Eamonn R Maher

List of Publications by Year
in descending order

479 papers	56,887 citations	797 122 h-index	1801 217 g-index
531 all docs	531 docs citations	531 times ranked	51629 citing authors

#	Article	IF	Citations
1	Cost-effectiveness model of renal cell carcinoma (RCC) surveillance in hereditary leiomyomatosis and renal cell carcinoma (HLRCC). Journal of Medical Genetics, 2023, 60, 41-47.	1.5	4
2	UK recommendations forSDHAgermline genetic testing and surveillance in clinical practice. Journal of Medical Genetics, 2023, 60, 107-111.	1.5	4
3	Combining clinical, radiological and genetic approaches to pneumothorax management. Thorax, 2022, 77, 196-198.	2.7	2
4	International initiative for a curated SDHB variant database improving the diagnosis of hereditary paraganglioma and pheochromocytoma. Journal of Medical Genetics, 2022, 59, 785-792.	1.5	5
5	SDHC phaeochromocytoma and paraganglioma: A UKâ€wide case series. Clinical Endocrinology, 2022, 96, 499-512.	1.2	7
6	Quantifying evidence toward pathogenicity for rare phenotypes: The case of succinate dehydrogenase genes, SDHB and SDHD. Genetics in Medicine, 2022, 24, 41-50.	1.1	5
7	ImprintSeq, a novel tool to interrogate DNA methylation at human imprinted regions and diagnose multilocus imprinting disturbance. Genetics in Medicine, 2022, 24, 463-474.	1.1	8
8	Multilocus Inherited Neoplasia Allele Syndrome (MINAS): an update. European Journal of Human Genetics, 2022, 30, 265-270.	1.4	12
9	Evaluation of tumour surveillance protocols and outcomes in von Hippel-Lindau disease in a national health service. British Journal of Cancer, 2022, 126, 1339-1345.	2.9	4
10	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	1.8	14
11	Elongin C (ELOC/TCEB1)-associated von Hippelâ€“Lindau disease. Human Molecular Genetics, 2022, 31, 2728-2737.	1.4	11
12	Investigating the role of somatic sequencing platforms for phaeochromocytoma and paraganglioma in a large UK cohort. Clinical Endocrinology, 2022, 97, 448-459.	1.2	4
13	Fetal central nervous system anomalies: When should we offer exome sequencing?. Prenatal Diagnosis, 2022, 42, 736-743.	1.1	16
14	Frequency of pathogenic germline variants in cancer susceptibility genes in 1336 renal cell carcinoma cases. Human Molecular Genetics, 2022, 31, 3001-3011.	1.4	9
15	Large scale genotypeâ€•and phenotypeâ€•driven machine learning in Von Hippelâ€Lindau disease. Human Mutation, 2022, 43, 1268-1285.	1.1	6
16	Comparison of methylation episignatures in KMT2B- and KMT2D-related human disorders. Epigenomics, 2022, 14, 537-547.	1.0	10
17	Molecular Basis of Beckwithâ€“Wiedemann Syndrome Spectrum with Associated Tumors and Consequences for Clinical Practice. Cancers, 2022, 14, 3083.	1.7	13
18	Constitutional de novo deletion CNV encompassing REST predisposes to diffuse hyperplastic perilobar nephroblastomatosis (HPLN). Journal of Medical Genetics, 2021, 58, 581-585.	1.5	3

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19	Pathogenic germline variants in patients with features of hereditary renal cell carcinoma: Evidence for further locus heterogeneity. Genes Chromosomes and Cancer, 2021, 60, 5-16.	1.5	10
20	Evidence that autosomal recessive spastic cerebral palsy-1 (CPSQ1) is caused by a missense variant in HPDL. Brain Communications, 2021, 3, fcab002.	1.5	8
21	The role of [68ÂGa]Ga-DOTATATE PET/CT in wild-type KIT/PDGFRA gastrointestinal stromal tumours (GIST). EJNMMI Research, 2021, 11, 5.	1.1	4
22	Evidence to Support the Clinical Utility of Prenatal Exome Sequencing in Evaluation of the Fetus with Congenital Anomalies. BJOG: an International Journal of Obstetrics and Gynaecology, 2021, 128, e39-e50.	1.1	23
23	The Diagnostic Yield of Prenatal Genetic Technologies in Congenital Heart Disease: A Prospective Cohort Study. Fetal Diagnosis and Therapy, 2021, 48, 112-119.	0.6	10
24	Predisposition to cancer in children and adolescents. The Lancet Child and Adolescent Health, 2021, 5, 142-154.	2.7	53
25	Familial wild-type gastrointestinal stromal tumour in association with germline truncating variants in both SDHA and PALB2. European Journal of Human Genetics, 2021, 29, 1139-1145.	1.4	1
26	Quantitative evidence evaluation for singleton rare missense variants in rare distinctive adult-onset phenotypes: the exemplar of SDHB and SDHD. Molecular Genetics and Metabolism, 2021, 132, S37.	0.5	0
27	Isolatedâ€•and <scp>Beckwithâ€Wiedemann</scp> syndrome relatedâ€•lateralised overgrowth (hemihypertrophy): Clinical and molecular correlations in 94 individuals. Clinical Genetics, 2021, 100, 292-297.	1.0	7
28	A systematic review assessing the existence of pneumothorax-only variants of FLCN. Implications for lifelong surveillance of renal tumours. European Journal of Human Genetics, 2021, 29, 1595-1600.	1.4	12
29	Wilms tumour surveillance in at-risk children: Literature review and recommendations from the SIOP-Europe Host Genome Working Group and SIOP Renal Tumour Study Group. European Journal of Cancer, 2021, 153, 51-63.	1.3	25
30	Investigation and Management of Apparently Sporadic Central Nervous System Haemangioblastoma for Evidence of Von Hippelâ€“Lindau Disease. Genes, 2021, 12, 1414.	1.0	2
31	Metabolic Drivers in Hereditary Cancer Syndromes. Annual Review of Cancer Biology, 2020, 4, 77-97.	2.3	32
32	Fumarate Metabolic Signature for the Detection of Reed Syndrome in Humans. Clinical Cancer Research, 2020, 26, 391-396.	3.2	11
33	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	0.8	270
34	Hereditary Leiomyomatosis and Renal Cell Cancer: Clinical, Molecular, and Screening Features in a Cohort of 185 Affected Individuals. European Urology Oncology, 2020, 3, 764-772.	2.6	39
35	Genetic stratification of inherited and sporadic phaeochromocytoma and paraganglioma: implications for precision medicine. Human Molecular Genetics, 2020, 29, R128-R137.	1.4	21
36	A review of the tumour spectrum of germline succinate dehydrogenase gene mutations: Beyond phaeochromocytoma and paraganglioma. Clinical Endocrinology, 2020, 93, 528-538.	1.2	36

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37	Dysregulation at multiple points of the kynurenine pathway is a ubiquitous feature of renal cancer: implications for tumour immune evasion. British Journal of Cancer, 2020, 123, 137-147.	2.9	17
38	Cancer Surveillance Guideline for individuals with PTEN hamartoma tumour syndrome. European Journal of Human Genetics, 2020, 28, 1387-1393.	1.4	63
39	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	13.7	338
40	Cancer prevention with aspirin in hereditary colorectal cancer (Lynch syndrome), 10-year follow-up and registry-based 20-year data in the CAPP2 study: a double-blind, randomised, placebo-controlled trial. Lancet, The, 2020, 395, 1855-1863.	6.3	220
41	A recurrent pathogenic variant in <scp>TPM2</scp> reveals further phenotypic and genetic heterogeneity in multiple pterygium syndromeâ€related disorders. Clinical Genetics, 2020, 97, 908-914.	1.0	5
42	Characterization of renal cell carcinomaâ€associated constitutional chromosome abnormalities by genome sequencing. Genes Chromosomes and Cancer, 2020, 59, 333-347.	1.5	10
43	Ensuring that COVID-19 research is inclusive: guidance from the NIHR INCLUDE project. BMJ Open, 2020, 10, e043634.	0.8	24
44	Exome Sequencing for Prenatal Detection of Genetic Abnormalities in Fetal Ultrasound Anomalies: An Economic Evaluation. Fetal Diagnosis and Therapy, 2020, 47, 554-564.	0.6	11
45	Familial Kidney Cancer: Implications of New Syndromes and Molecular Insights. European Urology, 2019, 76, 754-764.	0.9	80
46	SDHC epi-mutation testing in gastrointestinal stromal tumours and related tumours in clinical practice. Scientific Reports, 2019, 9, 10244.	1.6	20
47	Human Genetics and Fetal Disease: Assessment of the Fetal Genome. , 2019, , 36-47.		0
48	Prenatal exome sequencing analysis in fetal structural anomalies detected by ultrasonography (PAGE): a cohort study. Lancet, The, 2019, 393, 747-757.	6.3	443
49	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	6.0	178
50	MethylCal: Bayesian calibration of methylation levels. Nucleic Acids Research, 2019, 47, e81-e81.	6.5	5
51	OTULIN deficiency in ORAS causes cell typeâ€specific LUBAC degradation, dysregulated TNF signalling and cell death. EMBO Molecular Medicine, 2019, 11, .	3.3	80
52	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. Genetical Research, 2019, 101, e3.	0.3	17
53	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	2.6	45
54	von Hippel-Lindau Disease: an Update. Current Genetic Medicine Reports, 2019, 7, 227-235.	1.9	8

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55	Clinical Practice Guidance: Surveillance for phaeochromocytoma and paraganglioma in paediatric succinate dehydrogenase gene mutation carriers. Clinical Endocrinology, 2019, 90, 499-505.	1.2	25
56	Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. Nature Reviews Genetics, 2019, 20, 235-248.	7.7	291
57	Molecular autopsy by trio exome sequencing (ES) and postmortem examination in fetuses and neonates with prenatally identified structural anomalies. Genetics in Medicine, 2019, 21, 1065-1073.	1.1	47
58	Beckwithâ€“Wiedemann Spectrum. , 2019, , 623-633.		0
59	Penetrance estimates for BRCA1, BRCA2 (also applied to Lynch syndrome) based on presymptomatic testing: a new unbiased method to assess risk?. Journal of Medical Genetics, 2018, 55, 442-448.	1.5	1
60	Revisiting Wilms tumour surveillance in Beckwithâ€“Wiedemann syndrome with IC2 methylation loss, reply. European Journal of Human Genetics, 2018, 26, 471-472.	1.4	13
61	Hereditary renal cell carcinoma syndromes: diagnosis, surveillance and management. World Journal of Urology, 2018, 36, 1891-1898.	1.2	105
62	Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	4.3	388
63	Tumour risks and genotypeâ€“phenotype correlations associated with germline variants in succinate dehydrogenase subunit genes SDHB, SDHC and SDHD. Journal of Medical Genetics, 2018, 55, 384-394.	1.5	177
64	CNVs affecting cancer predisposing genes (CPGs) detected as incidental findings in routine germline diagnostic chromosomal microarray (CMA) testing. Journal of Medical Genetics, 2018, 55, 89-96.	1.5	7
65	Translating In Vivo Metabolomic Analysis of Succinate Dehydrogenaseâ€“Deficient Tumors Into Clinical Utility. JCO Precision Oncology, 2018, 2, 1-12.	1.5	22
66	Rapid disease progression in a patient with mismatch repair-deficient and cortisol secreting adrenocortical carcinoma treated with pembrolizumab. Seminars in Oncology, 2018, 45, 151-155.	0.8	19
67	Bayesian approach to determining penetrance of pathogenic SDH variants. Journal of Medical Genetics, 2018, 55, 729-734.	1.5	44
68	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
69	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. American Journal of Human Genetics, 2018, 103, 3-18.	2.6	46
70	Homozygous loss-of-function mutations in SLC26A7 cause goitrous congenital hypothyroidism. JCI Insight, 2018, 3, .	2.3	44
71	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	2.6	26
72	PLAA Mutations Cause a Lethal Infantile Epileptic Encephalopathy by Disrupting Ubiquitin-Mediated Endolysosomal Degradation of Synaptic Proteins. American Journal of Human Genetics, 2017, 100, 706-724.	2.6	37

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73	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	0.7	36
74	<scp>SDHA</scp> related tumorigenesis: a new case series and literature review for variant interpretation and pathogenicity. Molecular Genetics & Genomic Medicine, 2017, 5, 237-250.	0.6	46
75	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	2.6	343
76	Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
77	Estimating the human mutation rate from autozygous segments reveals population differences in human mutational processes. Nature Communications, 2017, 8, 303.	5.8	81
78	A case of a metastatic SDHA mutated paraganglioma re-presenting twenty-three years after initial surgery. Endocrine-Related Cancer, 2017, 24, L69-L71.	1.6	10
79	Clinical and Molecular Features of Renal and Pheochromocytoma/Paraganglioma Tumor Association Syndrome (RAPTAS): Case Series and Literature Review. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 4013-4022.	1.8	35
80	Cover Image, Volume 173A, Number 7, July 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
81	Surveillance Recommendations for Children with Overgrowth Syndromes and Predisposition to Wilms Tumors and Hepatoblastoma. Clinical Cancer Research, 2017, 23, e115-e122.	3.2	140
82	Consensus Statement on next-generation-sequencing-based diagnostic testing of hereditary phaeochromocytomas and paragangliomas. Nature Reviews Endocrinology, 2017, 13, 233-247.	4.3	198
83	Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13.	1.0	101
84	Human biallelic MFN2 mutations induce mitochondrial dysfunction, upper body adipose hyperplasia, and suppression of leptin expression. ELife, 2017, 6, .	2.8	60
85	Characterization of endolymphatic sac tumors and von Hippelâ€“Lindau disease in the International Endolymphatic Sac Tumor Registry. Head and Neck, 2016, 38, E673-9.	0.9	48
86	Causes and Consequences of Multi-Locus Imprinting Disturbances in Humans. Trends in Genetics, 2016, 32, 444-455.	2.9	81
87	Diagnosis and Management of Hereditary Renal Cell Cancer. Recent Results in Cancer Research, 2016, 205, 85-104.	1.8	10
88	Von Hippel-Lindau Disease: Genetics and Role of Genetic Counseling in a Multiple Neoplasia Syndrome. Journal of Clinical Oncology, 2016, 34, 2172-2181.	0.8	132
89	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silverâ€“Russell and Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1377-1387.	1.4	68
90	The Deubiquitinase OTULIN Is an Essential Negative Regulator of Inflammation and Autoimmunity. Cell, 2016, 166, 1215-1230.e20.	13.5	259

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91	Fumarate is an epigenetic modifier that elicits epithelial-to-mesenchymal transition. Nature, 2016, 537, 544-547.	13.7	443
92	Comprehensive Screening of Eight Known Causative Genes in Congenital Hypothyroidism With Gland-in-Situ. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 4521-4531.	1.8	82
93	Phenotype, cancer risk, and surveillance in Beckwithâ€“Wiedemann syndrome depending on molecular genetic subgroups. American Journal of Medical Genetics, Part A, 2016, 170, 2248-2260.	0.7	163
94	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€“dystonia. Nature Communications, 2016, 7, 11601.	5.8	233
95	Expression and knockdown of zebrafish folliculin suggests requirement for embryonic brain morphogenesis. BMC Developmental Biology, 2016, 16, 23.	2.1	3
96	Defective Leukocyte Adhesion and Chemotaxis Contributes to Combined Immunodeficiency in Humans with Autosomal Recessive MST1 Deficiency. Journal of Clinical Immunology, 2016, 36, 117-122.	2.0	63
97	Multilocus Inherited Neoplasia Alleles Syndrome. JAMA Oncology, 2016, 2, 373.	3.4	43
98	Health and population effects of rare gene knockouts in adult humans with related parents. Science, 2016, 352, 474-477.	6.0	272
99	Germline ESR2 mutation predisposes to medullary thyroid carcinoma and causes up-regulation of RET expression. Human Molecular Genetics, 2016, 25, 1836-1845.	1.4	28
100	Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793.	1.4	44
101	Hypoxia, Hypoxia-inducible Transcription Factors, and Renal Cancer. European Urology, 2016, 69, 646-657.	0.9	249
102	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123.	1.8	174
103	Profiling of Somatic Mutations in Phaeochromocytoma and Paraganglioma by Targeted Next Generation Sequencing Analysis. International Journal of Endocrinology, 2015, 2015, 1-8.	0.6	64
104	Renal Cell Carcinoma: Overview. , 2015, , 337-344.		0
105	Germline Mutations in the CDKN2B Tumor Suppressor Gene Predispose to Renal Cell Carcinoma. Cancer Discovery, 2015, 5, 723-729.	7.7	88
106	A truncating TPO mutation (Y55X) in patients with hypothyroidism and total iodide organification defect. Endocrine Research, 2015, 40, 146-150.	0.6	8
107	Heterogeneous Genetic Background of the Association of Pheochromocytoma/Paraganglioma and Pituitary Adenoma: Results From a Large Patient Cohort. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E531-E541.	1.8	145
108	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. Clinical Epigenetics, 2015, 7, 23.	1.8	23

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109	Recurrent chromosomal gains and heterogeneous driver mutations characterise papillary renal cancer evolution. Nature Communications, 2015, 6, 6336.	5.8	100
110	15 YEARS OF PARAGANGLIOMA: Genetics and mechanism of pheochromocytomaâ€“paraganglioma syndromes characterized by germline SDHB and SDHD mutations. Endocrine-Related Cancer, 2015, 22, T71-T82.	1.6	52
111	Obesity, Aspirin, and Risk of Colorectal Cancer in Carriers of Hereditary Colorectal Cancer: A Prospective Investigation in the CAPP2 Study. Journal of Clinical Oncology, 2015, 33, 3591-3597.	0.8	91
112	Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. Nature Communications, 2015, 6, 8086.	5.8	134
113	VHL, the story of a tumour suppressor gene. Nature Reviews Cancer, 2015, 15, 55-64.	12.8	572
114	A clinical and genetic analysis of multiple primary cancer referrals to genetics services. European Journal of Human Genetics, 2015, 23, 581-587.	1.4	21
115	A nonsense thyrotropin receptor gene mutation (R609X) is associated with congenital hypothyroidism and heart defects. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 1101-5.	0.4	10
116	A deletion including exon 2 of the TSHR gene is associated with thyroid dysgenesis and severe congenital hypothyroidism. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 731-5.	0.4	10
117	An essential splice site mutation (c.317+1G>A) in the TSHR gene leads to severe thyroid dysgenesis. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 1021-5.	0.4	7
118	A common thyroid peroxidase gene mutation (G319R) in Turkish patients with congenital hypothyroidism could be due to a founder effect. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 383-7.	0.4	12
119	A truncating DUOX2 mutation (R434X) causes severe congenital hypothyroidism. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 323-7.	0.4	17
120	Germline mutations in RYR1 are associated with foetal akinesia deformation sequence/lethal multiple pterygium syndrome. Acta Neuropathologica Communications, 2014, 2, 148.	2.4	23
121	Hereditary leiomyomatosis and renal cell cancer (HLRCC): renal cancer risk, surveillance and treatment. Familial Cancer, 2014, 13, 637-644.	0.9	251
122	Germline FH Mutations Presenting With Pheochromocytoma. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2046-E2050.	1.8	147
123	Knockdown of Slingshot 2 (SSH2) serine phosphatase induces Caspase3 activation in human carcinoma cell lines with the loss of the Birtâ€“Hoggâ€“DubÃ© tumour suppressor gene (FLCN). Oncogene, 2014, 33, 956-965.	2.6	8
124	One Base Deletion (c.2422delT) in the TPO Gene Causes Severe Congenital Hypothyroidism. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2014, 6, 169-173.	0.4	13
125	CDKN1C mutations: two sides of the same coin. Trends in Molecular Medicine, 2014, 20, 614-622.	3.5	89
126	Clinical utility gene card for: von Hippelâ€“Lindau (VHL). European Journal of Human Genetics, 2014, 22, 572-572.	1.4	15

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127	How does altering the resolution of chromosomal microarray analysis in the prenatal setting affect the rates of pathological and uncertain findings?. Journal of Maternal-Fetal and Neonatal Medicine, 2014, 27, 649-657.	0.7	26
128	BAC Chromosomal Microarray for Prenatal Detection of Chromosome Anomalies in Fetal Ultrasound Anomalies: An Economic Evaluation. Fetal Diagnosis and Therapy, 2014, 36, 49-58.	0.6	4
129	Phaeochromocytoma and paraganglioma: next-generation sequencing and evolving Mendelian syndromes. Clinical Medicine, 2014, 14, 440-444.	0.8	7
130	Exome sequencing improves genetic diagnosis of structural fetal abnormalities revealed by ultrasound. Human Molecular Genetics, 2014, 23, 3269-3277.	1.4	164
131	The tumor susceptibility gene TMEM127â€Š is mutated in renal cell carcinomas and modulates endolysosomal function. Human Molecular Genetics, 2014, 23, 2428-2439.	1.4	55
132	Clinical utility gene card for: Beckwithâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	1.4	50
133	Novel truncating thyroglobulin gene mutations associated with congenital hypothyroidism. Endocrine, 2014, 45, 206-212.	1.1	25
134	A Practical Guide to Human Cancer Genetics. , 2014, , .		8
135	Three Different Cone Opsin Gene Array Mutational Mechanisms; Genotype-Phenotype Correlation and Functional Investigation of Cone Opsin Variants. Human Mutation, 2014, 35, n/a-n/a.	1.1	53
136	Methylation analysis and diagnostics of Beckwith-Wiedemann syndrome in 1,000 subjects. Clinical Epigenetics, 2014, 6, 11.	1.8	100
137	Aberrant DNA hypermethylation of SDHC: a novel mechanism of tumor development in Carney triad. Endocrine-Related Cancer, 2014, 21, 567-577.	1.6	161
138	Telomerase reverse transcriptase promoter mutations in tumors originating from the adrenal gland and extra-adrenal paraganglia. Endocrine-Related Cancer, 2014, 21, 653-661.	1.6	39
139	Urinary System. , 2014, , 137-144.		0
140	A combination of mutations in AKR1D1 and SKIV2L in a family with severe infantile liver disease. Orphanet Journal of Rare Diseases, 2013, 8, 74.	1.2	17
141	Thyroid dyshormonogenesis is mainly caused by <scp>TPO</scp> mutations in consanguineous community. Clinical Endocrinology, 2013, 79, 275-281.	1.2	47
142	The development of a clinical screening tool for tumour predisposition syndromes in childhood cancer patients. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 52-52.	0.0	0
143	Cancer of the Kidney and Urogenital Tract. , 2013, , 1-17.		0
144	The development of a clinical screening instrument for tumour predisposition syndromes in childhood cancer patients. European Journal of Cancer, 2013, 49, 3247-3254.	1.3	18

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145	Acrocallosal syndrome: Identification of a novel KIF7 mutation and evidence for oligogenic inheritance. European Journal of Medical Genetics, 2013, 56, 39-42.	0.7	21
146	Genomics and epigenomics of renal cell carcinoma. Seminars in Cancer Biology, 2013, 23, 10-17.	4.3	78
147	Evaluation of <scp>SDHB</scp>,<scp> SDHD</scp> and <scp>VHL</scp> gene susceptibility testing in the assessment of individuals with nonâ€syndromic phaeochromocytoma, paraganglioma and head and neck paraganglioma. Clinical Endocrinology, 2013, 78, 898-906.	1.2	62
148	Methylation profiling and evaluation of demethylating therapy in renal cell carcinoma. Clinical Epigenetics, 2013, 5, 16.	1.8	33
149	Epimutation profiling in Beckwith-Wiedemann syndrome: relationship with assisted reproductive technology. Clinical Epigenetics, 2013, 5, 23.	1.8	42
150	Combined <scp>NGS</scp> Approaches Identify Mutations in the Intraflagellar Transport Gene IFT140 in Skeletal Ciliopathies with Early Progressive Kidney Disease. Human Mutation, 2013, 34, 714-724.	1.1	120
151	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	1.1	114
152	The use of chromosomal microarray in prenatal diagnosis. The Obstetrician and Gynaecologist, 2013, 15, 80-84.	0.2	7
153	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	2.6	196
154	DNA methylation profiling distinguishes histological subtypes of renal cell carcinoma. Epigenetics, 2013, 8, 252-267.	1.3	40
155	DNA methylation profiles of long- and short-term glioblastoma survivors. Epigenetics, 2013, 8, 149-156.	1.3	108
156	Pheochromocytoma. Clinical Chemistry, 2013, 59, 466-472.	1.5	29
157	Perlman Syndrome: Overgrowth, Wilms Tumor Predisposition andDIS3L2. , 2013, 163, n/a-n/a.		1
158	Genes, assisted reproductive technology and trans-illumination. Epigenomics, 2013, 5, 331-340.	1.0	11
159	Perlman Syndrome: Overgrowth, Wilms Tumor Predisposition and DIS3L2. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 106-113.	0.7	40
160	Use of prenatal chromosomal microarray: prospective cohort study and systematic review and meta-analysis. Ultrasound in Obstetrics and Gynecology, 2013, 41, 610-620.	0.9	249
161	Functional epigenetic approach identifies frequently methylated genes in Ewing sarcoma. Epigenetics, 2013, 8, 1198-1204.	1.3	38
162	Increased Rate of Phenocopies in All Age Groups in BRCA1/BRCA2 Mutation Kindred, but Increased Prospective Breast Cancer Risk Is Confined to BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 2269-2276.	1.1	13

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163	A Comprehensive Next Generation Sequencingâ€“Based Genetic Testing Strategy To Improve Diagnosis of Inherited Pheochromocytoma and Paraganglioma. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1248-E1256.	1.8	92
164	Comparison of the clinical scoring systems in Silverâ€“Russell syndrome and development of modified diagnostic criteria to guide molecular genetic testing. Journal of Medical Genetics, 2013, 50, 635-639.	1.5	22
165	Canadian guideline on genetic screening for hereditary renal cell cancers. Canadian Urological Association Journal, 2013, 7, 319.	0.3	30
166	<scp>UBE</scp> 2 <scp>QL</scp> 1 is Disrupted by a Constitutional Translocation Associated with Renal Tumor Predisposition and is a Novel Candidate Renal Tumor Suppressor Gene. Human Mutation, 2013, 34, 1650-1661.	1.1	18
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