

# Sandra D'Alfonso

## List of Publications by Year in descending order

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164  
papers

15,245  
citations

28274

55  
h-index

19749

117  
g-index

173  
all docs

173  
docs citations

173  
times ranked

20158  
citing authors

#	ARTICLE	IF	CITATIONS
1	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. <i>Nature</i> , 2011, 476, 214-219.	27.8	2,400
2	Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis. <i>Nature Genetics</i> , 2013, 45, 1353-1360.	21.4	1,213
3	Multiple sclerosis genomic map implicates peripheral immune cells and microglia in susceptibility. <i>Science</i> , 2019, 365, .	12.6	710
4	Cytokine gene polymorphism in human disease: on-line databases. <i>Genes and Immunity</i> , 1999, 1, 3-19.	4.1	560
5	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	8.1	517
6	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	21.4	494
7	Functional variants in the B-cell gene BANK1 are associated with systemic lupus erythematosus. <i>Nature Genetics</i> , 2008, 40, 211-216.	21.4	436
8	A polymorphic variation in a putative regulation box of the TNFA promoter region. <i>Immunogenetics</i> , 1994, 39, 150-4.	2.4	317
9	Transancestral mapping and genetic load in systemic lupus erythematosus. <i>Nature Communications</i> , 2017, 8, 16021.	12.8	314
10	Class II HLA interactions modulate genetic risk for multiple sclerosis. <i>Nature Genetics</i> , 2015, 47, 1107-1113.	21.4	312
11	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331.	8.1	308
12	Overexpression of the Cytokine BAFF and Autoimmunity Risk. <i>New England Journal of Medicine</i> , 2017, 376, 1615-1626.	27.0	301
13	Fine-Mapping the Genetic Association of the Major Histocompatibility Complex in Multiple Sclerosis: HLA and Non-HLA Effects. <i>PLoS Genetics</i> , 2013, 9, e1003926.	3.5	250
14	Cytokine gene polymorphism in human disease: on-line databases, Supplement 1. <i>Genes and Immunity</i> , 2001, 2, 61-70.	4.1	248
15	Cytokine gene polymorphism in human disease: on-line databases, Supplement 2. <i>Genes and Immunity</i> , 2002, 3, 313-330.	4.1	225
16	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	21.4	223
17	High frequency of <i>TARDBP</i> gene mutations in Italian patients with amyotrophic lateral sclerosis. <i>Human Mutation</i> , 2009, 30, 688-694.	2.5	184
18	Network-Based Multiple Sclerosis Pathway Analysis with GWAS Data from 15,000 Cases and 30,000 Controls. <i>American Journal of Human Genetics</i> , 2013, 92, 854-865.	6.2	164

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19	Functional Analysis of a New Polymorphism in the Human TNF $\beta$ Gene Promoter. <i>Scandinavian Journal of Immunology</i> , 1995, 42, 501-504.	2.7	161
20	Concordance, disease progression, and heritability of coeliac disease in Italian twins. <i>Gut</i> , 2006, 55, 803-808.	12.1	155
21	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. <i>Journal of Medical Genetics</i> , 2010, 47, 190-194.	3.2	152
22	The multistep hypothesis of ALS revisited. <i>Neurology</i> , 2018, 91, e635-e642.	1.1	146
23	Extensive genetics of ALS. <i>Neurology</i> , 2012, 79, 1983-1989.	1.1	145
24	STAT4 associates with systemic lupus erythematosus through two independent effects that correlate with gene expression and act additively with IRF5 to increase risk. <i>Annals of the Rheumatic Diseases</i> , 2009, 68, 1746-1753.	0.9	138
25	Genetic association of miRNA-146a with systemic lupus erythematosus in Europeans through decreased expression of the gene. <i>Genes and Immunity</i> , 2012, 13, 268-274.	4.1	132
26	Replication of recently identified systemic lupus erythematosus genetic associations: a case-control study. <i>Arthritis Research and Therapy</i> , 2009, 11, R69.	3.5	131
27	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2014, 23, 2220-2231.	2.9	123
28	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2019, 85, 470-481.	5.3	118
29	Low-Frequency and Rare-Coding Variation Contributes to Multiple Sclerosis Risk. <i>Cell</i> , 2018, 175, 1679-1687.e7.	28.9	115
30	Cognitive impairment across ALS clinical stages in a population-based cohort. <i>Neurology</i> , 2019, 93, e984-e994.	1.1	115
31	Kallikrein genes are associated with lupus and glomerular basement membrane-specific antibody-induced nephritis in mice and humans. <i>Journal of Clinical Investigation</i> , 2009, 119, 911-923.	8.2	114
32	A loss-of-function variant of PTPN22 is associated with reduced risk of systemic lupus erythematosus. <i>Human Molecular Genetics</i> , 2008, 18, 569-579.	2.9	106
33	A systemic sclerosis and systemic lupus erythematosus pan-meta-GWAS reveals new shared susceptibility loci. <i>Human Molecular Genetics</i> , 2013, 22, 4021-4029.	2.9	104
34	Systemic lupus erythematosus candidate genes in the Italian population: Evidence for a significant association with interleukin-10. <i>Arthritis and Rheumatism</i> , 2000, 43, 120-128.	6.7	103
35	ALS phenotype is influenced by age, sex, and genetics. <i>Neurology</i> , 2020, 94, e802-e810.	1.1	99
36	New polymorphisms in the IL-10 promoter region. <i>Genes and Immunity</i> , 2000, 1, 231-233.	4.1	98

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37	A genome screen for multiple sclerosis in Sardinian multiplex families. <i>European Journal of Human Genetics</i> , 2001, 9, 621-626.	2.8	95
38	Two single-nucleotide polymorphisms in the 5' and 3' ends of the osteopontin gene contribute to susceptibility to systemic lupus erythematosus. <i>Arthritis and Rheumatism</i> , 2005, 52, 539-547.	6.7	94
39	Refining genetic associations in multiple sclerosis. <i>Lancet Neurology</i> , The, 2008, 7, 567-569.	10.2	90
40	Neuropeptide S Receptor 1 Gene Polymorphism Is Associated With Susceptibility to Inflammatory Bowel Disease. <i>Gastroenterology</i> , 2007, 133, 808-817.	1.3	87
41	Analysis of autosomal genes reveals gene×sex interactions and higher total genetic risk in men with systemic lupus erythematosus. <i>Annals of the Rheumatic Diseases</i> , 2012, 71, 694-699.	0.9	87
42	Novel optineurin mutations in patients with familial and sporadic amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2011, 82, 1239-1243.	1.9	86
43	Next Generation Sequencing of Pooled Samples: Guideline for Variants™ Filtering. <i>Scientific Reports</i> , 2016, 6, 33735.	3.3	81
44	Multiple sclerosis risk loci and disease severity in 7,125 individuals from 10 studies. <i>Neurology: Genetics</i> , 2016, 2, e87.	1.9	76
45	C9ORF72 repeat expansion in a large Italian ALS cohort: evidence of a founder effect. <i>Neurobiology of Aging</i> , 2012, 33, 2528.e7-2528.e14.	3.1	74
46	Ubiquilin 2 mutations in Italian patients with amyotrophic lateral sclerosis and frontotemporal dementia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 183-187.	1.9	74
47	A genome screen for multiple sclerosis in Italian families. <i>Genes and Immunity</i> , 2001, 2, 205-210.	4.1	70
48	IL12A, MPHOSPH9/CDK2AP1 and RGS1 are novel multiple sclerosis susceptibility loci. <i>Genes and Immunity</i> , 2010, 11, 397-405.	4.1	70
49	Association between polymorphisms in the TNF region and systemic lupus erythematosus in the Italian population. <i>Tissue Antigens</i> , 1996, 47, 551-555.	1.0	68
50	Genetic and physical interaction of the B-cell systemic lupus erythematosus-associated genes BANK1 and BLK. <i>Annals of the Rheumatic Diseases</i> , 2012, 71, 136-142.	0.9	67
51	HLA×multiple sclerosis association in Continental Italy and correlation with disease prevalence in Europe. <i>Journal of Neuroimmunology</i> , 2004, 150, 178-185.	2.3	66
52	Osteopontin gene haplotypes correlate with multiple sclerosis development and progression. <i>Journal of Neuroimmunology</i> , 2005, 163, 172-178.	2.3	66
53	A Candidate-Interactome×Aggregate Analysis of Genome-Wide Association Data in Multiple Sclerosis. <i>PLoS ONE</i> , 2013, 8, e63300.	2.5	66
54	The Genetic Association of Variants in CD6, TNFRSF1A and IRF8 to Multiple Sclerosis: A Multicenter Case-Control Study. <i>PLoS ONE</i> , 2011, 6, e18813.	2.5	63

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55	Mitochondrial DNA sequence variation in multiple sclerosis. <i>Neurology</i> , 2015, 85, 325-330.	1.1	60
56	Association of a Locus in the <i>CAMTA1</i> Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2016, 73, 812.	9.0	57
57	Immunoproteasome LMP2 60HH Variant Alters MBP Epitope Generation and Reduces the Risk to Develop Multiple Sclerosis in Italian Female Population. <i>PLoS ONE</i> , 2010, 5, e9287.	2.5	56
58	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. <i>Neuron</i> , 2021, 109, 448-460.e4.	8.1	56
59	Identification of a new putative functional IL18 gene variant through an association study in systemic lupus erythematosus. <i>Human Molecular Genetics</i> , 2009, 18, 3739-3748.	2.9	54
60	Genetic variants are major determinants of CSF antibody levels in multiple sclerosis. <i>Brain</i> , 2015, 138, 632-643.	7.6	54
61	ATXN-2 CAG repeat expansions are interrupted in ALS patients. <i>Human Genetics</i> , 2011, 130, 575-580.	3.8	52
62	<i>ATXN2</i> polyQ intermediate repeats are a modifier of ALS survival. <i>Neurology</i> , 2015, 84, 251-258.	1.1	52
63	HLA-class I markers and multiple sclerosis susceptibility in the Italian population. <i>Genes and Immunity</i> , 2010, 11, 173-180.	4.1	51
64	A novel peripherin gene (PRPH) mutation identified in one sporadic amyotrophic lateral sclerosis patient. <i>Neurobiology of Aging</i> , 2011, 32, 552.e1-552.e6.	3.1	49
65	A 3' untranslated region variant is associated with impaired expression of <i>CD226</i> in T and natural killer T cells and is associated with susceptibility to systemic lupus erythematosus. <i>Arthritis and Rheumatism</i> , 2010, 62, 3404-3414.	6.7	48
66	Association of Variants in the <i>SPTLC1</i> Gene With Juvenile Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2021, 78, 1236.	9.0	46
67	Chitotriosidase and lysosomal enzymes as potential biomarkers of disease progression in amyotrophic lateral sclerosis: A survey clinic-based study. <i>Journal of the Neurological Sciences</i> , 2015, 348, 245-250.	0.6	45
68	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. <i>Journal of Neurology</i> , 2015, 262, 1376-1378.	3.6	44
69	Epstein-Barr virus genetic variants are associated with multiple sclerosis. <i>Neurology</i> , 2015, 84, 1362-1368.	1.1	44
70	HLA alleles modulate EBV viral load in multiple sclerosis. <i>Journal of Translational Medicine</i> , 2018, 16, 80.	4.4	44
71	Polymorphisms in the genes coding for iron binding and transporting proteins are associated with disability, severity, and early progression in multiple sclerosis. <i>BMC Medical Genetics</i> , 2012, 13, 70.	2.1	42
72	Replication of the TNFSF4 (OX40L) promoter region association with systemic lupus erythematosus. <i>Genes and Immunity</i> , 2009, 10, 248-253.	4.1	41

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73	SOD1 gene mutations in Italian patients with Sporadic Amyotrophic Lateral Sclerosis (ALS). <i>Neuromuscular Disorders</i> , 2006, 16, 800-804.	0.6	40
74	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. <i>Neurobiology of Aging</i> , 2016, 43, 180.e1-180.e5.	3.1	40
75	Variations of the perforin gene in patients with multiple sclerosis. <i>Genes and Immunity</i> , 2008, 9, 438-444.	4.1	39
76	Linkage analysis of multiple sclerosis with candidate region markers in Sardinian and Continental Italian families. <i>European Journal of Human Genetics</i> , 1999, 7, 377-385.	2.8	38
77	Genetics of Multiple Sclerosis. <i>Molecular Diagnosis and Therapy</i> , 2002, 2, 37-58.	3.3	37
78	Variations in the coding and regulatory sequences of the angiogenin (ANG) gene are not associated to ALS (amyotrophic lateral sclerosis) in the Italian population. <i>Journal of the Neurological Sciences</i> , 2007, 258, 123-127.	0.6	37
79	Association tests with systemic lupus erythematosus (SLE) of IL10 markers indicate a direct involvement of a CA repeat in the 5' regulatory region. <i>Genes and Immunity</i> , 2002, 3, 454-463.	4.1	36
80	Power estimation for non-standardized multisite studies. <i>NeuroImage</i> , 2016, 134, 281-294.	4.2	36
81	Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. <i>Neurobiology of Aging</i> , 2013, 34, 1517.e9-1517.e10.	3.1	35
82	Prolactin and prolactin receptor gene polymorphisms in multiple sclerosis and systemic lupus erythematosus. <i>Human Immunology</i> , 2003, 64, 274-284.	2.4	34
83	Rare Variants in the <i>TREX1</i> Gene and Susceptibility to Autoimmune Diseases. <i>BioMed Research International</i> , 2013, 2013, 1-6.	1.9	34
84	Bias in association studies of systemic lupus erythematosus susceptibility due to geographical variation in the frequency of a programmed cell death 1 polymorphism across Europe. <i>Genes and Immunity</i> , 2007, 8, 138-146.	4.1	33
85	Fine mapping and conditional analysis identify a new mutation in the autoimmunity susceptibility gene BLK that leads to reduced half-life of the BLK protein. <i>Annals of the Rheumatic Diseases</i> , 2012, 71, 1219-1226.	0.9	33
86	An intragenic polymorphism in the human tumor necrosis factor alpha (TNFA) chain-encoding gene. <i>Immunogenetics</i> , 1996, 44, 321-322.	2.4	32
87	Promoter Insertion/Deletion in the <i>IRF5</i> Gene Is Highly Associated with Susceptibility to Systemic Lupus Erythematosus in Distinct Populations, But Exerts a Modest Effect on Gene Expression in Peripheral Blood Mononuclear Cells. <i>Journal of Rheumatology</i> , 2010, 37, 574-578.	2.0	32
88	Association of osteopontin regulatory polymorphisms with systemic sclerosis. <i>Human Immunology</i> , 2011, 72, 930-934.	2.4	32
89	Functional variations modulating PRKCA expression and alternative splicing predispose to multiple sclerosis. <i>Human Molecular Genetics</i> , 2014, 23, 6746-6761.	2.9	32
90	The Impact of Osteopontin Gene Variations on Multiple Sclerosis Development and Progression. <i>Clinical and Developmental Immunology</i> , 2012, 2012, 1-6.	3.3	31

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91	No evidence for a role of rare <i>CYP27B1</i> functional variations in multiple sclerosis. <i>Annals of Neurology</i> , 2013, 73, 433-437.	5.3	31
92	Gender-specific influence of the chromosome 16 chemokine gene cluster on the susceptibility to Multiple Sclerosis. <i>Journal of the Neurological Sciences</i> , 2008, 267, 86-90.	0.6	30
93	Multiple Polymorphisms Affect Expression and Function of the Neuropeptide S Receptor (NPSR1). <i>PLoS ONE</i> , 2011, 6, e29523.	2.5	30
94	Analysis of hnrNPA1, A2/B1, and A3 genes in patients with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2013, 34, 2695.e11-2695.e12.	3.1	30
95	No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. <i>Neurobiology of Aging</i> , 2011, 32, 966-967.	3.1	28
96	Association of Genetic Markers with CSF Oligoclonal Bands in Multiple Sclerosis Patients. <i>PLoS ONE</i> , 2013, 8, e64408.	2.5	27
97	A non-synonymous SNP within membrane metalloendopeptidase-like 1 (MMEL1) is associated with multiple sclerosis. <i>Genes and Immunity</i> , 2010, 11, 660-664.	4.1	25
98	IL12B Polymorphism and Type 1 Diabetes in the Italian Population: A Case-Control Study. <i>Diabetes</i> , 2002, 51, 1649-1650.	0.6	24
99	ICOS gene haplotypes correlate with IL10 secretion and multiple sclerosis evolution. <i>Journal of Neuroimmunology</i> , 2007, 186, 193-198.	2.3	24
100	PXKlocus in systemic lupus erythematosus: fine mapping and functional analysis reveals novel susceptibility gene ABHD6. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, e14-e14.	0.9	24
101	Lack of support for association between the KIF1B rs10492972[C] variant and multiple sclerosis. <i>Nature Genetics</i> , 2010, 42, 469-470.	21.4	23
102	Mutational Analysis of Known ALS Genes in an Italian Population-Based Cohort. <i>Neurology</i> , 2021, 96, e600-e609.	1.1	23
103	MICA and MICB microsatellite alleles in HLA extended haplotypes. <i>International Journal of Immunogenetics</i> , 2001, 28, 523-530.	1.2	22
104	The Osteopontin Gene +1239A/C Single Nucleotide Polymorphism is Associated with Type 1 Diabetes Mellitus in the Italian Population. <i>International Journal of Immunopathology and Pharmacology</i> , 2010, 23, 263-269.	2.1	21
105	Epstein-Barr virus-associated immune reconstitution inflammatory syndrome as possible cause of fulminant multiple sclerosis relapse after natalizumab interruption. <i>Journal of Neuroimmunology</i> , 2018, 319, 9-12.	2.3	21
106	The Length of SNCA Rep1 Microsatellite May Influence Cognitive Evolution in Parkinson's Disease. <i>Frontiers in Neurology</i> , 2018, 9, 213.	2.4	21
107	Fas Gene Polymorphisms Are Not Associated With Systemic Lupus Erythematosus, Multiple Sclerosis And Hiv Infection. <i>Disease Markers</i> , 1998, 13, 221-225.	1.3	20
108	A sequence variation in the MOG gene is involved in multiple sclerosis susceptibility in Italy. <i>Genes and Immunity</i> , 2008, 9, 7-15.	4.1	20

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109	Altered biosynthesis of tumour necrosis factor (TNF) alpha is involved in postburn hypertrophic scars. <i>Burns</i> , 1994, 20, 118-121.	1.9	19
110	Association study of a new polymorphism in the PECAM-1 gene in multiple sclerosis. <i>Journal of Neuroimmunology</i> , 2000, 104, 174-178.	2.3	19
111	Regional spreading of symptoms at diagnosis as a prognostic marker in amyotrophic lateral sclerosis: a population-based study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 291-297.	1.9	18
112	The IL12B gene does not confer susceptibility to coeliac disease. <i>Tissue Antigens</i> , 2002, 59, 70-72.	1.0	17
113	A whole genome screen for linkage disequilibrium in multiple sclerosis performed in a continental Italian population. <i>Journal of Neuroimmunology</i> , 2003, 143, 97-100.	2.3	17
114	Analysis of the KIFAP3 gene in amyotrophic lateral sclerosis: a multicenter survival study. <i>Neurobiology of Aging</i> , 2014, 35, 2420.e13-2420.e14.	3.1	16
115	Metal(loid)s role in the pathogenesis of amyotrophic lateral sclerosis: Environmental, epidemiological, and genetic data. <i>Environmental Research</i> , 2021, 192, 110292.	7.5	16
116	The Impact of Lifetime Alcohol and Cigarette Smoking Loads on Multiple Sclerosis Severity. <i>Frontiers in Neurology</i> , 2019, 10, 866.	2.4	14
117	Association of Systemic Lupus Erythematosus Clinical Features with European Population Genetic Substructure. <i>PLoS ONE</i> , 2011, 6, e29033.	2.5	14
118	HLA class I in acute promyelocytic leukemia (APL): possible correlation with clinical outcome. <i>Leukemia</i> , 2000, 14, 393-398.	7.2	13
119	Early onset frontotemporal dementia with psychiatric presentation due to the C9ORF72 hexanucleotide repeat expansion: a case report. <i>BMC Neurology</i> , 2014, 14, 228.	1.8	13
120	Inverse correlation of genetic risk score with age at onset in bout-onset and progressive-onset multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2015, 21, 1463-1467.	3.0	13
121	HLA supratypes in an Italian population. <i>Immunogenetics</i> , 1994, 39, 114-20.	2.4	12
122	Distribution of Tumor Necrosis Factor Alleles (NcoI RFLP) and their Relationship to HLA Haplotypes in an Italian Population. <i>Human Heredity</i> , 1993, 43, 103-110.	0.8	11
123	Identification of single nucleotide variations in the coding and regulatory regions of the myelin-associated glycoprotein gene and study of their association with multiple sclerosis. <i>Journal of Neuroimmunology</i> , 2002, 126, 196-204.	2.3	11
124	Association study of <i>IRAK-M</i> and <i>SIGIRR</i> genes with SLE in a large European-descent population. <i>Lupus</i> , 2012, 21, 1166-1171.	1.6	11
125	Genetic burden of common variants in progressive and bout-onset multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2014, 20, 802-811.	3.0	11
126	The natural history of an HLA haplotype and its recombinants. <i>Immunogenetics</i> , 1998, 48, 8-15.	2.4	10



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127	The burden of multiple sclerosis variants in continental Italians and Sardinians. <i>Multiple Sclerosis Journal</i> , 2015, 21, 1385-1395.	3.0	10
128	The first case of the <i>TARDBP</i> p.G294V mutation in a homozygous state: is a single pathogenic allele sufficient to cause ALS?. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2020, 21, 273-279.	1.7	10
129	Gametic association of HSP70 promoter region alleles and their inclusion in extended HLA haplotypes. <i>Tissue Antigens</i> , 1993, 42, 62-66.	1.0	9
130	Association of HLA class I markers with multiple sclerosis in the Italian and UK population: evidence of two independent protective effects. <i>Journal of Medical Genetics</i> , 2011, 48, 485-492.	3.2	9
131	Association of the CBLB gene with multiple sclerosis: new evidence from a replication study in an Italian population. <i>Journal of Medical Genetics</i> , 2011, 48, 210-211.	3.2	9
132	Novel association of acid phosphatase locus 1* <i>C</i> allele with systemic lupus erythematosus. <i>Human Immunology</i> , 2012, 73, 107-110.	2.4	9
133	The -346T polymorphism of the SH2D1A gene is a risk factor for development of autoimmunity/lymphoproliferation in males with defective Fas function. <i>Human Immunology</i> , 2012, 73, 585-592.	2.4	9
134	A Census of Tandemly Repeated Polymorphic Loci in Genic Regions Through the Comparative Integration of Human Genome Assemblies. <i>Frontiers in Genetics</i> , 2018, 9, 155.	2.3	9
135	Vitamin D Supplementation Modulates ICOS+ and ICOS <sup>~</sup> Regulatory T Cell in Siblings of Children With Type 1 Diabetes. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e4767-e4777.	3.6	9
136	Familial clustering in Italian progressive-onset and bout-onset multiple sclerosis. <i>Neurological Sciences</i> , 2014, 35, 789-791.	1.9	8
137	Analysis of the GCG repeat length in NIPA1 gene in C9orf72-mediated ALS in a large Italian ALS cohort. <i>Neurological Sciences</i> , 2019, 40, 2537-2540.	1.9	7
138	VPS54 genetic analysis in ALS Italian cohort. <i>European Journal of Neurology</i> , 2011, 18, e41-e42.	3.3	6
139	ATNX2 is not a regulatory gene in Italian amyotrophic lateral sclerosis patients with C9ORF72 GGGGCC expansion. <i>Neurobiology of Aging</i> , 2016, 39, 218.e5-218.e8.	3.1	6
140	C9ORF72 Repeat Expansion Affects the Proteome of Primary Skin Fibroblasts in ALS. <i>International Journal of Molecular Sciences</i> , 2021, 22, 10385.	4.1	6
141	HLA Genotyping: Methods for the Identification of the HLA-DQ2,-DQ8 Heterodimers Implicated in Celiac Disease (CD) Susceptibility. <i>Methods in Molecular Biology</i> , 2015, 1326, 79-92.	0.9	5
142	A case of late-onset OCD developing PLS and FTD. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2018, 19, 463-465.	1.7	5
143	Validation of an Algorithm to Detect Multiple Sclerosis Cases in Administrative Health Databases in Piedmont (Italy): An Application to the Estimate of Prevalence by Age and Urbanization Level. <i>Neuroepidemiology</i> , 2021, 55, 119-125.	2.3	5
144	A practical approach to HLA-DR genomic typing by heteroduplex analysis and a selective cleavage at position 86. <i>Human Immunology</i> , 1994, 40, 41-50.	2.4	4

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145	Nonparametric linkage analysis of alcohol dependence with chromosome 1 and 7 markers. <i>Genetic Epidemiology</i> , 1999, 17, S127-S131.	1.3	4
146	Mutations in the lamin B1 gene are not present in multiple sclerosis. <i>European Journal of Neurology</i> , 2009, 16, 544-546.	3.3	4
147	No evidence of association of the rare nsSNP rs35667974 in IFIH1 with multiple sclerosis. <i>Journal of Neuroimmunology</i> , 2010, 221, 112-114.	2.3	4
148	Coeliac disease mimicking Amyotrophic Lateral Sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2015, 16, 277-279.	1.7	4
149	Expanding the genetic spectrum of primary familial brain calcification due to SLC20A2 mutations: a case series. <i>Neurogenetics</i> , 2021, 22, 65-70.	1.4	4
150	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. <i>SSRN Electronic Journal</i> , 0, , .	0.4	4
151	An Investigation of the Role of Common and Rare Variants in a Large Italian Multiplex Family of Multiple Sclerosis Patients. <i>Genes</i> , 2021, 12, 1607.	2.4	4
152	QUANTITATIVE EXPRESSION OF HLA CLASS I MOLECULES IN ACUTE NON-LYMPHOBLASTIC LEUKAEMIA CELLS. <i>International Journal of Immunogenetics</i> , 1993, 20, 165-173.	1.2	3
153	Genomic and functional evaluation of TNFSF14 in multiple sclerosis susceptibility. <i>Journal of Genetics and Genomics</i> , 2021, 48, 497-507.	3.9	3
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163	The Impact of Life-Time Alcohol and Smoking Load on Multiple Sclerosis Severity. SSRN Electronic Journal, 0, , .	0.4	0
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