Stanislas Lyonnet

List of Publications by Year
in descending order

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282 papers	27,663 citations	4388 86 h-index	6995 154 g-index
285 all docs	285 docs citations	285 times ranked	28050 citing authors

#	Article	IF	Citations
1	Lamin A Truncation in Hutchinson-Gilford Progeria. Science, 2003, 300, 2055-2055.	12.6	1,247
2	Holt-Oram syndrome is caused by mutations in TBX5, a member of the Brachyury (T) gene family. Nature Genetics, 1997, 15, 21-29.	21.4	859
3	Hirschsprung disease, associated syndromes and genetics: a review. Journal of Medical Genetics, 2007, 45, 1-14.	3.2	848
4	Somatic and germline activating mutations of the ALK kinase receptor in neuroblastoma. Nature, 2008, 455, 967-970.	27.8	787
5	Polyalanine expansion and frameshift mutations of the paired-like homeobox gene PHOX2B in congenital central hypoventilation syndrome. Nature Genetics, 2003, 33, 459-461.	21.4	771
6	Mutations of the RET proto-oncogene in Hirschsprung's disease. Nature, 1994, 367, 378-380.	27.8	750
7	SOX10 mutations in patients with Waardenburg-Hirschsprung disease. Nature Genetics, 1998, 18, 171-173.	21.4	733
8	Mutation of the endothelin-3 gene in the Waardenburg-Hirschsprung disease (Shah-Waardenburg) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	21.4	425
9	Mutations in STAT3 and IL12RB1 impair the development of human IL-17â€“producing T cells. Journal of Experimental Medicine, 2008, 205, 1543-1550.	8.5	406
10	Hirschsprung disease, associated syndromes, and genetics: a review. Journal of Medical Genetics, 2001, 38, 729-739.	3.2	394
11	Claudin-1 gene mutations in neonatal sclerosing cholangitis associated with ichthyosis: A tight junction disease. Gastroenterology, 2004, 127, 1386-1390.	1.3	378
12	Targeted therapy in patients with PIK3CA-related overgrowth syndrome. Nature, 2018, 558, 540-546.	27.8	374
13	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	21.4	364
14	Diversity of RET proto-oncogene mutations in familial and sporadic Hirschsprung disease. Human Molecular Genetics, 1995, 4, 1381-1386.	2.9	342
15	miR-122, a paradigm for the role of microRNAs in the liver. Journal of Hepatology, 2008, 48, 648-656.	3.7	330
16	Mutant WD-repeat protein in triple-A syndrome. Nature Genetics, 2000, 26, 332-335.	21.4	304
17	Germline Mutations of the Pairedâ€“Like Homeobox 2B (PHOX2B) Gene in Neuroblastoma. American Journal of Human Genetics, 2004, 74, 761-764.	6.2	288
18	Germline deletion of the miR-17âˆ¼92 cluster causes skeletal and growth defects in humans. Nature Genetics, 2011, 43, 1026-1030.	21.4	275

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19	Segregation at three loci explains familial and population risk in Hirschsprung disease. Nature Genetics, 2002, 31, 89-93.	21.4	269
20	Array-based comparative genomic hybridisation identifies high frequency of cryptic chromosomal rearrangements in patients with syndromic autism spectrum disorders. Journal of Medical Genetics, 2006, 43, 843-849.	3.2	267
21	Mutations in TCF4, Encoding a Class I Basic Helix-Loop-Helix Transcription Factor, Are Responsible for Pitt-Hopkins Syndrome, a Severe Epileptic Encephalopathy Associated with Autonomic Dysfunction. American Journal of Human Genetics, 2007, 80, 988-993.	6.2	264
22	Nuclear Outsourcing of RNA Interference Components to Human Mitochondria. PLoS ONE, 2011, 6, e20746.	2.5	249
23	Pleiotropic Effects of CEP290 (NPHP6) Mutations Extend to Meckel Syndrome. American Journal of Human Genetics, 2007, 81, 170-179.	6.2	248
24	Genotypeâ€“phenotype correlations in Down syndrome identified by array CGH in 30 cases of partial trisomy and partial monosomy chromosome 21. European Journal of Human Genetics, 2009, 17, 454-466.	2.8	240
25	Only four genes (EDA1, EDAR, EDARADD, and WNT10A) account for 90% of hypohidrotic/anhidrotic ectodermal dysplasia cases. Human Mutation, 2011, 32, 70-72.	2.5	240
26	Diverse phenotypes associated with exon 10 mutations of the RET proto-oncogene. Human Molecular Genetics, 1994, 3, 2163-2168.	2.9	239
27	Differential Contributions of Rare and Common, Coding and Noncoding Ret Mutations to Multifactorial Hirschsprung Disease Liability. American Journal of Human Genetics, 2010, 87, 60-74.	6.2	230
28	PHOX2B Genotype Allows for Prediction of Tumor Risk in Congenital Central Hypoventilation Syndrome. American Journal of Human Genetics, 2005, 76, 421-426.	6.2	222
29	The Meckel-Gruber Syndrome Gene, MKS3, Is Mutated in Joubert Syndrome. American Journal of Human Genetics, 2007, 80, 186-194.	6.2	217
30	Mutation of the endothelin-receptor B gene in Waardenburg-Hirschsprung disease. Human Molecular Genetics, 1995, 4, 2407-2409.	2.9	214
31	Germline mutations of the RET ligand GDNF are not sufficient to cause Hirschsprung disease. Nature Genetics, 1996, 14, 345-347.	21.4	203
32	A human model for multigenic inheritance: Phenotypic expression in Hirschsprung disease requires both the RET gene and a new 9q31 locus. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 268-273.	7.1	203
33	Comparison of Clinical Presentations and Outcomes Between Patients With TGFBR2 and FBN1 Mutations in Marfan Syndrome and Related Disorders. Circulation, 2009, 120, 2541-2549.	1.6	203
34	KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	21.4	203
35	Phenotypic spectrum of CHARGE syndrome in fetuses with CHD7 truncating mutations correlates with expression during human development. Journal of Medical Genetics, 2005, 43, 211-317.	3.2	199
36	Mutation update for theCSB/ERCC6andCSA/ERCC8genes involved in Cockayne syndrome. Human Mutation, 2010, 31, 113-126.	2.5	193

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37	A gene for Hirschsprung disease maps to the proximal long arm of chromosome 10. Nature Genetics, 1993, 4, 346-350.	21.4	190
38	Nineteen Years of National Screening for Congenital Hypothyroidism: Familial Cases with Thyroid Dysgenesis Suggest the Involvement of Genetic Factors. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 2009-2014.	3.6	190
39	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	2.9	190
40	Homozygous silencing of T-box transcription factor EOMES leads to microcephaly with polymicrogyria and corpus callosum agenesis. Nature Genetics, 2007, 39, 454-456.	21.4	181
41	Occurrence of myeloproliferative disorder in patients with Noonan syndrome. Journal of Pediatrics, 1997, 130, 885-889.	1.8	176
42	Heterozygous endothelin receptor B (EDNRB) mutations in isolated Hirschsprung disease. Human Molecular Genetics, 1996, 5, 355-357.	2.9	174
43	Matthew-Wood Syndrome Is Caused by Truncating Mutations in the Retinol-Binding Protein Receptor Gene STRA6. American Journal of Human Genetics, 2007, 80, 1179-1187.	6.2	174
44	Neurological Phenotype in Waardenburg Syndrome Type 4 Correlates with Novel SOX10 Truncating Mutations and Expression in Developing Brain. American Journal of Human Genetics, 2000, 66, 1496-1503.	6.2	172
45	Disruption of a long distance regulatory region upstream of SOX9 in isolated disorders of sex development. Journal of Medical Genetics, 2011, 48, 825-830.	3.2	162
46	Inversion of the circadian rhythm of melatonin in the Smith-Magenis syndrome. Journal of Pediatrics, 2001, 139, 111-116.	1.8	158
47	Long-range regulation at the SOX9 locus in development and disease. Journal of Medical Genetics, 2009, 46, 649-656.	3.2	148
48	Mutation of the RET ligand, neurturin, supports multigenic inheritance in Hirschsprung disease [published erratum appears in Hum Mol Genet 1998 Oct;7(11):1831]. Human Molecular Genetics, 1998, 7, 1449-1452.	2.9	145
49	CHARGE Syndrome Includes Hypogonadotropic Hypogonadism and Abnormal Olfactory Bulb Development. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 5621-5626.	3.6	142
50	Pierre Robin Sequence: A series of 117 consecutive cases. Journal of Pediatrics, 2001, 139, 588-590.	1.8	136
51	Molecular consequences of PHOX2B missense, frameshift and alanine expansion mutations leading to autonomic dysfunction. Human Molecular Genetics, 2005, 14, 3697-3708.	2.9	135
52	A novel loss-of-function mutation in TTF-2 is associated with congenital hypothyroidism, thyroid agenesis and cleft palate. Human Molecular Genetics, 2002, 11, 2051-2059.	2.9	134
53	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
54	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132

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55	Polyalanine expansions in human. Human Molecular Genetics, 2004, 13, R235-R243.	2.9	131
56	Clinical homogeneity and genetic heterogeneity in Weill-Marchesani syndrome. , 2003, 123A, 204-207.		130
57	Splice-mediated insertion of an Alu sequence inactivates ornithine delta-aminotransferase: a role for Alu elements in human mutation.. Proceedings of the National Academy of Sciences of the United States of America, 1991, 88, 815-819.	7.1	129
58	Contiguous Gene Deletion within Chromosome Arm 10q Is Associated with Juvenile Polyposis of Infancy, Reflecting Cooperation between the BMPR1A and PTEN Tumor-Suppressor Genes. American Journal of Human Genetics, 2006, 78, 1066-1074.	6.2	127
59	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	2.5	126
60	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	2.4	125
61	TCTN3 Mutations Cause Mohr-Majewski Syndrome. American Journal of Human Genetics, 2012, 91, 372-378.	6.2	123
62	Contribution of rare and common variants determine complex diseasesâ€"Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	2.0	119
63	Functional Loss of Semaphorin 3C and/or Semaphorin 3D and Their Epistatic Interaction with Ret Are Critical to Hirschsprung Disease Liability. American Journal of Human Genetics, 2015, 96, 581-596.	6.2	118
64	Noonan Syndrome: Relationships between Genotype, Growth, and Growth Factors. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 300-306.	3.6	117
65	Spectrum of NSD1 mutations in Sotos and Weaver syndromes. Journal of Medical Genetics, 2003, 40, 436-440.	3.2	116
66	Mutations of the RET-GDNF Signaling Pathway in Ondine's Curse. American Journal of Human Genetics, 1998, 62, 715-717.	6.2	115
67	Exome Sequencing Identifies PDE4D Mutations as Another Cause of Acrodysostosis. American Journal of Human Genetics, 2012, 90, 740-745.	6.2	115
68	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	2.5	113
69	Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. American Journal of Human Genetics, 2016, 98, 971-980.	6.2	113
70	Estimating the age of rare disease mutations: the example of Triple-A syndrome. Journal of Medical Genetics, 2004, 41, 445-449.	3.2	112
71	Prevalence of 22q11 deletion in fetuses with conotruncal cardiac defects: A 6-year prospective study. Journal of Pediatrics, 2001, 138, 520-524.	1.8	110
72	Paradoxical NSD1 Mutations in Beckwith-Wiedemann Syndrome and 11p15 Anomalies in Sotos Syndrome. American Journal of Human Genetics, 2004, 74, 715-720.	6.2	110

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73	Temporal bone anomaly proposed as a major criteria for diagnosis of CHARGE syndrome. American Journal of Medical Genetics Part A, 2001, 99, 124-127.	2.4	109
74	PAX2 mutations in oligomeganephronia. Kidney International, 2001, 59, 457-462.	5.2	106
75	Large-Scale Deletions and SMADIP1 Truncating Mutations in Syndromic Hirschsprung Disease with Involvement of Midline Structures. American Journal of Human Genetics, 2001, 69, 1370-1377.	6.2	105
76	XNP mutation in a large family with Juberg-Marsidi syndrome. Nature Genetics, 1996, 12, 359-360.	21.4	101
77	Molecular analysis of pericentrin gene (PCNT) in a series of 24 Seckel/microcephalic osteodysplastic primordial dwarfism type II (MOPD II) families. Journal of Medical Genetics, 2010, 47, 797-802.	3.2	100
78	Various mechanisms cause RET-mediated signaling defects in Hirschsprung's disease.. Journal of Clinical Investigation, 1998, 101, 1415-1423.	8.2	99
79	Peripheral neuropathy with hypomyelination, chronic intestinal pseudo-obstruction and deafness: A developmental ?neural crest syndrome? related to a SOX10 mutation. Annals of Neurology, 2000, 48, 671-676.	5.3	98
80	Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. Human Molecular Genetics, 2014, 23, 2279-2289.	2.9	98
81	Mortality Associated with Neurofibromatosis 1: A Cohort Study of 1895 Patients in 1980-2006 in France. Orphanet Journal of Rare Diseases, 2011, 6, 18.	2.7	96
82	Prevalence of the microdeletion 22q11 in newborn infants with congenital conotruncal cardiac anomalies. European Journal of Pediatrics, 1998, 157, 881-884.	2.7	93
83	Spectrum ofMKS1andMKS3mutations in Meckel syndrome: a genotype-phenotype correlation. Human Mutation, 2007, 28, 523-524.	2.5	92
84	ALDH1A3 Mutations Cause Recessive Anophthalmia and Microphthalmia. American Journal of Human Genetics, 2013, 92, 265-270.	6.2	92
85	A duplication in the L1CAM gene associated with Xâ€“linked hydrocephalus. Nature Genetics, 1993, 4, 421-425.	21.4	91
86	Genetics of limb anomalies in humans. Trends in Genetics, 1999, 15, 409-417.	6.7	91
87	EFTUD2haploinsufficiency leads to syndromic oesophageal atresia. Journal of Medical Genetics, 2012, 49, 737-746.	3.2	89
88	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255.	21.4	88
89	Co-segregation of MEN2 and Hirschsprung's disease: The same mutation ofRET with both gain and loss-of-function?. Human Mutation, 1999, 13, 331-336.	2.5	87
90	Familial Forms of Thyroid Dysgenesis among Infants with Congenital Hypothyroidism. New England Journal of Medicine, 2000, 343, 441-442.	27.0	87

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91	MECP2 mutation in non-fatal, non-progressive encephalopathy in a male. Journal of Medical Genetics, 2001, 38, 171-174.	3.2	87
92	Human neural crest cells display molecular and phenotypic hallmarks of stem cells. Human Molecular Genetics, 2008, 17, 3411-3425.	2.9	87
93	Xq28 duplication presenting with intestinal and bladder dysfunction and a distinctive facial appearance. European Journal of Human Genetics, 2009, 17, 434-443.	2.8	87
94	microRNAs in diseases: from candidate to modifier genes. Clinical Genetics, 2010, 77, 306-313.	2.0	87
95	Rokitansky Syndrome: Clinical Experience and Results of Sigmoid Vaginoplasty in 23 Young Girls. Journal of Urology, 2007, 177, 1107-1111.	0.4	86
96	Identification of 23TGFBR2and 6TGFBR1gene mutations and genotype-phenotype investigations in 457 patients with Marfan syndrome type I and II, Loeys-Dietz syndrome and related disorders. Human Mutation, 2008, 29, E284-E295.	2.5	86
97	CpG dinucleotides are mutation hot spots in phenylketonuria. Genomics, 1989, 5, 936-939.	2.9	85
98	PHOX2B Germline and Somatic Mutations in Late-Onset Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2008, 177, 906-911.	5.6	85
99	Functional Effects of PTPN11 (SHP2) Mutations Causing LEOPARD Syndrome on Epidermal Growth Factor-Induced Phosphoinositide 3-Kinase/AKT/Glycogen Synthase Kinase 3Î² Signaling. Molecular and Cellular Biology, 2010, 30, 2498-2507.	2.3	85
100	REEP1 mutations in SPG31: Frequency, mutational spectrum, and potential association with mitochondrial morpho-functional dysfunction. Human Mutation, 2011, 32, 1118-1127.	2.5	83
101	Genetic characterization of congenital tufting enteropathy: epcam associated phenotype and involvement of SPINT2 in the syndromic form. Human Genetics, 2014, 133, 299-310.	3.8	83
102	TP63 gene mutation in ADULT syndrome. European Journal of Human Genetics, 2001, 9, 642-645.	2.8	82
103	Mutations in KIAA0586 Cause Lethal Ciliopathies Ranging from a Hydrolethalus Phenotype to Short-Rib Polydactyly Syndrome. American Journal of Human Genetics, 2015, 97, 311-318.	6.2	82
104	Molecular Bases of Human Neurocristopathies. , 2006, 589, 213-234.		79
105	PAX2 mutations in renalâ€“coloboma syndrome: mutational hotspot and germline mosaicism. European Journal of Human Genetics, 2000, 8, 820-826.	2.8	77
106	Vestibular anomalies in CHARGE syndrome: investigations on and consequences for postural development. European Journal of Pediatrics, 2000, 159, 569-574.	2.7	75
107	A Human Homeotic Transformation Resulting from Mutations in PLCB4 and GNAI3 Causes Auriculocondylar Syndrome. American Journal of Human Genetics, 2012, 90, 907-914.	6.2	75
108	Ret in human development and oncogenesis. BioEssays, 1997, 19, 389-395.	2.5	74

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109	Spectrum of Mutations of theAAASGene in Allgrove Syndrome: Lack of Mutations in Six Kindreds with Isolated Resistance to Corticotropin. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5433-5437.	3.6	72
110	Noradrenergic neuronal development is impaired by mutation of the proneural HASH-1 gene in congenital central hypoventilation syndrome (Ondine's curse). Human Molecular Genetics, 2003, 12, 3173-3180.	2.9	72
111	Atypical findings in Kabuki syndrome: Report of 8 patients in a series of 20 and review of the literature. American Journal of Medical Genetics, Part A, 2004, 129A, 64-68.	1.2	72
112	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	8.8	72
113	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	3.8	71
114	An overview of isolated and syndromic oesophageal atresia. Clinical Genetics, 2007, 71, 392-399.	2.0	70
115	Identification of Novel Craniofacial Regulatory Domains Located far Upstream ofSOX9and Disrupted in Pierre Robin Sequence. Human Mutation, 2014, 35, 1011-1020.	2.5	69
116	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 2015, 125, 3585-3599.	8.2	69
117	Stabilization of RNA during laser capture microdissection by performing experiments under argon atmosphere or using ethanol as a solvent in staining solutions. Rna, 2008, 14, 2698-2704.	3.5	68
118	New clinical and therapeutic perspectives in Currarino syndrome (study of 29 cases). Journal of Pediatric Surgery, 2006, 41, 126-131.	1.6	67
119	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain, 2018, 141, 1998-2013.	7.6	67
120	Endothelin-3 Gene Mutations in Isolated and Syndromic Hirschsprung Disease. European Journal of Human Genetics, 1997, 5, 247-251.	2.8	67
121	Dysmorphic phenotype and neurological impairment in 22 retinoblastoma patients with constitutional cytogenetic 13q deletion. Clinical Genetics, 1999, 55, 478-482.	2.0	66
122	MMP21 is mutated in human heterotaxy and is required for normal left-right asymmetry in vertebrates. Nature Genetics, 2015, 47, 1260-1263.	21.4	65
123	Phenotype and genotype analysis of a French cohort of 119 patients with CHARGE syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 417-430.	1.6	65
124	Genetics and Early Disturbances of Breathing Control: The Genetics of Childhood Disease and Development: A Series of Review Articles. Pediatric Research, 2004, 55, 729-733.	2.3	64
125	Germline mutations of the paired-like homeobox 2B (PHOX2B) gene in neuroblastoma. Cancer Letters, 2005, 228, 51-58.	7.2	63
126	Delineation of Late Onset Hypoventilation Associated with Hypothalamic Dysfunction Syndrome. Pediatric Research, 2008, 64, 689-694.	2.3	63

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127	Mutations of the ImprintedCDKN1CGene as a Cause of the Overgrowth Beckwith-Wiedemann Syndrome: Clinical Spectrum and Functional Characterization. Human Mutation, 2015, 36, 894-902.	2.5	62
128	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
129	Mutations in Endothelin 1 Cause Recessive Auriculocondylar Syndrome and Dominant Isolated Question-Mark Ears. American Journal of Human Genetics, 2013, 93, 1118-1125.	6.2	59
130	Homozygosity Mapping of a Locus for a Novel Syndromic Ichthyosis to Chromosome 3q27â€“q28. Journal of Investigative Dermatology, 2002, 119, 70-76.	0.7	58
131	Mutation in a primate-conserved retrotransposon reveals a noncoding RNA as a mediator of infantile encephalopathy. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 4980-4985.	7.1	58
132	Extensive investigation of the IGF2/H19 imprinting control region reveals novel OCT4/SOX2 binding site defects associated with specific methylation patterns in Beckwith-Wiedemann syndrome. Human Molecular Genetics, 2014, 23, 5763-5773.	2.9	58
133	Interaction between a chromosome 10RETenhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. Human Mutation, 2009, 30, 771-775.	2.5	57
134	The C20orf133 gene is disrupted in a patient with Kabuki syndrome. Journal of Medical Genetics, 2007, 44, 562-569.	3.2	56
135	DCDC2Mutations Cause Neonatal Sclerosing Cholangitis. Human Mutation, 2016, 37, 1025-1029.	2.5	56
136	Expression of theRET proto-oncogene in human Embryos. , 1998, 80, 481-486.		55
137	Expression of thePAX2 gene in human embryos and exclusion in the CHARGE syndrome. American Journal of Medical Genetics Part A, 2000, 93, 85-88.	2.4	55
138	A novel automated strategy for screening cryptic telomeric rearrangements in children with idiopathic mental retardation. European Journal of Human Genetics, 2001, 9, 319-327.	2.8	55
139	Evaluation of methods for amplification of picogram amounts of total RNA for whole genome expression profiling. BMC Genomics, 2009, 10, 246.	2.8	54
140	Functional Characterization of Three Mutations of the Endothelin B Receptor Gene in Patients With Hirschsprungâ€™s Disease: Evidence for Selective Loss of Gi Coupling. Molecular Medicine, 2001, 7, 115-124.	4.4	53
141	Adams-Oliver syndrome and hepatoportal sclerosis: Occasional association or common mechanism?. American Journal of Medical Genetics, Part A, 2005, 135A, 186-189.	1.2	52
142	Deletion of Pten in the mouse enteric nervous system induces ganglioneuromatosis and mimics intestinal pseudoobstruction. Journal of Clinical Investigation, 2009, 119, 3586-3596.	8.2	52
143	Nineteen Years of National Screening for Congenital Hypothyroidism: Familial Cases with Thyroid Dysgenesis Suggest the Involvement of Genetic Factors. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 2009-2014.	3.6	52
144	A gene for Holtâ€“Oram syndrome maps to the distal long arm of chromosome 12. Nature Genetics, 1994, 6, 405-408.	21.4	51

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145	Epistasis between RET and BBS mutations modulates enteric innervation and causes syndromic Hirschsprung disease. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 13921-13926.	7.1	51
146	ZEB2 zinc-finger missense mutations lead to hypomorphic alleles and a mild Mowatâ€“Wilson syndrome. Human Molecular Genetics, 2013, 22, 2652-2661.	2.9	51
147	PHOX2B gene mutation in a patient with late-onset central hypoventilation. Pediatric Pulmonology, 2004, 38, 349-351.	2.0	50
148	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	2.5	50
149	Expression of the SMADIP1 gene during early human development. Mechanisms of Development, 2002, 114, 187-191.	1.7	49
150	Linkage and mutational analysis of familial thyroid dysgenesis demonstrate genetic heterogeneity implicating novel genes. European Journal of Human Genetics, 2005, 13, 232-239.	2.8	49
151	CLMP Is Required for Intestinal Development, and Loss-of-Function Mutations Cause Congenital Short-Bowel Syndrome. Gastroenterology, 2012, 142, 453-462.e3.	1.3	49
152	Understanding the basis of auriculocondylar syndrome: Insights from human, mouse and zebrafish genetic studies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 306-317.	1.6	48
153	Identification of a novel ARL13B variant in a Joubert syndrome-affected patient with retinal impairment and obesity. European Journal of Human Genetics, 2015, 23, 621-627.	2.8	48
154	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531.	6.2	47
155	Thyroid Hemiagenesis Is a Rare Variant of Thyroid Dysgenesis with a Familial Component but without Pax8 Mutations in a Cohort of 22 Cases. Pediatric Research, 2005, 57, 908-913.	2.3	46
156	ISL1 Directly Regulates FGF10 Transcription during Human Cardiac Outflow Formation. PLoS ONE, 2012, 7, e30677.	2.5	46
157	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. Journal of Medical Genetics, 2011, 48, 417-421.	3.2	45
158	Antenatal spectrum of CHARGE syndrome in 40 fetuses withCHD7mutations. Journal of Medical Genetics, 2012, 49, 698-707.	3.2	45
159	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. Journal of Medical Genetics, 2013, 50, 174-186.	3.2	44
160	Three Copies of Four Interferon Receptor Genes Underlie a Mild Type I Interferonopathy in Down Syndrome. Journal of Clinical Immunology, 2020, 40, 807-819.	3.8	44
161	Heterozygous Bile Salt Export Pump Deficiency: A Possible Genetic Predisposition to Transient Neonatal Cholestasis. Journal of Pediatric Gastroenterology and Nutrition, 2006, 42, 114-116.	1.8	40
162	Exclusion ofWNT4 as a major gene in Rokitansky-KÃ¼ster-Hauser anomaly. American Journal of Medical Genetics, Part A, 2005, 137A, 98-99.	1.2	39

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163	Clinical and molecular overlap in overgrowth syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 137C, 4-11.	1.6	39
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