Sandra Leistner-Segal

List of Publications by Year
in descending order

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Version: 2024-02-01

102	1,915	257450	315739
papers	citations	24 h-index	38 g-index

114	114	114	2975
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Sanfilippo syndrome type B: Analysis of patients diagnosed by the <scp>MPS</scp> Brazil Network. American Journal of Medical Genetics, Part A, 2022, 188, 760-767.	1.2	3
2	Updated birth prevalence and relative frequency of mucopolysaccharidoses across Brazilian regions. Genetics and Molecular Biology, 2021, 44, e20200138.	1.3	13
3	Prevalence of thrombophilia-associated genetic risk factors in blood donors of a regional hospital in southern Brazil. Hematology, Transfusion and Cell Therapy, 2021, , .	0.2	1
4	Genotypeâ€“phenotype studies in a large cohort of Brazilian patients with Hunter syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 349-356.	1.6	6
5	Demographic, clinical, and ancestry characterization of a large cluster of mucopolysaccharidosis IV A in the Brazilian Northeast region. American Journal of Medical Genetics, Part A, 2021, 185, 2929-2940.	1.2	5
6	Detection of Mosaic Variants in Mothers of MPS II Patients by Next Generation Sequencing. Frontiers in Molecular Biosciences, 2021, 8, 789350.	3.5	0
7	Methylation of BDNF and SLC6A4 Gene Promoters in Brazilian Patients With Temporal Lobe Epilepsy Presenting or Not Psychiatric Comorbidities. Frontiers in Integrative Neuroscience, 2021, 15, 764742.	2.1	5
8	Genetic Analysis of Hereditary Ataxias in Peru Identifies SCA10 Families with Incomplete Penetrance. Cerebellum, 2020, 19, 208-215.	2.5	3
9	Glatiramer acetate increases T- and B -regulatory cells and decreases granulocyte-macrophage colony-stimulating factor (GM-CSF) in an animal model of multiple sclerosis. Journal of Neuroimmunology, 2020, 345, 577281.	2.3	11
10	Is the gut microbiota dysbiotic in patients with classical homocystinuria?. Biochimie, 2020, 173, 3-11.	2.6	10
11	Diagnosis of Mucopolysaccharidoses. Diagnostics, 2020, 10, 172.	2.6	46
12	Friedreich Ataxia: Diagnostic Yield and Minimal Frequency in South Brazil. Cerebellum, 2019, 18, 147-151.	2.5	5
13	Back Cover, Volume 40, Issue 8. Human Mutation, 2019, 40, ii-ii.	2.5	0
14	Phenotype-oriented NGS panels for mucopolysaccharidoses: Validation and potential use in the diagnostic flowchart. Genetics and Molecular Biology, 2019, 42, 207-214.	1.3	11
15	Analysis of Predictive Biomarkers in Patients With Lung Adenocarcinoma From Southern Brazil Reveals a Distinct Profile From Other Regions of the Country. Journal of Global Oncology, 2019, 5, 1-9.	0.5	13
16	Polymorphic variants (p.Ser141Ser and p.Arg737Gly) at the NAGLU gene are really indicative of pseudodeficiency alleles?. Italian Journal of Pediatrics, 2019, 45, 60.	2.6	0
17	Molecular characterization of a large group of Mucopolysaccharidosis type IIIC patients reveals the evolutionary history of the disease. Human Mutation, 2019, 40, 1084-1100.	2.5	17
18	Information and Diagnosis Networks â€“ tools to improve diagnosis and treatment for patients with rare genetic diseases. Genetics and Molecular Biology, 2019, 42, 155-164.	1.3	9

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19	Population medical genetics: translating science to the community. Genetics and Molecular Biology, 2019, 42, 312-320.	1.3	8
20	MPS Brazil Network: A summary of all mucopolysaccharidosis type IIIB patients. Molecular Genetics and Metabolism, 2019, 126, S88.	1.1	1
21	Identification of MPS clusters in Latin America: An opportunity for targeted health care programs. Molecular Genetics and Metabolism, 2019, 126, S87-S88.	1.1	0
22	Spectrum of GALNS mutations and haplotype study in Brazilian patients with Mucopolysaccharidosis type IVA. Meta Gene, 2018, 16, 77-84.	0.6	6
23	Gaucheroma mimicking hepatocellular carcinoma in a cirrhotic type I Gaucher disease patient. Molecular Genetics and Metabolism, 2018, 123, S135.	1.1	0
24	Val66Met polymorphism association with serum BDNF and inflammatory biomarkers in major depression. World Journal of Biological Psychiatry, 2018, 19, 402-409.	2.6	28
25	TP53 p.Arg337His germline mutation prevalence in Southern Brazil: Further evidence for mutation testing in young breast cancer patients. PLoS ONE, 2018, 13, e0209934.	2.5	10
26	Recent advances in molecular testing to improve early diagnosis in children with mucopolysaccharidoses. Expert Review of Molecular Diagnostics, 2018, 18, 855-866.	3.1	10
27	Nonneurological Involvement in Late-Onset Friedreich Ataxia (LOFA): Exploring the Phenotypes. Cerebellum, 2017, 16, 253-256.	2.5	22
28	Elevation of glycosaminoglycans in the amniotic fluid of a fetus with mucopolysaccharidosis VII. Prenatal Diagnosis, 2017, 37, 435-439.	2.3	20
29	Hematopoietic stem cell transplantation for patients with mucopolysaccharidosis type II. Molecular Genetics and Metabolism, 2017, 120, S77.	1.1	0
30	LC/MS/MS measurement of glycosaminoglycans in amniotic fluid of a MPS VII fetus. Molecular Genetics and Metabolism, 2017, 120, S77.	1.1	3
31	Hematopoietic Stem Cell Transplantation for Patients with Mucopolysaccharidosis II. Biology of Blood and Marrow Transplantation, 2017, 23, 1795-1803.	2.0	80
32	Can clinical subtypes contribute to genetic studies on major depression?. Australasian Psychiatry, 2017, 25, 633-634.	0.7	0
33	Intellectual Disability in a Birth Cohort: Prevalence, Etiology, and Determinants at the Age of 4 Years. Public Health Genomics, 2016, 19, 290-297.	1.0	25
34	Current molecular genetics strategies for the diagnosis of lysosomal storage disorders. Expert Review of Molecular Diagnostics, 2016, 16, 113-123.	3.1	13
35	Prevalence of thrombophilia and thrombotic events inpatients with Fabry disease in a reference center forlysosomal disorders in Southern Brazil. Clinical and Biomedical Research, 2016, 36, 23-26.	0.1	0
36	AnÃ¡lise comparativa entre as metodologias de PCR metilaÃ§Ã£o-especÃfica (MSP), Southern blot (SB) e FISH utilizadas no diagnÃ³stico genÃ©tico molecular de pacientes com suspeita clÃnica das sÃndromes de Prader-Willi ou Angelman. Clinical and Biomedical Research, 2016, 36, 71-79.	0.1	0

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37	Diagnostic and treatment strategies in mucopolysaccharidosis VI. The Application of Clinical Genetics, 2015, 8, 245.	3.0	31
38	What can HPA axis-linked genes tell us about anxiety disorders in adolescents?. Trends in Psychiatry and Psychotherapy, 2015, 37, 232-237.	0.8	15
39	MPS I and MPS II: Minimal estimated incidence in Brazil and comparison to the rest of the world. Molecular Genetics and Metabolism, 2015, 114, S43.	1.1	1
40	Genetic causes of intellectual disability in a birth cohort: A populationâ€based study. American Journal of Medical Genetics, Part A, 2015, 167, 1204-1214.	1.2	39
41	Identification of a premature stop codon mutation in the PHGDH gene in severe Neuâ€Laxova syndromeâ€"evidence for phenotypic variability. American Journal of Medical Genetics, Part A, 2015, 167, 1323-1329.	1.2	22
42	Diagnosis and therapy options in mucopolysaccharidosis II (Hunter syndrome). Expert Opinion on Orphan Drugs, 2015, 3, 141-150.	0.8	5
43	Genomic Instability in Human Lymphocytes from Male Users of Crack Cocaine. International Journal of Environmental Research and Public Health, 2014, 11, 10003-10015.	2.6	17
44	Cardiac disease as the presenting feature of mucopolysaccharidosis type IIIA: A case report. Molecular Genetics and Metabolism Reports, 2014, 1, 422-424.	1.1	9
45	A Community-Based Study of Mucopolysaccharidosis Type VI in Brazil: The Influence of Founder Effect, Endogamy and Consanguinity. Human Heredity, 2014, 77, 189-196.	0.8	31
46	Anxiety disorders and anxiety-related traits and serotonin transporter gene-linked polymorphic region (5-HTTLPR) in adolescents. Psychiatric Genetics, 2014, 24, 176-180.	1.1	5
47	Is interictal EEG activity a biomarker for mood disorders in temporal lobe epilepsy?. Clinical Neurophysiology, 2014, 125, 1952-1958.	1.5	14
48	Mucopolysaccharidosis type II: Identification of 30 novel mutations among Latin American patients. Molecular Genetics and Metabolism, 2014, 111, 133-138.	1.1	40
49	Mineralocorticoid receptor genotype moderates the association between physical neglect and serum BDNF. Journal of Psychiatric Research, 2014, 59, 8-13.	3.1	12
50	Biotinidase deficiency: clinical and genetic studies of 38 Brazilian patients. BMC Medical Genetics, 2014, 15, 96.	2.1	16
51	Lack of association between thrombophilic gene variants and recurrent pregnancy loss. Human Fertility, 2014, 17, 99-105.	1.7	17
52	Tryptophan hydroxylase 2 (TPH2) gene polymorphisms and psychiatric comorbidities in temporal lobe epilepsy. Epilepsy and Behavior, 2014, 32, 59-63.	1.7	21
53	Body composition in patients with classical homocystinuria: body mass relates to homocysteine and choline metabolism. Gene, 2014, 546, 443-447.	2.2	15
54	Molecular testing of 163 patients with Morquio A (Mucopolysaccharidosis IVA) identifies 39 novel GALNS mutations. Molecular Genetics and Metabolism, 2014, 112, 160-170.	1.1	52

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55	Fast and robust protocol for prenatal diagnosis of mucopolysaccharidosis Type II. Clinical and Biomedical Research, 2014, 34, 371-373.	0.1	1
56	Analysis of KIR gene frequencies and HLA class I genotypes in breast cancer and control group. Human Immunology, 2013, 74, 1130-1133.	2.4	33
57	Important aspects in the molecular diagnosis of mucopolysaccharidoses. Journal of Inherited Metabolic Disease, 2013, 36, 905-906.	3.6	1
58	Identification of a novel missense mutation in Brazilian patient with a severe form of mucopolysaccharidosis type IVA. Gene, 2013, 517, 112-115.	2.2	6
59	Extension of the molecular analysis to the promoter region of the iduronate 2-sulfatase gene reveals genomic alterations in mucopolysaccharidosis type II patients with normal coding sequence. Gene, 2013, 526, 150-154.	2.2	7
60	Associations between parenting behavior and anxiety in a rodent model and a clinical sample: relationship to peripheral BDNF levels. Translational Psychiatry, 2012, 2, e195-e195.	4.8	80
61	Prevalence of 185delAG and 5382insC mutations in BRCA1, and 6174delT in BRCA2 in women of Ashkenazi Jewish origin in southern Brazil. Genetics and Molecular Biology, 2012, 35, 599-602.	1.3	26
62	Severe phenotype in MPS II patients associated with a large deletion including contiguous genes. American Journal of Medical Genetics, Part A, 2012, 158A, 1055-1059.	1.2	16
63	Serotonin gene polymorphisms and psychiatry comorbidities in temporal lobe epilepsy. Epilepsy Research, 2012, 99, 260-266.	1.6	25
64	Response to: A functional 5-HT1A variant and comorbid anxiety. Epilepsy Research, 2012, 100, 201.	1.6	0
65	Is puberty a trigger for 5HTTLPR polymorphism association with depressive symptoms?. Journal of Psychiatric Research, 2012, 46, 831-833.	3.1	5
66	Evidence of association between Val66Met polymorphism at BDNF gene and anxiety disorders in a community sample of children and adolescents. Neuroscience Letters, 2011, 502, 197-200.	2.1	25
67	Genetic studies in a cluster of Mucopolysaccharidosis Type VI patients in Northeast Brazil. Molecular Genetics and Metabolism, 2011, 104, 603-607.	1.1	34
68	Mucopolysaccharidoses in northern Brazil: Targeted mutation screening and urinary glycosaminoglycan excretion in patients undergoing enzyme replacement therapy. Genetics and Molecular Biology, 2011, 34, 410-415.	1.3	7
69	Detection of human bocavirus and human metapneumovirus by real-time PCR from patients with respiratory symptoms in Southern Brazil. Memorias Do Instituto Oswaldo Cruz, 2011, 106, 56-60.	1.6	37
70	The multidimensional evaluation and treatment of anxiety in children and adolescents: rationale, design, methods and preliminary findings. Revista Brasileira De Psiquiatria, 2011, 33, 181-195.	1.7	42
71	Are MPS II heterozygotes actually asymptomatic? A study based on clinical and biochemical data, Xâ€inactivation analysis and imaging evaluations. American Journal of Medical Genetics, Part A, 2011, 155, 50-57.	1.2	12
72	Serotonin transporter gene (5HTT) polymorphisms and temporal lobe epilepsy. Epilepsy Research, 2011, 95, 152-157.	1.6	26

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73	No major clinical impact of Val66Met BDNF gene polymorphism on temporal lobe epilepsy. Epilepsy Research, 2010, 88, 108-111.	1.6	15
74	Panic disorder and serotonergic genes (SLC6A4, HTR1A and HTR2A): Association and interaction with childhood trauma and parenting. Neuroscience Letters, 2010, 485, 11-15.	2.1	34
75	The BDNF Val66Met polymorphism is an independent risk factor for high lethality in suicide attempts of depressed patients. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2010, 34, 940-944.	4.8	46
76	Emerging research groups studying Brazilian psychiatric genetics. Revista Brasileira De Psiquiatria, 2010, 32, 91-92.	1.7	1
77	Clinical and biochemical studies in mucopolysaccharidosis type II carriers. Journal of Inherited Metabolic Disease, 2009, 32, 732-738.	3.6	12
78	Novel allelic variants in the human serotonin transporter gene linked polymorphism (5-HTTLPR) among depressed patients with suicide attempt. Neuroscience Letters, 2009, 451, 79-82.	2.1	14
79	Preliminary evidence of association between EFHC2, a gene implicated in fear recognition, and harm avoidance. Neuroscience Letters, 2009, 452, 84-86.	2.1	14
80	Analysis of the R72P polymorphism of the TP53 gene in patients with invasive ductal breast carcinoma. Molecular Medicine Reports, 2009, 2, 793-7.	2.4	4
81	Identification of Î² thalassemia mutations in South Brazilians. Annals of Hematology, 2008, 87, 381-384.	1.8	25
82	Lack of association between the serotonin transporter promoter polymorphism (5-HTTLPR) and personality traits in asymptomatic patients with panic disorder. Neuroscience Letters, 2008, 431, 173-178.	2.1	15
83	Prenatal diagnosis of fetal chromosomal abnormalities: report of an 18-year experience in a Brazilian public hospital. Genetics and Molecular Biology, 2008, 31, 829-833.	1.3	10
84	Prevalence of the serpin peptidase inhibitor (alpha-1-antitrypsin) PI*S and PI*Z alleles in Brazilian children with liver disease. Genetics and Molecular Biology, 2008, 31, 423-426.	1.3	1
85	Lack of association between the Serotonin Transporter Promoter Polymorphism (5-HTTLPR) and Panic Disorder: a systematic review and meta-analysis. Behavioral and Brain Functions, 2007, 3, 41.	3.3	59
86	A clinical study of 77 patients with mucopolysaccharidosis type II. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 63-70.	1.5	112
87	Molecular Analysis of the p53 Gene in Patients with Intestinal Metaplasia of the Cardia and Barrett's Esophagus: Characterization by Sequencing. Digestive Diseases and Sciences, 2007, 52, 2183-2185.	2.3	0
88	Analysis of R213R and 13494 gâ†'a polymorphisms of the p53 gene in individuals with esophagitis, intestinal metaplasia of the cardia and Barrettâ€™s Esophagus compared with a control group. Genomic Medicine, 2007, 1, 57-63.	0.3	6
89	Association between suicide attempts in south Brazilian depressed patients with the serotonin transporter polymorphism. Psychiatry Research, 2006, 143, 289-291.	3.3	16
90	The role of methylenetetrahydrofolate reductase in acute lymphoblastic leukemia in a Brazilian mixed population. Leukemia Research, 2006, 30, 477-481.	0.8	61

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91	TP53 gene R72P polymorphism analysis in patients with Barrett esophagus. Cancer Genetics and Cytogenetics, 2006, 170, 76-77.	1.0	3
92	Further cases of â€œneighborâ€•mutations in mucopolysaccharidosis type II. American Journal of Medical Genetics, Part A, 2006, 140A, 1684-1686.	1.2	5
93	Mucopolysaccharidosis type VI: Identification of novel mutations on the arylsulphatase B gene in South American patients. Journal of Inherited Metabolic Disease, 2005, 28, 1027-1034.	3.6	28
94	Clinical and biochemical study of 28 patients with mucopolysaccharidosis type VI. Clinical Genetics, 2004, 66, 208-213.	2.0	103
95	Metabolic effects and the methylenetetrahydrofolate reductase (MTHFR) polymorphism associated with neural tube defects in southern Brazil. Birth Defects Research Part A: Clinical and Molecular Teratology, 2004, 70, 459-463.	1.6	35
96	p53 protein overexpression and p53 mutation analysis in patients with intestinal metaplasia of the cardia and Barrett's esophagus. Cancer Letters, 2004, 210, 213-218.	7.2	20
97	Mucopolysaccharidosis VII: clinical, biochemical and molecular investigation of a Brazilian family. Clinical Genetics, 2003, 64, 172-175.	2.0	11
98	Identification and characterization of 13 new mutations in mucopolysaccharidosis type I patients. Molecular Genetics and Metabolism, 2003, 78, 37-43.	1.1	75
99	Identification of a Novel Mutation in the ARSB Gene That Is Frequent Among Brazilian MPSVI Patients. Genetic Testing and Molecular Biomarkers, 2003, 7, 347-349.	1.7	19
100	Molecular analysis of thePi*Z allele in patients with liver disease. American Journal of Medical Genetics Part A, 2001, 104, 287-290.	2.4	16
101	Pseudodeficiency of arylsulphatase A: Strategy for clarification of genotype in families of subjects with low ASA activity and neurological symptoms. Journal of Inherited Metabolic Disease, 1995, 18, 710-716.	3.6	24
102	The Contribution of Molecular Techniques in Prenatal Diagnosis and Post mortem Fetus with Multiple Malformation. , 0, , .		0