Paolo Fortina

List of Publications by Citations

210 papers	9,710 citations	47 h-index	92 g-index
225 ext. papers	10,990 ext. citations	6.7 avg, IF	5.45 L-index

#	Paper	IF	Citations
210	The reverse Warburg effect: aerobic glycolysis in cancer associated fibroblasts and the tumor stroma. *Cell Cycle*, **2009**, 8, 3984-4001	4.7	890
209	Connexin-26 mutations in sporadic and inherited sensorineural deafness. *Lancet, The*, **1998**, 351, 394-8	40	542
208	The complex transcriptional landscape of the anucleate human platelet. *BMC Genomics*, **2013**, 14, 1	4.5	480
207	Connexin26 mutations associated with the most common form of non-syndromic neurosensory autosomal recessive deafness (DFNB1) in Mediterraneans. *Human Molecular Genetics*, **1997**, 6, 1605-9	5.6	442
206	High carrier frequency of the 35delG deafness mutation in European populations. Genetic Analysis Consortium of GJB2 35delG. *European Journal of Human Genetics*, **2000**, 8, 19-23	5.3	318
205	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	11.5	307
204	Seventy-five genetic loci influencing the human red blood cell. *Nature*, **2012**, 492, 369-75	50.4	257
203	Integrated cell isolation and polymerase chain reaction analysis using silicon microfilter chambers. *Analytical Biochemistry*, **1998**, 257, 95-100	3.1	231
202	Mitochondrial DNA mutations and mitochondrial abnormalities in dilated cardiomyopathy. *American Journal of Pathology*, **1998**, 153, 1501-10	5.8	190
201	Melanoma adapts to RAF/MEK inhibitors through FOXD3-mediated upregulation of ERBB3. *Journal of Clinical Investigation*, **2013**, 123, 2155-68	15.9	189
200	Loss of stromal caveolin-1 leads to oxidative stress, mimics hypoxia and drives inflammation in the tumor microenvironment, conferring the "reverse Warburg effect": a transcriptional informatics analysis with validation. *Cell Cycle*, **2010**, 9, 2201-19	4.7	188
199	Nanobiotechnology: the promise and reality of new approaches to molecular recognition. *Trends in Biotechnology*, **2005**, 23, 168-73	15.1	182
198	Functional significance of macrophage-derived exosomes in inflammation and pain. *Pain*, **2014**, 155, 1527-1539	7	172
197	Human platelet microRNA-mRNA networks associated with age and gender revealed by integrated plateletomics. *Blood*, **2014**, 123, e37-45	2.2	155
196	p21CIP1 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, 19035-9	11.5	147
195	Evaluation of DNA Fragment Sizing and Quantification by the Agilent 2100 Bioanalyzer. *Clinical Chemistry*, **2000**, 46, 1851-1853	5.5	123
194	Applications of nanoparticles to diagnostics and therapeutics in colorectal cancer. *Trends in Biotechnology*, **2007**, 25, 145-52	15.1	119

193	Fabrication of plastic microchips by hot embossing. *Lab on A Chip*, **2002**, 2, 1-4	7.2	108
192	Degenerate oligonucleotide primed-polymerase chain reaction and capillary electrophoretic analysis of human DNA on microchip-based devices. *Analytical Biochemistry*, **1998**, 257, 101-6	3.1	107
191	Roles of Ecatenin signaling in phenotypic expression and proliferation of articular cartilage superficial zone cells. *Laboratory Investigation*, **2011**, 91, 1739-52	5.9	100
190	Adsorption and Surface Diffusion of DNA Oligonucleotides at Liquid/Solid Interfaces. *Langmuir*, **1997**, 13, 320-329	4	100
189	Genome-wide analysis of neuroblastomas using high-density single nucleotide polymorphism arrays. *PLoS ONE*, **2007**, 2, e255	3.7	95
188	ChIP sequencing of cyclin D1 reveals a transcriptional role in chromosomal instability in mice. *Journal of Clinical Investigation*, **2012**, 122, 833-43	15.9	93
187	Stat5 promotes metastatic behavior of human prostate cancer cells in vitro and in vivo. *Endocrine-Related Cancer*, **2010**, 17, 481-93	5.7	92
186	Linkage of DFNB1 to Non-Syndromic Neurosensory Autosomal-Recessive Deafness in Mediterranean Families. *European Journal of Human Genetics*, **1997**, 5, 83-88	5.3	86
185	Microchip module for blood sample preparation and nucleic acid amplification reactions. *Genome Research*, **2001**, 11, 405-12	9.7	84
184	ChIP sequencing of cyclin D1 reveals a transcriptional role in chromosomal instability in mice. *Journal of Clinical Investigation*, **2013**, 123, 2332-2332	15.9	78
183	Genetic analysis in Italian families with inflammatory bowel disease supports linkage to the IBD1 locus--a GISC study. *European Journal of Human Genetics*, **1999**, 7, 567-73	5.3	73
182	Nucleic acid detection using non-radioactive labelling methods. *Molecular and Cellular Probes*, **1995**, 9, 145-56	3.3	67
181	Human alpha-thalassemia syndromes: detection of molecular defects. *American Journal of Hematology*, **1996**, 53, 81-91	7.1	66
180	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, 172-9	15.9	66
179	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	9.3	64
178	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, 1959-72	5.8	63
177	The human platelet: strong transcriptome correlations among individuals associate weakly with the platelet proteome. *Biology Direct*, **2014**, 9, 3	7.2	62
176	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	3.7	62

175	Micropillar array chip for integrated white blood cell isolation and PCR. *New Biotechnology*, **2005**, 21, 157-62		60
174	Angiotensin converting enzyme gene deletion allele is independently and strongly associated with coronary atherosclerosis and myocardial infarction. *Heart*, **1995**, 74, 584-91	5.1	60
173	Transcriptomic profiling of 39 commonly-used neuroblastoma cell lines. *Scientific Data*, **2017**, 4, 170033	8.2	56
172	Analytical ancestry: "firsts" in fluorescent labeling of nucleosides, nucleotides, and nucleic acids. *Clinical Chemistry*, **2009**, 55, 670-83	5.5	55
171	Cyclin D1 induction of Dicer governs microRNA processing and expression in breast cancer. *Nature Communications*, **2013**, 4, 2812	17.4	53
170	Concordance study of 3 direct-to-consumer genetic-testing services. *Clinical Chemistry*, **2011**, 57, 518-21	5.5	52
169	Analysis of Clinically Relevant Single-Nucleotide Polymorphisms by Use of Microelectronic Array Technology. *Clinical Chemistry*, **2002**, 48, 2124-2130	5.5	52
168	Genetic modifiers of thalassemia and clinical severity as assessed by age at first transfusion. *Haematologica*, **2012**, 97, 989-93	6.6	51
167	Detection of Activating Estrogen Receptor Gene () Mutations in Single Circulating Tumor Cells. *Clinical Cancer Research*, **2017**, 23, 6086-6093	12.9	50
166	Molecular diagnostics: hurdles for clinical implementation. *Trends in Molecular Medicine*, **2002**, 8, 264-6	11.5	50
165	Detection and Characterization of Circulating Tumor Associated Cells in Metastatic Breast Cancer. *International Journal of Molecular Sciences*, **2016**, 17,	6.3	50
164	Genotyping on a thermal gradient DNA chip. *Genome Research*, **2003**, 13, 467-75	9.7	47
163	High-resolution SNP arrays in mental retardation diagnostics: how much do we gain?. *European Journal of Human Genetics*, **2010**, 18, 178-85	5.3	44
162	Revealing genes associated with vitellogenesis in the liver of the zebrafish (Danio rerio) by transcriptome profiling. *BMC Genomics*, **2009**, 10, 141	4.5	44
161	Current perspectives in protein array technology. *Annals of Clinical Biochemistry*, **2006**, 43, 457-67	2.2	44
160	Beta-thalassemia microelectronic chip: a fast and accurate method for mutation detection. *Clinical Chemistry*, **2004**, 50, 73-9	5.5	42
159	E2F Reporting Reveals Efficacious Schedules of MEK1/2-CDK4/6 Targeting and mTOR-S6 Resistance Mechanisms. *Cancer Discovery*, **2018**, 8, 568-581	24.4	41
158	Different hematological phenotypes caused by the interaction of triplicated alpha-globin genes and heterozygous beta-thalassemia. *American Journal of Hematology*, **1997**, 55, 83-8	7.1	41

157	Association of RB/p16-pathway perturbations with DCIS recurrence: dependence on tumor versus tissue microenvironment. *American Journal of Pathology*, **2011**, 179, 1171-8	5.8	40
156	Whole-exome sequencing of DNA from peripheral blood mononuclear cells (PBMC) and EBV-transformed lymphocytes from the same donor. *BMC Genomics*, **2011**, 12, 464	4.5	39
155	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, 337-44	2.2	39
154	Gene expression profiling during the transition to failure in TNF-alpha over-expressing mice demonstrates the development of autoimmune myocarditis. *Journal of Molecular and Cellular Cardiology*, **2004**, 36, 515-30	5.8	39
153	Recessive mutation in tetraspanin CD151 causes Kindler syndrome-like epidermolysis bullosa with multi-systemic manifestations including nephropathy. *Matrix Biology*, **2018**, 66, 22-33	11.4	39
152	Exosomal □6 integrin is required for monocyte M2 polarization in prostate cancer. *Matrix Biology*, **2018**, 70, 20-35	11.4	36
151	Developmental dysplasia of the hip: linkage mapping and whole exome sequencing identify a shared variant in CX3CR1 in all affected members of a large multigeneration family. *Journal of Bone and Mineral Research*, **2013**, 28, 2540-9	6.3	36
150	Microchip-based Devices for Molecular Diagnosis of Genetic Diseases. *Molecular Diagnosis and Therapy*, **1996**, 1, 183-200		36
149	Selective inhibition of Ph-positive ALL cell growth through kinase-dependent and -independent effects by CDK6-specific PROTACs. *Blood*, **2020**, 135, 1560-1573	2.2	35
148	NF□ 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, 2024-33	2.4	35
147	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	4.3	34
146	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, 1777-93	6.1	34
145	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	5.5	34
144	Homozygosity for the V37I Connexin 26 mutation in three unrelated children with sensorineural hearing loss. *Clinical Genetics*, **2002**, 61, 459-64	4	34
143	Sensitivity, reproducibility, and accuracy in short tandem repeat genotyping using capillary array electrophoresis. *Genome Research*, **1996**, 6, 893-903	9.7	34
142	Kinase-independent role of cyclin D1 in chromosomal instability and mammary tumorigenesis. *Oncotarget*, **2015**, 6, 8525-38	3.3	34
141	Next-generation sequencing in the clinic. *Nature Biotechnology*, **2013**, 31, 990-2	44.5	33
140	STAT5A/B gene locus undergoes amplification during human prostate cancer progression. *American Journal of Pathology*, **2013**, 182, 2264-75	5.8	33

139	Reversine enhances generation of progenitor-like cells by dedifferentiation of annulus fibrosus cells. *Tissue Engineering - Part A*, **2010**, 16, 1443-55	3.9	33
138	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	6.2	32
137	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	2.2	32
136	RB loss contributes to aggressive tumor phenotypes in MYC-driven triple negative breast cancer. *Cell Cycle*, **2015**, 14, 109-22	4.7	30
135	Parallel molecular genetic analysis. *European Journal of Human Genetics*, **1998**, 6, 417-29	5.3	30
134	Increased amplification efficiency of microchip-based PCR by dynamic surface passivation. *BioTechniques*, **2004**, 36, 248-52	2.5	30
133	Simple two-color array-based approach for mutation detection. *European Journal of Human Genetics*, **2000**, 8, 884-94	5.3	30
132	Duchenne/Becker muscular dystrophy carrier detection using quantitative PCR and fluorescence-based strategies. *American Journal of Medical Genetics Part A*, **1993**, 48, 200-8		30
131	CARD15 genotyping in inflammatory bowel disease patients by multiplex pyrosequencing. *Clinical Chemistry*, **2003**, 49, 1675-9	5.5	29
130	In vivo MAPK reporting reveals the heterogeneity in tumoral selection of resistance to RAF inhibitors. *Cancer Research*, **2013**, 73, 7101-10	10.1	28
129	Ethanol potentiates HIV-1 gp120-induced apoptosis in human neurons via both the death receptor and NMDA receptor pathways. *Virology*, **2005**, 334, 59-73	3.6	28
128	Test Pricing and Reimbursement in Genomic Medicine: Towards a General Strategy. *Public Health Genomics*, **2016**, 19, 352-363	1.9	28
127	Clinical exome performance for reporting secondary genetic findings. *Clinical Chemistry*, **2015**, 61, 213-20	5.5	27
126	Surface effects on PCR reactions in multichip microfluidic platforms. *Biomedical Microdevices*, **2004**, 6, 75-80	3.7	27
125	Kinetics of heterogeneous hybridization on indium tin oxide surfaces with and without an applied potential. *Electrophoresis*, **2002**, 23, 1551-7	3.6	27
124	RB and p53 cooperate to prevent liver tumorigenesis in response to tissue damage. *Gastroenterology*, **2011**, 141, 1439-50	13.3	26
123	Detection of the most common mutations causing beta-thalassemia in Mediterraneans using a multiplex amplification refractory mutation system (MARMS). *Genome Research*, **1992**, 2, 163-6	9.7	26
122	Rapid sizing of polymorphic microsatellite markers by capillary array electrophoresis. *Journal of Chromatography A*, **1997**, 781, 295-305	4.5	25

121	Establishment of an orthotopic patient-derived xenograft mouse model using uveal melanoma hepatic metastasis. *Journal of Translational Medicine*, **2017**, 15, 145	8.5	24
120	The frame-shift mutation of the NOD2/CARD15 gene is significantly increased in ulcerative colitis: an *IG-IBD study. *Gastroenterology*, **2004**, 126, 625-7	13.3	24
119	Single-Cell Genomics. *Clinical Chemistry*, **2019**, 65, 972-985	5.5	23
118	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	4.3	23
117	Polymorphic Changes in the 5′ Flanking Region of Factor VII Have a Combined Effect on Promoter Strength. *Thrombosis and Haemostasis*, **2002**, 88, 763-767	7	23
116	The retinoblastoma tumor suppressor pathway modulates the invasiveness of ErbB2-positive breast cancer. *Oncogene*, **2014**, 33, 3980-91	9.2	22
115	System for preparing microhybridization arrays on glass slides. *Analytical Chemistry*, **1998**, 70, 5085-92	7.8	22
114	Fluorescence-based DNA minisequence analysis for detection of known single-base changes in genomic DNA. *Molecular and Cellular Probes*, **1995**, 9, 175-82	3.3	22
113	Cyclin D1-mediated microRNA expression signature predicts breast cancer outcome. *Theranostics*, **2018**, 8, 2251-2263	12.1	21
112	Bridging genomics research between developed and developing countries: the Genomic Medicine Alliance. *Personalized Medicine*, **2014**, 11, 615-623	2.2	21
111	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	4.3	21
110	Glucocorticoids paradoxically facilitate steroid resistance in T cell acute lymphoblastic leukemias and thymocytes. *Journal of Clinical Investigation*, **2020**, 130, 863-876	15.9	21
109	Autosomal recessive congenital ichthyosis: Genomic landscape and phenotypic spectrum in a cohort of 125 consanguineous families. *Human Mutation*, **2019**, 40, 288-298	4.7	21
108	Novel oncogene-induced metastatic prostate cancer cell lines define human prostate cancer progression signatures. *Cancer Research*, **2013**, 73, 978-89	10.1	20
107	Microarray Technology and Applications: An All-Language Literature Survey Including Books and Patents. *Clinical Chemistry*, **2001**, 47, 1479-1482	5.5	20
106	A newly-characterized alpha-thalassaemia-1 deletion removes the entire alpha-like globin gene cluster in an Italian family. *British Journal of Haematology*, **1991**, 78, 529-34	4.5	20
105	The future of laboratory medicine - a 2014 perspective. *Clinica Chimica Acta*, **2015**, 438, 284-303	6.2	19
104	Miniaturized detection technology in molecular diagnostics. *Expert Review of Molecular Diagnostics*, **2005**, 5, 549-59	3.8	19

103	A pilot C282Y hemochromatosis screening in Italian newborns by TaqMan technology. *Genetic Testing and Molecular Biomarkers*, **2000**, 4, 177-81		19
102	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, 67-9	1.4	19
101	Allelic association of microsatellites of 6p in Italian hemochromatosis patients. *Human Genetics*, **1996**, 97, 476-81	6.3	19
100	Performance of exome sequencing for pharmacogenomics. *Personalized Medicine*, **2014**, 12, 109-115	2.2	18
99	Variants in genes involved in functional pathways associated with hypertension in African Americans. *Clinical and Translational Science*, **2010**, 3, 279-86	4.9	17
98	Region-specific detection of neuroblastoma loss of heterozygosity at multiple loci simultaneously using a SNP-based tag-array platform. *Genome Research*, **2005**, 15, 1168-76	9.7	17
97	Rapid detection of medium chain acyl-CoA dehydrogenase gene mutations by non-radioactive, single strand conformation polymorphism minigels. *Journal of Medical Genetics*, **1994**, 31, 551-4	5.8	17
96	Hearts lacking caveolin-1 develop hypertrophy with normal cardiac substrate metabolism. *Cell Cycle* , **2008**, 7, 2509-18	4.7	16
95	Substrate uptake and metabolism are preserved in hypertrophic caveolin-3 knockout hearts. *American Journal of Physiology - Heart and Circulatory Physiology*, **2008**, 295, H657-66	5.2	16
94	Opportunities for near-infrared thermal ablation of colorectal metastases by guanylyl cyclase C-targeted gold nanoshells. *Future Oncology*, **2006**, 2, 705-16	3.6	16
93	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, 312-20	4.7	16
92	Combined segregation and linkage analysis of inflammatory bowel disease in the IBD1 region using severity to characterise Crohn's disease and ulcerative colitis. On behalf of the GISC. *European Journal of Human Genetics*, **2000**, 8, 846-52	5.3	16
91	Small extracellular vesicles modulated by V3 integrin induce neuroendocrine differentiation in recipient cancer cells. *Journal of Extracellular Vesicles*, **2020**, 9, 1761072	16.4	15
90	Next generation sequencing in cancer: opportunities and challenges for precision cancer medicine. *Scandinavian Journal of Clinical and Laboratory Investigation*, **2016**, 245, S84-91	2	15
89	The retinoblastoma tumor suppressor modulates DNA repair and radioresponsiveness. *Clinical Cancer Research*, **2014**, 20, 5468-5482	12.9	15
88	Fluorescence-based, multiplex allele-specific PCR (MASPCR) detection of the delta F508 deletion in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. *Molecular and Cellular Probes*, **1992**, 6, 353-6	3.3	15
87	Inherited Interleukin 2-Inducible T-Cell (ITK) Kinase Deficiency in Siblings With Epidermodysplasia Verruciformis and Hodgkin Lymphoma. *Clinical Infectious Diseases*, **2019**, 68, 1938-1941	11.6	15
86	Targeting CDK6 and BCL2 Exploits the "MYB Addiction" of Ph Acute Lymphoblastic Leukemia. *Cancer Research*, **2018**, 78, 1097-1109	10.1	15

85	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	5.3	14
84	Caveolin-1 overexpression enhances androgen-dependent growth and proliferation in the mouse prostate. *International Journal of Biochemistry and Cell Biology*, **2011**, 43, 1318-29	5.6	14
83	Direct-access genetic testing: the view from Europe. *Nature Reviews Genetics*, **2011**, 12, 670	30.1	14
82	Genotyping beta-globin gene mutations on copolymer-coated glass slides with the ligation detection reaction. *Clinical Chemistry*, **2008**, 54, 1657-63	5.5	14
81	Identification of APC gene mutations in colorectal cancer using universal microarray-based combinatorial sequencing-by-hybridization. *Human Mutation*, **2004**, 24, 261-71	4.7	14
80	A greater than 200 kb deletion removing the entire beta-like globin gene cluster in a family of Irish descent. *Hemoglobin*, **1991**, 15, 23-41	0.6	14
79	A large deletion encompassing the entire alpha-like globin gene cluster in a family of northern European extraction. *Nucleic Acids Research*, **1988**, 16, 11223-35	20.1	14
78	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	2.4	14
77	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	4.7	14
76	Regulation of miR106b cluster through the RB pathway: mechanism and functional targets. *Cell Cycle*, **2013**, 12, 98-111	4.7	13
75	The presurgical management with erythrocytapheresis of a patient with a high-oxygen-affinity, unstable Hb variant (Hb Bryn Mawr). *Transfusion*, **1997**, 37, 703-7	2.9	13
74	Correlation between transfusion requirement, blood volume and haemoglobin level in homozygous beta-thalassaemia. *Acta Haematologica*, **1980**, 64, 103-8	2.7	13
73	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, 127-46	1.4	12
72	Analysis of CARD15 gene variants in Italian pediatric patients with inflammatory bowel diseases. *Journal of Pediatrics*, **2005**, 147, 272-3	3.6	12
71	Leber's 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		12
70	Fluorescent approaches to diagnosis of Lesch-Nyhan syndrome and quantitative analysis of carrier status. *Molecular and Cellular Probes*, **1993**, 7, 311-24	3.3	12
69	Four-laser scanning confocal system for microarray analysis. *BioTechniques*, **2002**, 32, 346-8, 350, 352, 354	2.5	11
68	Donor splice-site mutation in CUL4B is likely cause of X-linked intellectual disability. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2294-9	2.5	10

67	Mutation detection using ligase chain reaction in passivated silicon-glass microchips and microchip capillary electrophoresis. *BioTechniques*, **2004**, 37, 392, 394, 396-8	2.5	10
66	RNA-Binding Protein HuR Promotes Th17 Cell Differentiation and Can Be Targeted to Reduce Autoimmune Neuroinflammation. *Journal of Immunology*, **2020**, 204, 2076-2087	5.3	9
65	Nanotechnology: improving clinical testing?. *Clinical Chemistry*, **2010**, 56, 1384-9	5.5	9
64	Institutional Profile: Golden Helix Institute of Biomedical Research: interdisciplinary research and educational activities in pharmacogenomics and personalized medicine. *Pharmacogenomics*, **2012**, 13, 387-92	2.6	9
63	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	2.2	9
62	Epigenomic profiling of neuroblastoma cell lines. *Scientific Data*, **2020**, 7, 116	8.2	9
61	alpha-thalassemia caused by a 16 bp deletion in the 3' untranslated region of the alpha 2-globin gene including the first nucleotide of the poly A signal sequence. *Hemoglobin*, **1997**, 21, 121-30	0.6	8
60	Combinatorial sequencing-by-hybridization: analysis of the NF1 gene. *Genetic Testing and Molecular Biomarkers*, **2006**, 10, 8-17		8
59	Protein microarrays: a literature survey. *Clinical Chemistry*, **2003**, 49, 2109	5.5	8
58	Microchips: An All-Language Literature Survey Including Books and Patents. *Clinical Chemistry*, **2002**, 48, 1620-1622	5.5	8
57	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	6.3	8
56	Nanotechnology and Applications: An All-Language Literature Survey Including Books and Patents. *Clinical Chemistry*, **2002**, 48, 662-665	5.5	8
55	Infundibulopelvic stenosis, multicystic kidney, and calyectasis in a kindred: clinical observations and genetic analysis. *American Journal of Medical Genetics Part A*, **1995**, 59, 218-24		7
54	A distinct GM-CSF T helper cell subset requires T-bet to adopt a T1 phenotype and promote neuroinflammation. *Science Immunology*, **2020**, 5,	28	7
53	Sighting acute myocardial infarction through platelet gene expression. *Scientific Reports*, **2019**, 9, 19574	4.9	7
52	iSeqQC: a tool for expression-based quality control in RNA sequencing. *BMC Bioinformatics*, **2020**, 21, 56	3.6	6
51	Association of a polymorphic variant of the adiponectin gene with insulin resistance in african americans. *Clinical and Translational Science*, **2008**, 1, 194-9	4.9	6
50	Vestibular and hearing loss in genetic and metabolic disorders. *Current Opinion in Neurology*, **1999**, 12, 35-9	7.1	6

49	A novel autosomal recessive GJB2-associated disorder: Ichthyosis follicularis, bilateral severe sensorineural hearing loss, and punctate palmoplantar keratoderma. *Human Mutation*, **2019**, 40, 217-229	4.7	6
48	Clinical genomics: when whole genome sequencing is like a whole-body CT scan. *Clinical Chemistry*, **2014**, 60, 1390-2	5.5	5
47	Mammary gland selective excision of c-jun identifies its role in mRNA splicing. *Cancer Research*, **2012**, 72, 1023-34	10.1	5
46	Absence of the fragile X CGG trinucleotide repeat expansion in girls diagnosed with a pervasive developmental disorder. *Journal of Pediatrics*, **1998**, 133, 363-5	3.6	5
45	Beyond Microtechnology Nanotechnology in Molecular Diagnosis **2007**, 187-197		5
44	Linkage of ulcerative colitis to the pericentromeric region of chromosome 16 in Italian inflammatory bowel disease families is independent of the presence of common CARD15 mutations. *Journal of Medical Genetics*, **2003**, 40, 837-41	5.8	5
43	Maple syrup urine disease (MSUD): screening for known mutations in Italian patients. *Journal of Inherited Metabolic Disease*, **1994**, 17, 652-60	5.4	5
42	Diagnosis of Duchenne/Becker muscular dystrophy and quantitative identification of carrier status by use of entangled solution capillary electrophoresis. *Clinical Chemistry*, **1997**, 43, 745-51	5.5	5
41	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	2.1	4
40	Mechanisms of endothelial cell attachment, proliferation, and differentiation on 4 types of platinum-based endovascular coils. *World Neurosurgery*, **2014**, 82, 684-95	2.1	4
39	Nanostructured luminescently labeled nucleic acids. *Luminescence*, **2017**, 32, 132-141	2.5	4
38	Response to 'Europe and direct-to-consumer genetic tests'. *Nature Reviews Genetics*, **2012**, 13, 146-146	30.1	4
37	In the Presence of other Inherited or Acquired High-risk Situations, the FV Cambridge Mutation May Be an Additional Thrombophilic Risk Factor, through Its Effect on APC Sensitivity. *Thrombosis and Haemostasis*, **2000**, 83, 963-964	7	4
36	RFLPs of the phenylalanine hydroxylase gene in the Italian population. *Journal of Inherited Metabolic Disease*, **1989**, 12, 162-5	5.4	4
35	Whole-Transcriptome Analysis by RNA Sequencing for Genetic Diagnosis of Mendelian Skin Disorders in the Context of Consanguinity. *Clinical Chemistry*, **2021**, 67, 876-888	5.5	4
34	Genomic test validation for incidental findings. *Clinical Chemistry*, **2014**, 60, 292-3	5.5	3
33	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, 883-7	4.6	3
32	Analytical ancestry: evolution of the array in analysis. *Clinical Chemistry*, **2010**, 56, 1797-803	5.5	3

31	Hb Osler [beta 145(HC2)Tyr-->Asp] results from posttranslational modification. *Hemoglobin*, **1997**, 21, 109-20	0.6	3
30	Multiplex pyrosequencing for DNA variation analysis. *Methods in Molecular Biology*, **2007**, 373, 75-88	1.4	3
29	DOP-PCR amplification of whole genomic DNA and microchip-based capillary electrophoresis. *Methods in Molecular Biology*, **2001**, 163, 211-9	1.4	3
28	Nanotechnology and applications: an all-language literature survey including books and patents. *Clinical Chemistry*, **2002**, 48, 662-5	5.5	3
27	MicroRNAs as new players in the genomic galaxy and disease puzzles. *Clinical and Translational Science*, **2008**, 1, 50-6	4.9	2
26	Fundamental Studies of DNA Adsorption and Hybridization on Solid Surfaces. *ACS Symposium Series* , **1999**, 190-204	0.4	2
25	Targeting chemotherapy to de-condensed H3K27me3-marked chromatin of AML cells enhances leukemia suppression.. *Cancer Research*, **2021**,	10.1	2
24	MicroRNA-139 Expression Is Dispensable for the Generation of Influenza-Specific CD8 T Cell Responses.. *Journal of Immunology*, **2022**,	5.3	2
23	Beyond mRNAs and Mirnas: Unraveling the Full-Spectrum of the Normal Human Platelet Transcriptome Through Next-Generation Sequencing. *Blood*, **2012**, 120, 3298-3298	2.2	2
22	Genetic heterogeneity of heritable ectopic mineralization disorders in a large international cohort.. *Genetics in Medicine*, **2021**,	8.1	2
21	Genetic Predisposition to Numerous Large Ulcerating Basal Cell Carcinomas and Response to Immune Therapy. *International Journal of Dermatology and Venereology*, **2021**, 4, 70-75	0.5	2
20	Validation of a Miniaturized Permeability Assay Compatible with CRISPR-Mediated Genome-Wide Screen. *Scientific Reports*, **2019**, 9, 14238	4.9	1
19	A tandem duplication of chromosome 21 in a newborn showing a phenotype inconsistent with Down syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1043-5	2.5	1
18	Analysis of short tandem repeat markers by capillary array electrophoresis. *Methods in Molecular Biology*, **2001**, 163, 151-61	1.4	1
17	Mapping of Deletional Forms of □and □Thalassemia. *Annals of the New York Academy of Sciences*, **1990**, 612, 480-484	6.5	1
16	Interaction of rare illegitimate recombination event and a poly A addition site mutation resulting in a severe form of alpha thalassemia. *Blood*, **1994**, 83, 3356-62	2.2	1
15	High-Throughput Sequencing of the Human Platelet Transcriptome. *Blood*, **2010**, 116, 481-481	2.2	1
14	Allelic association of microsatellites of 6p in Italian hemochromatosis patients. *Human Genetics*, **1996**, 97, 476-481	6.3	1

13	Identification of FVIII gene mutations in patients with hemophilia A using new combinatorial sequencing by hybridization. *Indian Journal of Human Genetics*, **2008**, 14, 55-64		1
12	Epigenomic profiling of neuroblastoma cell lines		1
11	The Complex Transcriptional Landscape of the Human Platelet. *Blood*, **2012**, 120, 390-390	2.2	1
10	The Genomic Medicine Alliance: A Global Effort to Facilitate the Introduction of Genomics into Healthcare in Developing Nations **2018**, 173-188		1
9	Novel PTCH1 and concurrent TP53 mutations in four patients with numerous non-syndromic basal cell carcinomas: The paradigm of oncogenic synergy. *Experimental Dermatology*, **2021**,	4	1
8	A de novo supernumerary genomic discontinuous ring chromosome 21 in a child with mild intellectual disability. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1425-31	2.5	0
7	The Use of Microelectronic-Based Techniques in Molecular Diagnostic Assays **2010**, 513-526		
6	Large deletions encompassing the entire alpha- and beta-like globin gene clusters in humans. *Progress in Clinical and Biological Research*, **1989**, 316B, 151-9		
5	Recent Developments in Miniaturized PCR-Microchips, Microarrays and Microdroplets. *Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine*, **2012**, 23, 76-9	2.4	
4	Surface Modification and Hybridization on a Thermal Gradient DNA Chip **2001**, 585-586		
3	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	2.2	
2	Towards a Reference Human Platelet Transcriptome: Evaluation Of Inter-Individual Correlations and Its Relationship With a Platelet Proteome. *Blood*, **2013**, 122, 2297-2297	2.2	
1	Applied Genomics and Public Health Cancer Genomics **2020**, 53-72		