Keren Machol

List of Publications by Year
in descending order

10	307	1307594 7	1372567 10
papers	citations	h-index	g-index

10	10	10	877
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. American Journal of Human Genetics, 2016, 99, 934-941.	6.2	111
2	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
3	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. Science Advances, 2020, 6, eaax0021.	10.3	56
4	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
5	Corneal clouding, cataract, and colobomas with a novel missense mutation in B4GALT7â€”a review of eye anomalies in the linkeropathy syndromes. American Journal of Medical Genetics, Part A, 2016, 170, 2711-2718.	1.2	19
6	Hearing loss in individuals with osteogenesis imperfecta in North America: Results from a multicenter study. American Journal of Medical Genetics, Part A, 2020, 182, 697-704.	1.2	17
7	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	1.2	8
8	Evidence that <scp>FGFRL1</scp> contributes to congenital diaphragmatic hernia development in humans. American Journal of Medical Genetics, Part A, 2021, 185, 836-840.	1.2	8
9	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
10	Molecular alterations due to Col5a1 haploinsufficiency in a mouse model of classic Ehlersâ€“Danlos syndrome. Human Molecular Genetics, 2022, 31, 1325-1335.	2.9	1