

# Sandra Leistner-Segal

## List of Publications by Year in descending order

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Version: 2024-02-01

102  
papers

1,915  
citations

257450

24  
h-index

315739

38  
g-index

114  
all docs

114  
docs citations

114  
times ranked

2975  
citing authors

#	ARTICLE	IF	CITATIONS
1	A clinical study of 77 patients with mucopolysaccharidosis type II. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2007, 96, 63-70.	1.5	112
2	Clinical and biochemical study of 28 patients with mucopolysaccharidosis type VI. <i>Clinical Genetics</i> , 2004, 66, 208-213.	2.0	103
3	Associations between parenting behavior and anxiety in a rodent model and a clinical sample: relationship to peripheral BDNF levels. <i>Translational Psychiatry</i> , 2012, 2, e195-e195.	4.8	80
4	Hematopoietic Stem Cell Transplantation for Patients with Mucopolysaccharidosis II. <i>Biology of Blood and Marrow Transplantation</i> , 2017, 23, 1795-1803.	2.0	80
5	Identification and characterization of 13 new mutations in mucopolysaccharidosis type I patients. <i>Molecular Genetics and Metabolism</i> , 2003, 78, 37-43.	1.1	75
6	The role of methylenetetrahydrofolate reductase in acute lymphoblastic leukemia in a Brazilian mixed population. <i>Leukemia Research</i> , 2006, 30, 477-481.	0.8	61
7	Lack of association between the Serotonin Transporter Promoter Polymorphism (5-HTTLPR) and Panic Disorder: a systematic review and meta-analysis. <i>Behavioral and Brain Functions</i> , 2007, 3, 41.	3.3	59
8	Molecular testing of 163 patients with Morquio A (Mucopolysaccharidosis IVA) identifies 39 novel GALNS mutations. <i>Molecular Genetics and Metabolism</i> , 2014, 112, 160-170.	1.1	52
9	The BDNF Val66Met polymorphism is an independent risk factor for high lethality in suicide attempts of depressed patients. <i>Progress in Neuro-Psychopharmacology and Biological Psychiatry</i> , 2010, 34, 940-944.	4.8	46
10	Diagnosis of Mucopolysaccharidoses. <i>Diagnostics</i> , 2020, 10, 172.	2.6	46
11	The multidimensional evaluation and treatment of anxiety in children and adolescents: rationale, design, methods and preliminary findings. <i>Revista Brasileira De Psiquiatria</i> , 2011, 33, 181-195.	1.7	42
12	Mucopolysaccharidosis type II: Identification of 30 novel mutations among Latin American patients. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 133-138.	1.1	40
13	Genetic causes of intellectual disability in a birth cohort: A population-based study. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1204-1214.	1.2	39
14	Detection of human bocavirus and human metapneumovirus by real-time PCR from patients with respiratory symptoms in Southern Brazil. <i>Memorias Do Instituto Oswaldo Cruz</i> , 2011, 106, 56-60.	1.6	37
15	Metabolic effects and the methylenetetrahydrofolate reductase (MTHFR) polymorphism associated with neural tube defects in southern Brazil. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2004, 70, 459-463.	1.6	35
16	Panic disorder and serotonergic genes (SLC6A4, HTR1A and HTR2A): Association and interaction with childhood trauma and parenting. <i>Neuroscience Letters</i> , 2010, 485, 11-15.	2.1	34
17	Genetic studies in a cluster of Mucopolysaccharidosis Type VI patients in Northeast Brazil. <i>Molecular Genetics and Metabolism</i> , 2011, 104, 603-607.	1.1	34
18	Analysis of KIR gene frequencies and HLA class I genotypes in breast cancer and control group. <i>Human Immunology</i> , 2013, 74, 1130-1133.	2.4	33

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19	A Community-Based Study of Mucopolysaccharidosis Type VI in Brazil: The Influence of Founder Effect, Endogamy and Consanguinity. <i>Human Heredity</i> , 2014, 77, 189-196.	0.8	31
20	Diagnostic and treatment strategies in mucopolysaccharidosis VI. <i>The Application of Clinical Genetics</i> , 2015, 8, 245.	3.0	31
21	Mucopolysaccharidosis type VI: Identification of novel mutations on the arylsulphatase B gene in South American patients. <i>Journal of Inherited Metabolic Disease</i> , 2005, 28, 1027-1034.	3.6	28
22	Val66Met polymorphism association with serum BDNF and inflammatory biomarkers in major depression. <i>World Journal of Biological Psychiatry</i> , 2018, 19, 402-409.	2.6	28
23	Serotonin transporter gene (5HTT) polymorphisms and temporal lobe epilepsy. <i>Epilepsy Research</i> , 2011, 95, 152-157.	1.6	26
24	Prevalence of 185delAG and 5382insC mutations in BRCA1, and 6174delT in BRCA2 in women of Ashkenazi Jewish origin in southern Brazil. <i>Genetics and Molecular Biology</i> , 2012, 35, 599-602.	1.3	26
25	Identification of $\beta^2$ thalassemia mutations in South Brazilians. <i>Annals of Hematology</i> , 2008, 87, 381-384.	1.8	25
26	Evidence of association between Val66Met polymorphism at BDNF gene and anxiety disorders in a community sample of children and adolescents. <i>Neuroscience Letters</i> , 2011, 502, 197-200.	2.1	25
27	Serotonin gene polymorphisms and psychiatry comorbidities in temporal lobe epilepsy. <i>Epilepsy Research</i> , 2012, 99, 260-266.	1.6	25
28	Intellectual Disability in a Birth Cohort: Prevalence, Etiology, and Determinants at the Age of 4 Years. <i>Public Health Genomics</i> , 2016, 19, 290-297.	1.0	25
29	Pseudodeficiency of arylsulphatase A: Strategy for clarification of genotype in families of subjects with low ASA activity and neurological symptoms. <i>Journal of Inherited Metabolic Disease</i> , 1995, 18, 710-716.	3.6	24
30	Identification of a premature stop codon mutation in the <i>PHGDH</i> gene in severe Neurolaxova syndrome—evidence for phenotypic variability. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1323-1329.	1.2	22
31	Nonneurological Involvement in Late-Onset Friedreich Ataxia (LOFA): Exploring the Phenotypes. <i>Cerebellum</i> , 2017, 16, 253-256.	2.5	22
32	Tryptophan hydroxylase 2 (TPH2) gene polymorphisms and psychiatric comorbidities in temporal lobe epilepsy. <i>Epilepsy and Behavior</i> , 2014, 32, 59-63.	1.7	21
33	p53 protein overexpression and p53 mutation analysis in patients with intestinal metaplasia of the cardia and Barrett's esophagus. <i>Cancer Letters</i> , 2004, 210, 213-218.	7.2	20
34	Elevation of glycosaminoglycans in the amniotic fluid of a fetus with mucopolysaccharidosis VII. <i>Prenatal Diagnosis</i> , 2017, 37, 435-439.	2.3	20
35	Identification of a Novel Mutation in the ARSB Gene That Is Frequent Among Brazilian MPSVI Patients. <i>Genetic Testing and Molecular Biomarkers</i> , 2003, 7, 347-349.	1.7	19
36	Genomic Instability in Human Lymphocytes from Male Users of Crack Cocaine. <i>International Journal of Environmental Research and Public Health</i> , 2014, 11, 10003-10015.	2.6	17

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37	Lack of association between thrombophilic gene variants and recurrent pregnancy loss. <i>Human Fertility</i> , 2014, 17, 99-105.	1.7	17
38	Molecular characterization of a large group of Mucopolysaccharidosis type IIIc patients reveals the evolutionary history of the disease. <i>Human Mutation</i> , 2019, 40, 1084-1100.	2.5	17
39	Molecular analysis of the Pi*Z allele in patients with liver disease. <i>American Journal of Medical Genetics Part A</i> , 2001, 104, 287-290.	2.4	16
40	Association between suicide attempts in south Brazilian depressed patients with the serotonin transporter polymorphism. <i>Psychiatry Research</i> , 2006, 143, 289-291.	3.3	16
41	Severe phenotype in MPS II patients associated with a large deletion including contiguous genes. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1055-1059.	1.2	16
42	Biotinidase deficiency: clinical and genetic studies of 38 Brazilian patients. <i>BMC Medical Genetics</i> , 2014, 15, 96.	2.1	16
43	Lack of association between the serotonin transporter promoter polymorphism (5-HTTLPR) and personality traits in asymptomatic patients with panic disorder. <i>Neuroscience Letters</i> , 2008, 431, 173-178.	2.1	15
44	No major clinical impact of Val66Met BDNF gene polymorphism on temporal lobe epilepsy. <i>Epilepsy Research</i> , 2010, 88, 108-111.	1.6	15
45	Body composition in patients with classical homocystinuria: body mass relates to homocysteine and choline metabolism. <i>Gene</i> , 2014, 546, 443-447.	2.2	15
46	What can HPA axis-linked genes tell us about anxiety disorders in adolescents?. <i>Trends in Psychiatry and Psychotherapy</i> , 2015, 37, 232-237.	0.8	15
47	Novel allelic variants in the human serotonin transporter gene linked polymorphism (5-HTTLPR) among depressed patients with suicide attempt. <i>Neuroscience Letters</i> , 2009, 451, 79-82.	2.1	14
48	Preliminary evidence of association between EFHC2, a gene implicated in fear recognition, and harm avoidance. <i>Neuroscience Letters</i> , 2009, 452, 84-86.	2.1	14
49	Is interictal EEG activity a biomarker for mood disorders in temporal lobe epilepsy?. <i>Clinical Neurophysiology</i> , 2014, 125, 1952-1958.	1.5	14
50	Current molecular genetics strategies for the diagnosis of lysosomal storage disorders. <i>Expert Review of Molecular Diagnostics</i> , 2016, 16, 113-123.	3.1	13
51	Analysis of Predictive Biomarkers in Patients With Lung Adenocarcinoma From Southern Brazil Reveals a Distinct Profile From Other Regions of the Country. <i>Journal of Global Oncology</i> , 2019, 5, 1-9.	0.5	13
52	Updated birth prevalence and relative frequency of mucopolysaccharidoses across Brazilian regions. <i>Genetics and Molecular Biology</i> , 2021, 44, e20200138.	1.3	13
53	Clinical and biochemical studies in mucopolysaccharidosis type II carriers. <i>Journal of Inherited Metabolic Disease</i> , 2009, 32, 732-738.	3.6	12
54	Are MPS II heterozygotes actually asymptomatic? A study based on clinical and biochemical data, Xa€inactivation analysis and imaging evaluations. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 50-57.	1.2	12

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55	Mineralocorticoid receptor genotype moderates the association between physical neglect and serum BDNF. <i>Journal of Psychiatric Research</i> , 2014, 59, 8-13.	3.1	12
56	Mucopolysaccharidosis VII: clinical, biochemical and molecular investigation of a Brazilian family. <i>Clinical Genetics</i> , 2003, 64, 172-175.	2.0	11
57	Phenotype-oriented NGS panels for mucopolysaccharidoses: Validation and potential use in the diagnostic flowchart. <i>Genetics and Molecular Biology</i> , 2019, 42, 207-214.	1.3	11
58	Glatiramer acetate increases T- and B-regulatory cells and decreases granulocyte-macrophage colony-stimulating factor (GM-CSF) in an animal model of multiple sclerosis. <i>Journal of Neuroimmunology</i> , 2020, 345, 577281.	2.3	11
59	Prenatal diagnosis of fetal chromosomal abnormalities: report of an 18-year experience in a Brazilian public hospital. <i>Genetics and Molecular Biology</i> , 2008, 31, 829-833.	1.3	10
60	TP53 p.Arg337His germline mutation prevalence in Southern Brazil: Further evidence for mutation testing in young breast cancer patients. <i>PLoS ONE</i> , 2018, 13, e0209934.	2.5	10
61	Recent advances in molecular testing to improve early diagnosis in children with mucopolysaccharidoses. <i>Expert Review of Molecular Diagnostics</i> , 2018, 18, 855-866.	3.1	10
62	Is the gut microbiota dysbiotic in patients with classical homocystinuria?. <i>Biochimie</i> , 2020, 173, 3-11.	2.6	10
63	Cardiac disease as the presenting feature of mucopolysaccharidosis type IIIA: A case report. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 422-424.	1.1	9
64	Information and Diagnosis Networks “ tools to improve diagnosis and treatment for patients with rare genetic diseases. <i>Genetics and Molecular Biology</i> , 2019, 42, 155-164.	1.3	9
65	Population medical genetics: translating science to the community. <i>Genetics and Molecular Biology</i> , 2019, 42, 312-320.	1.3	8
66	Mucopolysaccharidoses in northern Brazil: Targeted mutation screening and urinary glycosaminoglycan excretion in patients undergoing enzyme replacement therapy. <i>Genetics and Molecular Biology</i> , 2011, 34, 410-415.	1.3	7
67	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. <i>Gene</i> , 2013, 526, 150-154.	2.2	7
68	Analysis of R213R and 13494 G>A polymorphisms of the p53 gene in individuals with esophagitis, intestinal metaplasia of the cardia and Barrett’s Esophagus compared with a control group. <i>Genomic Medicine</i> , 2007, 1, 57-63.	0.3	6
69	Identification of a novel missense mutation in Brazilian patient with a severe form of mucopolysaccharidosis type IVA. <i>Gene</i> , 2013, 517, 112-115.	2.2	6
70	Spectrum of GALNS mutations and haplotype study in Brazilian patients with Mucopolysaccharidosis type IVA. <i>Meta Gene</i> , 2018, 16, 77-84.	0.6	6
71	Genotype-phenotype studies in a large cohort of Brazilian patients with Hunter syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2021, 187, 349-356.	1.6	6
72	Further cases of “neighbor” mutations in mucopolysaccharidosis type II. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1684-1686.	1.2	5

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73	Is puberty a trigger for 5HTTLPR polymorphism association with depressive symptoms?. Journal of Psychiatric Research, 2012, 46, 831-833.	3.1	5
74	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
75	Diagnosis and therapy options in mucopolysaccharidosis II (Hunter syndrome). Expert Opinion on Orphan Drugs, 2015, 3, 141-150.	0.8	5
76	Friedreich Ataxia: Diagnostic Yield and Minimal Frequency in South Brazil. Cerebellum, 2019, 18, 147-151.	2.5	5
77	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
78	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
79	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
80	TP53 gene R72P polymorphism analysis in patients with Barrett esophagus. Cancer Genetics and Cytogenetics, 2006, 170, 76-77.	1.0	3
81	LC/MS/MS measurement of glycosaminoglycans in amniotic fluid of a MPS VII fetus. Molecular Genetics and Metabolism, 2017, 120, S77.	1.1	3
82	Genetic Analysis of Hereditary Ataxias in Peru Identifies SCA10 Families with Incomplete Penetrance. Cerebellum, 2020, 19, 208-215.	2.5	3
83	Sanfilippo syndrome type B: Analysis of patients diagnosed by the <sc>MPS</sc> Brazil Network. American Journal of Medical Genetics, Part A, 2022, 188, 760-767.	1.2	3
84	Important aspects in the molecular diagnosis of mucopolysaccharidoses. Journal of Inherited Metabolic Disease, 2013, 36, 905-906.	3.6	1
85	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
86	MPS Brazil Network: A summary of all mucopolysaccharidosis type IIIB patients. Molecular Genetics and Metabolism, 2019, 126, S88.	1.1	1
87	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
88	Emerging research groups studying Brazilian psychiatric genetics. Revista Brasileira De Psiquiatria, 2010, 32, 91-92.	1.7	1
89	Fast and robust protocol for prenatal diagnosis of mucopolysaccharidosis Type II. Clinical and Biomedical Research, 2014, 34, 371-373.	0.1	1
90	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

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91	Molecular Analysis of the p53 Gene in Patients with Intestinal Metaplasia of the Cardia and Barrett's Esophagus: Characterization by Sequencing. <i>Digestive Diseases and Sciences</i> , 2007, 52, 2183-2185.	2.3	0
92	The Contribution of Molecular Techniques in Prenatal Diagnosis and Post mortem Fetus with Multiple Malformation. , 0, , .		0
93	Response to: A functional 5-HT1A variant and comorbid anxiety. <i>Epilepsy Research</i> , 2012, 100, 201.	1.6	0
94	Hematopoietic stem cell transplantation for patients with mucopolysaccharidosis type II. <i>Molecular Genetics and Metabolism</i> , 2017, 120, S77.	1.1	0
95	Can clinical subtypes contribute to genetic studies on major depression?. <i>Australasian Psychiatry</i> , 2017, 25, 633-634.	0.7	0
96	Gaucheroma mimicking hepatocellular carcinoma in a cirrhotic type I Gaucher disease patient. <i>Molecular Genetics and Metabolism</i> , 2018, 123, S135.	1.1	0
97	Back Cover, Volume 40, Issue 8. <i>Human Mutation</i> , 2019, 40, ii-ii.	2.5	0
98	Polymorphic variants (p.Ser141Ser and p.Arg737Gly) at the NAGLU gene are really indicative of pseudodeficiency alleles?. <i>Italian Journal of Pediatrics</i> , 2019, 45, 60.	2.6	0
99	Identification of MPS clusters in Latin America: An opportunity for targeted health care programs. <i>Molecular Genetics and Metabolism</i> , 2019, 126, S87-S88.	1.1	0
100	Prevalence of thrombophilia and thrombotic events inpatients with Fabry disease in a reference center forlysosomal disorders in Southern Brazil. <i>Clinical and Biomedical Research</i> , 2016, 36, 23-26.	0.1	0
101	Análise comparativa entre as metodologias de PCR metilase-específica (MSP), Southern blot (SB) e FISH utilizadas no diagnóstico genético molecular de pacientes com suspeita clínica das síndromes de Prader-Willi ou Angelman. <i>Clinical and Biomedical Research</i> , 2016, 36, 71-79.	0.1	0
102	Detection of Mosaic Variants in Mothers of MPS II Patients by Next Generation Sequencing. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 789350.	3.5	0