

Rima Slim

List of Publications by Year in descending order

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papers

3,451
citations

172457

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docs citations

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times ranked

2826
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#	ARTICLE	IF	CITATIONS
1	A defect in harmonin, a PDZ domain-containing protein expressed in the inner ear sensory hair cells, underlies Usher syndrome type 1C. <i>Nature Genetics</i> , 2000, 26, 51-55.	21.4	449
2	Mutations in NALP7 cause recurrent hydatidiform moles and reproductive wastage in humans. <i>Nature Genetics</i> , 2006, 38, 300-302.	21.4	419
3	The infervers autoinflammatory mutation online registry: update with new genes and functions. <i>Human Mutation</i> , 2008, 29, 803-808.	2.5	239
4	Genetic Mapping of a Maternal Locus Responsible for Familial Hydatidiform Moles. <i>Human Molecular Genetics</i> , 1999, 8, 667-667.	2.9	130
5	Maternal alleles acquiring paternal methylation patterns in biparental complete hydatidiform moles. <i>Human Molecular Genetics</i> , 2003, 12, 1405-1413.	2.9	129
6	A proposed new contiguous gene syndrome on 8q consists of Branchio-Oto-Renal (BOR) syndrome, Duane syndrome, a dominant form of hydrocephalus and trapeze aplasia; implications for the mapping of the BOR gene. <i>Human Molecular Genetics</i> , 1994, 3, 1859-1866.	2.9	121
7	A newly identified locus for Usher syndrome type I, USH1E, maps to chromosome 21q21. <i>Human Molecular Genetics</i> , 1997, 6, 27-31.	2.9	101
8	A novel approach identifies new differentially methylated regions (DMRs) associated with imprinted genes. <i>Genome Research</i> , 2011, 21, 465-476.	5.5	101
9	A familial case of recurrent hydatidiform molar pregnancies with biparental genomic contribution. <i>Human Genetics</i> , 1999, 105, 112-115.	3.8	92
10	NLRP7 mutations in women with diploid androgenetic and triploid moles: a proposed mechanism for mole formation. <i>Human Molecular Genetics</i> , 2009, 18, 888-897.	2.9	84
11	Women heterozygous for NALP7/NLRP7 mutations are at risk for reproductive wastage: report of two novel mutations. <i>Human Mutation</i> , 2007, 28, 741-741.	2.5	78
12	Genetics and Epigenetics of Recurrent Hydatidiform Moles: Basic Science and Genetic Counselling. <i>Current Obstetrics and Gynecology Reports</i> , 2014, 3, 55-64.	0.8	76
13	Biallelic PADI6 variants linking infertility, miscarriages, and hydatidiform moles. <i>European Journal of Human Genetics</i> , 2018, 26, 1007-1013.	2.8	69
14	Causative Mutations and Mechanism of Androgenetic Hydatidiform Moles. <i>American Journal of Human Genetics</i> , 2018, 103, 740-751.	6.2	69
15	NLRP7 in the spectrum of reproductive wastage: rare non-synonymous variants confer genetic susceptibility to recurrent reproductive wastage. <i>Journal of Medical Genetics</i> , 2011, 48, 540-548.	3.2	68
16	The genetics of hydatidiform moles: new lights on an ancient disease. <i>Clinical Genetics</i> , 2006, 71, 25-34.	2.0	65
17	Targeted next-generation sequencing identifies a homozygous nonsense mutation in ABHD12, the gene underlying PHARC, in a family clinically diagnosed with Usher syndrome type 3. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 59.	2.7	61
18	NLRP7, a Nucleotide Oligomerization Domain-like Receptor Protein, Is Required for Normal Cytokine Secretion and Co-localizes with Golgi and the Microtubule-organizing Center. <i>Journal of Biological Chemistry</i> , 2011, 286, 43313-43323.	3.4	60

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19	Report of four new patients with protein-truncating mutations in C6orf221/KHDC3L and colocalization with NLRP7. <i>European Journal of Human Genetics</i> , 2013, 21, 957-964.	2.8	59
20	A Human Pseudoautosomal Gene Encodes the ANT3 ADP/ATP Translocase and Escapes X-Inactivation. <i>Genomics</i> , 1993, 16, 26-33.	2.9	55
21	NLRP7 and KHDC3L, the two maternal-effect proteins responsible for recurrent hydatidiform moles, co-localize to the oocyte cytoskeleton. <i>Human Reproduction</i> , 2015, 30, 159-169.	0.9	55
22	The genetics of recurrent hydatidiform moles: new insights and lessons from a comprehensive analysis of 113 patients. <i>Modern Pathology</i> , 2018, 31, 1116-1130.	5.5	51
23	A cytokine receptor gene cluster in the X-Y pseudoautosomal region?. <i>Blood</i> , 1993, 82, 22-28.	1.4	45
24	NLRP7 and the Genetics of Hydatidiform Moles: Recent Advances and New Challenges. <i>Frontiers in Immunology</i> , 2013, 4, 242.	4.8	40
25	The human placental protein 14 (PP14) gene is localized on chromosome 9q34. <i>Human Genetics</i> , 1991, 86, 515-518.	3.8	39
26	The genetics of recurrent hydatidiform moles in China: correlations between NLRP7 mutations, molar genotypes and reproductive outcomes. <i>Molecular Human Reproduction</i> , 2011, 17, 612-619.	2.8	39
27	Live births in women with recurrent hydatidiform mole and two NLRP7 mutations. <i>Reproductive BioMedicine Online</i> , 2015, 31, 120-124.	2.4	36
28	A strong founder effect for two NLRP7 mutations in the Indian population: an intriguing observation. <i>Clinical Genetics</i> , 2009, 76, 292-295.	2.0	34
29	Familial molar tissues due to mutations in the inflammatory gene, NALP7, have normal postzygotic DNA methylation. <i>Human Genetics</i> , 2006, 120, 390-395.	3.8	31
30	Antagonism of EG-VEGF Receptors as Targeted Therapy for Choriocarcinoma Progression <i>In Vitro</i> and <i>In Vivo</i> . <i>Clinical Cancer Research</i> , 2017, 23, 7130-7140.	7.0	31
31	Comprehensive genotype-phenotype correlations between NLRP7 mutations and the balance between embryonic tissue differentiation and trophoblastic proliferation. <i>Journal of Medical Genetics</i> , 2014, 51, 623-634.	3.2	28
32	Two novel mutations in the KHDC3L gene in Asian patients with recurrent hydatidiform mole. <i>Human Genome Variation</i> , 2016, 3, 16027.	0.7	26
33	Evidence of a genetic heterogeneity of familial hydatidiform moles. <i>Placenta</i> , 2005, 26, 5-9.	1.5	23
34	Pathogenic variant in NLRP7 (19q13.42) associated with recurrent gestational trophoblastic disease: Data from early embryo development observed during <i>in vitro</i> fertilization. <i>Clinical and Experimental Reproductive Medicine</i> , 2017, 44, 40.	1.5	23
35	Recurrent triploid and dispermic conceptions in patients with NLRP7 mutations. <i>Placenta</i> , 2011, 32, 409-412.	1.5	22
36	Construction of a Yeast Artificial Chromosome Contig Spanning the Pseudoautosomal Region and Isolation of 25 New Sequence-Tagged Sites. <i>Genomics</i> , 1993, 16, 691-697.	2.9	21

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37	The genomic architecture of NLRP7 is Alu rich and predisposes to disease-associated large deletions. <i>European Journal of Human Genetics</i> , 2016, 24, 1445-1452.	2.8	21
38	Patients with familial biparental hydatidiform moles have normal methylation at imprinted genes. <i>European Journal of Human Genetics</i> , 2005, 13, 486-490.	2.8	20
39	NLRP7 inter-domain interactions: the NACHT-associated domain is the physical mediator for oligomeric assembly. <i>Molecular Human Reproduction</i> , 2014, 20, 990-1001.	2.8	20
40	Comprehensive analysis of 204 sporadic hydatidiform moles: revisiting risk factors and their correlations with the molar genotypes. <i>Modern Pathology</i> , 2020, 33, 880-892.	5.5	19
41	The Usher syndrome in the Lebanese population and further refinement of the USH2A candidate region. <i>Human Genetics</i> , 1998, 103, 193-198.	3.8	17
42	Detailed gene and allele content analysis of three homozygous KIR haplotypes. <i>Tissue Antigens</i> , 2006, 68, 72-77.	1.0	17
43	TMED2/p24 ¹ is expressed in all gestational stages of human placentas and in choriocarcinoma cell lines. <i>Placenta</i> , 2012, 33, 214-219.	1.5	17
44	Novel pathogenic variants in <i>NLRP7</i> , <i>NLRP5</i> and <i>PADI6</i> in patients with recurrent hydatidiform moles and reproductive failure. <i>Clinical Genetics</i> , 2021, 99, 823-828.	2.0	17
45	Relative order determination of four Yp cosmids on metaphase and interphase chromosomes by two-color competitive in situ hybridization. <i>Human Genetics</i> , 1991, 88, 21-26.	3.8	16
46	Microcephaly, cutis verticis gyrata of the scalp, retinitis pigmentosa, cataracts, sensorineural deafness, and mental retardation in two brothers. <i>American Journal of Medical Genetics Part A</i> , 2001, 98, 244-249.	2.4	16
47	NLRP7 Promotes Choriocarcinoma Growth and Progression through the Establishment of an Immunosuppressive Microenvironment. <i>Cancers</i> , 2021, 13, 2999.	3.7	16
48	Differential expression of E-cadherin, β -catenin, and Lewis x between invasive hydatidiform moles and post-molar choriocarcinomas. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2013, 462, 653-663.	2.8	15
49	Recurrent Pregnancy Loss in a Woman With NLRP7 Mutation. <i>International Journal of Gynecological Pathology</i> , 2013, 32, 399-405.	1.4	15
50	The neurofibromatosis 1 gene transcripts expressed in peripheral nerve and neurofibromas bear the additional exon located in the GAP domain. <i>Biochemical and Biophysical Research Communications</i> , 1992, 188, 851-857.	2.1	14
51	NLRP7 and the genetics of post-molar choriocarcinomas in Senegal. <i>Molecular Human Reproduction</i> , 2012, 18, 52-56.	2.8	14
52	Rearrangements between irradiated chromosomes in three-species radiation hybrid cell lines revealed by two-color in situ hybridization. <i>Human Genetics</i> , 1993, 92, 11-17.	3.8	12
53	Prenatal identification of an isochromosome for the short arm of the Y i(Yp), by cytogenetic and Molecular analyses. <i>Prenatal Diagnosis</i> , 1994, 14, 23-28.	2.3	11
54	A novel VPS13B mutation in two brothers with Cohen syndrome, cutis verticis gyrata and sensorineural deafness. <i>European Journal of Human Genetics</i> , 2009, 17, 1076-1079.	2.8	11

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55	Molecular Genetics of the Usher Syndrome in Lebanon: Identification of 11 Novel Protein Truncating Mutations by Whole Exome Sequencing. PLoS ONE, 2014, 9, e107326.	2.5	10
56	A novel NLRP7 protein-truncating mutation associated with discordant and divergent p57 immunostaining in diploid biparental and triploid digynic moles. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2020, 477, 309-315.	2.8	9
57	A novel 5-bp deletion in Clarin 1 in a family with Usher syndrome. Ophthalmic Genetics, 2011, 32, 245-249.	1.2	8
58	Recurrent triploid digynic conceptions and mature ovarian teratomas: Are they different manifestations of the same genetic defect?. Genes Chromosomes and Cancer, 2017, 56, 832-840.	2.8	7
59	A protein-truncating mutation in <i>CCNB3</i> in a patient with recurrent miscarriages and failure of meiosis I. Journal of Medical Genetics, 2022, 59, 568-570.	3.2	7
60	The human homologue (PEG3) of the mouse paternally expressed gene 3 (Peg3) is maternally imprinted but not mutated in women with familial recurrent hydatidiform molar pregnancies. Journal of the Society for Gynecologic Investigation, 2001, 8, 305-313.	1.7	7
61	Familial Hydatidiform Molar Pregnancy: The Germline Imprinting Defect Hypothesis?. , 2006, 301, 229-241.		6
62	The genetics of recurrent hydatidiform moles in Mexico: further evidence of a strong founder effect for one mutation in NLRP7 and its widespread. Journal of Assisted Reproduction and Genetics, 2021, 38, 1879-1886.	2.5	5
63	Assignments of 37 YAC clones to R-banded chromosomes by fluorescent in situ hybridization. Cytogenetic and Genome Research, 1994, 65, 104-107.	1.1	4
64	Two families from New England with usher syndrome type IC with distinct haplotypes. American Journal of Ophthalmology, 2001, 131, 355-358.	3.3	4
65	Map refinement of the Usher syndrome type 1B gene, MYO7A, relative to 11q13.5 microsatellite markers. Clinical Genetics, 1998, 54, 155-158.	2.0	4
66	Absence of KHDC3L mutations in Chinese patients with recurrent and sporadic hydatidiform moles. Cancer Genetics, 2013, 206, 327-329.	0.4	4
67	Circulating Tumor DNA: A Potential Novel Diagnostic Approach in Gestational Trophoblastic Neoplasia. EBioMedicine, 2016, 4, 11-12.	6.1	4
68	A bioinformatics transcriptome meta-analysis highlights the importance of trophoblast differentiation in the pathology of hydatidiform moles. Placenta, 2018, 65, 29-36.	1.5	4
69	Further refinement of Pendred syndrome locus by homozygosity analysis to a 0.8 cM interval flanked by D7S496 and D7S2425.. Journal of Medical Genetics, 1998, 35, 202-204.	3.2	3
70	Hydatidiform Moles. , 2019, , 485-497.		3
71	Microsatellite DNA Genotyping and Flow Cytometry Ploidy Analyses of Formalin-fixed Paraffin-embedded Hydatidiform Molar Tissues. Journal of Visualized Experiments, 2019, , .	0.3	2