Ali Abdul Salam Awni Al Kaissi

List of Publications by Year
in descending order

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122	1,012	567281	552781
papers	citations	15 h-index	26 g-index

126	126	126	1727
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. Nature Genetics, 2011, 43, 197-203.	21.4	229
2	CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. American Journal of Human Genetics, 2017, 101, 391-403.	6.2	35
3	Distinctive spine abnormalities in patients with Goldenhar syndrome: tomographic assessment. European Spine Journal, 2015, 24, 594-599.	2.2	30
4	Musculo-Skeletal Abnormalities in Patients with Marfan Syndrome. Clinical Medicine Insights: Arthritis and Musculoskeletal Disorders, 2013, 6, CMAMD.S10279.	1.2	27
5	Atlanto-axial segmentation defects and os odontoideum in two male siblings with opsismodysplasia. Skeletal Radiology, 2009, 38, 293-296.	2.0	24
6	Persistent torticollis, facial asymmetry, grooved tongue, and dolicho-odontoid process in connection with atlas malformation complex in three family subjects. European Spine Journal, 2007, 16, 265-270.	2.2	22
7	The Diversity of the Clinical Phenotypes in Patients With Fibrodysplasia Ossificans Progressiva. Journal of Clinical Medicine Research, 2016, 8, 246-253.	1.2	22
8	The diagnosis and management of patients with idiopathic osteolysis. Pediatric Rheumatology, 2011, 9, 31.	2.1	20
9	Caudal regression syndrome and popliteal webbing in connection with maternal diabetes mellitus: a case report and literature review. Cases Journal, 2008, 1, 407.	0.4	19
10	Mid-diaphyseal Endosteal Thickening With Subsequent Medullary Narrowing in a Patient With Hallermann-Streiff Syndrome. Journal of Clinical Medicine Research, 2011, 3, 328-30.	1.2	19
11	A novel mutation in ATRX associated with intellectual disability, syndromic features, and osteosarcoma. Pediatric Blood and Cancer, 2017, 64, e26522.	1.5	18
12	The Managment of cervical spine abnormalities in children with spondyloepiphyseal dysplasia congenita. Medicine (United States), 2019, 98, e13780.	1.0	17
13	Craniocervical junction malformation in a child with Oromandibular-limb hypogenesis-MÃ¶bius syndrome. Orphanet Journal of Rare Diseases, 2007, 2, 2.	2.7	16
14	Osteochondritis dissecans and Osgood Schlatter disease in a family with Stickler syndrome. Pediatric Rheumatology, 2009, 7, 4.	2.1	16
15	Asymmetrical skull, ptosis, hypertelorism, high nasal bridge, clefting, umbilical anomalies, and skeletal anomalies in sibs: Is Carnevale syndrome a separate entity?. American Journal of Medical Genetics, Part A, 2007, 143A, 349-354.	1.2	15
16	Extra phenotypic features in a girl with Miller syndrome. Clinical Dysmorphology, 2011, 20, 66-72.	0.3	15
17	Broad Spectrum of Skeletal Malformation Complex in Patients with Cleidocranial Dysplasia Syndrome: Radiographic and Tomographic Study. Clinical Medicine Insights: Arthritis and Musculoskeletal Disorders, 2013, 6, CMAMD.S11933.	1.2	15
18	Schmid's Type of Metaphyseal Chondrodysplasia: Diagnosis and Management. Orthopaedic Surgery, 2018, 10, 241-246.	1.8	15

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19	Treatment of Varus Deformities of the Lower Limbs in Patients with Achondroplasia and Hypochondroplasia. The Open Orthopaedics Journal, 2013, 7, 33-39.	0.2	14
20	Corrections of Lower Limb Deformities in Patients with Diastrophic Dysplasia. Orthopaedic Surgery, 2014, 6, 274-279.	1.8	13
21	The management of knee dislocation in a child with Larsen syndrome. Clinics, 2011, 66, 1295-1299.	1.5	13
22	Hypohidrotic ectodermal dysplasia with tibial aplasia. Clinical Dysmorphology, 2002, 11, 175-178.	0.3	12
23	Robinow syndrome: Report of two cases and review of the literature. Journal of Medical Imaging and Radiation Oncology, 2007, 51, 83-86.	0.6	12
24	Premature Osteoarthritis as Presenting Sign of Type II Collagenopathy: A Case Report and Literature Review. Seminars in Arthritis and Rheumatism, 2013, 42, 355-360.	3.4	12
25	Reconstruction of Limb Deformities in Patients with Thrombocytopeniaâ€absent Radius Syndrome. Orthopaedic Surgery, 2015, 7, 50-56.	1.8	12
26	Windswept lower limb deformities in patients with hypophosphataemic rickets. Swiss Medical Weekly, 2013, 143, w13904.	1.6	12
27	Familial vertebral segmentation defects, Sprengel anomaly, and omovertebral bone with variable expressivity. American Journal of Medical Genetics, Part A, 2005, 138A, 374-378.	1.2	11
28	Occipitoatlantoaxial junction malformation and early onset senile ankylosing vertebral hyperostosis in a girl with MURCS association. American Journal of Medical Genetics, Part A, 2009, 149A, 470-474.	1.2	11
29	A novel form of ischio-vertebral syndrome. Skeletal Radiology, 2007, 36, 77-81.	2.0	9
30	A novel syndrome resembling Desbuquois dysplasia. American Journal of Medical Genetics, Part A, 2005, 132A, 68-75.	1.2	8
31	Progressive non-infectious anterior vertebral fusion, split cord malformation and situs inversus visceralis. BMC Musculoskeletal Disorders, 2006, 7, 94.	1.9	8
32	Progressive acetabular dysplasia in a boy with mucopolysaccharoidosis type IV A (Morquio syndrome): a case report. Cases Journal, 2008, 1, 410.	0.4	8
33	Advanced ossification of the carpal bones, and monkey wrench appearance of the femora, features suggestive of a propable mild form of desbeqious dysplasia: a case report and review of the literature. Cases Journal, 2009, 2, 45.	0.4	8
34	Evidence of Reduced Bone Turnover and Disturbed Mineralization Process in a Boy with Stickler Syndrome. Calcified Tissue International, 2010, 86, 126-131.	3.1	8
35	Radiographic and Tomographic Analysis in Patients with Stickler Syndrome Type I. International Journal of Medical Sciences, 2013, 10, 1250-1258.	2.5	8
36	Congenital contractures and distinctive phenotypic features consistent with Stuve-Wiedmann syndrome in a male infant. Cases Journal, 2008, 1, 121.	0.4	7

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37	Ball and socket ankle joint in connection with bilateral tarsal synostosis in a boy with congenital absence of the portal vain: a novel malformation complex. Cases Journal, 2008, 1, 76.	0.4	7
38	Significant traumatic atrophy of the spinal cord in connection with severe cervical vertebral body hypoplasia in a boy with Larsen syndrome: a case report and review of the literature. Cases Journal, 2009, 2, 6729.	0.4	7
39	A child with splitâ€hand/foot associated with tibial hemimelia (SHFLD syndrome) and thrombocytopenia maps to chromosome region 17p13.3. American Journal of Medical Genetics, Part A, 2014, 164, 2338-2343.	1.2	7
40	Spine malformation complex in 3 diverse syndromic entities. Medicine (United States), 2016, 95, e5505.	1.0	7
41	Bone matrix hypermineralization associated with low bone turnover in a case of Nasu-Hakola disease. Bone, 2019, 123, 48-55.	2.9	7
42	Skeletal phenotype/genotype in progressive pseudorheumatoid chondrodysplasia. Clinical Rheumatology, 2020, 39, 553-560.	2.2	7
43	Lower limbs deformities in patients with McCune-Albright syndrome: Tomography and treatment. African Journal of Paediatric Surgery, 2016, 13, 125.	0.6	7
44	Progressive Congenital Torticollis in VATER Association Syndrome. Spine, 2006, 31, E376-E378.	2.0	6
45	Arthrogryposis multiplex congenital in a child manifesting phenotypic features resembling dysosteosclerosis/osteosclerosis malformation complex; 3DCT scan analysis of the skull base. Cases Journal, 2008, 1, 56.	0.4	6
46	How frequent is osteogenesis imperfecta in patients with idiopathic osteoporosis?. Medicine (United) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.0	6
47	Atlanto-axial rotatory fixation in a girl with Spondylocarpotarsal synostosis syndrome. Scoliosis, 2006, 1, 15.	0.4	5
48	Achondroplasia manifesting as enchondromatosis and ossification of the spinal ligaments: a case report. Journal of Medical Case Reports, 2008, 2, 263.	0.8	5
49	Progressive noninfectious anterior vertebral fusion in a girl with axial mesodermal dysplasia spectrum. Clinical Dysmorphology, 2008, 17, 65-68.	0.3	5
50	Femoral-tibial-synostosis in a child with Roberts syndrome (Pseudothalidomide): a case report. Cases Journal, 2008, 1, 109.	0.4	5
51	Significant ophthalmoarthropathy associated with ectodermal dysplasia in a child with Marshall-Stickler overlap: a case report. Cases Journal, 2008, 1, 270.	0.4	5
52	Acroform type of enchondromatosis associated with severe vertebral involvement and facial dysmorphism in a boy with a new variant of enchondromatosis type I1 of Spranger: case report and a review of the literature. Cases Journal, 2008, 1, 324.	0.4	5
53	Professional awareness is needed to distinguish between child physical abuse from other disorders that can mimic signs of abuse (Skull base sclerosis in infant manifesting features of infantile cortical) Tj ETQq1 1 0.784314 rgBT /Over		
54	Is Webbing (Pterygia) a Constant Feature in Patients with Escobar Syndrome?. Orthopaedic Surgery, 2013, 5, 297-301.	1.8	5

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55	Agenesis of the Corpus Callosum and Skeletal Deformities in Two Unrelated Patients: Analysis via MRI and Radiography. Case Reports in Orthopedics, 2014, 2014, 1-5.	0.3	5
56	The constellation of skeletal deformities in a family with mixed types of mucopolysaccharidoses. Medicine (United States), 2016, 95, e4561.	1.0	5
57	Clinical and Genetic Heterogeneity in Six Tunisian Families With Horizontal Gaze Palsy With Progressive Scoliosis: A Retrospective Study of 13 Cases. Frontiers in Pediatrics, 2020, 8, 172.	1.9	5
58	Can Multiple Hereditary Exostoses Overlap With Mesomelic Dysplasia?. Journal of Clinical Medicine Research, 2016, 8, 605-609.	1.2	5
59	Craniovertebral malformation complex in a child with Weismann-Netter-Stuhl syndrome. Jornal De Pediatria, 2006, 82, 236-239.	2.0	5
60	Corrections of diverse forms of lower limb deformities in patients with mucopolysaccharidosis type IVA (Morquio syndrome). African Journal of Paediatric Surgery, 2016, 13, 88.	0.6	5
61	Windswept deformity in a patient with Schwartz-Jampel syndrome. Swiss Medical Weekly, 2012, 142, w13519.	1.6	5
62	Unusual facies, thumb hypoplasia, distinctive spinal fusions and extraspinal mobility limitation, in a pair of monozygotic twins. Clinical Dysmorphology, 2007, 16, 151-155.	0.3	4
63	Vertebral hyperostosis, ankylosed vertebral fracture and atlantoaxial rotatory subluxation in an elderly patient with a history of infantile idiopathic scoliosis; a case report. Journal of Medical Case Reports, 2007, 1, 25.	0.8	4
64	Ischiopubic and odontoid synchondrosis in a boy with progressive pseudorheumatoid chondrodysplasia. Pediatric Rheumatology, 2007, 5, 19.	2.1	4
65	A hypoplastic atlas and long odontoid process in a girl manifesting phenotypic features resembling spondyloepimetaphyseal dysplasia joint laxity syndrome. Skeletal Radiology, 2008, 37, 469-473.	2.0	4
66	A patient with Melorheostosis manifesting with features similar to tricho-dento-osseous syndrome: a case report. Journal of Medical Case Reports, 2008, 2, 51.	0.8	4
67	The aetiology behind torticollis and variable spine defects in patients with MÃ¼llerian duct/renal aplasia-cervicothoracic somite dysplasia syndrome: 3D CT scan analysis. European Spine Journal, 2011, 20, 1720-1727.	2.2	4
68	Reâ€alignmentâ€procedures for Skeletal Dysplasia in Three Patients with Genetically Diverse Syndromes. Orthopaedic Surgery, 2013, 5, 33-39.	1.8	4
69	Tomographic Study of the Malformation Complex in Correlation With the Genotype in Patients With Robinow Syndrome: Review Article. Journal of Investigative Medicine High Impact Case Reports, 2020, 8, 232470962091177.	0.6	4
70	Fractures in connection with an atypical form of craniodiaphyseal dysplasia: case report of a boy and his mother. Clinics, 2012, 67, 1505-1509.	1.5	4
71	Progressive vertebral fusion in a girl with spinal enchondromatosis. European Journal of Radiology Extra, 2007, 63, 125-129.	0.1	3
72	A novel malformation complex of bilateral and symmetric preaxial radial ray-thumb aplasia and lower limb defects with minimal facial dysmorphic features: a case report and literature review. Cases Journal, 2008, 1, 271.	0.4	3

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73	Outward bulging of the right parietal bone in connection with fibrous dysplasia in an infant: a case report. Cases Journal, 2008, 1, 347.	0.4	3
74	Swellings over the Limbs as the Earliest Feature in a Patient with Osteogenesis Imperfecta Type V. Case Reports in Orthopedics, 2014, 2014, 1-4.	0.3	3
75	Spinal and pelvic corrections in a patient with spondylocostal dysplasia syndrome and hemimyelomeningocele. African Journal of Paediatric Surgery, 2014, 11, 341.	0.6	3
76	Progressive Collapse of the Thoracic Cage. American Journal of Medicine, 2016, 129, e1-e4.	1.5	3
77	Muscle Weakness. Journal of Investigative Medicine High Impact Case Reports, 2017, 5, 232470961668958.	0.6	3
78	Craniosynostosis, Scheuermann's disease, and intellectual disability resembling Shprintzenâ€“Goldberg syndrome. Medicine (United States), 2017, 96, e6199.	1.0	3
79	Neonatal Death Dwarfism in a Girl with Distinctive Bone Dysplasia Compatible with Grebe Chondrodysplasia: Analysis by CT Scan-based Phenotype. Journal of Clinical Imaging Science, 2014, 4, 53.	1.1	3
80	Subtotal amelia in a child with autosomal recessive hypohidrotic ectodermal dysplasia. African Health Sciences, 2005, 5, 270-5.	0.7	3
81	Diffuse Skull Base/Cervical Fusion Syndromes in Two Siblings With Spondylocostal Dysostosis Syndrome. Spine, 2008, 33, E425-E428.	2.0	2
82	Distinctive tomographic features of atlantoaxial dislocation in a boy with acromesomelic dysplasia du Pan syndrome. Clinical Dysmorphology, 2009, 18, 122-126.	0.3	2
83	Synophyrs, curly eyelashes and Ptyrigium colli in a girl with Desbuquois dysplasia: a case report and review of the literature. Cases Journal, 2009, 2, 7873.	0.4	2
84	Facial dysmorphism associated with distinctive spine abnormalities in a girl and her mother. Clinical Dysmorphology, 2011, 20, 182-186.	0.3	2
85	Reconstruction of bilateral tibial aplasia and split hand-foot syndrome in a father and daughter. African Journal of Paediatric Surgery, 2014, 11, 3.	0.6	2
86	Correction of the axial and appendicular deformities in a patient with Silver-Russel syndrome. African Journal of Paediatric Surgery, 2015, 12, 36.	0.6	2
87	Spinal and extraspinal deformities in a patient with dysspondyloenchondromatosis. GMS German Medical Science, 2013, 11, Doc06.	2.7	2
88	Distinctive Tomographic Abnormalities of the Craniocervical Region in a Patient with Osteogensis Imperfecta Type IV B. Clinics, 2010, 65, 647-649.	1.5	2
89	Dolicho-odontoid in a boy with pseudoachondroplasia. European Journal of Orthopaedic Surgery and Traumatology, 2008, 18, 297-301.	1.4	1
90	Persistent cloaca associated with a duplicated left leg: a novel disorganization-like syndrome. Clinical Dysmorphology, 2008, 17, 137-139.	0.3	1

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91	Progressive joint limitations as the first alarming signs in a boy with short â€“ limbed dwarfism: A case report. Cases Journal, 2008, 1, 112.	0.4	1
92	Tomographic assessment of the spine in children with spondylocostal dysotosis syndrome. Clinics, 2010, 65, 953-959.	1.5	1
93	Distinctive Vertebral Abnormalities in a Patient with VACTERL Association. RoFo Fortschritte Auf Dem Gebiet Der Rontgenstrahlen Und Der Bildgebenden Verfahren, 2012, 184, 62-65.	1.3	1
94	Spinal Exostosis in a Boy with Multiple Hereditary Exostoses. Case Reports in Orthopedics, 2013, 2013, 1-4.	0.3	1
95	Axial correction of the lower limb deformities in a girl with anauxetic dysplasia. Musculoskeletal Surgery, 2014, 98, 71-75.	1.5	1
96	Extended phenotypes in a boy and his mother with otoâ€‰palatoâ€‰digitalâ€‰syndrome type <scp>II</scp>. Clinical Case Reports (discontinued), 2015, 3, 762-766.	0.5	1
97	Bilateral and Symmetrical Anteromedial Bowing of the Lower Limbs in a Patient with Neurofibromatosis Type-I. Case Reports in Orthopedics, 2015, 2015, 1-4.	0.3	1
98	Turning the backbone into an ankylosed concrete-like structure. Medicine (United States), 2018, 97, e0278.	1.0	1
99	Massive Axial and Appendicular Skeletal Deformities in Connection with Gorham-Stout Syndrome. Medicines (Basel, Switzerland), 2019, 6, 54.	1.4	1
100	Progressive Deformity of the Lower Limbs in a Patient with KID (Keratitis-Ichthyosis-Deafness) Syndrome. Case Reports in Orthopedics, 2020, 2020, 1-5.	0.3	1
101	Upper Limb Pathology in Children with Mucopolysaccharidoses. TravmatologiÃ¢ I OrtopediÃ¢ Rossii, 2021, 27, 34-43.	0.5	1
102	A constellation of orthopaedic deformities in connection with cartilage oligomeric matrix protein mutation. African Journal of Paediatric Surgery, 2019, 16, 23.	0.6	1
103	Arthrogryposis is a descriptive term, not a specific disease entity: escobar syndrome is an Example. Minerva Pediatrics, 2020, , .	0.4	1
104	Torticollis in Connection with Spine Phenotype. Diagnostics, 2022, 12, 1672.	2.6	1
105	Distinctive spinal changes in two patients with unusual forms of autosomal dominant endosteal hyperostosis: a case series. Journal of Medical Case Reports, 2007, 1, 142.	0.8	0
106	Progressive anterior knee pain associated with patellar instability in a 57-year-old father and his daughter. Skeletal Radiology, 2010, 39, 921-921.	2.0	0
107	Dysmorphic facies and diffuse posterior spine ankylosis in a patient with unusual form of spondyloenchondrodysplasia (Spranger type IV). European Spine Journal, 2013, 22, 409-415.	2.2	0
108	Are parents of children with Cockayne syndrome manifesting features of the disorder?. Medicine (United States), 2017, 96, e8970.	1.0	0

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109	Leri-Weill Dyschondrosteosis Syndrome: Analysis via 3DCT Scan. Medicines (Basel, Switzerland), 2019, 6, 60.	1.4	0
110	The phenotypic spectrum in a patient with Glycine to Serine mutation in the <i>COL2A1</i> gene: overview study. AIMS Molecular Science, 2021, 8, 76-85.	0.5	0
111	Clinical Phenotype and Bone Biopsy Characteristics in a Child with Proteus Syndrome. Calcified Tissue International, 2021, 109, 586-595.	3.1	0
112	The clinical and radiographic phenotypic characterization of a girl with multiple malformation complex resembling Idaho Syndrome. Gazzetta Medica Italiana Archivio Per Le Scienze Mediche, 2021, 180, .	0.1	0
113	Occipito-Vertebral Dissociation in Connection with Extensive Cervical Spine Malsegmentation in a Boy with MÃ¶bius Syndrome. Clinics, 2009, 64, 1034-1036.	1.5	0
114	Severe Skew Foot Deformity in a Patient With Freeman-Sheldon Syndrome. Journal of Clinical Medicine Research, 2011, 3, 265-7.	1.2	0
115	Distinctive Skeletal Abnormalities With No Microdeletions or Microduplications on Array-CGH in a Boy With Mohr Syndrome (Oro-Facial-Digital Type II). Journal of Clinical Medicine Research, 2015, 7, 1002-1006.	1.2	0
116	Progressive non-infectious anterior vertebral fusion in a baby with Saethre-Chotzen-acrocephalosyndactyly type III syndrome. Pediatric Traumatology, Orthopaedics and Reconstructive Surgery, 2015, 3, 32-35.	0.3	0
117	Bilateral coxa vara and tibia vara associated with severe short stature in a girl manifesting a constellation of bone lesions with exclusive involvement of the lower limbs. Pediatric Traumatology, Orthopaedics and Reconstructive Surgery, 2018, 6, 63-69.	0.3	0
118	Unilateral lytic changes over the weight-bearing joint causing severe destruction of ankle joint (atypical Charcot joint) in a girl with congenital insensitivity to pain without anhidrosis (hereditary) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 Orthopaedics and Reconstructive Surgery, 2019, 7, 81-86.	0.3	0
119	Varus deformity of the left lower extremity causing degenerative lesion of the posterior horn of the left medial meniscus in a patient with Paget's disease of bone. GMS German Medical Science, 2014, 12, Doc13.	2.7	0
120	Infantile systemic hyalinosis: Variable grades of severity. African Journal of Paediatric Surgery, 2021, 18, 224-230.	0.6	0
121	The articular and the craniocervical abnormalities are of confusing age of onset in patients with Maroteaux-Lamy disease (MPS VI). Minerva Pediatrics, 2020, , .	0.4	0
122	Coxa vara in conjunction with metaphyseal dysostosis. Journal of the College of Physicians and Surgeons--Pakistan: JCPSP, 2012, 22, 466-9.	0.4	0