients with congenital scoliosis: A single-center study. 9, ○
- 70 Early-onset osteoporosis: Rare monogenic forms elucidate the complexity of disease pathogenesis beyond type I collagen. ○
- 69 Dysplasias in the Child's Spine. **2022**,
- 68 Examining craniofacial variation among crispant and mutant zebrafish models of human skeletal diseases. ○
- 67 Exploration of the fetal skeleton by ultra-low-dose computed tomography: guidelines from the Fetal Imaging Task Force of the European Society of Paediatric Radiology.
- 66 Growing Pains: The Need for Engineered Platforms to Study Growth Plate Biology. 2200471 1
- 65 Clinical characteristics of 10 Chinese patients with melorheostosis and identification of a somatic MAP2K1 variant in one case.
- 64 Ellis-Van Creveld Syndrome: Clinical and Molecular Analysis of 50 Individuals. *jmedgenet-2022-108435* ○
- 63 Computational biology insights into genotype-clinical phenotype-protein phenotype relationships between novel SLC26A2 variants identified in inherited skeletal dysplasias. **2022**, 65, 104595
- 62 Exome sequencing revealed USP9X and COL2A1 mutations in a large family with multiple epiphyseal dysplasia. **2022**, 163, 116508 ○
- 61 Craniofacial and dental phenotype of two girls with osteogenesis imperfecta due to mutations in CRTAP. **2022**, 164, 116516
- 60 Bibliography. **2022**, 447-452 ○
- 59 Osteopetrosis: Gene-Based Nosology and Significance & SLC29A3, & Dysosteosclerosis. ○

- 58 Exploring and expanding the phenotype and genotype diversity in seven Chinese families with spondylo-epi-metaphyseal dysplasia. 13, 0
- 57 Biallelic variants in CHST3 cause Spondyloepiphyseal dysplasia with joint dislocations in three Pakistani kindreds. **2022**, 23, 0
- 56 Prenatal Cases Reflect the Complexity of the COL1A1/2 Associated Osteogenesis Imperfecta. **2022**, 13, 1578 0
- 55 High Bone Mass Disorders: New Insights from Connecting the Clinic and the Bench. 0
- 54 A Comprehensive Review on Collagen Type I Development of Biomaterials for Tissue Engineering: From Biosynthesis to Bioscaffold. **2022**, 10, 2307 6
- 53 Biallelic loss of function variants in EXOC6B are associated with impaired primary ciliogenesis and cause spondylo-epi-metaphyseal dysplasia with joint laxity type 3. 1
- 52 Case report: A homozygous ADAMTSL2 missense variant causes geleophysic dysplasia with high similarity to Weill-Marchesani syndrome. 13, 0
- 51 A Neonate with Dysmorphic Features and Respiratory Distress. **2022**, 23, e703-e707 0
- 50 Chronic pain in adults with osteogenesis imperfecta and its relationship to appraisal, coping, and quality of life: A cross-sectional study. **2022**, 101, e30256 0
- 49 A novel MTX2 gene splice site variant resulting in exon skipping, causing the recently described mandibuloacral dysplasia progeroid syndrome. 0
- 48 Proceedings of the 2022 Santa Fe Bone Symposium: Current Concepts in the Care of Patients with Osteoporosis and Metabolic Bone Diseases. **2022**, 0
- 47 Künstliche Intelligenz bei der Diagnose Seltener Erkrankungen: die Entwicklung der Phänotyp-Analyse. 1
- 46 Congenital systemic chondrodysplasia in a white lion (Panthera leo). 0
- 45 SLC4A2, another gene involved in acid-base balancing machinery of osteoclasts, causes osteopetrosis. **2022**, 116603 0
- 44 The incorporation of next-generation sequencing into pediatric care. **2022**, 0
- 43 Displasia tanatofica tipo II, una entidad congñita inusual. Reporte de caso. **2022**, 35, 0
- 42 Antenatal Phenotype of Desbuquois Dysplasia. 0
- 41 Novel missense COL2A1 variant in a fetus with achondrogenesis type II. **2022**, 9, 0

- 40 Natural history and genetic spectrum of the Turkish metaphyseal dysplasia cohort, including rare types caused by biallelic COL10A1, COL2A1, and LBR variants. **2022**, 116614 ○
- 39 Genetic analysis of 55 cases with fetal skeletal dysplasia. **2022**, 17, ○
- 38 Osteopetrosis: Gene-based nosology and significance dysosteosclerosis. **2022**, 116615 ○
- 37 Genetic testing and diagnostic strategies of fetal skeletal dysplasia: a preliminary study in Wu Han, China . ○
- 36 Fetal skeletal dysplasia cohort of a single tertiary referral center in Istanbul, Turkey. ○
- 35 A multi-omics study to characterize the transdifferentiation of human dermal fibroblasts to osteoblast-like cells. 9, ○
- 34 Novel SRY-box transcription factor 9 variant in campomelic dysplasia and the location of missense and nonsense variants along the protein domains: A case report. 10, ○
- 33 Mutant MESD links cellular stress to type I collagen aggregation in osteogenesis imperfecta type XX. **2023**, 115, 81-106 ○
- 32 Chiari I Malformations and the Heritable Disorders of Connective Tissue. **2023**, 34, 61-65 ○
- 31 A case of congenital multiple epiphyseal dysplasia from the Late Migration Period graveyard in Drnholec (Czech Republic). **2023**, 40, 33-40 ○
- 30 First case report of Nager syndrome patient from Georgia. **2022**, 10, 2050313X2211442 ○
- 29 A Patient with Bone Fragility, Multiple Fractures, Osteosarcoma, and the Variant c.143A>G in the IFITM5 Gene: A Case Report. Volume 14, 453-458 ○
- 28 Prenatal diagnosis of ALPL gene mutations in recurrent fetal skeletal dysplasia. **2022**, 61, 1065-1068 ○
- 27 Conclusion of diagnostic odysseys due to inversions disruptingGLI3andFBN1. jmedgenet-2022-108753 ○
- 26 Accuracy of ICD-10 Coding for Femoral Head Bearing Surfaces in Hip Arthroplasty. **2022**, ○
- 25 PCYT1A Missense Variant in Vizslas with Disproportionate Dwarfism. **2022**, 13, 2354 ○
- 24 Genome sequencing identifies a large non-coding region deletion of SNX10 causing autosomal recessive osteopetrosis. ○
- 23 Evaluation of first trimester ultrasound fetal biometry ratios femur length/biparietal diameter, femur length/abdominal circumference and femur length/foot for the screening of skeletal dysplasia. ○

- 22 Clinical variability in DYNC2H1-related skeletal ciliopathies includes Ellis-van Creveld syndrome. ○
- 21 A pictorial review of the radiographic skeletal findings in Morquio syndrome (mucopolysaccharidosis type IV). ○
- 20 Clinical and molecular characterization of a patient with MBTPS1 related spondyloepiphyseal dysplasia: Evidence of pathogenicity for a synonymous variant. 10, ○
- 19 IPSC reprogramming of two patients with spondyloepimetaphyseal dysplasia (SEMD, biglycan type). **2023**, 67, 103024 ○
- 18 Growth plate extracellular matrix defects and short stature in children. **2022**, 27, 247-255 ○
- 17 Schimke immunosseous dysplasia: an ultra-rare disease. a 20-year case series from the tertiary hospital in the Czech Republic. **2023**, 49, ○
- 16 Reduced glycolysis links resting zone chondrocyte proliferation in the growth plate. ○
- 15 Exome sequencing in fetuses with short long bones detected by ultrasonography: A retrospective cohort study. 14, ○
- 14 Skeletal Dysplasia Families: A Stepwise Approach to Diagnosis. **2023**, 43, ○
- 13 IPSC reprogramming of two patients with spondyloepiphyseal dysplasia congenita (SEDC). **2023**, 69, 103080 ○
- 12 A Chinese case of CHST3-related skeletal dysplasia and a systematic review. ○
- 11 From Genetics to Clinical Implications: A Study of 675 Dutch Osteogenesis Imperfecta Patients. **2023**, 13, 281 ○
- 10 Micro Ribonucleic Acid 9a (miR 9a) Antagonist Normalizes Bone Metabolism in Osteogenesis Imperfecta (OI) Mice Model. **2023**, 11, 465 ○
- 9 Nosology of genetic skeletal disorders: 2023 revision. **2023**, 191, 1164-1209 ○
- 8 Surgical Treatment of Chondrodysplasia Punctata Tibial-Metacarpal Type Until Skeletal Maturity. **2022**, 12, ○
- 7 Bisphosphonate treatment at s pondylo-ocular syndrome due to a novel compound heterozygote variant in XYLT2 and review of the literature. ○
- 6 Brain and craniovertebral junction in patients with achondroplasia using low dose dynamic computed tomography. **2023**, 4, 131-143 ○
- 5 Examining craniofacial variation among crispant and mutant zebrafish models of human skeletal diseases. ○

- 4 SP7: from Bone Development to Skeletal Disease. **2023**, 21, 241-252 ○
- 3 Lrp5 p. Val667Met Variant Compromises Bone Mineral Density and Matrix Properties in Osteoporosis. ○
- 2 Unequal Impact of COL1A1 and COL1A2 Variants on Dentinogenesis Imperfecta. 002203452311545 ○
- 1 Standardized growth charts for children with osteogenesis imperfecta. ○