Hamish S Scott

List of Publications by Citations

261 papers	15,569 citations	67 h-index	118 g-index
282 ext. papers	17,534 ext. citations	7.2 avg, IF	5.97 L-index

#	Paper	IF	Citations
261	Positional cloning of the APECED gene. *Nature Genetics*, **1997**, 17, 393-8	36.3	1105
260	Use of within-array replicate spots for assessing differential expression in microarray experiments. *Bioinformatics*, **2005**, 21, 2067-75	7.2	1079
259	TWEAK, a new secreted ligand in the tumor necrosis factor family that weakly induces apoptosis. *Journal of Biological Chemistry*, **1997**, 272, 32401-10	5.4	522
258	A network-biology perspective of microRNA function and dysfunction in cancer. *Nature Reviews Genetics*, **2016**, 17, 719-732	30.1	440
257	Heritable GATA2 mutations associated with familial myelodysplastic syndrome and acute myeloid leukemia. *Nature Genetics*, **2011**, 43, 1012-7	36.3	424
256	RANK signals from CD4(+)3(-) inducer cells regulate development of Aire-expressing epithelial cells in the thymic medulla. *Journal of Experimental Medicine*, **2007**, 204, 1267-72	16.6	378
255	Dodecamer repeat expansion in cystatin B gene in progressive myoclonus epilepsy. *Nature*, **1997**, 386, 847-51	50.4	316
254	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, 1364-72	2.2	305
253	Gene dosage--limiting role of Aire in thymic expression, clonal deletion, and organ-specific autoimmunity. *Journal of Experimental Medicine*, **2004**, 200, 1015-26	16.6	254
252	Meiotic and epigenetic defects in Dnmt3L-knockout mouse spermatogenesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 4068-73	11.5	233
251	Autoimmune regulator is expressed in the cells regulating immune tolerance in thymus medulla. *Biochemical and Biophysical Research Communications*, **1999**, 257, 821-5	3.4	229
250	Autoimmune polyendocrine syndrome type 1 and NALP5, a parathyroid autoantigen. *New England Journal of Medicine*, **2008**, 358, 1018-28	59.2	225
249	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, 293-8	4.3	221
248	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, 1283-91	2.2	216
247	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. *Nature Genetics*, **2013**, 45, 1226-1231	36.3	205
246	A common mutation in Sardinian autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy patients. *Human Genetics*, **1998**, 103, 428-34	6.3	192
245	Autoantigen-specific interactions with CD4+ thymocytes control mature medullary thymic epithelial cell cellularity. *Immunity*, **2008**, 29, 451-63	32.3	190

244	The autoimmune regulator protein has transcriptional transactivating properties and interacts with the common coactivator CREB-binding protein. *Journal of Biological Chemistry*, **2000**, 275, 16802-9	5.4	168
243	Loss of LKB1 kinase activity in Peutz-Jeghers syndrome, and evidence for allelic and locus heterogeneity. *American Journal of Human Genetics*, **1998**, 63, 1641-50	11	166
242	Medullary thymic epithelial cells expressing Aire represent a unique lineage derived from cells expressing claudin. *Nature Immunology*, **2007**, 8, 304-11	19.1	165
241	Molecular genetics of mucopolysaccharidosis type I: diagnostic, clinical, and biological implications. *Human Mutation*, **1995**, 6, 288-302	4.7	162
240	Insertion of beta-satellite repeats identifies a transmembrane protease causing both congenital and childhood onset autosomal recessive deafness. *Nature Genetics*, **2001**, 27, 59-63	36.3	159
239	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, 1884-93	6.1	155
238	Aire regulates the transfer of antigen from mTECs to dendritic cells for induction of thymic tolerance. *Blood*, **2011**, 118, 2462-72	2.2	153
237	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, 645-55	11	149
236	Cloning of the sulphamidase gene and identification of mutations in Sanfilippo A syndrome. *Nature Genetics*, **1995**, 11, 465-7	36.3	141
235	Modifiers of epigenetic reprogramming show paternal effects in the mouse. *Nature Genetics*, **2007**, 39, 614-22	36.3	140
234	Identification and characterization of two putative human arginine methyltransferases (HRMT1L1 and HRMT1L2). *Genomics*, **1998**, 48, 330-40	4.3	138
233	GATA2 is required for lymphatic vessel valve development and maintenance. *Journal of Clinical Investigation*, **2015**, 125, 2979-94	15.9	136
232	Common mutations in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy patients of different origins. *Molecular Endocrinology*, **1998**, 12, 1112-9		133
231	Human alpha-L-iduronidase: cDNA isolation and expression. *Proceedings of the National Academy of Sciences of the United States of America*, **1991**, 88, 9695-9	11.5	131
230	The transmembrane serine protease (TMPRSS3) mutated in deafness DFNB8/10 activates the epithelial sodium channel (ENaC) in vitro. *Human Molecular Genetics*, **2002**, 11, 2829-36	5.6	124
229	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. *Blood*, **2016**, 127, 1017-23	2.2	117
228	APECED mutations in the autoimmune regulator (AIRE) gene. *Human Mutation*, **2001**, 18, 205-11	4.7	106
227	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, 3902-18	5.3	103

226	Structure and sequence of the human alpha-L-iduronidase gene. *Genomics*, **1992**, 13, 1311-3	4.3	100
225	Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 1. *Clinical Cancer Research*, **2017**, 23, e46-e53	12.9	95
224	Integrative analysis of RUNX1 downstream pathways and target genes. *BMC Genomics*, **2008**, 9, 363	4.5	93
223	Mucopolysaccharidosis type I: identification of 8 novel mutations and determination of the frequency of the two common alpha-L-iduronidase mutations (W402X and Q70X) among European patients. *Human Molecular Genetics*, **1994**, 3, 861-6	5.6	93
222	Guidelines for human gene nomenclature (1997). HUGO Nomenclature Committee. *Genomics*, **1997**, 45, 468-71	4.3	91
221	Mutation analyses of North American APS-1 patients. *Human Mutation*, **1999**, 13, 69-74	4.7	90
220	The lymphotoxin pathway regulates Aire-independent expression of ectopic genes and chemokines in thymic stromal cells. *Journal of Immunology*, **2008**, 180, 5384-92	5.3	87
219	Two isoforms of a human intersectin (ITSN) protein are produced by brain-specific alternative splicing in a stop codon. *Genomics*, **1998**, 53, 369-76	4.3	86
218	APECED: a monogenic autoimmune disease providing new clues to self-tolerance. *Trends in Immunology*, **1998**, 19, 384-6		83
217	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, 3824-32	5.3	83
216	Integrative genomic analysis reveals cancer-associated mutations at diagnosis of CML in patients with high-risk disease. *Blood*, **2018**, 132, 948-961	2.2	80
215	Knobloch syndrome: novel mutations in COL18A1, evidence for genetic heterogeneity, and a functionally impaired polymorphism in endostatin. *Human Mutation*, **2004**, 23, 77-84	4.7	79
214	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, 1853-64	5.6	79
213	Modulation of Aire regulates the expression of tissue-restricted antigens. *Molecular Immunology*, **2008**, 45, 25-33	4.3	78
212	Molecular cloning and characterization of a novel gene family of four ancient conserved domain proteins (ACDP). *Gene*, **2003**, 306, 37-44	3.8	78
211	OR34-6 A Novel Mechanism of SDH-Deficient Tumorigenesis and Implications for Genetic Testing in Patients with Pheochromocytoma-Paraganglioma. *Journal of the Endocrine Society*, **2019**, 3,	0.4	78
210	Interferon autoantibodies associated with AIRE deficiency decrease the expression of IFN-stimulated genes. *Blood*, **2008**, 112, 2657-66	2.2	75
209	Statistical modeling of sequencing errors in SAGE libraries. *Bioinformatics*, **2004**, 20 Suppl 1, i31-9	7.2	75

208	Redefining epithelial progenitor potential in the developing thymus. *European Journal of Immunology*, **2007**, 37, 2411-8	6.1	74
207	ARMC5 mutations are common in familial bilateral macronodular adrenal hyperplasia. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E1784-92	5.6	73
206	Post-Aire maturation of thymic medullary epithelial cells involves selective expression of keratinocyte-specific autoantigens. *Frontiers in Immunology*, **2012**, 3, 19	8.4	73
205	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, 242-6	10.7	73
204	A comparative analysis of algorithms for somatic SNV detection in cancer. *Bioinformatics*, **2013**, 29, 2223-30	7.3	71
203	alpha-L-iduronidase mutations (Q70X and P533R) associate with a severe Hurler phenotype. *Human Mutation*, **1992**, 1, 333-9	4.7	71
202	Sequential phases in the development of Aire-expressing medullary thymic epithelial cells involve distinct cellular input. *European Journal of Immunology*, **2008**, 38, 942-7	6.1	69
201	Autoimmune regulator deficiency results in decreased expression of CCR4 and CCR7 ligands and in delayed migration of CD4+ thymocytes. *Journal of Immunology*, **2009**, 183, 7682-91	5.3	68
200	AIRE's CARD revealed, a new structure for central tolerance provokes transcriptional plasticity. *Journal of Biological Chemistry*, **2008**, 283, 1723-1731	5.4	68
199	Cloning and expression of the gene involved in Sanfilippo B syndrome (mucopolysaccharidosis III B). *Human Molecular Genetics*, **1996**, 5, 771-7	5.6	68
198	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndrome-like phenotype and hyperactivated MAPK signaling in humans and mice. *Journal of Clinical Investigation*, **2011**, 121, 3479-91	15.9	68
197	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, 2006-21	9.7	68
196	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, 4250-9	2.2	67
195	Novel missense mutations of TMPRSS3 in two consanguineous Tunisian families with non-syndromic autosomal recessive deafness. *Human Mutation*, **2001**, 18, 101-8	4.7	67
194	Expression of Aire and the early wave of apoptosis in spermatogenesis. *Journal of Immunology*, **2008**, 180, 1338-43	5.3	66
193	Expression of autoimmune regulator gene (AIRE) and T regulatory cells in human thymomas. *Clinical and Experimental Immunology*, **2007**, 149, 504-12	6.2	66
192	A common mutation for mucopolysaccharidosis type I associated with a severe Hurler syndrome phenotype. *Human Mutation*, **1992**, 1, 103-8	4.7	64
191	Ablation and regeneration of tolerance-inducing medullary thymic epithelial cells after cyclosporine, cyclophosphamide, and dexamethasone treatment. *Journal of Immunology*, **2009**, 183, 823-31	5.3	63

190	Two novel JAK2 exon 12 mutations in JAK2V617F-negative polycythaemia vera patients. *Leukemia*, **2008**, 22, 870-3	10.7	63
189	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	6	62
188	Decreased phosphatidylethanolamine binding protein expression correlates with Abeta accumulation in the Tg2576 mouse model of Alzheimer's disease. *Neurobiology of Aging*, **2006**, 27, 614-23	5.6	60
187	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, 386-92	6.3	60
186	Long-term clinical progress in bone marrow transplanted mucopolysaccharidosis type I patients with a defined genotype. *Journal of Inherited Metabolic Disease*, **1993**, 16, 1024-33	5.4	60
185	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	6.3	58
184	Cloning of two human homologs of the Drosophila single-minded gene SIM1 on chromosome 6q and SIM2 on 21q within the Down syndrome chromosomal region. *Genome Research*, **1997**, 7, 615-24	9.7	57
183	Recommendations for Childhood Cancer Screening and Surveillance in DNA Repair Disorders. *Clinical Cancer Research*, **2017**, 23, e23-e31	12.9	56
182	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, 842-7	11	56
181	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, 2234-8	2.2	55
180	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, 1088-97	2.2	55
179	An integrated genetic and functional analysis of the role of type II transmembrane serine proteases (TMPRSSs) in hearing loss. *Human Mutation*, **2008**, 29, 130-41	4.7	55
178	Genomic subtyping and therapeutic targeting of acute erythroleukemia. *Nature Genetics*, **2019**, 51, 694-704	30.5	54
177	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, 17383-97	5.4	54
176	Molecular defects in Sanfilippo syndrome type A. *Human Molecular Genetics*, **1997**, 6, 787-91	5.6	54
175	Isolation and characterization of the mouse Aire gene. *Biochemical and Biophysical Research Communications*, **1999**, 255, 483-90	3.4	54
174	Mice deficient for the type II transmembrane serine protease, TMPRSS1/hepsin, exhibit profound hearing loss. *American Journal of Pathology*, **2007**, 171, 608-16	5.8	53
173	Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 2 and Related Disorders. *Clinical Cancer Research*, **2017**, 23, e54-e61	12.9	52

172	Recommendations for Surveillance for Children with Leukemia-Predisposing Conditions. *Clinical Cancer Research*, **2017**, 23, e14-e22	12.9	52
171	Cloning of a human RNA editing deaminase (ADARB1) of glutamate receptors that maps to chromosome 21q22.3. *Genomics*, **1997**, 41, 210-7	4.3	50
170	Mutations in the TMPRSS3 gene are a rare cause of childhood nonsyndromic deafness in Caucasian patients. *Journal of Molecular Medicine*, **2002**, 80, 124-31	5.5	50
169	Identification of a pathogenic variant in TREX1 in early-onset cerebral systemic lupus erythematosus by Whole-exome sequencing. *Arthritis and Rheumatology*, **2014**, 66, 3382-6	9.5	48
168	Deep sequencing analysis of the developing mouse brain reveals a novel microRNA. *BMC Genomics*, **2011**, 12, 176	4.5	46
167	Spliceosome mutations in hematopoietic malignancies. *Nature Genetics*, **2011**, 44, 9-10	36.3	46
166	Myeloid neoplasms with germline DDX41 mutation. *International Journal of Hematology*, **2017**, 106, 163-174	2.3	44
165	Hematopoietic defects in the Ts1Cje mouse model of Down syndrome. *Blood*, **2009**, 113, 1929-37	2.2	44
164	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	4.5	43
163	A serial analysis of gene expression profile of the Alzheimer's disease Tg2576 mouse model. *Neurotoxicity Research*, **2010**, 17, 360-79	4.3	43
162	Vinclozolin exposure in utero induces postpubertal prostatitis and reduces sperm production via a reversible hormone-regulated mechanism. *Endocrinology*, **2010**, 151, 783-92	4.8	42
161	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, 20652-63	5.4	42
160	Structure and sequence of the human sulphamidase gene. *DNA Research*, **1996**, 3, 269-71	4.5	42
159	Nedd4-WW domain-binding protein 5 (Ndfip1) is associated with neuronal survival after acute cortical brain injury. *Journal of Neuroscience*, **2006**, 26, 7234-44	6.6	41
158	Axonemal beta heavy chain dynein DNAH9: cDNA sequence, genomic structure, and investigation of its role in primary ciliary dyskinesia. *Genomics*, **2001**, 72, 21-33	4.3	41
157	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, 1791-8	5.6	40
156	Mutation analysis of 19 North American mucopolysaccharidosis type I patients: identification of two additional frequent mutations. *Human Mutation*, **1994**, 3, 275-82	4.7	40
155	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, 140-7	6.3	39

154	Novel mutations of TMPRSS3 in four DFNB8/B10 families segregating congenital autosomal recessive deafness. *Journal of Medical Genetics*, **2001**, 38, 396-400	5.8	39
153	Revealing Missing Human Protein Isoforms Based on Ab Initio Prediction, RNA-seq and Proteomics. *Scientific Reports*, **2015**, 5, 10940	4.9	38
152	RUNX1-mutated families show phenotype heterogeneity and a somatic mutation profile unique to germline predisposed AML. *Blood Advances*, **2020**, 4, 1131-1144	7.8	37
151	Increased IL-17A secretion in response to Candida albicans in autoimmune polyendocrine syndrome type 1 and its animal model. *European Journal of Immunology*, **2011**, 41, 235-45	6.1	37
150	AML1 interconnected pathways of leukemogenesis. *Cancer Investigation*, **2003**, 21, 105-36	2.1	36
149	Secondary leukemia in patients with germline transcription factor mutations (RUNX1, GATA2, CEBPA). *Blood*, **2020**, 136, 24-35	2.2	35
148	Gene network disruptions and neurogenesis defects in the adult Ts1Cje mouse model of Down syndrome. *PLoS ONE*, **2010**, 5, e11561	3.7	34
147	A cSNP Map and Database for Human Chromosome 21. *Genome Research*, **2001**, 11, 300-307	9.7	34
146	GATA2 monoallelic expression underlies reduced penetrance in inherited GATA2-mutated MDS/AML. *Leukemia*, **2018**, 32, 2502-2507	10.7	33
145	The susceptibility of Aire(-/-) mice to experimental myasthenia gravis involves alterations in regulatory T cells. *Journal of Autoimmunity*, **2011**, 36, 16-24	15.5	33
144	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, 71-9	4.3	33
143	Differential effects on gene transcription and hematopoietic differentiation correlate with GATA2 mutant disease phenotypes. *Leukemia*, **2018**, 32, 194-202	10.7	32
142	Familial vasopressin-sensitive ACTH-independent macronodular adrenal hyperplasia (VPs-AIMAH): clinical studies of three kindreds. *Clinical Endocrinology*, **2009**, 70, 883-91	3.4	32
141	Thymic deletion and regulatory T cells prevent antimyeloperoxidase GN. *Journal of the American Society of Nephrology: JASN*, **2013**, 24, 573-85	12.7	31
140	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	18.3	31
139	A case of Aromatase deficiency due to a novel CYP19A1 mutation. *BMC Endocrine Disorders*, **2014**, 14, 16	3.3	30
138	TMPRSS3, a type II transmembrane serine protease mutated in non-syndromic autosomal recessive deafness. *Frontiers in Bioscience - Landmark*, **2008**, 13, 1557-67	2.8	30
137	Splice factor mutations and alternative splicing as drivers of hematopoietic malignancy. *Immunological Reviews*, **2015**, 263, 257-78	11.3	29

136	MicroSAGE is highly representative and reproducible but reveals major differences in gene expression among samples obtained from similar tissues. *Genome Biology*, **2003**, 4, R17	18.3	29
135	Huntington disease-linked locus D4S111 exposed as the alpha-L-iduronidase gene. *Somatic Cell and Molecular Genetics*, **1991**, 17, 421-5		29
134	Autoimmune regulator controls T cell help for pathogenetic autoantibody production in collagen-induced arthritis. *Arthritis and Rheumatism*, **2009**, 60, 1683-93		28
133	Genetic regulators of myelopoiesis and leukemic signaling identified by gene profiling and linear modeling. *Journal of Leukocyte Biology*, **2006**, 80, 433-47	6.5	28
132	Cloning and characterization of a putative human glycerol 3-phosphate permease gene (SLC37A1 or G3PP) on 21q22.3: mutation analysis in two candidate phenotypes, DFNB10 and a glycerol kinase deficiency. *Genomics*, **2000**, 70, 190-200	4.3	28
131	Ectrodactyly and Lethal Pulmonary Acinar Dysplasia Associated with Homozygous FGFR2 Mutations Identified by Exome Sequencing. *Human Mutation*, **2016**, 37, 955-63	4.7	28
130	DNA methylation signatures of the AIRE promoter in thymic epithelial cells, thymomas and normal tissues. *Molecular Immunology*, **2011**, 49, 518-26	4.3	27
129	Mutations among Italian mucopolysaccharidosis type I patients. *Journal of Inherited Metabolic Disease*, **1997**, 20, 803-6	5.4	27
128	Differential gene expression studies to explore the molecular pathophysiology of Down syndrome. *Brain Research Reviews*, **2001**, 36, 265-74		27
127	A tale of two siblings: two cases of AML arising from a single pre-leukemic DNMT3A mutant clone. *Leukemia*, **2015**, 29, 2101-4	10.7	26
126	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, 683-97	5.1	26
125	A four-gene LincRNA expression signature predicts risk in multiple cohorts of acute myeloid leukemia patients. *Leukemia*, **2018**, 32, 263-272	10.7	25
124	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, 22-9	4.3	25
123	Morquio A syndrome: cloning, sequence, and structure of the human N-acetylgalactosamine 6-sulfatase (GALNS) gene. *Genomics*, **1994**, 22, 652-4	4.3	25
122	Autoimmune hepatitis in a murine autoimmune polyendocrine syndrome type 1 model is directed against multiple autoantigens. *Hepatology*, **2015**, 61, 1295-305	11.2	23
121	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, 616-23	6.3	23
120	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	6.5	22
119	Many BCR-ABL1 compound mutations reported in chronic myeloid leukemia patients may actually be artifacts due to PCR-mediated recombination. *Blood*, **2014**, 124, 153-5	2.2	22

118	Tmprss3 loss of function impairs cochlear inner hair cell Kcnma1 channel membrane expression. *Human Molecular Genetics*, **2013**, 22, 1289-99	5.6	22
117	Estimating the proportion of microarray probes expressed in an RNA sample. *Nucleic Acids Research* , **2010**, 38, 2168-76	20.1	21
116	Identification of a novel member of the CLIC family, CLIC6, mapping to 21q22.12. *Gene*, **2003**, 320, 31-40	3.8	21
115	Reduced thymic Aire expression and abnormal NF-kappa B2 signaling in a model of systemic autoimmunity. *Journal of Immunology*, **2009**, 182, 2690-9	5.3	20
114	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	4.3	20
113	PCR of a VNTR linked to mucopolysaccharidosis type I and Huntington disease. *Nucleic Acids Research*, **1991**, 19, 6348	20.1	20
112	The mutational burden of therapy-related myeloid neoplasms is similar to primary myelodysplastic syndrome but has a distinctive distribution. *Leukemia*, **2019**, 33, 2842-2853	10.7	19
111	Linearization and purification of BAC DNA for the development of transgenic mice. *Transgenic Research*, **1999**, 8, 147-50	3.3	19
110	Two novel mutations causing mucopolysaccharidosis type I detected by single strand conformational analysis of the alpha-L-iduronidase gene. *Human Molecular Genetics*, **1993**, 2, 1311-2	5.6	19
109	Mucopolysaccharidosis type I (Hurler syndrome): linkage disequilibrium indicates the presence of a major allele. *Human Genetics*, **1992**, 88, 701-2	6.3	19
108	Self-reverting mutations partially correct the blood phenotype in a Diamond Blackfan anemia patient. *Haematologica*, **2017**, 102, e506-e509	6.6	17
107	CBG Santiago: a novel CBG mutation. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E151-5	5.6	17
106	A comparative analysis of transcribed genes in the mouse hypothalamus and neocortex reveals chromosomal clustering. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 14972-7	11.5	17
105	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, 213-23	1.9	17
104	C21orf5, a novel human chromosome 21 gene, has a Caenorhabditis elegans ortholog (pad-1) required for embryonic patterning. *Genomics*, **2000**, 68, 30-40	4.3	17
103	Integrating massively parallel sequencing into diagnostic workflows and managing the annotation and clinical interpretation challenge. *Human Mutation*, **2014**, 35, 413-23	4.7	16
102	DNMT3L is a regulator of X chromosome compaction and post-meiotic gene transcription. *PLoS ONE*, **2011**, 6, e18276	3.7	16
101	Transplantation of autoimmune regulator-encoding bone marrow cells delays the onset of experimental autoimmune encephalomyelitis. *European Journal of Immunology*, **2010**, 40, 3499-509	6.1	15

100	What is expanded in progressive myoclonus epilepsy?. *Nature Genetics*, **1997**, 17, 17	36.3	15
99	Localization of a novel human RNA-editing deaminase (hRED2 or ADARB2) to chromosome 10p15. *Human Genetics*, **1997**, 100, 398-400	6.3	15
98	Derivation of an endogenous small RNA from double-stranded Sox4 sense and natural antisense transcripts in the mouse brain. *Genomics*, **2016**, 107, 88-99	4.3	14
97	BCR-ABL1 kinase domain mutations may persist at very low levels for many years and lead to subsequent TKI resistance. *British Journal of Cancer*, **2013**, 109, 1593-8	8.7	14
96	Autoimmune regulator (AIRE)-deficient CD8+CD28low regulatory T lymphocytes fail to control experimental colitis. *Proceedings of the National Academy of Sciences of the United States of America* , **2011**, 108, 12437-42	11.5	14
95	PCR of a KpnI RFLP in the alpha-L-iduronidase (IDUA) gene. *Nucleic Acids Research*, **1991**, 19, 5796	20.1	14
94	Poor prognosis in familial acute myeloid leukaemia with combined biallelic CEBPA mutations and downstream events affecting the ATM, FLT3 and CDX2 genes. *British Journal of Haematology*, **2010**, 150, 382-5	4.5	13
93	Genetic heterogeneity of granulocytes for the JAK2 V617F mutation in essential thrombocythaemia: implications for mutation detection in peripheral blood. *Pathology*, **2006**, 38, 336-42	1.6	13
92	Localization of Idd11 is not associated with thymus and nkt cell abnormalities in NOD mice. *Diabetes*, **2005**, 54, 3453-7	0.9	13
91	Construction of a 2.5-Mb integrated physical and gene map of distal 21q22.3. *Genomics*, **1998**, 49, 1-13	4.3	13
90	A novel germline mutation in a family with ataxia-pancytopenia syndrome and pediatric acute lymphoblastic leukemia. *Haematologica*, **2019**, 104, e318-e321	6.6	11
89	Aberrant RAG-mediated recombination contributes to multiple structural rearrangements in lymphoid blast crisis of chronic myeloid leukemia. *Leukemia*, **2020**, 34, 2051-2063	10.7	11
88	Ultrastructure of medullary thymic epithelial cells of autoimmune regulator (Aire)-deficient mice. *Immunology and Cell Biology*, **2010**, 88, 50-6	5	11
87	SAGE analysis of genes differentially expressed in presymptomatic TgSOD1G93A transgenic mice identified cellular processes involved in early stage of ALS pathology. *Journal of Molecular Neuroscience*, **2010**, 41, 172-82	3.3	11
86	A synonymous GATA2 variant underlying familial myeloid malignancy with striking intrafamilial phenotypic variability. *British Journal of Haematology*, **2020**, 190, e297-e301	4.5	10
85	Clonal and lineage analysis of somatic DNMT3A and JAK2 mutations in a chronic phase polycythemia vera patient. *British Journal of Haematology*, **2012**, 156, 268-70	4.5	10
84	Thyroxine treatments do not correct inner ear defects in tmprss1 mutant mice. *NeuroReport*, **2010**, 21, 897-901	1.7	10
83	Immunochemical characterization of a novel mitochondrially located protein encoded by a nuclear gene within the DFNB8/10 critical region on 21q22.3. *Biochemical and Biophysical Research Communications*, **1997**, 238, 806-10	3.4	10

82	NKT cell development in the absence of the autoimmune regulator gene (Aire). *European Journal of Immunology*, **2008**, 38, 2689-96	6.1	10
81	Genomic organisation and nervous system expression of radial spoke protein 3. *Gene*, **2004**, 336, 15-23	3.8	10
80	Linkage, but not gene order, of homologous loci, including alpha-L-iduronidase (Idua), is conserved in the Huntington disease region of the mouse and human genomes. *Mammalian Genome*, **1992**, 3, 23-7	3.2	10
79	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	10.7	10
78	Characterisation of a compound in-cis GATA2 germline mutation in a pedigree presenting with myelodysplastic syndrome/acute myeloid leukemia with concurrent thrombocytopenia. *Leukemia*, **2015**, 29, 1795-7	10.7	9
77	and germ line variants predict response and identify CML patients with the greatest risk of imatinib failure. *Blood Advances*, **2017**, 1, 1369-1381	7.8	9
76	Global transcript expression profiling by Serial Analysis of Gene Expression (SAGE). *Genetic Engineering*, **2001**, 23, 201-19		9
75	Structure of the human Lanosterol synthase gene and its analysis as a candidate for holoprosencephaly (HPE1). *Human Genetics*, **1999**, 105, 489-95	6.3	9
74	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	9.7	9
73	Rare and novel epidermal growth factor receptor mutations in non-small-cell lung cancer and lack of clinical response to gefitinib in two cases. *Journal of Thoracic Oncology*, **2012**, 7, 941-2	8.9	8
72	GATA2 is a New Predisposition Gene for Familial Myelodysplastic Syndrome (MDS) and Acute Myeloid Leukemia (AML). *Blood*, **2010**, 116, LBA-3-LBA-3	2.2	8
71	Investigating the potential role of genetic and epigenetic variation of DNA methyltransferase genes in hyperplastic polyposis syndrome. *PLoS ONE*, **2011**, 6, e16831	3.7	8
70	The Genomic Landscape of Sporadic Prolactinomas. *Endocrine Pathology*, **2019**, 30, 318-328	4.2	7
69	Allan-Herndon-Dudley syndrome with unusual profound sensorineural hearing loss. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1872-6	2.5	7
68	Apparent 'JAK2-negative' polycythaemia vera due to compound mutations in exon 14. *British Journal of Haematology*, **2017**, 178, 333-336	4.5	7
67	Multiplex ligation-dependent probe amplification (MLPA) genotyping assay for mouse models of down syndrome. *Frontiers in Bioscience - Landmark*, **2007**, 12, 3010-6	2.8	7
66	An 86-bp VNTR within IDUA is the basis of the D4S111 polymorphic locus. *Genomics*, **1992**, 14, 1118-20	4.3	7
65	Targeted pharmacotherapy after somatic cancer mutation screening. *F1000Research*, **2016**, 5, 1551	3.6	7

64	Delayed diagnosis leading to accelerated-phase chronic eosinophilic leukemia due to a cytogenetically cryptic, imatinib-responsive TNIP1-PDFGRB fusion gene. *Leukemia*, **2016**, 30, 1402-5	10.7	6
63	Genetic Testing in Endocrinology. *Clinical Biochemist Reviews*, **2018**, 39, 17-28	7.3	6
62	Case report of whole genome sequencing in the XY female: identification of a novel SRY mutation and revision of a misdiagnosis of androgen insensitivity syndrome. *BMC Endocrine Disorders*, **2016**, 16, 58	3.3	6
61	Gene and protein expression profiles of JAK-STAT signalling pathway in the developing brain of the Ts1Cje down syndrome mouse model. *International Journal of Neuroscience*, **2019**, 129, 871-881	2	6
60	T cell receptor assessment in autoimmune disease requires access to the most adjacent immunologically active organ. *Journal of Autoimmunity*, **2017**, 81, 24-33	15.5	5
59	Metallophilic macrophages are fully developed in the thymus of autoimmune regulator (Aire)-deficient mice. *Histochemistry and Cell Biology*, **2009**, 131, 643-9	2.4	5
58	A pedigree with autosomal dominant thrombocytopenia, red cell macrocytosis, and an occurrence of t(12:21) positive pre-B acute lymphoblastic leukemia. *Blood Cells, Molecules, and Diseases*, **2007**, 39, 107-14	2.1	5
57	Identification and targeted management of a neurodegenerative disorder caused by biallelic mutations in SLC5A6. *Npj Genomic Medicine*, **2019**, 4, 28	6.2	5
56	A novel, somatic, transforming mutation in the extracellular domain of Epidermal Growth Factor Receptor identified in myeloproliferative neoplasm. *Scientific Reports*, **2017**, 7, 2467	4.9	4
55	Pseudodiastrophic dysplasia expands the known phenotypic spectrum of defects in proteoglycan biosynthesis. *Journal of Medical Genetics*, **2020**, 57, 454-460	5.8	4
54	In depth analysis of the Sox4 gene locus that consists of sense and natural antisense transcripts. *Data in Brief*, **2016**, 7, 282-90	1.2	4
53	Genome-wide gene expression profiling identifies overlap with malignant adrenocortical tumours and novel mechanisms of inefficient steroidogenesis in familial ACTH-independent macronodular adrenal hyperplasia. *Endocrine-Related Cancer*, **2012**, 19, L19-23	5.7	4
52	Diversity and clonotypic composition of influenza-specific CD8+ TCR repertoires remain unaltered in the absence of Aire. *European Journal of Immunology*, **2010**, 40, 849-58	6.1	4
51	No evidence for core-binding factor CBFbeta as a leukemia predisposing factor in chromosome 16q22-linked familial AML. *Leukemia*, **2004**, 18, 881	10.7	4
50	Chromosome band 16q22-linked familial AML: exclusion of candidate genes, and possible disease risk modification by NQO1 polymorphisms. *Genes Chromosomes and Cancer*, **2004**, 41, 278-82	5	4
49	Targeted pharmacotherapy after somatic cancer mutation screening. *F1000Research*, **2016**, 5, 1551	3.6	4
48	Insights into pituitary tumorigenesis: from Sanger sequencing to next-generation sequencing and beyond. *Expert Review of Endocrinology and Metabolism*, **2019**, 14, 399-418	4.1	4
47	GATA2 deficiency syndrome: A decade of discovery. *Human Mutation*, **2021**, 42, 1399-1421	4.7	4

46	ARMC5 is not implicated in familial hyperaldosteronism type II (FH-II). *Journal of Human Hypertension*, **2017**, 31, 857-859	2.6	3
45	Paternal mosaicism for a novel PBX1 mutation associated with recurrent perinatal death: Phenotypic expansion of the PBX1-related syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1273-1277	2.5	3
44	Presentation of m.3243A>G (MT-TL1; tRNALeu) variant with focal neurology in infancy. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2697-701	2.5	3
43	High Incidence of Mutated Cancer-Associated Genes at Diagnosis in CML Patients with Early Transformation to Blast Crisis. *Blood*, **2015**, 126, 600-600	2.2	3
42	NOMINATOR: Feasibility of genomic testing of rare cancers to match cancer to treatment.. *Journal of Clinical Oncology*, **2020**, 38, 103-103	2.2	3
41	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	6.6	3
40	Expanding the phenotype of NUP85 mutations beyond nephrotic syndrome to primary autosomal recessive microcephaly and Seckel syndrome spectrum disorders. *Human Molecular Genetics*, **2021**, 30, 2068-2081	5.6	3
39	Clinical implications of transient myeloproliferative disorder in a neonate without Down syndrome features. *Journal of Paediatrics and Child Health*, **2017**, 53, 1018-1020	1.3	2
38	RUNX1 mutations are rare in chronic phase polycythaemia vera. *British Journal of Haematology*, **2011**, 153, 672-5	4.5	2
37	A response to Suzuki et al. How pathogenic is the p.D104N/endostatin polymorphic allele of COL18A1 in Knobloch syndrome? *Human Mutation*, **2005**, 25, 316-316	4.7	2
36	Universal fluorescent labeling of PCR products for DHPLC analysis: reducing cost and increasing sample throughput. *BioTechniques*, **2005**, 39, 34, 36, 38, 40	2.5	2
35	Australian Familial Haematological Cancer Study - Findings from 15 Years of Aggregated Clinical, Genomic and Transcriptomic Data. *Blood*, **2019**, 134, 1439-1439	2.2	2
34	BCR-ABL Assay Sensitivity of MR4.5 Achieved in >90%, and MR5 in >75% of Samples, through mRNA Selection before qRT-PCR. *Blood*, **2015**, 126, 2777-2777	2.2	2
33	Expanded Phenotypic and Genetic Heterogeneity in the Clinical Spectrum of FPD-AML: Lymphoid Malignancies and Skin Disorders Are Common Features in Carriers of Germline RUNX1 Mutations. *Blood*, **2016**, 128, 1212-1212	2.2	2
32	Novel Fusion Genes at CML Diagnosis Reveal a Complex Pattern of Genomic Rearrangements and Sequence Inversions Associated with the Philadelphia Chromosome in Patients with Early Blast Crisis. *Blood*, **2016**, 128, 1219-1219	2.2	2
31	A putative role for the aryl hydrocarbon receptor (AHR) gene in a patient with cyclical Cushing's disease. *BMC Endocrine Disorders*, **2020**, 20, 18	3.3	2
30	Aberrant Splicing of in Families With Unexplained Succinate Dehydrogenase-Deficient Paragangliomas. *Journal of the Endocrine Society*, **2020**, 4, bvaa071	0.4	2
29	Correct application of variant classification guidelines in germline mutated disorders to assist clinical diagnosis. *Leukemia and Lymphoma*, **2020**, 61, 246-247	1.9	2

28	Autosomal dominant hypocalcaemia due to a novel CASR mutation: clinical and genetic implications. *Clinical Endocrinology*, **2016**, 85, 495-7	3.4	1
27	Rare and Common Germline Variants Contribute to Occurrence of Myelodysplastic Syndrome. *Blood*, **2015**, 126, 1644-1644	2.2	1
26	Metabolic Profiling of Adult Acute Myeloid Leukemia (AML). *Blood*, **2016**, 128, 1684-1684	2.2	1
25	Two monogenic disorders masquerading as one: severe congenital neutropenia with monocytosis and non-syndromic sensorineural hearing loss. *BMC Medical Genetics*, **2020**, 21, 35	2.1	1
24	Expression Profiling of Notch Signalling Pathway and Gamma-Secretase Activity in the Brain of Ts1Cje Mouse Model of Down Syndrome. *Journal of Molecular Neuroscience*, **2019**, 67, 632-642	3.3	1
23	8q13.1-q13.2 deletion associated with inferior cerebellar vermian hypoplasia and digital anomalies: a new syndrome?. *Pediatric Neurology*, **2015**, 52, 230-4.e1	2.9	0
22	Transcriptional profiling of the postnatal brain of the Ts1Cje mouse model of Down syndrome. *Genomics Data*, **2014**, 2, 314-7		0
21	RNA Splicing Defects in Cancer-Linked Genes Indicate Mutation or Focal Gene Deletion and Are Associated with TKI Resistance in CML. *Blood*, **2019**, 134, 662-662	2.2	0
20	Compound heterozygous variants in LAMC3 in association with posterior periventricular nodular heterotopia. *BMC Medical Genomics*, **2021**, 14, 64	3.7	0
19	Childhood acute myeloid leukemia shows a high level of germline predisposition. *Blood*, **2021**, 138, 2293-2298	22	0
18	RNF43 pathogenic Germline variant in a family with colorectal cancer. *Clinical Genetics*, **2022**, 101, 122-126	4	0
17	Pathogenic variants in cause recessive central conducting lymphatic anomaly with lymphedema.. *Science Translational Medicine*, **2022**, 14, eabm4869	17.5	0
16	The human genome project and its impact in medicine. *European Review*, **1996**, 4, 415	0.3	
15	Dodecamer Repeat Expansion in Progressive Myoclonus Epilepsy 1 **2006**, 121-141		
14	Therapy-Related Myeloid Neoplasms (T-MN) and Primary MDS (PMDS) Patients with Very Low (VL) or Low (L) IPSS-R Score Share Clinical and Biological Characteristics and Have Similar Outcome. *Blood*, **2018**, 132, 3078-3078	2.2	
13	Development of a Data Portal for Aggregation and Analysis of Genomics Data in Familial Platelet Disorder with Predisposition to Myeloid Malignancy - the RUNX1.DB. *Blood*, **2018**, 132, 5241-5241	2.2	
12	An RNA-Based Next Generation Sequencing (NGS) Strategy Detects More Cancer Gene Mutations Than a DNA-Based Approach for the Prediction and Assessment of Resistance in CML. *Blood*, **2019**, 134, 2918-2918	2.2	
11	Familial Clustering of Hematological Malignancies: Harbingers of Wider Germline Cancer Susceptibility. *Blood*, **2019**, 134, 3794-3794	2.2	

10	Aberrant Activation of Epidermal Growth Factor Receptor in MPN May Respond to the Kinase Inhibitor Gefitinib. *Blood*, **2014**, 124, 1882-1882	2.2
9	Clonal Diversity of Recurrently Mutated Genes in Myelodysplastic Syndromes. *Blood*, **2014**, 124, 4634-4634	2.2
8	The Frequency of Genetic Mutations in T-MN Is High and Comparable to Primary MDS but the Spectrum Is Different. *Blood*, **2016**, 128, 3157-3157	2.2
7	Detection of Low Level Nilotinib or Dasatinib Resistant BCR-ABL Mutations by Mass Spectrometry In CML Patients Who Fail Imatinib Is Highly Predictive of Their Subsequent Clonal Expansion When Treated with the Drug for Which Their Mutation Confers Resistance. *Blood*, **2010**, 116, 891-891	2.2
6	Mechanisms of Co-Operation of DNMT3A Mutations with JAK2 V617F Through Histone H4 Arginine 3 Provides New Insights in MPN Disease Pathogenesis. *Blood*, **2011**, 118, 2823-2823	2.2
5	Multiple Low Level Mutations Identifies Imatinib Resistant CML Patients At Risk of Poor Response to Second-Line Inhibitor Therapy, Irrespective of the Resistance Profile of the Mutations. *Blood*, **2011**, 118, 111-111	2.2
4	The patient's BCR-ABL1 Kinase Domain Mutation History Is Important for Decisions Regarding Tyrosine Kinase Inhibitor Therapy. *Blood*, **2012**, 120, 1692-1692	2.2
3	PCR-Mediated Recombination Can Lead To Artificial Chimera Formation, Which May Pose As BCR-ABL1 Compound Mutations. *Blood*, **2013**, 122, 4014-4014	2.2
2	Familial AML With Germline CEBPA Mutations: Extended Clinical Outcomes and Analysis Of Secondary Mutations Using Whole Exome Sequencing. *Blood*, **2013**, 122, 740-740	2.2
1	Laboratory quality assessment of candidate gene panel testing for acute myeloid leukaemia: a joint ALLG / RCPAQAP initiative. *Pathology*, **2021**, 53, 487-492	1.6