Mark David Ludman

List of Publications by Year
in descending order

17	1,235	759233 12	940533 16
papers	citations	h-index	g-index

17	17	17	2540
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Diagnostic yield of multigene panel testing in an Israeli cohort: enrichment of low-penetrance variants. Breast Cancer Research and Treatment, 2020, 181, 445-453.	2.5	7
2	The time-consuming demands of the practice of medical genetics in the era of advanced genomic testing. Genetics in Medicine, 2016, 18, 372-377.	2.4	35
3	Germline Mutations in MAP3K6 Are Associated with Familial Gastric Cancer. PLoS Genetics, 2014, 10, e1004669.	3.5	57
4	Prenatal bilateral adrenal calcifications, hypogonadism, and nephrotic syndrome: beyond Wolman disease. Prenatal Diagnosis, 2014, 34, 608-611.	2.3	2
5	Further evidence for germline BAP1 mutations predisposing to melanoma and malignant mesothelioma. Cancer Genetics, 2013, 206, 206-210.	0.4	81
6	Mosaic tetrasomy 5p resulting from an isochromosome 5p marker chromosome: Case report and review of literature. American Journal of Medical Genetics, Part A, 2012, 158A, 406-411.	1.2	13
7	Mutations in origin recognition complex gene ORC4 cause Meier-Gorlin syndrome. Nature Genetics, 2011, 43, 360-364.	21.4	156
8	Mutations in Centrosomal Protein CEP152 in Primary Microcephaly Families Linked to MCPH4. American Journal of Human Genetics, 2010, 87, 40-51.	6.2	174
9	Overlapping spectra of SMAD4 mutations in juvenile polyposis (JP) and JPâ€“HHT syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 333-339.	1.2	128
10	Novel mutations in the sacsin gene in ataxia patients from Maritime Canada. Journal of the Neurological Sciences, 2010, 288, 79-87.	0.6	18
11	Mutation in the Gene Encoding Ubiquitin Ligase LRSAM1 in Patients with Charcot-Marie-Tooth Disease. PLoS Genetics, 2010, 6, e1001081.	3.5	59
12	Mutations in mitochondrial carrier family gene SLC25A38 cause nonsyndromic autosomal recessive congenital sideroblastic anemia. Nature Genetics, 2009, 41, 651-653.	21.4	220
13	Mutation in Pyrroline-5-Carboxylate Reductase 1 Gene in Families with Cutis Laxa Type 2. American Journal of Human Genetics, 2009, 85, 120-129.	6.2	81
14	Is gene discovery research or diagnosis?. Genetics in Medicine, 2008, 10, 385-390.	2.4	7
15	Ehlers-Danlos syndrome type VIIA and VIIB result from splice-junction mutations or genomic deletions that involve exon 6 in theCOL1A1 andCOL1A2 genes of type I collagen. American Journal of Medical Genetics Part A, 1997, 72, 94-105.	2.4	117
16	New syndrome?: MCA/MR syndrome with multiple circumferential skin creases. , 1996, 62, 23-25.		23
17	Allogenic Bone Marrow Transplantation in Severe Gaucher Disease. Pediatric Research, 1992, 31, 503-507.	2.3	57