Stella Mitrani-Rosenbaum

List of Publications by Year
in descending order

62 papers	3,035 citations	218381 26 h-index	161609 54 g-index
62 all docs	62 docs citations	62 times ranked	3188 citing authors

#	Article	IF	Citations
1	Pre Clinical Assessment of AAVrh74.MCK.GNE Viral Vector Therapeutic Potential: Robust Activity Despite Lack of Consistent Animal Model for GNE Myopathy. Journal of Neuromuscular Diseases, 2022, 9, 179-192.	1.1	6
2	The Obesogenic and Glycemic Effect of Bariatric Surgery in a Family with a Melanocortin 4 Receptor Loss-of-Function Mutation. Metabolites, 2022, 12, 430.	1.3	3
3	The glycomic sialylation profile of GNE Myopathy muscle cells does not point to consistent hyposialylation of individual glycoconjugates. Neuromuscular Disorders, 2020, 30, 621-630.	0.3	11
4	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.	1.1	8
5	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.	1.4	8
6	237th ENMC International Workshop: GNE myopathy â€“ current and future research Hoofddorp, The Netherlands, 14â€“16 September 2018. Neuromuscular Disorders, 2019, 29, 401-410.	0.3	5
7	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.	1.9	39
8	Epigenetic changes as a common trigger of muscle weakness in congenital myopathies. Human Molecular Genetics, 2015, 24, 4636-4647.	1.4	44
9	Survival-apoptosis associated signaling in GNE myopathy-cultured myoblasts. Journal of Receptor and Signal Transduction Research, 2015, 35, 249-257.	1.3	13
10	Uncovering the Role of Hypermethylation by CTG Expansion in Myotonic Dystrophy Type 1ÂUsing Mutant Human Embryonic Stem Cells. Stem Cell Reports, 2015, 5, 221-231.	2.3	40
11	Gne depletion during zebrafish development impairs skeletal muscle structure and function. Human Molecular Genetics, 2014, 23, 3349-3361.	1.4	19
12	GNE myopathy: New name and new mutation nomenclature. Neuromuscular Disorders, 2014, 24, 387-389.	0.3	61
13	Correction of the Middle Eastern M712T Mutation Causing GNE Myopathy by Trans-Splicing. NeuroMolecular Medicine, 2014, 16, 322-331.	1.8	14
14	Variable Phenotypes of Knockin Mice Carrying the M712T Gne Mutation. NeuroMolecular Medicine, 2013, 15, 180-191.	1.8	26
15	Variable Myopathic Presentation in a Single Family with Novel Skeletal RYR1 Mutation. PLoS ONE, 2013, 8, e69296.	1.1	12
16	Sustained expression and safety of human GNE in normal mice after gene transfer based on AAV8 systemic delivery. Neuromuscular Disorders, 2012, 22, 1015-1024.	0.3	27
17	Detection of N-glycans on small amounts of glycoproteins in tissue samples and sodium dodecyl sulfateâ€“polyacrylamide gels. Analytical Biochemistry, 2012, 423, 253-260.	1.1	16
18	Colon cancer associated transcriptâ€1: A novel RNA expressed in malignant and preâ€malignant human tissues. International Journal of Cancer, 2012, 130, 1598-1606.	2.3	250

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19	Development of a MicroRNA-Based Molecular Assay for the Detection of Papillary Thyroid Carcinoma in Aspiration Biopsy Samples. Thyroid, 2011, 21, 111-118.	2.4	99
20	GNE Is Involved in the Early Development of Skeletal and Cardiac Muscle. PLoS ONE, 2011, 6, e21389.	1.1	18
21	The Proteomic Profile of Hereditary Inclusion Body Myopathy. PLoS ONE, 2011, 6, e16334.	1.1	45
22	Mutations and polymorphisms of the skeletal muscle ÃŽÂ±-actin gene (ACTA1). Human Mutation, 2009, 30, 1267-1277.	1.1	198
23	Attention deficit hyperactivity disorder in obese melanocortinâ€4â€receptor (MC4R) deficient subjects: A newly described expression of MC4R deficiency. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1547-1553.	1.1	33
24	The Hereditary Inclusion Body Myopathy Enigma and its Future Therapy. Neurotherapeutics, 2008, 5, 633-637.	2.1	27
25	Mitochondrial processes are impaired in hereditary inclusion body myopathy. Human Molecular Genetics, 2008, 17, 3663-3674.	1.4	49
26	UDP-N-Acetylglucosamine 2-Epimerase/N-Acetylmannosamine Kinase (GNE) Binds to Alpha-Actinin 1: Novel Pathways in Skeletal Muscle?. PLoS ONE, 2008, 3, e2477.	1.1	71
27	Hereditary inclusion body myopathy and other rimmed vacuolar myopathies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2007, 86, 243-253.	1.0	6
28	Distinctive patterns of microRNA expression in primary muscular disorders. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17016-17021.	3.3	458
29	Influence of UDP-GlcNAc 2-Epimerase/ManNAc Kinase Mutant Proteins on Hereditary Inclusion Body Myopathyâ€. Biochemistry, 2006, 45, 2968-2977.	1.2	58
30	Localization of UDP-GlcNAc 2-epimerase/ManAc kinase (GNE) in the Golgi complex and the nucleus of mammalian cells. Experimental Cell Research, 2005, 304, 365-379.	1.2	72
31	No overall hyposialylation in hereditary inclusion body myopathy myoblasts carrying the homozygous M712T GNE mutation. Biochemical and Biophysical Research Communications, 2005, 328, 221-226.	1.0	93
32	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, 105-109.	1.3	77
33	Insight into the intrinsic sensitivity of the PCR assay used to detect CMV infection in amniotic fluid specimens. Journal of Clinical Virology, 2004, 29, 260-270.	1.6	9
34	Mutations spectrum ofGNE in hereditary inclusion body myopathy sparing the quadriceps. Human Mutation, 2003, 21, 99-99.	1.1	110
35	Distal myopathy with rimmed vacuoles is allelic to hereditary inclusion body myopathy. Neurology, 2003, 61, 145-145.	1.5	3
36	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.	0.6	14

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37	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, 1169-1176.	1.0	18
38	Cloning and characterization of a human novel gene C9orf19 encoding a conserved putative protein with an SCP-like extracellular protein domain. Gene, 2002, 293, 141-148.	1.0	17
39	Monopaternal superfecundation of quintuplets after transfer of two embryos in an in vitro fertilization cycle. Fertility and Sterility, 2001, 76, 621-623.	0.5	11
40	The UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase gene is mutated in recessive hereditary inclusion body myopathy. Nature Genetics, 2001, 29, 83-87.	9.4	476
41	Physical and transcriptional map of the hereditary inclusion body myopathy locus on chromosome 9p12-p13. European Journal of Human Genetics, 2001, 9, 501-509.	1.4	21
42	Muscular dystrophy due to dysferlin deficiency in Libyan Jews. Brain, 2000, 123, 1229-1237.	3.7	88
43	X inactivation-specific transcript expression in mouse oocytes and zygotes. Molecular Human Reproduction, 2000, 6, 591-594.	1.3	4
44	Fine-Structure Mapping of the Hereditary Inclusion Body Myopathy Locus. Genomics, 1999, 55, 43-48.	1.3	24
45	Genetics of inclusion body myopathies. Current Opinion in Rheumatology, 1998, 10, 543-547.	2.0	27
46	Various types of herediary inclusion body myopathies map to chromosome 9p1-q1. Annals of Neurology, 1997, 41, 548-551.	2.8	55
47	RhD genotype determination by single sperm cell analysis. American Journal of Obstetrics and Gynecology, 1996, 174, 1300-1305.	0.7	12
48	Diagnosing and preventing inherited diseases. Molecular Human Reproduction, 1996, 2, 60-62.	1.3	23
49	Presence of infective Epstein-Barr virus in the urine of patients with infectious mononucleosis. Journal of Medical Virology, 1994, 44, 229-233.	2.5	10
50	Simultaneous detection of three common sexually transmitted agents by polymerase chain reaction. American Journal of Obstetrics and Gynecology, 1994, 171, 784-790.	0.7	12
51	Genetics: Preimplantation diagnosis of cystic fibrosis by simultaneous detection of the W1282X and Î”F508 mutations*. Human Reproduction, 1994, 9, 1676-1680.	0.4	28
52	Differential Cooperation of a Carcinogen with Human Papillomavirus Type 6 and 16 DNAs in in vitro Oncogenic Transformation. Intervirology, 1992, 33, 76-85.	1.2	10
53	Differential expression of HPV types 6 and 11 in condylomas and cervical preneoplastic lesions. Virus Research, 1992, 25, 23-36.	1.1	4
54	Eczema Herpeticum Induced by Sun Exposure. International Journal of Dermatology, 1992, 31, 298-299.	0.5	13

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55	Detection of human papillomavirus (HPV) DNA in focal epithelial hyperplasia. Journal of Oral Pathology and Medicine, 1989, 18, 172-177.	1.4	56
56	Papillomaviruses in lesions of the lower genital tract in Israeli patients. European Journal of Cancer & Clinical Oncology, 1988, 24, 725-731.	0.9	9
57	Integration and transcription of human papillomavirus type 6 recombinant DNA in mouse cells. Virus Research, 1987, 8, 335-347.	1.1	2
58	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.	2.3	1
59	Interaction of Herpes Simplex Virus with Human Cell Lines at Various Stages of Lymphoid Differentiation. Intervirology, 1981, 16, 33-42.	1.2	8
60	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, 583-590.	2.3	40
61	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, 1481-1491.	2.0	18
62	Hereditary inclusion body myopathies. , 0, , 492-498.		6