

Mark David Ludman

List of Publications by Year in descending order

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17
papers

1,235
citations

759233

12
h-index

940533

16
g-index

17
all docs

17
docs citations

17
times ranked

2540
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in mitochondrial carrier family gene SLC25A38 cause nonsyndromic autosomal recessive congenital sideroblastic anemia. <i>Nature Genetics</i> , 2009, 41, 651-653.	21.4	220
2	Mutations in Centrosomal Protein CEP152 in Primary Microcephaly Families Linked to MCPH4. <i>American Journal of Human Genetics</i> , 2010, 87, 40-51.	6.2	174
3	Mutations in origin recognition complex gene ORC4 cause Meier-Gorlin syndrome. <i>Nature Genetics</i> , 2011, 43, 360-364.	21.4	156
4	Overlapping spectra of SMAD4 mutations in juvenile polyposis (JP) and JP+HHT syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 333-339.	1.2	128
5	Ehlers-Danlos syndrome type VIIA and VIIB result from splice-junction mutations or genomic deletions that involve exon 6 in the COL1A1 and COL1A2 genes of type I collagen. <i>American Journal of Medical Genetics Part A</i> , 1997, 72, 94-105.	2.4	117
6	Mutation in Pyrroline-5-Carboxylate Reductase 1 Gene in Families with Cutis Laxa Type 2. <i>American Journal of Human Genetics</i> , 2009, 85, 120-129.	6.2	81
7	Further evidence for germline BAP1 mutations predisposing to melanoma and malignant mesothelioma. <i>Cancer Genetics</i> , 2013, 206, 206-210.	0.4	81
8	Mutation in the Gene Encoding Ubiquitin Ligase LRSAM1 in Patients with Charcot-Marie-Tooth Disease. <i>PLoS Genetics</i> , 2010, 6, e1001081.	3.5	59
9	Allogenic Bone Marrow Transplantation in Severe Gaucher Disease. <i>Pediatric Research</i> , 1992, 31, 503-507.	2.3	57
10	Germline Mutations in MAP3K6 Are Associated with Familial Gastric Cancer. <i>PLoS Genetics</i> , 2014, 10, e1004669.	3.5	57
11	The time-consuming demands of the practice of medical genetics in the era of advanced genomic testing. <i>Genetics in Medicine</i> , 2016, 18, 372-377.	2.4	35
12	New syndrome?: MCA/MR syndrome with multiple circumferential skin creases. , 1996, 62, 23-25.		23
13	Novel mutations in the sarsin gene in ataxia patients from Maritime Canada. <i>Journal of the Neurological Sciences</i> , 2010, 288, 79-87.	0.6	18
14	Mosaic tetrasomy 5p resulting from an isochromosome 5p marker chromosome: Case report and review of literature. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 406-411.	1.2	13
15	Is gene discovery research or diagnosis?. <i>Genetics in Medicine</i> , 2008, 10, 385-390.	2.4	7
16	Diagnostic yield of multigene panel testing in an Israeli cohort: enrichment of low-penetrance variants. <i>Breast Cancer Research and Treatment</i> , 2020, 181, 445-453.	2.5	7
17	Prenatal bilateral adrenal calcifications, hypogonadism, and nephrotic syndrome: beyond Wolman disease. <i>Prenatal Diagnosis</i> , 2014, 34, 608-611.	2.3	2