Hamish S Scott

List of Publications by Year
in descending order

275	19,015	10351 72	13727 129
papers	citations	h-index	g-index

282	282	282	22413
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Positional cloning of the APECED gene. Nature Genetics, 1997, 17, 393-398.	9.4	1,294
2	Use of within-array replicate spots for assessing differential expression in microarray experiments. Bioinformatics, 2005, 21, 2067-2075.	1.8	1,250
3	TWEAK, a New Secreted Ligand in the Tumor Necrosis Factor Family That Weakly Induces Apoptosis. Journal of Biological Chemistry, 1997, 272, 32401-32410.	1.6	603
4	A network-biology perspective of microRNA function and dysfunction in cancer. Nature Reviews Genetics, 2016, 17, 719-732.	7.7	579
5	Heritable GATA2 mutations associated with familial myelodysplastic syndrome and acute myeloid leukemia. Nature Genetics, 2011, 43, 1012-1017.	9.4	524
6	RANK signals from CD4+3â˜ inducer cells regulate development of Aire-expressing epithelial cells in the thymic medulla. Journal of Experimental Medicine, 2007, 204, 1267-1272.	4.2	434
7	Dodecamer repeat expansion in cystatin B gene in progressive myoclonus epilepsy. Nature, 1997, 386, 847-851.	13.7	366
8	In vitro analyses of known and novel RUNX1/AML1 mutations in dominant familial platelet disorder with predisposition to acute myelogenous leukemia: implications for mechanisms of pathogenesis. Blood, 2002, 99, 1364-1372.	0.6	348
9	Gene Dosageâ€“limiting Role of Aire in Thymic Expression, Clonal Deletion, and Organ-specific Autoimmunity. Journal of Experimental Medicine, 2004, 200, 1015-1026.	4.2	271
10	Isolation and Initial Characterization of a Novel Zinc Finger Gene, DNMT3L, on 21q22.3, Related to the Cytosine-5- Methyltransferase 3 Gene Family. Genomics, 2000, 65, 293-298.	1.3	270
11	Autoimmune Polyendocrine Syndrome Type 1 and NALP5, a Parathyroid Autoantigen. New England Journal of Medicine, 2008, 358, 1018-1028.	13.9	270
12	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. Nature Genetics, 2013, 45, 1226-1231.	9.4	270
13	Autoimmune Regulator Is Expressed in the Cells Regulating Immune Tolerance in Thymus Medulla. Biochemical and Biophysical Research Communications, 1999, 257, 821-825.	1.0	263
14	Meiotic and epigenetic defects in Dnmt3L-knockout mouse spermatogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 4068-4073.	3.3	261
15	Loss-of-function germline GATA2 mutations in patients with MDS/AML or MonoMAC syndrome and primary lymphedema reveal a key role for GATA2 in the lymphatic vasculature. Blood, 2012, 119, 1283-1291.	0.6	244
16	Autoantigen-Specific Interactions with CD4+ Thymocytes Control Mature Medullary Thymic Epithelial Cell Cellularity. Immunity, 2008, 29, 451-463.	6.6	219
17	A common mutation in Sardinian autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy patients. Human Genetics, 1998, 103, 428-434.	1.8	217
18	The Autoimmune Regulator Protein Has Transcriptional Transactivating Properties and Interacts with the Common Coactivator CREB-binding Protein. Journal of Biological Chemistry, 2000, 275, 16802-16809.	1.6	212

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19	Insertion of Î²-satellite repeats identifies a transmembrane protease causing both congenital and childhood onset autosomal recessive deafness. Nature Genetics, 2001, 27, 59-63.	9.4	208
20	Medullary thymic epithelial cells expressing Aire represent a unique lineage derived from cells expressing claudin. Nature Immunology, 2007, 8, 304-311.	7.0	199
21	Loss of LKB1 Kinase Activity in Peutz-Jeghers Syndrome, and Evidence for Allelic and Locus Heterogeneity. American Journal of Human Genetics, 1998, 63, 1641-1650.	2.6	194
22	Molecular genetics of muccpolysaccharidosis type I: Diagnostic, clinical, and biological implications. Human Mutation, 1995, 6, 288-302.	1.1	186
23	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. Blood, 2016, 127, 1017-1023.	0.6	179
24	GATA2 is required for lymphatic vessel valve development and maintenance. Journal of Clinical Investigation, 2015, 125, 2979-2994.	3.9	177
25	The Phenotypic Spectrum of GLI3 Morphopathies Includes Autosomal Dominant Preaxial Polydactyly Type-IV and Postaxial Polydactyly Type-A/B; No Phenotype Prediction from the Position of GLI3 Mutations. American Journal of Human Genetics, 1999, 65, 645-655.	2.6	175
26	Aire regulates the transfer of antigen from mTECs to dendritic cells for induction of thymic tolerance. Blood, 2011, 118, 2462-2472.	0.6	174
27	RNA and protein expression of the murine autoimmune regulator gene (Aire) in normal, RelB-deficient and in NOD mouse. European Journal of Immunology, 2000, 30, 1884-1893.	1.6	168
28	Modifiers of epigenetic reprogramming show paternal effects in the mouse. Nature Genetics, 2007, 39, 614-622.	9.4	154
29	The transmembrane serine protease (TMPRSS3) mutated in deafness DFNB8/10 activates the epithelial sodium channel (ENaC) in vitro. Human Molecular Genetics, 2002, 11, 2829-2836.	1.4	153
30	Cloning of the sulphamidase gene and identification of mutations in Sanfilippo A syndrome. Nature Genetics, 1995, 11, 465-467.	9.4	152
31	Integrative genomic analysis reveals cancer-associated mutations at diagnosis of CML in patients with high-risk disease. Blood, 2018, 132, 948-961.	0.6	152
32	Identification and Characterization of Two Putative Human Arginine Methyltransferases (HRMT1L1 and) Tj ETQq0 0 0 rgBT /Overlock 10	1.3	151
33	Common Mutations in Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy Patients of Different Origins. Molecular Endocrinology, 1998, 12, 1112-1119.	3.7	150
34	Human alpha-L-iduronidase: cDNA isolation and expression.. Proceedings of the National Academy of Sciences of the United States of America, 1991, 88, 9695-9699.	3.3	137
35	Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 1. Clinical Cancer Research, 2017, 23, e46-e53.	3.2	133
36	APECED mutations in the autoimmune regulator (AIRE) gene. Human Mutation, 2001, 18, 205-211.	1.1	124

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37	Post-Aire Maturation of Thymic Medullary Epithelial Cells Involves Selective Expression of Keratinocyte-Specific Autoantigens. Frontiers in Immunology, 2012, 3, 19.	2.2	123
38	Aire-Deficient C57BL/6 Mice Mimicking the Common Human 13-Base Pair Deletion Mutation Present with Only a Mild Autoimmune Phenotype. Journal of Immunology, 2009, 182, 3902-3918.	0.4	117
39	Integrative analysis of RUNX1 downstream pathways and target genes. BMC Genomics, 2008, 9, 363.	1.2	116
40	Mucopolysaccharidosis type I: identification of 8 novel mutations and determination of the frequency of the two common Î±-L-iduronidase mutations (W402X and Q70X) among European patients. Human Molecular Genetics, 1994, 3, 861-866.	1.4	111
41	NOMENCLATURE. Genomics, 1997, 45, 468-471.	1.3	109
42	Mutation analyses of North American APS-1 patients. , 1999, 13, 69-74.		109
43	Structure and sequence of the human Î±-l-iduronidase gene. Genomics, 1992, 13, 1311-1313.	1.3	108
44	RUNX1-mutated families show phenotype heterogeneity and a somatic mutation profile unique to germline predisposed AML. Blood Advances, 2020, 4, 1131-1144.	2.5	102
45	Mice trisomic for a bacterial artificial chromosome with the single-minded 2 gene (Sim2) show phenotypes similar to some of those present in the partial trisomy 16 mouse models of Down syndrome. Human Molecular Genetics, 2000, 9, 1853-1864.	1.4	99
46	Interferon autoantibodies associated with AIRE deficiency decrease the expression of IFN-stimulated genes. Blood, 2008, 112, 2657-2666.	0.6	98
47	Molecular cloning and characterization of a novel gene family of four ancient conserved domain proteins (ACDP). Gene, 2003, 306, 37-44.	1.0	97
48	Genomic subtyping and therapeutic targeting of acute erythroleukemia. Nature Genetics, 2019, 51, 694-704.	9.4	97
49	The Lymphotoxin Pathway Regulates Aire-Independent Expression of Ectopic Genes and Chemokines in Thymic Stromal Cells. Journal of Immunology, 2008, 180, 5384-5392.	0.4	96
50	ARMC5Mutations Are Common in Familial Bilateral Macronodular Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1784-E1792.	1.8	96
51	HENMT1 and piRNA Stability Are Required for Adult Male Germ Cell Transposon Repression and to Define the Spermatogenic Program in the Mouse. PLoS Genetics, 2015, 11, e1005620.	1.5	95
52	APECED: a monogenic autoimmune disease providing new clues to self-tolerance. Trends in Immunology, 1998, 19, 384-386.	7.5	93
53	Two Isoforms of a Human Intersectin (ITSN) Protein Are Produced by Brain-Specific Alternative Splicing in a Stop Codon. Genomics, 1998, 53, 369-376.	1.3	93
54	Statistical modeling of sequencing errors in SAGE libraries. Bioinformatics, 2004, 20, i31-i39.	1.8	93

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55	Recommendations for Childhood Cancer Screening and Surveillance in DNA Repair Disorders. Clinical Cancer Research, 2017, 23, e23-e31.	3.2	93
56	Modulation of Aire regulates the expression of tissue-restricted antigens. Molecular Immunology, 2008, 45, 25-33.	1.0	92
57	A Specific Anti-Aire Antibody Reveals Aire Expression Is Restricted to Medullary Thymic Epithelial Cells and Not Expressed in Periphery. Journal of Immunology, 2008, 180, 3824-3832.	0.4	92
58	Autoimmune Regulator Deficiency Results in Decreased Expression of CCR4 and CCR7 Ligands and in Delayed Migration of CD4+ Thymocytes. Journal of Immunology, 2009, 183, 7682-7691.	0.4	90
59	Knobloch syndrome: Novel mutations inCOL18A1, evidence for genetic heterogeneity, and a functionally impaired polymorphism in endostatin. Human Mutation, 2004, 23, 77-84.	1.1	89
60	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndromeâ€“like phenotype and hyperactivated MAPK signaling in humans and mice. Journal of Clinical Investigation, 2011, 121, 3479-3491.	3.9	89
61	Redefining epithelial progenitor potential in the developing thymus. European Journal of Immunology, 2007, 37, 2411-2418.	1.6	86
62	Sensitive Detection of BCR-ABL1 Mutations in Patients With Chronic Myeloid Leukemia After Imatinib Resistance Is Predictive of Outcome During Subsequent Therapy. Journal of Clinical Oncology, 2011, 29, 4250-4259.	0.8	86
63	A comparative analysis of algorithms for somatic SNV detection in cancer. Bioinformatics, 2013, 29, 2223-2230.	1.8	86
64	Novel RUNX1 mutations in familial platelet disorder with enhanced risk for acute myeloid leukemia: clues for improved identification of the FPD/AML syndrome. Leukemia, 2010, 24, 242-246.	3.3	85
65	Expression of autoimmune regulator gene (AIRE) and T regulatory cells in human thymomas. Clinical and Experimental Immunology, 2007, 149, 504-512.	1.1	83
66	Ablation and Regeneration of Tolerance-Inducing Medullary Thymic Epithelial Cells after Cyclosporine, Cyclophosphamide, and Dexamethasone Treatment. Journal of Immunology, 2009, 183, 823-831.	0.4	83
67	The Mouse Brain Transcriptome by SAGE: Differences in Gene Expression between P30 Brains of the Partial Trisomy 16 Mouse Model of Down Syndrome (Ts65Dn) and Normals. Genome Research, 2000, 10, 2006-2021.	2.4	81
68	AIRE's CARD Revealed, a New Structure for Central Tolerance Provokes Transcriptional Plasticity. Journal of Biological Chemistry, 2008, 283, 1723-1731.	1.6	80
69	Recommendations for Surveillance for Children with Leukemia-Predisposing Conditions. Clinical Cancer Research, 2017, 23, e14-e22.	3.2	80
70	Secondary leukemia in patients with germline transcription factor mutations (RUNX1, GATA2, CEBPA). Blood, 2020, 136, 24-35.	0.6	79
71	Î±-L-iduronidase mutations (Q70X and P533R) associate with a severe Hurler phenotype. Human Mutation, 1992, 1, 333-339.	1.1	78
72	Myeloid neoplasms with germline DDX41 mutation. International Journal of Hematology, 2017, 106, 163-174.	0.7	77

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73	Novel missense mutations of TMPRSS3 in two consanguineous Tunisian families with non-syndromic autosomal recessive deafness. Human Mutation, 2001, 18, 101-108.	1.1	76
74	Two novel JAK2 exon 12 mutations in JAK2V617F-negative polycythaemia vera patients. Leukemia, 2008, 22, 870-873.	3.3	76
75	Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 2 and Related Disorders. Clinical Cancer Research, 2017, 23, e54-e61.	3.2	76
76	Expression of Aire and the Early Wave of Apoptosis in Spermatogenesis. Journal of Immunology, 2008, 180, 1338-1343.	0.4	75
77	Cloning and expression of the gene involved in Sanfilippo B syndrome (mucopolysaccharidosis III B). Human Molecular Genetics, 1996, 5, 771-777.	1.4	74
78	Sequential phases in the development of Aireâ€expressing medullary thymic epithelial cells involve distinct cellular input. European Journal of Immunology, 2008, 38, 942-947.	1.6	74
79	A common mutation for mucopolysaccharidosis type I associated with a severe Hurler syndrome phenotype. Human Mutation, 1992, 1, 103-108.	1.1	73
80	Tmprss3, a Transmembrane Serine Protease Deficient in Human DFNB8/10 Deafness, Is Critical for Cochlear Hair Cell Survival at the Onset of Hearing. Journal of Biological Chemistry, 2011, 286, 17383-17397.	1.6	71
81	An integrated genetic and functional analysis of the role of type II transmembrane serine proteases (TMPRSSs) in hearing loss. Human Mutation, 2008, 29, 130-141.	1.1	70
82	A testis-specific gene, TPTE , encodes a putative transmembrane tyrosine phosphatase and maps to the pericentromeric region of human chromosomes 21 and 13, and to chromosomes 15, 22, and Y. Human Genetics, 1999, 105, 399-409.	1.8	69
83	Poor response to second-line kinase inhibitors in chronic myeloid leukemia patients with multiple low-level mutations, irrespective of their resistance profile. Blood, 2012, 119, 2234-2238.	0.6	69
84	Long-term clinical progress in bone marrow transplanted mucopolysaccharidosis type I patients with a defined genotype. Journal of Inherited Metabolic Disease, 1993, 16, 1024-1033.	1.7	67
85	Decreased phosphatidylethanolamine binding protein expression correlates with AÎ² accumulation in the Tg2576 mouse model of Alzheimer's disease. Neurobiology of Aging, 2006, 27, 614-623.	1.5	67
86	Cloning of Two Human Homologs of the Drosophila single-minded Gene SIM1 on Chromosome 6q and SIM2 on 21q Within the Down Syndrome Chromosomalâ€‰Region. Genome Research, 1997, 7, 615-624.	2.4	66
87	Mice Deficient for the Type II Transmembrane Serine Protease, TMPRSS1/hepsin, Exhibit Profound Hearing Loss. American Journal of Pathology, 2007, 171, 608-616.	1.9	66
88	Short-term inhibition of p53 combined with keratinocyte growth factor improves thymic epithelial cell recovery and enhances T-cell reconstitution after murine bone marrow transplantation. Blood, 2010, 115, 1088-1097.	0.6	66
89	Mutations in the TMPRSS3 gene are a rare cause of childhood nonsyndromic deafness in Caucasian patients. Journal of Molecular Medicine, 2002, 80, 124-131.	1.7	65
90	Identification and characterization of a novel cyclic nucleotide phosphodiesterase gene (PDE9A) that maps to 21q22.3: alternative splicing of mRNA transcripts, genomic structure and sequence. Human Genetics, 1998, 103, 386-392.	1.8	64

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91	A PCR Amplification Method Reveals Instability of the Dodecamer Repeat in Progressive Myoclonus Epilepsy (EPM1) and No Correlation between the Size of the Repeat and Age at Onset. American Journal of Human Genetics, 1998, 62, 842-847.	2.6	62
92	Spliceosome mutations in hematopoietic malignancies. Nature Genetics, 2012, 44, 9-10.	9.4	61
93	Functional transcriptome analysis of the postnatal brain of the Ts1Cje mouse model for Down syndrome reveals global disruption of interferon-related molecular networks. BMC Genomics, 2014, 15, 624.	1.2	61
94	Brief Report: Identification of a Pathogenic Variant in TREX1 in Earlyâ€Onset Cerebral Systemic Lupus Erythematosus by Wholeâ€Exome Sequencing. Arthritis and Rheumatology, 2014, 66, 3382-3386.	2.9	61
95	Molecular Defects in Sanfilippo Syndrome Type A. Human Molecular Genetics, 1997, 6, 787-791.	1.4	60
96	Isolation and Characterization of the MouseAireGene. Biochemical and Biophysical Research Communications, 1999, 255, 483-490.	1.0	60
97	Proteomic and Metabolomic Analyses of Mitochondrial Complex I-deficient Mouse Model Generated by Spontaneous B2 Short Interspersed Nuclear Element (SINE) Insertion into NADH Dehydrogenase (Ubiquinone) Fe-S Protein 4 (Ndufs4) Gene. Journal of Biological Chemistry, 2012, 287, 20652-20663.	1.6	58
98	Hematopoietic defects in the Ts1Cje mouse model of Down syndrome. Blood, 2009, 113, 1929-1937.	0.6	56
99	Novel mutations of TMPRSS3 in four DFNB8/B10 families segregating congenital autosomal recessive deafness. Journal of Medical Genetics, 2001, 38, 396-400.	1.5	55
100	Cloning of a Human RNA Editing Deaminase (ADARB1) of Glutamate Receptors That Maps to Chromosome 21q22.3. Genomics, 1997, 41, 210-217.	1.3	54
101	A Serial Analysis of Gene Expression Profile of the Alzheimerâ€™s Disease Tg2576 Mouse Model. Neurotoxicity Research, 2010, 17, 360-379.	1.3	54
102	Deep sequencing analysis of the developing mouse brain reveals a novel microRNA. BMC Genomics, 2011, 12, 176.	1.2	53
103	Revealing Missing Human Protein Isoforms Based on Ab Initio Prediction, RNA-seq and Proteomics. Scientific Reports, 2015, 5, 10940.	1.6	51
104	Nedd4-WW Domain-Binding Protein 5 (Ndfip1) Is Associated with Neuronal Survival after Acute Cortical Brain Injury. Journal of Neuroscience, 2006, 26, 7234-7244.	1.7	49
105	GATA2 monoallelic expression underlies reduced penetrance in inherited GATA2-mutated MDS/AML. Leukemia, 2018, 32, 2502-2507.	3.3	48
106	Axonemal Beta Heavy Chain Dynein DNAH9: cDNA Sequence, Genomic Structure, and Investigation of Its Role in Primary Ciliary Dyskinesia. Genomics, 2001, 72, 21-33.	1.3	47
107	Isolation and characterization of the UBASH3A gene on 21q22.3 encoding a potential nuclear protein with a novel combination of domains. Human Genetics, 2001, 108, 140-147.	1.8	47
108	Vinclozolin Exposure in Utero Induces Postpubertal Prostatitis and Reduces Sperm Production via a Reversible Hormone-Regulated Mechanism. Endocrinology, 2010, 151, 783-792.	1.4	46

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109	A cSNP Map and Database for Human Chromosome 21. Genome Research, 2001, 11, 300-307.	2.4	46
110	Structure and Sequence of the Human Sulphamidase Gene. DNA Research, 1996, 3, 269-271.	1.5	45
111	Isolation and Characterization of a Human Chromosome 21q22.3 Gene (WDR4) and Its Mouse Homologue That Code for a WD-Repeat Protein. Genomics, 2000, 68, 71-79.	1.3	45
112	Gene Network Disruptions and Neurogenesis Defects in the Adult Ts1Cje Mouse Model of Down Syndrome. PLoS ONE, 2010, 5, e11561.	1.1	44
113	Differential effects on gene transcription and hematopoietic differentiation correlate with GATA2 mutant disease phenotypes. Leukemia, 2018, 32, 194-202.	3.3	44
114	Altered Spacing of Promoter Elements Due to the Dodecamer Repeat Expansion Contributes to Reduced Expression of the Cystatin B Gene in EPM1. Human Molecular Genetics, 1999, 8, 1791-1798.	1.4	43
115	Splice factor mutations and alternative splicing as drivers of hematopoietic malignancy. Immunological Reviews, 2015, 263, 257-278.	2.8	43
116	The mutational burden of therapy-related myeloid neoplasms is similar to primary myelodysplastic syndrome but has a distinctive distribution. Leukemia, 2019, 33, 2842-2853.	3.3	43
117	Mutation analysis of 19 North American mucopolysaccharidosis type I patients: Identification of two additional frequent mutations. Human Mutation, 1994, 3, 275-282.	1.1	42
118	Familial vasopressinâ€sensitive ACTHâ€independent macronodular adrenal hyperplasia (VPsâ€AIMAH): clinical studies of three kindreds. Clinical Endocrinology, 2009, 70, 883-891.	1.2	41
119	Increased ILâ€17A secretion in response to Candida albicans in autoimmune polyendocrine syndrome type 1 and its animal model. European Journal of Immunology, 2011, 41, 235-245.	1.6	41
120	AML1 Interconnected Pathways of Leukemogenesis. Cancer Investigation, 2003, 21, 105-136.	0.6	39
121	TMPRSS3, a type II transmembrane serine protease mutated in non-syndromic autosomal recessive deafness. Frontiers in Bioscience - Landmark, 2008, 13, 1557.	3.0	39
122	The susceptibility of Aireâ^'/â^' mice to experimental myasthenia gravis involves alterations in regulatory T cells. Journal of Autoimmunity, 2011, 36, 16-24.	3.0	38
123	Genetic regulators of myelopoiesis and leukemic signaling identified by gene profiling and linear modeling. Journal of Leukocyte Biology, 2006, 80, 433-447.	1.5	37
124	MicroSAGE is highly representative and reproducible but reveals major differences in gene expression among samples obtained from similar tissues. Genome Biology, 2003, 4, R17.	13.9	36
125	A four-gene LincRNA expression signature predicts risk in multiple cohorts of acute myeloid leukemia patients. Leukemia, 2018, 32, 263-272.	3.3	36
126	Thymic Deletion and Regulatory T Cells Prevent Antimyeloperoxidase GN. Journal of the American Society of Nephrology: JASN, 2013, 24, 573-585.	3.0	35

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127	A case of Aromatase deficiency due to a novel CYP19A1 mutation. BMC Endocrine Disorders, 2014, 14, 16.	0.9	35
128	Mutations among Italian mucopolysaccharidosis type I patients. Journal of Inherited Metabolic Disease, 1997, 20, 803-806.	1.7	34
129	Education and promiscuity. Nature, 2002, 420, 468-469.	13.7	34
130	Autoimmune regulator controls T cell help for pathogenetic autoantibody production in collagenâ€induced arthritis. Arthritis and Rheumatism, 2009, 60, 1683-1693.	6.7	34
131	Molecular networks involved in mouse cerebral corticogenesis and spatio-temporal regulation of Sox4 and Sox11 novel antisense transcripts revealed by transcriptome profiling. Genome Biology, 2009, 10, R104.	13.9	34
132	Differential gene expression studies to explore the molecular pathophysiology of Down syndrome. Brain Research Reviews, 2001, 36, 265-274.	9.1	33
133	Spatiotemporal Regulation of Multiple Overlapping Sense and Novel Natural Antisense Transcripts at the Nrgn and Camk2n1 Gene Loci during Mouse Cerebral Corticogenesis. Cerebral Cortex, 2011, 21, 683-697.	1.6	33
134	Germline variants in familial pituitary tumour syndrome genes are common in young patients and families with additional endocrine tumours. European Journal of Endocrinology, 2017, 176, 635-644.	1.9	33
135	Huntington disease-linked locusD4S111 exposed as the ?-l-iduronidase gene. Somatic Cell and Molecular Genetics, 1991, 17, 421-425.	0.7	32
136	A tale of two siblings: two cases of AML arising from a single pre-leukemic DNMT3A mutant clone. Leukemia, 2015, 29, 2101-2104.	3.3	32
137	Autoimmune hepatitis in a murine autoimmune polyendocrine syndrome type 1 model is directed against multiple autoantigens. Hepatology, 2015, 61, 1295-1305.	3.6	32
138	Targeted gene panels identify a high frequency of pathogenic germline variants in patients diagnosed with a hematological malignancy and at least one other independent cancer. Leukemia, 2021, 35, 3245-3256.	3.3	32
139	Identification of a novel member of the CLIC family, CLIC6, mapping to 21q22.12. Gene, 2003, 320, 31-40.	1.0	31
140	Many BCR-ABL1 compound mutations reported in chronic myeloid leukemia patients may actually be artifacts due to PCR-mediated recombination. Blood, 2014, 124, 153-155.	0.6	31
141	Cloning and Characterization of a Putative Human Glycerol 3-Phosphate Permease Gene (SLC37A1 or) Tj ETQq1 1 0.784314 rgBT /Ov Deficiency. Genomics, 2000, 70, 190-200.	1.3	30
142	DNA methylation signatures of the AIRE promoter in thymic epithelial cells, thymomas and normal tissues. Molecular Immunology, 2011, 49, 518-526.	1.0	30
143	Ectrodactyly and Lethal Pulmonary Acinar Dysplasia Associated with HomozygousFGFR2Mutations Identified by Exome Sequencing. Human Mutation, 2016, 37, 955-963.	1.1	30
144	GATA2 deficiency syndrome: A decade of discovery. Human Mutation, 2021, 42, 1399-1421.	1.1	30

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145	Morquio A Syndrome: Cloning, Sequence, and Structure of the Human N-Acetylgalactosamine 6-Sulfatase (GALNS) Gene. Genomics, 1994, 22, 652-654.	1.3	29
146	Refined Localization of Autosomal Recessive Nonsyndromic Deafness DFNB10 Locus Using 34 Novel Microsatellite Markers, Genomic Structure, and Exclusion of Six Known Genes in the Region. Genomics, 2000, 68, 22-29.	1.3	29
147	Reduced Thymic Aire Expression and Abnormal NF-Î²B2 Signaling in a Model of Systemic Autoimmunity. Journal of Immunology, 2009, 182, 2690-2699.	0.4	29
148	The RUNX1 database (RUNX1db): establishment of an expert curated RUNX1 registry and genomics database as a public resource for familial platelet disorder with myeloid malignancy. Haematologica, 2021, 106, 3004-3007.	1.7	29
149	Tmprss3 loss of function impairs cochlear inner hair cell Kcnma1 channel membrane expression. Human Molecular Genetics, 2013, 22, 1289-1299.	1.4	28
150	Aberrant RAG-mediated recombination contributes to multiple structural rearrangements in lymphoid blast crisis of chronic myeloid leukemia. Leukemia, 2020, 34, 2051-2063.	3.3	27
151	Self-reverting mutations partially correct the blood phenotype in a Diamond Blackfan anemia patient. Haematologica, 2017, 102, e506-e509.	1.7	26
152	Characterization of a Novel Gene, C21orf2, on Human Chromosome 21q22.3 and Its Exclusion as the APECED Gene by Mutation Analysis. Genomics, 1998, 47, 64-70.	1.3	25
153	C21orf5, a Novel Human Chromosome 21 Gene, Has a Caenorhabditis elegans Ortholog (pad-1) Required for Embryonic Patterning. Genomics, 2000, 68, 30-40.	1.3	25
154	PCR of a VNTR linked to mucopolysaccharidosis type I and Huntington disease. Nucleic Acids Research, 1991, 19, 6348-6348.	6.5	24
155	Isolation of a human gene (HES1) with homology to an Escherichia coli and a zebrafish protein that maps to chromosome 21q22.3. Human Genetics, 1997, 99, 616-623.	1.8	24
156	CBG Santiago: A Novel CBG Mutation. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E151-E155.	1.8	24
157	Integrating Massively Parallel Sequencing into Diagnostic Workflows and Managing the Annotation and Clinical Interpretation Challenge. Human Mutation, 2014, 35, 413-423.	1.1	23
158	Linearization and purification of BAC DNA for the development of transgenic mice. Transgenic Research, 1999, 8, 147-150.	1.3	22
159	Estimating the proportion of microarray probes expressed in an RNA sample. Nucleic Acids Research, 2010, 38, 2168-2176.	6.5	21
160	Mucopolysaccharidosis type I (Hurler syndrome): Linkage disequilibrium indicates the presence of a major allele. Human Genetics, 1992, 88, 701-702.	1.8	20
161	The epilepsy, the protease inhibitor and the dodecamer: progressive myoclonus epilepsy, cystatin b and a 12-mer repeat expansion. Cytogenetic and Genome Research, 2003, 100, 213-223.	0.6	20
162	DNMT3L Is a Regulator of X Chromosome Compaction and Post-Meiotic Gene Transcription. PLoS ONE, 2011, 6, e18276.	1.1	20

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