Stella Mitrani-Rosenbaum

List of Publications by Year in Descending Order

61 papers	2,505 citations	24 h-index	49 g-index
62 ext. papers	2,820 ext. citations	5.4 avg, IF	3.82 L-index

#	Paper	IF	Citations
61	The Obesogenic and Glycemic Effect of Bariatric Surgery in a Family with a Melanocortin 4 Receptor Loss-of-Function Mutation. *Metabolites*, **2022**, 12, 430	5.6	
60	The glycomic sialylation profile of GNE Myopathy muscle cells does not point to consistent hyposialylation of individual glycoconjugates. *Neuromuscular Disorders*, **2020**, 30, 621-630	2.9	3
59	Upregulation of Hallmark Muscle Genes Protects GneM743T/M743T Mutated Knock-In Mice From Kidney and Muscle Phenotype. *Journal of Neuromuscular Diseases*, **2020**, 7, 119-136	5	5
58	Pax7, Pax3 and Mamstr genes are involved in skeletal muscle impaired regeneration of dy2J/dy2J mouse model of Lama2-CMD. *Human Molecular Genetics*, **2019**, 28, 3369-3390	5.6	4
57	237th ENMC International Workshop: GNE myopathy - current and future research Hoofddorp, The Netherlands, 14-16 September 2018. *Neuromuscular Disorders*, **2019**, 29, 401-410	2.9	2
56	The Interaction of UDP-N-Acetylglucosamine 2-Epimerase/N-Acetylmannosamine Kinase (GNE) and Alpha-Actinin 2 Is Altered in GNE Myopathy M743T Mutant. *Molecular Neurobiology*, **2017**, 54, 2928-2938	6.2	24
55	Survival-apoptosis associated signaling in GNE myopathy-cultured myoblasts. *Journal of Receptor and Signal Transduction Research*, **2015**, 35, 249-57	2.6	6
54	Uncovering the Role of Hypermethylation by CTG Expansion in Myotonic Dystrophy Type 1 Using Mutant Human Embryonic Stem Cells. *Stem Cell Reports*, **2015**, 5, 221-31	8	29
53	Epigenetic changes as a common trigger of muscle weakness in congenital myopathies. *Human Molecular Genetics*, **2015**, 24, 4636-47	5.6	33
52	Gne depletion during zebrafish development impairs skeletal muscle structure and function. *Human Molecular Genetics*, **2014**, 23, 3349-61	5.6	12
51	GNE myopathy: new name and new mutation nomenclature. *Neuromuscular Disorders*, **2014**, 24, 387-9	2.9	45
50	Correction of the Middle Eastern M712T mutation causing GNE myopathy by trans-splicing. *NeuroMolecular Medicine*, **2014**, 16, 322-31	4.6	11
49	Variable phenotypes of knockin mice carrying the M712T Gne mutation. *NeuroMolecular Medicine*, **2013**, 15, 180-91	4.6	16
48	Variable myopathic presentation in a single family with novel skeletal RYR1 mutation. *PLoS ONE*, **2013**, 8, e69296	3.7	11
47	Colon cancer associated transcript-1: a novel RNA expressed in malignant and pre-malignant human tissues. *International Journal of Cancer*, **2012**, 130, 1598-606	7.5	219
46	Sustained expression and safety of human GNE in normal mice after gene transfer based on AAV8 systemic delivery. *Neuromuscular Disorders*, **2012**, 22, 1015-24	2.9	13
45	Detection of N-glycans on small amounts of glycoproteins in tissue samples and sodium dodecyl sulfate-polyacrylamide gels. *Analytical Biochemistry*, **2012**, 423, 253-60	3.1	15

44	Development of a microRNA-based molecular assay for the detection of papillary thyroid carcinoma in aspiration biopsy samples. *Thyroid*, **2011**, 21, 111-8	6.2	89
43	GNE is involved in the early development of skeletal and cardiac muscle. *PLoS ONE*, **2011**, 6, e21389	3.7	14
42	The proteomic profile of hereditary inclusion body myopathy. *PLoS ONE*, **2011**, 6, e16334	3.7	35
41	Mutations and polymorphisms of the skeletal muscle alpha-actin gene (ACTA1). *Human Mutation*, **2009**, 30, 1267-77	4.7	150
40	Mitochondrial processes are impaired in hereditary inclusion body myopathy. *Human Molecular Genetics*, **2008**, 17, 3663-74	5.6	38
39	UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase (GNE) binds to alpha-actinin 1: novel pathways in skeletal muscle?. *PLoS ONE*, **2008**, 3, e2477	3.7	48
38	Attention deficit hyperactivity disorder in obese melanocortin-4-receptor (MC4R) deficient subjects: a newly described expression of MC4R deficiency. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 1547-53	3.5	28
37	The hereditary inclusion body myopathy enigma and its future therapy. *Neurotherapeutics*, **2008**, 5, 633-7	6.4	24
36	Hereditary inclusion body myopathy and other rimmed vacuolar myopathies. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2007**, 86, 243-53	3	3
35	Distinctive patterns of microRNA expression in primary muscular disorders. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 17016-21	11.5	391
34	Influence of UDP-GlcNAc 2-epimerase/ManNAc kinase mutant proteins on hereditary inclusion body myopathy. *Biochemistry*, **2006**, 45, 2968-77	3.2	51
33	Localization of UDP-GlcNAc 2-epimerase/ManAc kinase (GNE) in the Golgi complex and the nucleus of mammalian cells. *Experimental Cell Research*, **2005**, 304, 365-79	4.2	62
32	No overall hyposialylation in hereditary inclusion body myopathy myoblasts carrying the homozygous M712T GNE mutation. *Biochemical and Biophysical Research Communications*, **2005**, 328, 221-6	3.4	76
31	The homozygous M712T mutation of UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase results in reduced enzyme activities but not in altered overall cellular sialylation in hereditary inclusion body myopathy. *FEBS Letters*, **2004**, 566, 105-9	3.8	61
30	Insight into the intrinsic sensitivity of the PCR assay used to detect CMV infection in amniotic fluid specimens. *Journal of Clinical Virology*, **2004**, 29, 260-70	14.5	9
29	Mutations spectrum of GNE in hereditary inclusion body myopathy sparing the quadriceps. *Human Mutation*, **2003**, 21, 99	4.7	97
28	Distal myopathy with rimmed vacuoles is allelic to hereditary inclusion body myopathy. *Neurology*, **2003**, 61, 145; author reply 145	6.5	
27	Establishment of the genomic structure and identification of thirteen single-nucleotide polymorphisms in the human RECK gene. *Cytogenetic and Genome Research*, **2002**, 97, 58-61	1.9	12

26	Cloning and characterization of a novel human gene RNF38 encoding a conserved putative protein with a RING finger domain. *Biochemical and Biophysical Research Communications*, **2002**, 294, 1169-76	3.4	16
25	Cloning and characterization of a human novel gene C9orf19 encoding a conserved putative protein with an SCP-like extracellular protein domain. *Gene*, **2002**, 293, 141-8	3.8	16
24	The UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase gene is mutated in recessive hereditary inclusion body myopathy. *Nature Genetics*, **2001**, 29, 83-7	36.3	407
23	Physical and transcriptional map of the hereditary inclusion body myopathy locus on chromosome 9p12-p13. *European Journal of Human Genetics*, **2001**, 9, 501-9	5.3	21
22	Monopaternal superfecundation of quintuplets after transfer of two embryos in an in vitro fertilization cycle. *Fertility and Sterility*, **2001**, 76, 621-3	4.8	9
21	Muscular dystrophy due to dysferlin deficiency in Libyan Jews. Clinical and genetic features. *Brain*, **2000**, 123 (Pt 6), 1229-37	11.2	71
20	X inactivation-specific transcript expression in mouse oocytes and zygotes. *Molecular Human Reproduction*, **2000**, 6, 591-4	4.4	4
19	Fine-structure mapping of the hereditary inclusion body myopathy locus. *Genomics*, **1999**, 55, 43-8	4.3	24
18	Genetics of inclusion body myopathies. *Current Opinion in Rheumatology*, **1998**, 10, 543-7	5.3	25
17	Various types of hereditary inclusion body myopathies map to chromosome 9p1-q1. *Annals of Neurology*, **1997**, 41, 548-51	9.4	51
16	Management of rhesus isoimmunization by preimplantation genetic diagnosis. *Molecular Human Reproduction*, **1996**, 2, 60-2	4.4	21
15	RhD genotype determination by single sperm cell analysis. *American Journal of Obstetrics and Gynecology*, **1996**, 174, 1300-5	6.4	9
14	Presence of infective Epstein-Barr virus in the urine of patients with infectious mononucleosis. *Journal of Medical Virology*, **1994**, 44, 229-33	19.7	7
13	Simultaneous detection of three common sexually transmitted agents by polymerase chain reaction. *American Journal of Obstetrics and Gynecology*, **1994**, 171, 784-90	6.4	10
12	Preimplantation diagnosis of cystic fibrosis by simultaneous detection of the W1282X and delta F508 mutations. *Human Reproduction*, **1994**, 9, 1676-80	5.7	26
11	Differential cooperation of a carcinogen with human papillomavirus type 6 and 16 DNAs in in vitro oncogenic transformation. *Intervirology*, **1992**, 33, 76-85	2.5	10
10	Differential expression of HPV types 6 and 11 in condylomas and cervical preneoplastic lesions. *Virus Research*, **1992**, 25, 23-36	6.4	4
9	Eczema herpeticum induced by sun exposure. *International Journal of Dermatology*, **1992**, 31, 298-9	1.7	12

8	Detection of human papillomavirus (HPV) DNA in focal epithelial hyperplasia. *Journal of Oral Pathology and Medicine*, **1989**, 18, 172-7	3.3	50
7	Papillomaviruses in lesions of the lower genital tract in Israeli patients. *European Journal of Cancer & Clinical Oncology*, **1988**, 24, 725-31		9
6	Integration and transcription of human papillomavirus type 6 recombinant DNA in mouse cells. *Virus Research*, **1987**, 8, 335-47	6.4	2
5	Hybridization between a human epithelial line, infectable by Epstein-Barr virus, and Burkitt lymphoma lines: membrane properties, superinfectability, inducibility and tumorigenicity. *International Journal of Cancer*, **1982**, 30, 593-600	7.5	1
4	Interaction of Herpes simplex virus with human cell lines at various stages of lymphoid differentiation. *Intervirology*, **1981**, 16, 33-42	2.5	5
3	Establishment in continuous culture of a T-lymphoid cell line (HD-Mar) from a patient with Hodgkin's lymphoma. *International Journal of Cancer*, **1980**, 25, 583-90	7.5	37
2	A comparative study of human cell lines derived from patients with lymphoma, leukemia and infectious mononucleosis: membrane properties, ultrastructure, and surface morphology. *Cancer*, **1977**, 40, 1481-91	6.4	18
1	Hereditary inclusion body myopathies492-498		4