Hitesh H Shah

List of Publications by Year
in descending order

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		331670	434195
94	2,681	21	31
papers	citations	h-index	g-index

95	95	95	2608
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Apache Hadoop YARN. , 2013, , .		1,314
2	Apache Tez. , 2015, , .		138
3	Duplications ofBHLHA9are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. Journal of Medical Genetics, 2012, 49, 119-125.	3.2	81
4	Analysis of the WISP3 gene in Indian families with progressive pseudorheumatoid dysplasia. American Journal of Medical Genetics, Part A, 2012, 158A, 2820-2828.	1.2	63
5	Deletions in PITX1 cause a spectrum of lower-limb malformations including mirror-image polydactyly. European Journal of Human Genetics, 2012, 20, 705-708.	2.8	63
6	Management of Severe Crouch Gait in Children and Adolescents With Cerebral Palsy. Journal of Pediatric Orthopaedics, 2010, 30, 832-839.	1.2	44
7	Congenital pseudarthrosis of the tibia: Management and complications. Indian Journal of Orthopaedics, 2012, 46, 616-626.	1.1	39
8	Quantitative Measures for Evaluating the Radiographic Outcome of Legg-CalvÃ©-Perthes Disease. Journal of Bone and Joint Surgery - Series A, 2013, 95, 354-361.	3.0	38
9	Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. Bone, 2018, 110, 368-377.	2.9	38
10	Perthes Disease. Orthopedic Clinics of North America, 2014, 45, 87-97.	1.2	37
11	Mutations in patients with osteogenesis imperfecta from consanguineous Indian families. European Journal of Medical Genetics, 2015, 58, 21-27.	1.3	37
12	Effect of Prophylactic Trochanteric Epiphyseodesis in Older Children With Perthes' Disease. Journal of Pediatric Orthopaedics, 2009, 29, 889-895.	1.2	35
13	Congenital Pseudarthrosis of the Tibia Treated With Intramedullary Rodding and Cortical Bone Grafting. Journal of Pediatric Orthopaedics, 2011, 31, 79-88.	1.2	35
14	Clinical utility of fetal autopsy and its impact on genetic counseling. Prenatal Diagnosis, 2015, 35, 685-691.	2.3	34
15	Congenital posteromedial bowing of the tibia: a retrospective analysis of growth abnormalities in the leg. Journal of Pediatric Orthopaedics Part B, 2009, 18, 120-128.	0.6	33
16	Identification of a novel LRRK1 mutation in a family with osteosclerotic metaphyseal dysplasia. Journal of Human Genetics, 2017, 62, 437-441.	2.3	33
17	Pre-axial mirror polydactyly associated with tibial deficiency: a study of the patterns of skeletal anomalies of the foot and leg. Journal of Children's Orthopaedics, 2007, 1, 49-54.	1.1	31
18	GALNS mutations in Indian patients with mucopolysaccharidosis IVA. American Journal of Medical Genetics, Part A, 2014, 164, 2793-2801.	1.2	31

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19	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. American Journal of Medical Genetics, Part A, 2016, 170, 410-417.	1.2	31
20	A novel sequence variant in SFRP4 causing Pyle disease. Journal of Human Genetics, 2017, 62, 575-576.	2.3	27
21	Mutation spectrum of COL1A1 and COL1A2 genes in Indian patients with osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2014, 164, 1482-1489.	1.2	24
22	What Factors Influence Union and Refracture of Congenital Pseudarthrosis of the Tibia? A Multicenter Long-term Study. Journal of Pediatric Orthopaedics, 2018, 38, e332-e337.	1.2	24
23	Environmental Tobacco and Wood Smoke Increase the Risk of Legg-CalvÃ©-Perthes Disease. Clinical Orthopaedics and Related Research, 2012, 470, 2369-2375.	1.5	23
24	The Sphericity Deviation Score. Journal of Pediatric Orthopaedics, 2014, 34, 522-528.	1.2	22
25	Recurrent and novel GLB1 mutations in India. Gene, 2015, 567, 173-181.	2.2	22
26	Novel and recurrent mutations in WISP3 and an atypical phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 2481-2484.	1.2	21
27	To What Extent Does Remodeling of the Proximal Femur and the Acetabulum Occur Between Disease Healing and Skeletal Maturity in Perthes Disease?. Journal of Pediatric Orthopaedics, 2008, 28, 711-716.	1.2	19
28	A child with Erysipelothrix arthritisâ€“beware of the little known. Asian Pacific Journal of Tropical Biomedicine, 2012, 2, 503-504.	1.2	19
29	Phenotype and genotype in patients with Larsen syndrome: clinical homogeneity and allelic heterogeneity in seven patients. BMC Medical Genetics, 2016, 17, 27.	2.1	18
30	Second family provides further evidence for causation of Steel syndrome by biallelic mutations in <scp>COL27A1</scp>. Clinical Genetics, 2017, 92, 323-326.	2.0	18
31	A novel multiple joint dislocation syndrome associated with a homozygous nonsense variant in the EXOC6B gene. European Journal of Human Genetics, 2016, 24, 1206-1210.	2.8	16
32	Evolution of Legg-CalvÃ©-Perthes disease following proximal femoral varus osteotomy performed in the avascular necrosis stage: A prospective study. Journal of Children's Orthopaedics, 2020, 14, 58-67.	1.1	16
33	Quadricepsplasty for congenital dislocation of the knee and congenital quadriceps contracture. Journal of Children's Orthopaedics, 2012, 6, 397-410.	1.1	15
34	Does proximal femoral varus osteotomy in Leggâ€“CalvÃ©â€“Perthes disease predispose to angular mal-alignment of the knee? A clinical and radiographic study at skeletal maturity. Journal of Children's Orthopaedics, 2013, 7, 205-211.	1.1	15
35	A novel mutation (g.106737G>T) in zone of polarizing activity regulatory sequence (ZRS) causes variable limb phenotypes in Werner mesomelia. American Journal of Medical Genetics, Part A, 2014, 164, 898-906.	1.2	15
36	Clinical and genetic spectrum of 104 Indian families with central nervous system white matter abnormalities. Clinical Genetics, 2021, 100, 542-550.	2.0	12

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37	Management of the Knee Problems in Spastic Cerebral Palsy. Indian Journal of Orthopaedics, 2019, 53, 53-62.	1.1	12
38	Exome Sequencing Identifies a Dominant <i>TNNT3</i> Mutation in a Large Family with Distal Arthrogryposis. Molecular Syndromology, 2014, 5, 218-228.	0.8	11
39	Ageâ€•and genderâ€•related reference ranges for thromboelastography from a healthy Indian population. International Journal of Laboratory Hematology, 2020, 42, 180-189.	1.3	11
40	Donor site morbidity following the harvesting of cortical bone graft from the tibia in children. Journal of Children's Orthopaedics, 2010, 4, 417-421.	1.1	10
41	Seven additional families with spondylocarpotarsal synostosis syndrome with novel biallelic deleterious variants in FLNB. Clinical Genetics, 2018, 94, 159-164.	2.0	10
42	Phenotyping and genotyping of skeletal dysplasias: Evolution of a center and a decade of experience in India. Bone, 2019, 120, 204-211.	2.9	10
43	Validation of Pediatric Self-Report Patient-Reported Outcomes Measurement Information System (PROMIS) Measures in Different Stages of Legg-CalvÃ©-Perthes Disease. Journal of Pediatric Orthopaedics, 2020, 40, 235-240.	1.2	9
44	Hemiepiphysiodesis using 2-holed reconstruction plate for correction of angular deformity of the knee in children. Journal of Orthopaedics, 2020, 20, 54-59.	1.3	8
45	Controversies in the management of pediatric neck femur fractures- a systematic review. Journal of Orthopaedics, 2021, 27, 92-102.	1.3	8
46	Second report of slipped capital femoral epiphysis in Rubinsteinâ€“Taybi syndrome. Clinical Dysmorphology, 2011, 20, 55-57.	0.3	7
47	Late presented case of distal humerus epiphyseal separation in a newborn. BMJ Case Reports, 2016, 2016, bcr2016215296.	0.5	7
48	The fate of the joint space in Leggâ€“CalvÃ©â€“Perthesâ€™ disease. Skeletal Radiology, 2013, 42, 341-345.	2.0	6
49	Pycnodysostosis: Novel Variants in CTSK and Occurrence of Giant Cell Tumor. Journal of Pediatric Genetics, 2018, 07, 009-013.	0.7	6
50	Biallelic variants p.Arg1133Cys and p.Arg1379Cys in COL2A1: Further delineation of phenotypic spectrum of recessive Type 2 collagenopathies. American Journal of Medical Genetics, Part A, 2020, 182, 338-347.	1.2	6
51	Surgical management of the congenital dislocation of the knee and hip in children presented after six months of age. International Orthopaedics, 2020, 44, 2635-2644.	1.9	6
52	Orthopedic manifestation as the presenting symptom of acute lymphoblastic leukemia. Journal of Orthopaedics, 2020, 22, 326-330.	1.3	6
53	Does the timing of treatment affect complications of pediatric femoral neck fractures?. Journal of Orthopaedics, 2020, 22, 207-212.	1.3	6
54	A syndrome of facial dysmorphism, cubital pterygium, short distal phalanges, swan neck deformity of fingers, and scoliosis. American Journal of Medical Genetics, Part A, 2014, 164, 1035-1040.	1.2	5

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55	Congenital pseudoarthrosis of the radius in Neurofibromatosis Type 1: An entity not to be missed!. Journal of Orthopaedics, 2020, 22, 427-430.	1.3	4
56	Demographics and Clinical Presentation of Early-Stage Legg-CalvÃ©-Perthes Disease: A Prospective, Multicenter, International Study. Journal of the American Academy of Orthopaedic Surgeons, The, 2021, 29, e85-e91.	2.5	4
57	Further characterization of acro-renal-uterine-mandibular syndrome. Clinical Dysmorphology, 2012, 21, 83-86.	0.3	3
58	Handedness in diplegic cerebral palsy. Developmental Neurorehabilitation, 2012, 15, 386-389.	1.1	3
59	An emerging ribosomopathy affecting the skeleton due to biallelic variations in NEPRO. American Journal of Medical Genetics, Part A, 2019, 179, 1709-1717.	1.2	3
60	Does early and aggressive management of significant extrusion of the femoral head affect the outcome of Perthesâ€™ disease with the age of onset younger than 7Âyears?. Musculoskeletal Surgery, 2022, 106, 325-335.	1.5	3
61	A rare combination of amniotic constriction band with osteogenesis imperfecta. BMJ Case Reports, 2015, 2015, bcr2015212400-bcr2015212400.	0.5	3
62	Posttraumatic Static Volar Intercalated Segment Instability - Iatrogenic or Missed Injury. Journal of Orthopaedic Case Reports, 2016, 6, 59-61.	0.1	3
63	A homozygous hypomorphicBNIP1variant causes an increase in autophagosomes and reduced autophagic flux and results in a spondyloâ€epiphyseal dysplasia. Human Mutation, 2022, 43, 625-642.	2.5	3
64	Growth retardation, intellectual disability, facial anomalies, cataract, thoracic hypoplasia, and skeletal abnormalities: A novel phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2941-2945.	1.2	2
65	Thorn-induced pseudotumour of the fibula. BMJ Case Reports, 2014, 2014, bcr2014204115-bcr2014204115.	0.5	2
66	Tibial hypoplasia with a bifid tibia: an unclassified tibial hemimelia. BMJ Case Reports, 2016, 2016, bcr2016216622.	0.5	2
67	Unclassified tibial hemimelia. BMJ Case Reports, 2016, 2016, bcr2016215305.	0.5	2
68	Growth, sexual development and menstrual issues among girls with cerebral palsy â€“ A cross sectional study in a tertiary care centre. Clinical Epidemiology and Global Health, 2019, 7, 367-371.	1.9	2
69	Demographic and Practice Variability Amongst Indian Centres in a Multicentre Prospective Observational Study on Developmental Dysplasia of the Hip. Indian Journal of Orthopaedics, 2021, 55, 1559-1567.	1.1	2
70	A review of skeletal dysplasia research in India. Journal of Postgraduate Medicine, 2018, 64, 98-103.	0.4	2
71	The â€œDiscoid Epiphysisâ€â€”An Uncommon Presentation of Legg-CalvÃ©-Perthes Disease. Journal of Pediatric Orthopaedics, 2022, 42, e570-e576.	1.2	2
72	Primary protrusio acetabuli in childhood. Pediatric Radiology, 2010, 40, 55-55.	2.0	1

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73	Loss of a condyle of the femur or tibia following septic arthritis in infancy: problems of management and testing of a hypothesis of pathogenesis. Journal of Children's Orthopaedics, 2012, 6, 319-325.	1.1	1
74	The fate of the hip in spondylo-epi-metaphyseal dysplasia: clinical and radiological evaluation of adults with SEMD Handigodu type. Skeletal Radiology, 2012, 41, 939-945.	2.0	1
75	Comparison of plaster-of-Paris casts and Woodcast splints for immobilization of the limb during serial manipulation and casting for idiopathic clubfoot in infants. Bone and Joint Journal, 2020, 102-B, 1399-1404.	4.4	1
76	A rare case of rubber band syndrome of wrist with distal radius and ulna fracture. Journal of Orthopaedics, 2020, 20, 60-62.	1.3	1
77	The spectrum of tibial pseudarthrosis with constriction band syndrome in children. Journal of Pediatric Orthopaedics Part B, 2021, Publish Ahead of Print, .	0.6	1
78	Pseudoachondroplasia: Phenotype and genotype in 11 Indian patients. American Journal of Medical Genetics, Part A, 2022, 188, 751-759.	1.2	1
79	Reliability of 3 Radiologic Classifications for the Severity of the Developmental Dysplasia of the Hip in Children Older Than 4 Years. Journal of Pediatric Orthopaedics, 2021, Publish Ahead of Print, 23-29.	1.2	1
80	An unusual presentation, novel treatment with Meropenem PMMA beads and complications of Klebsiella osteomyelitis in a healthy adult- A case report. Journal of Clinical Orthopaedics and Trauma, 2022, 24, 101719.	1.5	1
81	Radiologic Outcomes of Bilateral and Unilateral Perthes Disease: A Comparative Cohort Study. Journal of Pediatric Orthopaedics, 2022, 42, e168-e173.	1.2	1
82	Steel syndrome: Report of three patients, including monozygotic twins and review of clinical and mutation profiles. European Journal of Medical Genetics, 2022, 65, 104521.	1.3	1
83	SP6-38 Tobacco smoke and the risk of Perthes' disease in south west India: a case-control study. Journal of Epidemiology and Community Health, 2011, 65, A465-A465.	3.7	0
84	Hypoplasia/aplasia of pelvis, femora, fibulae, ulna, digits and nails. Clinical Dysmorphology, 2011, 20, 205-209.	0.3	0
85	Talonavicular Arthrodesis for the Treatment of Neurological Flat Foot Deformity in Pediatric Patients. Journal of Pediatric Orthopaedics, 2013, 33, e39.	1.2	0
86	A Rare Case of Congenital Tibiofemoral Fusion with Bilateral Proximal Femoral Focal Deficiency. JBJS Case Connector, 2017, 7, e22-e22.	0.3	0
87	Valgus-impacted fracture of neck of femur in a 12-year-old child. BMJ Case Reports, 2021, 14, e240707.	0.5	0
88	Slipped capital femoral epiphysis in a healed Perthes hip. BMJ Case Reports, 2021, 14, e243977.	0.5	0
89	Post-operative Hypertension following Correction of Flexion Deformity of the Knees in a Spastic Diplegic Child: A Case Report. Malaysian Orthopaedic Journal, 2016, 10, 46-48.	0.5	0
90	Fracture neck of femur in Factor XIII deficiency: Was better outcome possible?. Journal of Family Medicine and Primary Care, 2017, 6, 651.	0.9	0

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91	Knowledge among Mothersâ€™ of Children and Youth with Hemophilia-A Cross Sectional Survey at a Hemophilia Center. Indian Journal of Public Health Research and Development, 2018, 9, 27.	0.0	0
92	Management of Osteogenesis Imperfecta in India. , 2020, , 265-285.		0
93	Does the Deformity Index Reliably Predict the Shape of the Femoral Head at Healing of Legg-CalvÃ©-Perthes Disease?. Journal of Pediatric Orthopaedics, 2022, 42, e163-e167.	1.2	0
94	A systematic review of maternal diabetes and congenital skeletal malformation. Congenital Anomalies (discontinued), 2022, , .	0.6	0