Mohammad Shboul

List of Publications by Year
in descending order

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27	938	567247 15	642715 23
papers	citations	h-index	g-index

28	28	28	2567
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Bioenergetic analysis of agedâ€phenotype skin in a rare syndromic cutis laxa. Journal of Cosmetic Dermatology, 2021, 20, 2999-3006.	1.6	0
2	Clinical Phenotype and Bone Biopsy Characteristics in a Child with Proteus Syndrome. Calcified Tissue International, 2021, 109, 586-595.	3.1	0
3	Infantile systemic hyalinosis: Variable grades of severity. African Journal of Paediatric Surgery, 2021, 18, 224-230.	0.6	0
4	Skeletal phenotype/genotype in progressive pseudorheumatoid chondrodysplasia. Clinical Rheumatology, 2020, 39, 553-560.	2.2	7
5	Congenital insensitivity to pain with anhidrosis syndrome: A series from Jordan. Clinical Neurology and Neurosurgery, 2020, 189, 105636.	1.4	12
6	Mitchell-Riley syndrome iPSC exhibit reduced pancreatic endoderm differentiation due to an RFX6 mutation. Development (Cambridge), 2020, 147, .	2.5	10
7	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
8	Arthrogryposis is a descriptive term, not a specific disease entity: escobar syndrome is an Example. Minerva Pediatrics, 2020, , .	0.4	1
9	Leri-Weill Dyschondrosteosis Syndrome: Analysis via 3DCT Scan. Medicines (Basel, Switzerland), 2019, 6, 60.	1.4	0
10	Massive Axial and Appendicular Skeletal Deformities in Connection with Gorham-Stout Syndrome. Medicines (Basel, Switzerland), 2019, 6, 54.	1.4	1
11	Deficiency of lrp4 in zebrafish and human LRP4 mutation induce aberrant activation of Jaggedâ€“Notch signaling in fin and limb development. Cellular and Molecular Life Sciences, 2019, 76, 163-178.	5.4	21
12	Bone matrix hypermineralization associated with low bone turnover in a case of Nasu-Hakola disease. Bone, 2019, 123, 48-55.	2.9	7
13	Genome-wide CRISPR-Cas9 Screen Identifies Leukemia-Specific Dependence on a Pre-mRNA Metabolic Pathway Regulated by DCPS. Cancer Cell, 2018, 33, 386-400.e5.	16.8	99
14	Novel mutations in the ciliopathy-associated gene CPLANE1 (C5orf42) cause OFD syndrome type VI rather than Joubert syndrome. European Journal of Medical Genetics, 2018, 61, 585-595.	1.3	22
15	A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. ELife, 2018, 7, .	6.0	53
16	Long-Term Culture of Self-renewing Pancreatic Progenitors Derived from Human Pluripotent Stem Cells. Stem Cell Reports, 2017, 8, 1675-1688.	4.8	55
17	Loss of Iroquois homeobox transcription factors 3 and 5 in osteoblasts disrupts cranial mineralization. Bone Reports, 2016, 5, 86-95.	0.4	21
18	Loss of the scavenger mRNA decapping enzyme DCPS causes syndromic intellectual disability with neuromuscular defects. Human Molecular Genetics, 2015, 24, 3163-3171.	2.9	31

#	Article	IF	Citations
19	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. Neuron, 2014, 84, 1240-1257.	8.1	89
20	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	3.8	71
21	The snRNA-processing complex, Integrator, is required for ciliogenesis and dynein recruitment to the nuclear envelope via distinct mechanisms. Biology Open, 2013, 2, 1390-1396.	1.2	18
22	Nuclear-localized Asunder regulates cytoplasmic dynein localization via its role in the Integrator complex. Molecular Biology of the Cell, 2013, 24, 2954-2965.	2.1	23
23	Definition of the phenotypic spectrum of Temtamy preaxial brachydactyly syndrome associated with autosomal recessive CHYS1 mutations. Middle East Journal of Medical Genetics, 2012, 1, 64-70.	0.0	6
24	Human Asunder promotes dynein recruitment and centrosomal tethering to the nucleus at mitotic entry. Molecular Biology of the Cell, 2012, 23, 4713-4724.	2.1	19
25	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nature Genetics, 2012, 44, 709-713.	21.4	68
26	Loss of CHSY1, a Secreted FRINGE Enzyme, Causes Syndromic Brachydactyly in Humans via Increased NOTCH Signaling. American Journal of Human Genetics, 2010, 87, 768-778.	6.2	82
27	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211