Paolo Fortina

List of Publications by Year
in descending order

215 papers	12,143 citations	36299 51 h-index	31843 101 g-index
225 all docs	225 docs citations	225 times ranked	19079 citing authors

#	Article	IF	Citations
1	The reverse Warburg effect: Aerobic glycolysis in cancer associated fibroblasts and the tumor stroma. Cell Cycle, 2009, 8, 3984-4001.	2.6	1,130
2	The complex transcriptional landscape of the anucleate human platelet. BMC Genomics, 2013, 14, 1.	2.8	913
3	Connexin-26 mutations in sporadic and inherited sensorineural deafness. Lancet, The, 1998, 351, 394-398.	13.7	610
4	Connexin26 mutations associated with the most common form of non- syndromic neurosensory autosomal recessive deafness (DFNB1) in Mediterraneans. Human Molecular Genetics, 1997, 6, 1605-1609.	2.9	540
5	Analysis of 13 cell types reveals evidence for the expression of numerous novel primate- and tissue-specific microRNAs. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E1106-15.	7.1	376
6	High carrier frequency of the 35delG deafness mutation in European populations. European Journal of Human Genetics, 2000, 8, 19-23.	2.8	363
7	Seventy-five genetic loci influencing the human red blood cell. Nature, 2012, 492, 369-375.	27.8	320
8	Integrated Cell Isolation and Polymerase Chain Reaction Analysis Using Silicon Microfilter Chambers. Analytical Biochemistry, 1998, 257, 95-100.	2.4	275
9	Functional significance of macrophage-derived exosomes in inflammation and pain. Pain, 2014, 155, 1527-1539.	4.2	253
10	Mitochondrial DNA Mutations and Mitochondrial Abnormalities in Dilated Cardiomyopathy. American Journal of Pathology, 1998, 153, 1501-1510.	3.8	225
11	Nanobiotechnology: the promise and reality of new approaches to molecular recognition. Trends in Biotechnology, 2005, 23, 168-173.	9.3	221
12	Melanoma adapts to RAF/MEK inhibitors through FOXD3-mediated upregulation of ERBB3. Journal of Clinical Investigation, 2013, 123, 2155-2168.	8.2	215
13	Loss of stromal caveolin-1 leads to oxidative stress, mimics hypoxia and drives inflammation in the tumor microenvironment, conferring the â€œreverse Warburg effectâ€• A transcriptional informatics analysis with validation. Cell Cycle, 2010, 9, 2201-2219.	2.6	212
14	Human platelet microRNA-mRNA networks associated with age and gender revealed by integrated plateletomics. Blood, 2014, 123, e37-e45.	1.4	199
15	p21 ^{CIP1} attenuates Ras- and c-Myc-dependent breast tumor epithelial mesenchymal transition and cancer stem cell-like gene expression in vivo. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 19035-19039.	7.1	163
16	Evaluation of DNA Fragment Sizing and Quantification by the Agilent 2100 Bioanalyzer. Clinical Chemistry, 2000, 46, 1851-1853.	3.2	144
17	Applications of nanoparticles to diagnostics and therapeutics in colorectal cancer. Trends in Biotechnology, 2007, 25, 145-152.	9.3	140
18	Fabrication of plastic microchips by hot embossing. Lab on A Chip, 2002, 2, 1.	6.0	127

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19	Roles of Î²-catenin signaling in phenotypic expression and proliferation of articular cartilage superficial zone cells. Laboratory Investigation, 2011, 91, 1739-1752.	3.7	121
20	Degenerate Oligonucleotide Primedâ€“Polymerase Chain Reaction and Capillary Electrophoretic Analysis of Human DNA on Microchip-Based Devices. Analytical Biochemistry, 1998, 257, 101-106.	2.4	119
21	Transcriptomic profiling of 39 commonly-used neuroblastoma cell lines. Scientific Data, 2017, 4, 170033.	5.3	113
22	Adsorption and Surface Diffusion of DNA Oligonucleotides at Liquid/Solid Interfaces. Langmuir, 1997, 13, 320-329.	3.5	111
23	Stat5 promotes metastatic behavior of human prostate cancer cells in vitro and in vivo. Endocrine-Related Cancer, 2010, 17, 481-493.	3.1	109
24	ChIP sequencing of cyclin D1 reveals a transcriptional role in chromosomal instability in mice. Journal of Clinical Investigation, 2012, 122, 833-843.	8.2	106
25	Genome-Wide Analysis of Neuroblastomas using High-Density Single Nucleotide Polymorphism Arrays. PLoS ONE, 2007, 2, e255.	2.5	105
26	Microchip Module for Blood Sample Preparation and Nucleic Acid Amplification Reactions. Genome Research, 2001, 11, 405-412.	5.5	104
27	Linkage of DFNB1 to Non-Syndromic Neurosensory Autosomal-Recessive Deafness in Mediterranean Families. European Journal of Human Genetics, 1997, 5, 83-88.	2.8	88
28	Development of an Automated and Sensitive Microfluidic Device for Capturing and Characterizing Circulating Tumor Cells (CTCs) from Clinical Blood Samples. PLoS ONE, 2016, 11, e0147400.	2.5	82
29	Genetic analysis in Italian families with inflammatory bowel disease supports linkage to the IBD1 locus â€“ A GISC study. European Journal of Human Genetics, 1999, 7, 567-573.	2.8	81
30	Glanzmann thrombasthenia secondary to a Gly273-->Asp mutation adjacent to the first calcium-binding domain of platelet glycoprotein IIb.. Journal of Clinical Investigation, 1994, 93, 172-179.	8.2	79
31	Angiotensin converting enzyme gene deletion allele is independently and strongly associated with coronary atherosclerosis and myocardial infarction.. Heart, 1995, 74, 584-591.	2.9	78
32	The human platelet: strong transcriptome correlations among individuals associate weakly with the platelet proteome. Biology Direct, 2014, 9, 3.	4.6	77
33	Human Î±â€Thalassemia syndromes:Detection of molecular defects. American Journal of Hematology, 1996, 53, 81-91.	4.1	76
34	Nucleic acid detection using non-radioactive labelling methods. Molecular and Cellular Probes, 1995, 9, 145-156.	2.1	73
35	Transcription Factor Stat3 Stimulates Metastatic Behavior of Human Prostate Cancer Cells in Vivo, whereas Stat5b Has a Preferential Role in the Promotion of Prostate Cancer Cell Viability and Tumor Growth. American Journal of Pathology, 2010, 176, 1959-1972.	3.8	71
36	Effect of Hydrophobicity and Electrostatics on Adsorption and Surface Diffusion of DNA Oligonucleotides at Liquid/Solid Interfaces. Journal of Colloid and Interface Science, 1998, 203, 197-207.	9.4	69

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37	Analytical Ancestry: â€œFirstsâ€•in Fluorescent Labeling of Nucleosides, Nucleotides, and Nucleic Acids. Clinical Chemistry, 2009, 55, 670-683.	3.2	69
38	Detection of Activating Estrogen Receptor Gene (ESR1) Mutations in Single Circulating Tumor Cells. Clinical Cancer Research, 2017, 23, 6086-6093.	7.0	68
39	Digital mRNA profiling. Nature Biotechnology, 2008, 26, 293-294.	17.5	67
40	Micropillar array chip for integrated white blood cell isolation and PCR. New Biotechnology, 2005, 21, 157-162.	2.7	65
41	Genetic modifiers of Â-thalassemia and clinical severity as assessed by age at first transfusion. Haematologica, 2012, 97, 989-993.	3.5	64
42	Detection and Characterization of Circulating Tumor Associated Cells in Metastatic Breast Cancer. International Journal of Molecular Sciences, 2016, 17, 1665.	4.1	63
43	In Vivo E2F Reporting Reveals Efficacious Schedules of MEK1/2â€“CDK4/6 Targeting and mTORâ€“S6 Resistance Mechanisms. Cancer Discovery, 2018, 8, 568-581.	9.4	62
44	Molecular diagnostics: hurdles for clinical implementation. Trends in Molecular Medicine, 2002, 8, 264-266.	6.7	61
45	Genotyping on a Thermal Gradient DNA Chip. Genome Research, 2003, 13, 467-475.	5.5	60
46	Concordance Study of 3 Direct-to-Consumer Genetic-Testing Services. Clinical Chemistry, 2011, 57, 518-521.	3.2	60
47	Revealing genes associated with vitellogenesis in the liver of the zebrafish (Danio rerio) by transcriptome profiling. BMC Genomics, 2009, 10, 141.	2.8	59
48	Cyclin D1 induction of Dicer governs microRNA processing and expression in breast cancer. Nature Communications, 2013, 4, 2812.	12.8	57
49	Key questions about the future of laboratory medicine in the next decade of the 21st century: A report from the IFCC-Emerging Technologies Division. Clinica Chimica Acta, 2019, 495, 570-589.	1.1	56
50	High-resolution SNP arrays in mental retardation diagnostics: how much do we gain?. European Journal of Human Genetics, 2010, 18, 178-185.	2.8	54
51	Exosomal Î±vÎ²6 integrin is required for monocyte M2 polarization in prostate cancer. Matrix Biology, 2018, 70, 20-35.	3.6	54
52	Different hematological phenotypes caused by the interaction of triplicated Î±-globin genes and heterozygous Î²-thalassemia. , 1997, 55, 83-88.		53
53	Analysis of Clinically Relevant Single-Nucleotide Polymorphisms by Use of Microelectronic Array Technology. Clinical Chemistry, 2002, 48, 2124-2130.	3.2	53
54	Selective inhibition of Ph-positive ALL cell growth through kinase-dependent and -independent effects by CDK6-specific PROTACs. Blood, 2020, 135, 1560-1573.	1.4	53

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55	Current perspectives in protein array technology. Annals of Clinical Biochemistry, 2006, 43, 457-467.	1.6	49
56	Recessive mutation in tetraspanin CD151 causes Kindler syndrome-like epidermolysis bullosa with multi-systemic manifestations including nephropathy. Matrix Biology, 2018, 66, 22-33.	3.6	49
57	The Otto Aufranc Award: Identification of a 4 Mb Region on Chromosome 17q21 Linked to Developmental Dysplasia of the Hip in One 18-member, Multigeneration Family. Clinical Orthopaedics and Related Research, 2010, 468, 337-344.	1.5	47
58	Developmental Dysplasia of the Hip: Linkage Mapping and Whole Exome Sequencing Identify a Shared Variant in CX 3 CR 1 in All Affected Members of a Large Multigeneration Family. Journal of Bone and Mineral Research, 2013, 28, 2540-2549.	2.8	47
59	Single-Cell Genomics. Clinical Chemistry, 2019, 65, 972-985.	3.2	47
60	Whole-exome sequencing of DNA from peripheral blood mononuclear cells (PBMC) and EBV-transformed lymphocytes from the same donor. BMC Genomics, 2011, 12, 464.	2.8	46
61	Î²-Thalassemia Microelectronic Chip: A Fast and Accurate Method for Mutation Detection. Clinical Chemistry, 2004, 50, 73-79.	3.2	44
62	Association of RB/p16-Pathway Perturbations with DCIS Recurrence. American Journal of Pathology, 2011, 179, 1171-1178.	3.8	44
63	STAT5A/B Gene Locus Undergoes Amplification during Human Prostate Cancer Progression. American Journal of Pathology, 2013, 182, 2264-2275.	3.8	44
64	Dystrophic Epidermolysis Bullosa: COL7A1 Mutation Landscape in a Multi-Ethnic Cohort of 152 Extended Families with High Degree of Customary Consanguineous Marriages. Journal of Investigative Dermatology, 2017, 137, 660-669.	0.7	44
65	Autosomal recessive congenital ichthyosis: Genomic landscape and phenotypic spectrum in a cohort of 125 consanguineous families. Human Mutation, 2019, 40, 288-298.	2.5	43
66	Kinase-independent role of cyclin D1 in chromosomal instability and mammary tumorigenesis. Oncotarget, 2015, 6, 8525-8538.	1.8	43
67	Microchip-based devices for molecular diagnosis of genetic diseases. Molecular Diagnosis and Therapy, 1996, 1, 183-200.	1.1	42
68	Sensitivity, reproducibility, and accuracy in short tandem repeat genotyping using capillary array electrophoresis.. Genome Research, 1996, 6, 893-903.	5.5	42
69	Reversine Enhances Generation of Progenitor-like Cells by Dedifferentiation of Annulus Fibrosus Cells. Tissue Engineering - Part A, 2010, 16, 1443-1455.	3.1	42
70	Structure-Based Screen Identifies a Potent Small Molecule Inhibitor of Stat5a/b with Therapeutic Potential for Prostate Cancer and Chronic Myeloid Leukemia. Molecular Cancer Therapeutics, 2015, 14, 1777-1793.	4.1	42
71	Gene expression profiling during the transition to failure in TNF-Î± over-expressing mice demonstrates the development of autoimmune myocarditisâ˜†. Journal of Molecular and Cellular Cardiology, 2004, 36, 515-530.	1.9	41
72	Duchenne/becker muscular dystrophy carrier detection using quantitative PCR and fluorescence-based strategies. American Journal of Medical Genetics Part A, 1993, 48, 200-208.	2.4	39

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73	NFÎºB activation and stimulation of chemokine production in normal human macrophages by the gadolinium-based magnetic resonance contrast agent Omniscan: possible role in the pathogenesis of nephrogenic systemic fibrosis. Annals of the Rheumatic Diseases, 2010, 69, 2024-2033.	0.9	39
74	Next-generation sequencing in the clinic. Nature Biotechnology, 2013, 31, 990-992.	17.5	38
75	Homozygosity for the V37I Connexin 26 mutation in three unrelated children with sensorineural hearing loss. Clinical Genetics, 2002, 61, 459-464.	2.0	37
76	Test Pricing and Reimbursement in Genomic Medicine: Towards a General Strategy. Public Health Genomics, 2016, 19, 352-363.	1.0	37
77	Parallel molecular genetic analysis. European Journal of Human Genetics, 1998, 6, 417-429.	2.8	36
78	Increased amplification efficiency of microchip-based PCR by dynamic surface passivation. BioTechniques, 2004, 36, 248-252.	1.8	36
79	Glucocorticoids paradoxically facilitate steroid resistance in T cell acute lymphoblastic leukemias and thymocytes. Journal of Clinical Investigation, 2020, 130, 863-876.	8.2	36
80	Ethanol potentiates HIV-1 gp120-induced apoptosis in human neurons via both the death receptor and NMDA receptor pathways. Virology, 2005, 334, 59-73.	2.4	35
81	Diagnosis of Duchenne/Becker muscular dystrophy and quantitative identification of carrier status by use of entangled solution capillary electrophoresis. Clinical Chemistry, 1997, 43, 745-751.	3.2	34
82	Clinical Exome Performance for Reporting Secondary Genetic Findings. Clinical Chemistry, 2015, 61, 213-220.	3.2	34
83	A G-to-A mutation in IVS-3 of the human gamma fibrinogen gene causing afibrinogenemia due to abnormal RNA splicing. Blood, 2000, 96, 2501-2505.	1.4	33
84	RB loss contributes to aggressive tumor phenotypes in MYC-driven triple negative breast cancer. Cell Cycle, 2015, 14, 109-122.	2.6	33
85	Establishment of an orthotopic patient-derived xenograft mouse model using uveal melanoma hepatic metastasis. Journal of Translational Medicine, 2017, 15, 145.	4.4	33
86	A distinct GM-CSF ⁺ T helper cell subset requires T-bet to adopt a T _H 1 phenotype and promote neuroinflammation. Science Immunology, 2020, 5, .	11.9	33
87	Simple two-color array-based approach for mutation detection. European Journal of Human Genetics, 2000, 8, 884-894.	2.8	32
88	Small extracellular vesicles modulated by Î±VÎ²3Âintegrin induce neuroendocrine differentiation in recipient cancer cells. Journal of Extracellular Vesicles, 2020, 9, 1761072.	12.2	32
89	Epigenomic profiling of neuroblastoma cell lines. Scientific Data, 2020, 7, 116.	5.3	32
90	Multigene Next-Generation Sequencing Panel Identifies Pathogenic Variants in Patients with Unknown Subtype of Epidermolysis Bullosa: Subclassification with Prognostic Implications. Journal of Investigative Dermatology, 2017, 137, 2649-2652.	0.7	31

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91	Kinetics of heterogeneous hybridization on indium tin oxide surfaces with and without an applied potential. Electrophoresis, 2002, 23, 1551.	2.4	30
92	CARD15 Genotyping in Inflammatory Bowel Disease Patients by Multiplex Pyrosequencing. Clinical Chemistry, 2003, 49, 1675-1679.	3.2	30
93	In Vivo MAPK Reporting Reveals the Heterogeneity in Tumoral Selection of Resistance to RAF Inhibitors. Cancer Research, 2013, 73, 7101-7110.	0.9	30
94	Detection of the most common mutations causing beta-thalassemia in Mediterraneans using a multiplex amplification refractory mutation system (MARMS).. Genome Research, 1992, 2, 163-166.	5.5	29
95	Microarray Technology and Applications: An All-Language Literature Survey Including Books and Patents. Clinical Chemistry, 2001, 47, 1479-1482.	3.2	28
96	Surface Effects on PCR Reactions in Multichip Microfluidic Platforms. Biomedical Microdevices, 2004, 6, 75-80.	2.8	28
97	RB and p53 Cooperate to Prevent Liver Tumorigenesis in Response to Tissue Damage. Gastroenterology, 2011, 141, 1439-1450.	1.3	28
98	Performance of exome sequencing for pharmacogenomics. Personalized Medicine, 2015, 12, 109-115.	1.5	28
99	Gene-Targeted Next Generation Sequencing Identifies PNPLA1 Mutations in Patients with a Phenotypic Spectrum of Autosomal Recessive Congenital Ichthyosis: The Impact of Consanguinity. Journal of Investigative Dermatology, 2017, 137, 678-685.	0.7	28
100	Rapid sizing of polymorphic microsatellite markers by capillary array electrophoresis. Journal of Chromatography A, 1997, 781, 295-305.	3.7	27
101	The future of laboratory medicine â€" A 2014 perspective. Clinica Chimica Acta, 2015, 438, 284-303.	1.1	27
102	The frame-shift mutation of the NOD2/CARD15 gene is significantly increased in ulcerative colitis: An â^—IG-IBD study. Gastroenterology, 2004, 126, 625-627.	1.3	26
103	Cyclin D1-mediated microRNA expression signature predicts breast cancer outcome. Theranostics, 2018, 8, 2251-2263.	10.0	26
104	Novel Oncogeneâ€“Induced Metastatic Prostate Cancer Cell Lines Define Human Prostate Cancer Progression Signatures. Cancer Research, 2013, 73, 978-989.	0.9	25
105	A newly-characterized Î±-thalassaemia-1 deletion removes the entire Î±-like globin gene cluster in an Italian family. British Journal of Haematology, 1991, 78, 529-534.	2.5	24
106	System for Preparing Microhybridization Arrays on Glass Slides. Analytical Chemistry, 1998, 70, 5085-5092.	6.5	24
107	Polymorphic Changes in the 5â€™ Flanking Region of Factor VII Have a Combined Effect on Promoter Strength. Thrombosis and Haemostasis, 2002, 88, 763-767.	3.4	24
108	The retinoblastoma tumor suppressor pathway modulates the invasiveness of ErbB2-positive breast cancer. Oncogene, 2014, 33, 3980-3991.	5.9	24

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109	Whole exome sequencing identifies a germline MET mutation in two siblings with hereditary wild-type RET medullary thyroid cancer. Human Mutation, 2018, 39, 371-377.	2.5	24
110	Fluorescence-based DNA minisequence analysis for detection of known single-base changes in genomic DNA. Molecular and Cellular Probes, 1995, 9, 175-182.	2.1	23
111	Allelic association of microsatellites of 6p in Italian hemochromatosis patients. Human Genetics, 1996, 97, 476-481.	3.8	22
112	Analysis of 31 CFTR mutations by polymerase chain reaction/oligonucleotide ligation assay in a pilot screening of 4476 newborns for cystic fibrosis. Journal of Medical Screening, 1999, 6, 67-69.	2.3	22
113	Bridging genomics research between developed and developing countries: the Genomic Medicine Alliance. Personalized Medicine, 2014, 11, 615-623.	1.5	22
114	Inherited Interleukin 2â€“Inducible T-Cell (ITK) Kinase Deficiency in Siblings With Epidermodysplasia Verruciformis and Hodgkin Lymphoma. Clinical Infectious Diseases, 2019, 68, 1938-1941.	5.8	22
115	RNA-Binding Protein HuR Promotes Th17 Cell Differentiation and Can Be Targeted to Reduce Autoimmune Neuroinflammation. Journal of Immunology, 2020, 204, 2076-2087.	0.8	22
116	A Pilot C282Y Hemochromatosis Screening in Italian Newborns by TaqManâ„¢ Technology. Genetic Testing and Molecular Biomarkers, 2000, 4, 177-181.	1.7	21
117	Miniaturized detection technology in molecular diagnostics. Expert Review of Molecular Diagnostics, 2005, 5, 549-559.	3.1	20
118	Hearts lacking caveolin-1 develop hypertrophy with normal cardiac substrate metabolism. Cell Cycle, 2008, 7, 2509-2518.	2.6	20
119	Substrate uptake and metabolism are preserved in hypertrophic caveolin-3 knockout hearts. American Journal of Physiology - Heart and Circulatory Physiology, 2008, 295, H657-H666.	3.2	20
120	Fluorescent approaches to diagnosis of Lesch-Nyhan syndrome and quantitative analysis of carrier status. Molecular and Cellular Probes, 1993, 7, 311-324.	2.1	19
121	Variants in Genes Involved in Functional Pathways Associated with Hypertension in African Americans. Clinical and Translational Science, 2010, 3, 279-286.	3.1	19
122	The Retinoblastoma Tumor Suppressor Modulates DNA Repair and Radioresponsiveness. Clinical Cancer Research, 2014, 20, 5468-5482.	7.0	19
123	Autosomal recessive congenital ichthyosis: CERS3 mutations identified by a next generation sequencing panel targeting ichthyosis genes. European Journal of Human Genetics, 2017, 25, 1282-1285.	2.8	19
124	Targeting CDK6 and BCL2 Exploits the â€œMYB Addictionâ€•of Ph+ Acute Lymphoblastic Leukemia. Cancer Research, 2018, 78, 1097-1109.	0.9	19
125	Sighting acute myocardial infarction through platelet gene expression. Scientific Reports, 2019, 9, 19574.	3.3	19
126	Rapid detection of medium chain acyl-CoA dehydrogenase gene mutations by non-radioactive, single strand conformation polymorphism minigels.. Journal of Medical Genetics, 1994, 31, 551-554.	3.2	18

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127	Combined segregation and linkage analysis of inflammatory bowel disease in the IBD1 region using severity to characterise Crohn's disease and ulcerative colitis. European Journal of Human Genetics, 2000, 8, 846-852.	2.8	18
128	Pyrosequencing for detection of mutations in the connexin 26 (GJB2) and mitochondrial 12S RNA (MTRNR1) genes associated with hereditary hearing loss. Human Mutation, 2002, 20, 312-320.	2.5	18
129	Region-specific detection of neuroblastoma loss of heterozygosity at multiple loci simultaneously using a SNP-based tag-array platform. Genome Research, 2005, 15, 1168-1176.	5.5	18
130	Regulation of miR106b cluster through the RB pathway. Cell Cycle, 2013, 12, 98-111.	2.6	18
131	A large deletion encompassing the entire Î±-like globin gene cluster in a family of Northern European extraction. Nucleic Acids Research, 1988, 16, 11223-11235.	14.5	17
132	Opportunities for near-infrared thermal ablation of colorectal metastases by guanylyl cyclase C-targeted gold nanoshells. Future Oncology, 2006, 2, 705-716.	2.4	17
133	Next generation sequencing in cancer: opportunities and challenges for precision cancer medicine. Scandinavian Journal of Clinical and Laboratory Investigation, 2016, 76, S84-S91.	1.2	17
134	Artificial Intelligence-Powered Search Tools and Resources in the Fight Against COVID-19. Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine, 2020, 31, 106-116.	0.7	17
135	Correlation between Transfusion Requirement, Blood Volume and Haemoglobin Level in Homozygous Î²-Thalassaemia. Acta Haematologica, 1980, 64, 103-108.	1.4	16
136	Fluorescence-based, multiplex allele-specific PCR (MASPCR) detection of the Î”F508 deletion in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. Molecular and Cellular Probes, 1992, 6, 353-356.	2.1	16
137	Identification ofAPCgene mutations in colorectal cancer using universal microarray-based combinatorial sequencing-by-hybridization. Human Mutation, 2004, 24, 261-271.	2.5	16
138	Caveolin-1 overexpression enhances androgen-dependent growth and proliferation in the mouse prostate. International Journal of Biochemistry and Cell Biology, 2011, 43, 1318-1329.	2.8	16
139	Direct-access genetic testing: the view from Europe. Nature Reviews Genetics, 2011, 12, 670-670.	16.3	16
140	A novel autosomal recessiveGJB2-associated disorder: Ichthyosis follicularis, bilateral severe sensorineural hearing loss, and punctate palmoplantar keratoderma. Human Mutation, 2019, 40, 217-229.	2.5	16
141	Whole-Transcriptome Analysis by RNA Sequencing for Genetic Diagnosis of Mendelian Skin Disorders in the Context of Consanguinity. Clinical Chemistry, 2021, 67, 876-888.	3.2	16
142	Targeting Chemotherapy to Decondensed H3K27me3-Marked Chromatin of AML Cells Enhances Leukemia Suppression. Cancer Research, 2022, 82, 458-471.	0.9	16
143	A > 200 kb deletion removing the entire Î²-like globin gene cluster in a family of Irish Descent. Hemoglobin, 1991, 15, 23-41.	0.8	15
144	iSeqQC: a tool for expression-based quality control in RNA sequencing. BMC Bioinformatics, 2020, 21, 56.	2.6	15

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145	The presurgical management with erythrocytapheresis of a patient with a high-oxygen-affinity, unstable Hb variant (Hb Bryn Mawr). Transfusion, 1997, 37, 703-707.	1.6	14
146	Genotyping Î²-Globin Gene Mutations on Copolymer-Coated Glass Slides with the Ligation Detection Reaction. Clinical Chemistry, 2008, 54, 1657-1663.	3.2	14
147	Protein Microarrays: A Literature Survey. Clinical Chemistry, 2003, 49, 2109-2109.	3.2	13
148	Mutation detection using ligase chain reaction in passivated silicon-glass microchips and microchip capillary electrophoresis. BioTechniques, 2004, 37, 392-398.	1.8	13
149	Analysis of CARD15 Gene Variants in Italian Pediatric Patients with Inflammatory Bowel Diseases. Journal of Pediatrics, 2005, 147, 272-273.	1.8	13
150	Use of Linkage Analysis, Genome-Wide Association Studies, and Next-Generation Sequencing in the Identification of Disease-Causing Mutations. Methods in Molecular Biology, 2013, 1015, 127-146.	0.9	13
151	Donor spliceâ€site mutation in CUL4B is likely cause of Xâ€linked intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2294-2299.	1.2	13
152	Leberâ€²s Hereditary Optic Neuropathy (LHON)-Related Mitochondrial DNA Sequence Changes in Italian Patients Presenting with Sporadic Bilateral Optic Neuritis. Biochemical and Molecular Medicine, 1995, 56, 45-51.	1.4	12
153	Four-Laser Scanning Confocal System for Microarray Analysis. BioTechniques, 2002, 32, 346-354.	1.8	12
154	Interaction of rare illegitimate recombination event and a poly A addition site mutation resulting in a severe form of alpha thalassemia. Blood, 1994, 83, 3356-3362.	1.4	11
155	Non-radioactive detection of the most common mutations in the cystic fibrosis transmembrane conductance regulator gene by multiplex allele-specific polymerase chain reaction. Human Genetics, 1992, 90, 375-8.	3.8	10
156	Î±-Thalassemia Caused by a 16 BP Deletion in the 3â€² Untranslated Region of the Î±2-Globin Gene Including the First Nucleotide of the Poly a Signal Sequence. Hemoglobin, 1997, 21, 121-130.	0.8	10
157	Combinatorial Sequencing-by-Hybridization: Analysis of theNF1Gene. Genetic Testing and Molecular Biomarkers, 2006, 10, 8-17.	1.7	10
158	Nanotechnology: Improving Clinical Testing?. Clinical Chemistry, 2010, 56, 1384-1389.	3.2	10
159	Effects of genetic variation in protease activated receptor 4 after an acute coronary syndrome: Analysis from the TRACER trial. Blood Cells, Molecules, and Diseases, 2018, 72, 37-43.	1.4	10
160	Institutional Profile: Golden Helix Institute of Biomedical Research: interdisciplinary research and educational activities in pharmacogenomics and personalized medicine. Pharmacogenomics, 2012, 13, 387-392.	1.3	9
161	DOP-PCR Amplification of Whole Genomic DNA and Microchip-Based Capillary Electrophoresis. , 2001, 163, 211-219.		8
162	Applications of emerging technologies to the study of human genetics. European Journal of Human Genetics, 1998, 6, 2-3.	2.8	8

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163	Microchips: An All-Language Literature Survey Including Books and Patents. Clinical Chemistry, 2002, 48, 1620-1622.	3.2	8
164	Nanotechnology and Applications: An All-Language Literature Survey Including Books and Patents. Clinical Chemistry, 2002, 48, 662-665.	3.2	8
165	Infundibulopelvic stenosis, multicystic kidney, and calyectasis in a kindred: Clinical observations and genetic analysis. American Journal of Medical Genetics Part A, 1995, 59, 218-224.	2.4	7
166	Maple syrup urine disease (MSUD): Screening for known mutations in Italian patients. Journal of Inherited Metabolic Disease, 1994, 17, 652-660.	3.6	6
167	Association of a Polymorphic Variant of the Adiponectin Gene with Insulin Resistance in African Americans. Clinical and Translational Science, 2008, 1, 194-199.	3.1	6
168	Identification of a KRAS mutation in a patient with non-small cell lung cancer treated with chemoradiotherapy and panitumumab. Cancer Biology and Therapy, 2013, 14, 883-887.	3.4	6
169	Clinical Genomics: When Whole Genome Sequencing Is like a Whole-body CT Scan. Clinical Chemistry, 2014, 60, 1390-1392.	3.2	6
170	Vestibular and hearing loss in genetic and metabolic disorders. Current Opinion in Neurology, 1999, 12, 35-39.	3.6	6
171	A Î²-Catenin-TCF-Sensitive Locus Control Region Mediates GUCY2C Ligand Loss in Colorectal Cancer. Cellular and Molecular Gastroenterology and Hepatology, 2022, 13, 1276-1296.	4.5	6
172	Whole-transcriptome sequencingâ€“based concomitant detection of viral and human genetic determinants of cutaneous lesions. JCI Insight, 2022, 7, .	5.0	6
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