Donald Love

List of Publications by Year in descending order

159 papers	9,124 citations	61945 43 h-index	40954 93 g-index
159 all docs	159 docs citations	159 times ranked	9455 citing authors

#	Article	IF	Citations
1	Germ-line mutations of the RET proto-oncogene in multiple endocrine neoplasia type 2A. Nature, 1993, 363, 458-460.	13.7	1,886
2	A Prospective Study of Sudden Cardiac Death among Children and Young Adults. New England Journal of Medicine, 2016, 374, 2441-2452.	13.9	619
3	Validation of Zebrafish (Danio rerio) Reference Genes for Quantitative Real-time RT-PCR Normalization. Acta Biochimica Et Biophysica Sinica, 2007, 39, 384-390.	0.9	540
4	An autosomal transcript in skeletal muscle with homology to dystrophin. Nature, 1989, 339, 55-58.	13.7	501
5	Human dystrophin expression in mdx mice after intramuscular injection of DNA constructs. Nature, 1991, 352, 815-818.	13.7	501
6	A mutation in the Î± tropomyosin gene TPM3 associated with autosomal dominant nemaline myopathy. Nature Genetics, 1995, 9, 75-79.	9.4	315
7	Direct Interaction between Emerin and Lamin A. Biochemical and Biophysical Research Communications, 2000, 267, 709-714.	1.0	226
8	Utility of Post-Mortem Genetic Testing in Cases of Sudden Arrhythmic Death Syndrome. Journal of the American College of Cardiology, 2017, 69, 2134-2145.	1.2	219
9	A mutation in the gene TNFRSF11B encoding osteoprotegerin causes an idiopathic hyperphosphatasia phenotype. Human Molecular Genetics, 2002, 11, 2119-2127.	1.4	190
10	Prospective, population-based long QT molecular autopsy study of postmortem negative sudden death in 1 to 40 year olds. Heart Rhythm, 2011, 8, 412-419.	0.3	148
11	Chemical discovery and global gene expression analysis in zebrafish. Nature Biotechnology, 2003, 21, 879-883.	9.4	142
12	Reduction in enkephalin and substance P messenger RNA in the striatum of early grade Huntington's disease: A detailed cellularin situ hybridization study. Neuroscience, 1996, 72, 1023-1036.	1.1	141
13	Zebrafish: bridging the gap between development and disease. Human Molecular Genetics, 2000, 9, 2443-2449.	1.4	137
14	Characterization of a 4.8kb transcript from the Duchenne muscular dystrophy locus expressed in Schwannoma cells. Human Molecular Genetics, 1992, 1, 103-109.	1.4	125
15	Idiopathic Hyperphosphatasia andTNFRSF11BMutations: Relationships Between Phenotype and Genotype. Journal of Bone and Mineral Research, 2003, 18, 2095-2104.	3.1	113
16	Technology for high-throughput screens: the present and future using zebrafish. Current Opinion in Biotechnology, 2004, 15, 564-571.	3.3	102
17	Genetic linkage studies map the multiple endocrine neoplasia type 2 loci to a small interval on chromosome 10q11.2. Human Molecular Genetics, 1993, 2, 241-246.	1.4	100
18	Decreased neuronal nitric oxide synthase messenger RNA and somatostatin messenger RNA in the striatum of Huntington's disease. Neuroscience, 1996, 72, 1037-1047.	1.1	96

#	Article	IF	Citations
19	Trinucleotide (CAG) repeat length is positively correlated with the degree of DNA fragmentation in Huntington's disease striatum. Neuroscience, 1998, 87, 49-53.	1.1	94
20	Distribution of emerin and lamins in the heart and implications for Emery-Dreifuss muscular dystrophy. Human Molecular Genetics, 1999, 8, 353-359.	1.4	90
21	Identification of large gene deletions and duplications in KCNQ1 and KCNH2 in patients with long QT syndrome. Heart Rhythm, 2008, 5, 1275-1281.	0.3	79
22	Short interfering RNA-mediated gene targeting in the zebrafish. FEBS Letters, 2004, 561, 89-93.	1.3	74
23	Assessment of the predictive accuracy of five in silico prediction tools, alone or in combination, and two metaservers to classify long QT syndrome gene mutations. BMC Medical Genetics, 2015, 16, 34.	2.1	73
24	Retroviral-mediated transfer of a dystrophin minigene intomdxmouse myoblasts in vitro. FEBS Letters, 1992, 296, 128-134.	1.3	72
25	Gene flow on the ice: genetic differentiation among AdÃ©lie penguin colonies around Antarctica. Molecular Ecology, 2001, 10, 1645-1656.	2.0	71
26	Sequence structure and expression of a cloned Î²-glucosidase gene from an extreme thermophile. Molecular Genetics and Genomics, 1988, 213, 84-92.	2.4	69
27	Human dystrophin expression corrects the myopathic phenotype in transgenic mdx mice. Human Molecular Genetics, 1992, 1, 35-40.	1.4	67
28	Apparent association of mental retardation and specific patterns of deletions screened with probes cf56a and cf23a in Duchenne muscular dystrophy. American Journal of Medical Genetics Part A, 1991, 39, 437-441.	2.4	66
29	Phenotypic expansion and further characterisation of the 17q21.31 microdeletion syndrome. Journal of Medical Genetics, 2009, 46, 480-489.	1.5	66
30	Genetics and Potential Biotechnological Applications of Thermophilic and Extremely Thermophilic Microorganisms. Biotechnology and Genetic Engineering Reviews, 1987, 5, 199-244.	2.4	64
31	Localisation of a dystrophin-related autosomal gene to 6q24 in man, and to mouse chromosome 10 in the region of the dystrophia muscularis (dy) locus. Human Genetics, 1990, 85, 324-6.	1.8	64
32	Nucleotide sequence of a gene fromCaldocellum saccharolyticumencoding for exocellulase and endocellulase activity. Nucleic Acids Research, 1989, 17, 439-439.	6.5	61
33	Dystrophin and dystrophin-related proteins: A review of protein and RNA studies. Neuromuscular Disorders, 1993, 3, 5-21.	0.3	59
34	Role of gut microbiota in Crohnâ€™s disease. Expert Review of Gastroenterology and Hepatology, 2009, 3, 535-546.	1.4	57
35	Posthumous diagnosis of long QT syndrome from neonatal screening cards. Heart Rhythm, 2010, 7, 481-486.	0.3	56
36	Community detection of long QT syndrome with a clinical registry: An alternative to ECG screening programs?. Heart Rhythm, 2013, 10, 233-238.	0.3	56

#	Article	IF	Citations
37	Sequences of junction fragments in the deletion-prone region of the dystrophin gene. Genomics, 1991, 10, 57-67.	1.3	53
38	Single nucleotide polymorphisms in arrhythmia genes modify the risk of cardiac events and sudden death in long QT syndrome. Heart Rhythm, 2014, 11, 76-82.	0.3	53
39	Monoclonal antibodies against defined regions of the muscular dystrophy protein, dystrophin. FEBS Letters, 1990, 262, 237-240.	1.3	52
40	Association of vitamin D receptor gene polymorphisms with insulin resistance and response to vitamin D. Metabolism: Clinical and Experimental, 2012, 61, 293-301.	1.5	51
41	Human dystrophin gene transfer: production and expression of a functional recombinant DNA-based gene. Human Genetics, 1991, 88, 53-58.	1.8	47
42	Zebrafish as a model for long QT syndrome: the evidence and the means of manipulating zebrafish gene expression. Acta Physiologica, 2010, 199, 257-276.	1.8	47
43	Visualization, characterization and modulation of calcium signaling during the development of slow muscle cells in intact zebrafish embryos. International Journal of Developmental Biology, 2011, 55, 153-174.	0.3	46
44	The p.Ala510Val mutation in the SPG7 (paraplegin) gene is the most common mutation causing adult onset neurogenetic disease in patients of British ancestry. Journal of Neurology, 2013, 260, 1286-1294.	1.8	45
45	Evaluation of Bioinformatic Programmes for the Analysis of Variants within Splice Site Consensus Regions. Advances in Bioinformatics, 2016, 2016, 1-10.	5.7	43
46	Brain dopamineâ€serotonin vesicular transport disease presenting as a severe infantile hypotonic parkinsonian disorder. Journal of Inherited Metabolic Disease, 2016, 39, 305-308.	1.7	41
47	Dystrophin in Adult Zebrafish Muscle. Biochemical and Biophysical Research Communications, 2001, 286, 478-483.	1.0	38
48	The yield of postmortem genetic testing in sudden death cases with structural findings at autopsy. European Journal of Human Genetics, 2020, 28, 17-22.	1.4	38
49	Long QT molecular autopsy in sudden infant death syndrome. Archives of Disease in Childhood, 2014, 99, 635-640.	1.0	36
50	Expression of leucine genes from an extremely thermophilic bacterium in Escherichia coli. Molecular Genetics and Genomics, 1987, 210, 490-497.	2.4	35
51	A novel microdeletion syndrome at 9q21.13 characterised by mental retardation, speech delay, epilepsy and characteristic facial features. European Journal of Medical Genetics, 2013, 56, 163-170.	0.7	35
52	A YAC contig in Xp21 containing the adrenal hypoplasia congenita and glycerol kinase deficiency genes. Human Molecular Genetics, 1992, 1, 579-585.	1.4	33
53	Detection of Sequence Variations in the Adenomatous Polyposis Coli (APC) Gene Using Denaturing High-Performance Liquid Chromatography. Genetic Testing and Molecular Biomarkers, 2001, 5, 281-290.	1.7	32
54	Melanocortin-3 receptor gene variants in a Maori kindred with obesity and early onset type 2 diabetes. Diabetes Research and Clinical Practice, 2002, 58, 61-71.	1.1	32

#	Article	IF	Citations
55	Characterization of deletions in the dystrophin gene giving mild phenotypes. American Journal of Medical Genetics Part A, 1990, 37, 136-142.	2.4	31
56	Identification and expression analysis of kcnh2 genes in the zebrafish. Biochemical and Biophysical Research Communications, 2010, 396, 817-824.	1.0	30
57	Quantitative Real-Time RT-PCR (qRT-PCR) of Zebrafish Transcripts: Optimization of RNA Extraction, Quality Control Considerations, and Data Analysis. Cold Spring Harbor Protocols, 2009, 2009, pdb.prot5314.	0.2	29
58	Kiwifruit extracts inhibit cytokine production by lipopolysaccharide-activated macrophages, and intestinal epithelial cells isolated from IL10 gene deficient mice. Cellular Immunology, 2011, 270, 70-79.	1.4	29
59	Analysis of the TGF Î² functional pathway in epithelial ovarian carcinoma. British Journal of Cancer, 2001, 85, 687-691.	2.9	28
60	A Nationwide, Population-Based Prevalence Study of Genetic Muscle Disorders. Neuroepidemiology, 2019, 52, 128-135.	1.1	27
61	Elevated serum gastrin levels in Jervell and Lange-Nielsen syndrome: A marker of severe KCNQ1 dysfunction?. Heart Rhythm, 2011, 8, 551-554.	0.3	26
62	Distinct dystrophin mRNA species are expressed in embryonic and adult mouse skeletal muscle. FEBS Letters, 1988, 242, 47-52.	1.3	25
63	Sarcoglycans of the zebrafish: orthology and localization to the sarcolemma and myosepta of muscle. Biochemical and Biophysical Research Communications, 2003, 303, 488-495.	1.0	25
64	The clinical utility of molecular diagnostic testing for primary immune deficiency disorders: a case based review. Allergy, Asthma and Clinical Immunology, 2010, 6, 12.	0.9	25
65	Spectrum of mutations in sarcoglycan genes in the Mumbai region of western India: High prevalence of 525del T. Neurology India, 2009, 57, 406.	0.2	25
66	Comparative semi-automated analysis of (CAG) repeats in the Huntington disease gene: use of internal standards. Molecular and Cellular Probes, 1999, 13, 283-289.	0.9	24
67	Activin is a potent growth suppressor of epithelial ovarian cancer cells. Cancer Letters, 2009, 285, 157-165.	3.2	24
68	Effects of kiwifruit extracts on colonic gene and protein expression levels in IL-10 gene-deficient mice. British Journal of Nutrition, 2012, 108, 113-129.	1.2	24
69	Long QT molecular autopsy in sudden unexplained death in the young (1-40 years old): Lessons learnt from an eight year experience in New Zealand. PLoS ONE, 2018, 13, e0196078.	1.1	24
70	Molecular Cloning of a Î²-Glucosidase Gene from an Extremely Thermophilic Anaerobe in E. coli and B. subtilis. Nature Biotechnology, 1987, 5, 384-387.	9.4	23
71	Compound Heterozygous Inheritance of Mutations in Coenzyme Q8A Results in Autosomal Recessive Cerebellar Ataxia and Coenzyme Q10 Deficiency in a Female Sib-Pair. JIMD Reports, 2017, 42, 31-36.	0.7	21
72	Physical mapping distal to the DMD locus. Genomics, 1990, 8, 106-112.	1.3	20

#	Article	IF	Citations
73	The New Zealand Neuromuscular Disease Registry. Journal of Clinical Neuroscience, 2012, 19, 1749-1750.	0.8	20
74	Compound heterozygous SLC19A3 mutations further refine the critical promoter region for biotin-thiamine-responsive basal ganglia disease. Journal of Physical Education and Sports Management, 2017, 3, a001909.	0.5	20
75	Null Alleles at the Huntington Disease Locus: Implications for Diagnostics and CAG Repeat Instability. Genetic Testing and Molecular Biomarkers, 2000, 4, 55-60.	1.7	18
76	Amino-Terminal Microdeletion within theCNTNAP2Gene Associated with Variable Expressivity of Speech Delay. Case Reports in Genetics, 2012, 2012, 1-4.	0.1	17
77	Microduplication of 3p26.3 Implicated in Cognitive Development. Case Reports in Genetics, 2014, 2014, 1-6.	0.1	17
78	Detection of sudden death syndromes in New Zealand. New Zealand Medical Journal, 2016, 129, 67-74.	0.5	16
79	Novel mutation in the <scp>TMEM</scp>127 gene associated with phaeochromocytoma. Internal Medicine Journal, 2013, 43, 449-451.	0.5	14
80	Diabetic Dead-in-Bed Syndrome: A Possible Link to a Cardiac Ion Channelopathy. Case Reports in Medicine, 2014, 2014, 1-5.	0.3	14
81	Application of complementary luminescent and fluorescent imaging techniques to visualize nuclear and cytoplasmic Ca²⁺ signalling during the in vivo differentiation of slow muscle cells in zebrafish embryos under normal and dystrophic conditions. Clinical and Experimental Pharmacology and Physiology, 2012, 39, 78-86.	0.9	12
82	EGFR Mutation Testing of non-squamous NSCLC: Impact and Uptake during Implementation of Testing Guidelines in a Population-Based Registry Cohort from Northern New Zealand. Targeted Oncology, 2017, 12, 663-675.	1.7	12
83	Lung cancer mutation testing: a clinical retesting study of agreement between a real-time PCR and a mass spectrometry test. Oncotarget, 2017, 8, 101437-101451.	0.8	12
84	Modeling inflammatory bowel disease: the zebrafish as a way forward. Expert Review of Molecular Diagnostics, 2007, 7, 177-193.	1.5	11
85	Targeted mutagenesis of zebrafish: Use of zinc finger nucleases. Birth Defects Research Part C: Embryo Today Reviews, 2011, 93, 249-255.	3.6	11
86	The natural history of elevated tetradecenoylâ€Lâ€carnitine detected by newborn screening in New Zealand: implications for very long chain acylâ€CoA dehydrogenase deficiency screening and treatment. Journal of Inherited Metabolic Disease, 2016, 39, 409-414.	1.7	11
87	Observations on the Natural History of Camurati-Engelmann Disease. Journal of Bone and Mineral Research, 2019, 34, 875-882.	3.1	11
88	Penetrance and expressivity of the R858H CACNA1C variant in a fiveâ€generation pedigree segregating an arrhythmogenic channelopathy. Molecular Genetics & Genomic Medicine, 2019, 7, e00476.	0.6	11
89	Linkage analysis in families with autosomal recessive limb-girdle muscular dystrophy (LGMD) and 6q probes flanking the dystrophin-related sequence. American Journal of Medical Genetics Part A, 1991, 38, 140-146.	2.4	10
90	Citrullinaemia type I: A common mutation in the Pacific Island population. Journal of Paediatrics and Child Health, 2011, 47, 262-265.	0.4	10

#	Article	IF	Citations
91	Fluorescent Function-Spacer-Lipid Construct Labelling Allows for Real-Time in Vivo Imaging of Cell Migration and Behaviour in Zebrafish (Danio Rerio). Journal of Fluorescence, 2012, 22, 1055-1063.	1.3	10
92	Frequency and genetic spectrum of maturity-onset diabetes of the young (MODY) in southern New Zealand. Journal of Diabetes and Metabolic Disorders, 2013, 12, 46.	0.8	10
93	A Novel Glycine Decarboxylase Gene Mutation in an Indian Family With Nonketotic Hyperglycinemia. Journal of Child Neurology, 2014, 29, 122-127.	0.7	10
94	A Turner Syndrome Patient Carrying a Mosaic Distal X Chromosome Marker. Case Reports in Genetics, 2014, 2014, 1-5.	0.1	10
95	12q14 Microdeletions: Additional Case Series with Confirmation of a Macrocephaly Region. Case Reports in Genetics, 2015, 2015, 1-7.	0.1	10
96	Development of a cardiac inherited disease service and clinical registry: A 15-year perspective. American Heart Journal, 2019, 209, 126-130.	1.2	10
97	Molecular characterization of further dystrophin gene microsatellites. Molecular and Cellular Probes, 1995, 9, 361-370.	0.9	9
98	Structure and Location of the Murine Adrenoleukodystrophy Gene. Genomics, 1996, 32, 395-400.	1.3	9
99	Screening for anaplastic lymphoma kinase (ALK) gene rearrangements in nonâ€smallâ€cell lung cancer in New Zealand. Internal Medicine Journal, 2020, 50, 716-725.	0.5	9
100	In VivoTesting of MicroRNA-Mediated Gene Knockdown in Zebrafish. Journal of Biomedicine and Biotechnology, 2012, 2012, 1-7.	3.0	8
101	Delineation of 2q32q35 Deletion Phenotypes: Two Apparent â€œProximalâ€•and â€œDistalâ€•Syndromes. Case Reports in Genetics, 2013, 2013, 1-8.	0.1	8
102	A Case of 17q21.31 Microduplication and 7q31.33 Microdeletion, Associated with Developmental Delay, Microcephaly, and Mild Dysmorphic Features. Case Reports in Genetics, 2014, 2014, 1-6.	0.1	8
103	Simple Repeat-Primed PCR Analysis of the Myotonic Dystrophy Type 1 Gene in a Clinical Diagnostics Environment. Journal of Neurodegenerative Diseases, 2013, 2013, 1-8.	1.1	8
104	A multiple interval physical map of the pericentromeric region of human chromosome 10. Human Genetics, 1994, 93, 313-318.	1.8	7
105	Pure Duplication of the Distal Long Arm of Chromosome 15 with Ebstein Anomaly and Clavicular Anomaly. Case Reports in Genetics, 2011, 2011, 1-5.	0.1	7
106	Expression of a Mutant kcnj2 Gene Transcript in Zebrafish. , 2014, 2014, 1-14.		7
107	Array Comparative Genomic Hybridization Identifies a Heterozygous Deletion of the Entire KCNJ2 Gene as a Cause of Sudden Cardiac Death. Circulation: Cardiovascular Genetics, 2014, 7, 17-22.	5.1	7
108	Next-generation sequencing using microfluidic PCR enrichment for molecular autopsy. BMC Cardiovascular Disorders, 2019, 19, 174.	0.7	7

#	Article	IF	Citations
109	The Diagnosis of Choriocarcinoma in Molar Pregnancies: A Revised Approach in Clinical Testing. Journal of Clinical Medicine Research, 2015, 7, 961-966.	0.6	7
110	Modeling Human Disease by Gene Targeting. Methods in Cell Biology, 2004, 76, 593-612.	0.5	6
111	Pseudotrisomy 13 syndrome: Use of homozygosity mapping to target candidate genes. Gene, 2011, 486, 37-40.	1.0	6
112	Disease Modeling by Gene Targeting Using MicroRNAs. Methods in Cell Biology, 2011, 105, 419-436.	0.5	6
113	Bacterial artificial chromosomes (BACs)-on-Beadsâ„¢ as a diagnostic platform for the rapid aneuploidy screening of products of conception. Molecular Medicine Reports, 2013, 8, 650-654.	1.1	6
114	A Streamlined Protocol for Molecular Testing of the DMD Gene within a Diagnostic Laboratory: A Combination of Array Comparative Genomic Hybridization and Bidirectional Sequence Analysis. ISRN Neurology, 2013, 2013, 1-7.	1.5	6
115	Congestive myeloradiculopathy in a patient with Cowden syndrome. Journal of Clinical Neuroscience, 2015, 22, 431-433.	0.8	6
116	Merosin-deficient congenital muscular dystrophy: A novel homozygous mutation in the laminin-2 gene. Journal of Clinical Neuroscience, 2015, 22, 1983-1985.	0.8	6
117	Microarray testing in clinical diagnosis: an analysis of 5,300 New Zealand patients. Molecular Cytogenetics, 2016, 9, 29.	0.4	6
118	Automation of a primer design and evaluation pipeline for subsequent sequencing of the coding regions of all human Refseq genes. Bioinformation, 2012, 8, 365-368.	0.2	6
119	Global gene expression analysis in the zebrafish: the challenge and the promise. Drug Discovery Today: Technologies, 2004, 1, 79-84.	4.0	5
120	SNP Analysis and Whole Exome Sequencing: Their Application in the Analysis of a Consanguineous Pedigree Segregating Ataxia. Microarrays (Basel, Switzerland), 2015, 4, 490-502.	1.4	5
121	Array comparative genomic hybridization identifies a heterozygous deletion of exon 3 of theRYR2gene. Upsala Journal of Medical Sciences, 2015, 120, 190-197.	0.4	5
122	Two Novel GLDC Mutations in a Neonate with Nonketotic Hyperglycinemia. Journal of Pediatric Genetics, 2016, 05, 174-180.	0.3	5
123	Array-Based Identification of Copy Number Changes in a Diagnostic Setting : Simultaneous Gene - Focused and Low Resolution Whole Human Genome Analysis. Sultan Qaboos University Medical Journal, 2013, 13, 69-79.	0.3	5
124	Expression of a Mutant kcnj2 Gene Transcript in Zebrafish. , 2013, 2013, 324839.		5
125	Distal 5q deletion with associated parietal foramina. Clinical Dysmorphology, 2010, 19, 43-47.	0.1	4
126	Inheritance of a Ring Chromosome 21 in a Couple UndergoingIn VitroFertilization (IVF): A Case Report. Case Reports in Genetics, 2011, 2011, 1-5.	0.1	4

#	Article	IF	Citations
127	Developmental delay referrals and the roles of Fragile X testing and molecular karyotyping: A New Zealand perspective. Molecular Medicine Reports, 2013, 7, 1710-1714.	1.1	4
128	<i>NOS1AP</i> Polymorphisms Modify QTc Interval Duration But Not Cardiac Arrest Risk in Hypertrophic Cardiomyopathy. Journal of Cardiovascular Electrophysiology, 2015, 26, 1346-1351.	0.8	4
129	Impacts for Children Living with Genetic Muscle Disorders and their Parents â€" Findings from a Population-Based Study. Journal of Neuromuscular Diseases, 2018, 5, 341-352.	1.1	4
130	Splice Site Variants in the KCNQ1 and SCN5A Genes: Transcript Analysis as a Tool in Supporting Pathogenicity. Journal of Clinical Medicine Research, 2017, 9, 709-718.	0.6	4
131	A transient assay for recombination demonstrates that Arabidopsis SNM1 and XRCC3 enhance non-homologous recombination. Genetics and Molecular Research, 2011, 10, 2104-32.	0.3	4
132	Diagnostic Screening Workflow for Mutations in the BRCA1 and BRCA2 Genes. Sultan Qaboos University Medical Journal, 2015, 15, e58-70.	0.3	4
133	Whole organism approaches to chemical genomics: the promising role of zebrafish (Danio rerio). Expert Opinion on Drug Discovery, 2007, 2, 1389-1401.	2.5	3
134	A Novel 2.3 Mb Microduplication of 9q34.3 Inserted into 19q13.4 in a Patient with Learning Disabilities. Case Reports in Pediatrics, 2012, 2012, 1-7.	0.2	3
135	Whole Exome Sequencing Reveals Compound Heterozygosity for Ethnically Distinct PEX7 Mutations Responsible for Rhizomelic Chondrodysplasia Punctata, Type 1. Case Reports in Genetics, 2015, 2015, 1-4.	0.1	3
136	Genetic testing in Polynesian long QT syndrome probands reveals a lower diagnostic yield and an increased prevalence of rare variants. Heart Rhythm, 2020, 17, 1304-1311.	0.3	3
137	A Novel STK4 Mutation Impairs T Cell Immunity Through Dysregulation of Cytokine-Induced Adhesion and Chemotaxis Genes. Journal of Clinical Immunology, 2021, 41, 1839-1852.	2.0	3
138	Implications of a Chr7q21.11 Microdeletion and the Role of the PCLO Gene in Developmental Delay = Ø§Ù„Ø£Ø«Ø§Ø± Ø§Ù„Ù…Ø ªØ±Ø Ùˆ Ø¯Ùˆ Ø± Ø¬ÙŠÙ† PCLO ÙÙŠ ØµØ¹ÙˆØ¨Ø§Øª Ø§Ù„Ø ªØ¹Ù„Ù…. Sultan Qaboos University Medical Journal, 2013, 13, 306-310.	0.3	3
139	Hierarchical mutation screening protocol for theBRCA1 gene. Human Mutation, 2000, 16, 422-430.	1.1	2
140	Zebrafish: At the Nexus of Functional and Chemical Genomics. Biotechnology and Genetic Engineering Reviews, 2006, 22, 77-100.	2.4	2
141	Discovery of three related females who type XY at the amelogenin locus. Forensic Science International: Genetics Supplement Series, 2008, 1, 577-579.	0.1	2
142	The SCN5A gene in Brugada syndrome: mutations, variants, missense and nonsense. What's a clinician to do?. Heart Rhythm, 2010, 7, 50-51.	0.3	2
143	Indolent Medullary Thyroid Cancer with a <i>RET</i> Proto-Oncogene Cys618Phe Mutation Presenting As Sporadic Unilateral Pheochromocytoma in a 55-Year-Old Korean Woman. Thyroid, 2011, 21, 325-326.	2.4	2
144	Zebrafish dystrophin and utrophin genes: Dissecting transcriptional expression during embryonic development. International Journal of Molecular Medicine, 2011, 29, 338-48.	1.8	2

#	Article	IF	Citations
145	Gene Dosage Analysis in a Clinical Environment: Gene-Targeted Microarrays as the Platform-of-Choice. Microarrays (Basel, Switzerland), 2013, 2, 51-62.	1.4	2
146	Application of Massively Parallel Sequencing in the Clinical Diagnostic Testing of Inherited Cardiac Conditions. Medical Sciences (Basel, Switzerland), 2014, 2, 98-126.	1.3	2
147	The New Zealand Neuromuscular Disease Registry: Rate of diagnoses confirmed by molecular testing. Journal of Clinical Neuroscience, 2015, 22, 434-436.	0.8	2
148	Massively Parallel Sequencing of Genes Implicated in Heritable Cardiac Disorders: A Strategy for a Small Diagnostic Laboratory. Medical Sciences (Basel, Switzerland), 2017, 5, 22.	1.3	2
149	Predicting the Pathogenic Potential of BRCA1 and BRCA2 Gene Variants Identified in Clinical Genetic Testing. Sultan Qaboos University Medical Journal, 2015, 15, e218-25.	0.3	2
150	Impact and predictors of quality of life in adults diagnosed with a genetic muscle disorder: a nationwide population-based study. Quality of Life Research, 2022, 31, 1657-1666.	1.5	2
151	Exclusion of the gene responsible for facioscapulohumeral muscular dystrophy (FSH) at 6q23-q27. Journal of the Neurological Sciences, 1991, 102, 206-208.	0.3	1
152	Array comparative genomic hybridisation: a new tool in the diagnostic genetic armoury. New Zealand Medical Journal, 2010, 123, 50-61.	0.5	1
153	A new DNA marker, D6S129, identifies aHindIII polymorphismOn ChromoSome 6q. Nucleic Acids Research, 1991, 19, 4310-4310.	6.5	0
154	Localization of two new DNA markers on the linkage map of human chromosome 6q. Cytogenetic and Genome Research, 1992, 60, 216-218.	0.6	0
155	Chimerism detected in fraternal twins using ABI AmpFlSTRÂ® Identifiler. Forensic Science International: Genetics Supplement Series, 2009, 2, 226-227.	0.1	0
156	Application of Nutrigenomics in Gastrointestinal Health. , 2010, , 83-94.		0
157	The left and right atria finally express themselves. Heart Rhythm, 2014, 11, 272-273.	0.3	0
158	Tumour Mutation Profiling with High-throughput Multiplexed Genotyping: A Review of its Use for Guiding Targeted Cancer Therapy. Current Cancer Therapy Reviews, 2014, 9, 236-244.	0.2	0
159	Determination of Pathogenicity of Breast Cancer 1 Gene Variants using the American College of Medical Genetics and Genomics and the Association for Molecular Pathology Guidelines. Sultan Qaboos University Medical Journal, 2019, 19, 324.	0.3	0