Ephrat Levy-Lahad

List of Publications by Year
in descending order

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163 papers	15,173 citations	50276 46 h-index	18130 120 g-index
167 all docs	167 docs citations	167 times ranked	17806 citing authors

#	Article	IF	Citations
1	The annual ASHG dinner. American Journal of Human Genetics, 2022, 109, 377-378.	6.2	1
2	Germline Pathogenic Variants in BRCA1 and BRCA2: Malignancies Beyond Female Breast and Ovarian Cancers. Journal of Clinical Oncology, 2022, , JCO2200003.	1.6	0
3	Salt-Losing 21-Hydroxylase Deficiency Caused by Double Homozygosity for Two â€œMildâ€•Mutations. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e680-e686.	3.6	3
4	Performance comparison: exome sequencing as a single test replacing Sanger sequencing. Molecular Genetics and Genomics, 2021, 296, 653-663.	2.1	7
5	The landscape of autosomal-recessive pathogenic variants in European populations reveals phenotype-specific effects. American Journal of Human Genetics, 2021, 108, 608-619.	6.2	36
6	Breast cancer risk and hormone replacement therapy among BRCA carriers after risk-reducing salpingo-oophorectomy. European Journal of Cancer, 2021, 148, 95-102.	2.8	16
7	Preconception carrier screening yield: effect of variants of unknown significance in partners of carriers with clinically significant variants. Genetics in Medicine, 2020, 22, 646-653.	2.4	18
8	Israeli Position Paper. Chest, 2020, 158, 2278-2281.	0.8	5
9	NKX2-2 Mutation Causes Congenital Diabetes and Infantile Obesity With Paradoxical Glucose-Induced Ghrelin Secretion. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 3486-3495.	3.6	11
10	Presymptomatic Awareness of Germline Pathogenic BRCA Variants and Associated Outcomes in Women With Breast Cancer. JAMA Oncology, 2020, 6, 1460.	7.1	18
11	Age at diagnosis of cancer in 185delAG BRCA1 mutation carriers of diverse ethnicities: tentative evidence for modifier factors. Familial Cancer, 2020, 20, 189-194.	1.9	1
12	Hiding in Plain Sight â€” Somatic Mutation in Human Disease. New England Journal of Medicine, 2020, 383, 2680-2682.	27.0	16
13	A defect in GPI synthesis as a suggested mechanism for the role of ARV1 in intellectual disability and seizures. Neurogenetics, 2020, 21, 259-267.	1.4	9
14	Population Screening for Inherited Predisposition to Breast and Ovarian Cancer. Annual Review of Genomics and Human Genetics, 2020, 21, 373-412.	6.2	31
15	Israeli Position Paper: Triage Decisions for Severely Ill Patients During the COVID-19 Pandemic. Joint Commission of the Israel National Bioethics Council, the Ethics Bureau of the Israel Medical Association and Representatives from the Israeli Ministry of Health. Rambam Maimonides Medical Journal, 2020, 11, e0019.	1.0	4
16	Helicase-inactivating BRIP1 mutation yields Fanconi anemia with microcephaly and other congenital abnormalities. Journal of Physical Education and Sports Management, 2020, 6, a005652.	1.2	2
17	A Unique Presentation of XY Gonadal Dysgenesis in Frasier Syndrome due to WT1 Mutation and a Literature Review. Pediatric Endocrinology Reviews, 2020, 17, 302-307.	1.2	2
18	The yield of full BRCA1/2 genotyping in Israeli Arab high-risk breast/ovarian cancer patients. Breast Cancer Research and Treatment, 2019, 178, 231-237.	2.5	7

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19	Analysis of the genetic basis of height in large Jewish nuclear families. PLoS Genetics, 2019, 15, e1008082.	3.5	1
20	Homozygous stop-gain variant in LRRC32, encoding a TGFÎ² receptor, associated with cleft palate, proliferative retinopathy, and developmental delay. European Journal of Human Genetics, 2019, 27, 1315-1319.	2.8	7
21	Variable Features of Juvenile Polyposis Syndrome With Gastric Involvement Among Patients With a Large Genomic Deletion of BMPR1A. Clinical and Translational Gastroenterology, 2019, 10, e00054.	2.5	9
22	Haploseek: a 24-hour all-in-one method for preimplantation genetic diagnosis (PGD) of monogenic disease and aneuploidy. Genetics in Medicine, 2019, 21, 1390-1399.	2.4	35
23	Clinical testing of BRCA1 and BRCA2: a worldwide snapshot of technological practices. Npj Genomic Medicine, 2018, 3, 7.	3.8	44
24	Familial communication and cascade testing among relatives of BRCA population screening participants. Genetics in Medicine, 2018, 20, 1446-1454.	2.4	67
25	Off the street phasing (OTSP): free no hassle haplotype phasing for molecular PGD applications. Reproductive BioMedicine Online, 2018, 36, e34.	2.4	0
26	American Funding Cutback to East Jerusalem Hospitals: A Blow to the Health of the City. American Journal of Public Health, 2018, 108, 1624-1625.	2.7	2
27	Noninvasive paternal exclusion testing for cystic fibrosis in the first five to eight weeks of gestation. Scientific Reports, 2018, 8, 15941.	3.3	1
28	Essential Role ofBRCA2in Ovarian Development and Function. New England Journal of Medicine, 2018, 379, 1042-1049.	27.0	79
29	Mitochondrial PITRM1 peptidase loss-of-function in childhood cerebellar atrophy. Journal of Medical Genetics, 2018, 55, 599-606.	3.2	26
30	Vrk1 partial Knockdown in Mice Results in Reduced Brain Weight and Mild Motor Dysfunction, and Indicates Neuronal VRK1 Target Pathways. Scientific Reports, 2018, 8, 11265.	3.3	14
31	Impact of germline BRCA identification on subsequent breast cancer stage and therapy: Implications for routine screening.. Journal of Clinical Oncology, 2018, 36, 1584-1584.	1.6	1
32	Features of Patients With Hereditary Mixed Polyposis Syndrome Caused by Duplication of GREM1 and Implications for Screening and Surveillance. Gastroenterology, 2017, 152, 1876-1880.e1.	1.3	34
33	Vesicular acetylcholine transporter defect underlies devastating congenital myasthenia syndrome. Neurology, 2017, 88, 1021-1028.	1.1	25
34	Medical genetics in Israel's diverse population. Lancet, The, 2017, 389, 2453-2455.	13.7	9
35	Genomic analysis of inherited breast cancer among Palestinian women: Genetic heterogeneity and a founder mutation in TP53. International Journal of Cancer, 2017, 141, 750-756.	5.1	40
36	Population screening for BRCA1/BRCA2 mutations: lessons from qualitative analysis of the screening experience. Genetics in Medicine, 2017, 19, 628-634.	2.4	25

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37	Population screening for BRCA1/BRCA2 founder mutations in Ashkenazi Jews: proactive recruitment compared with self-referral. Genetics in Medicine, 2017, 19, 754-762.	2.4	44
38	Human Embryo Editing: Opportunities and Importance of Transnational Cooperation. Cell Stem Cell, 2017, 21, 423-426.	11.1	21
39	Cost-effectiveness of population based BRCA testing with varying Ashkenazi Jewish ancestry. American Journal of Obstetrics and Gynecology, 2017, 217, 578.e1-578.e12.	1.3	63
40	Brain calcifications and PCDH12 variants. Neurology: Genetics, 2017, 3, e166.	1.9	15
41	Abstract P3-08-06: Screening, management, cancer diagnoses, and outcomes of women with germline BRCA mutations in Israel: The Noga Clinic experience. , 2017, , .		0
42	Loss of function of PCDH12 underlies recessive microcephaly mimicking intrauterine infection. Neurology, 2016, 86, 2016-2024.	1.1	32
43	Expanding the phenotype of <scp>CRB2</scp> mutationsÂâ€“ÂA new ciliopathy syndrome?. Clinical Genetics, 2016, 90, 540-544.	2.0	18
44	A role forTENM1mutations in congenital general anosmia. Clinical Genetics, 2016, 90, 211-219.	2.0	59
45	Developmental neuropsychological assessment of 4- to 5-year-old children born following Preimplantation Genetic Diagnosis (PGD): A pilot study. Child Neuropsychology, 2016, 22, 458-471.	1.3	12
46	Inherited Breast Cancer. , 2016, , 315-327.		0
47	TODRA, a lncRNA at the RAD51 Locus, Is Oppositely Regulated to RAD51, and Enhances RAD51-Dependent DSB (Double Strand Break) Repair. PLoS ONE, 2015, 10, e0134120.	2.5	35
48	Establishment of Homozygote Mutant Human Embryonic Stem Cells by Parthenogenesis. PLoS ONE, 2015, 10, e0138893.	2.5	5
49	Minichromosome maintenance complex component 8 (MCM8) gene mutations result in primary gonadal failure. Journal of Medical Genetics, 2015, 52, 391-399.	3.2	97
50	Proposed Shift in Screening for Breast Cancerâ€”Reply. JAMA - Journal of the American Medical Association, 2015, 313, 525.	7.4	3
51	The Spinal Muscular Atrophy with Pontocerebellar Hypoplasia GeneVRK1Regulates Neuronal Migration through an Amyloid-Î² Precursor Protein-Dependent Mechanism. Journal of Neuroscience, 2015, 35, 936-942.	3.6	36
52	VRK1 regulates Cajal body dynamics and protects coilin from proteasomal degradation in cell cycle. Scientific Reports, 2015, 5, 10543.	3.3	33
53	Combined mineralocorticoid and glucocorticoid deficiency is caused by a novel founder nicotinamide nucleotide transhydrogenase mutation that alters mitochondrial morphology and increases oxidative stress. Journal of Medical Genetics, 2015, 52, 636-641.	3.2	26
54	Copy number variations in cryptogenic cerebral palsy. Neurology, 2015, 84, 1660-1668.	1.1	82

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55	Preimplantation genetic risk reduction: a new dilemma in the era of chromosomal microarrays and exome sequencing. Reproductive BioMedicine Online, 2015, 31, 706-710.	2.4	7
56	Uncovering the Role of Hypermethylation by CTG Expansion in Myotonic Dystrophy Type 1ÂUsing Mutant Human Embryonic Stem Cells. Stem Cell Reports, 2015, 5, 221-231.	4.8	40
57	Alphaâ€thalassemia intellectual disability: variable phenotypic expression among males with a recurrent nonsense mutationÂâ€“Âc.<scp>109C</scp>>T (p.<scp>R37X</scp>). Clinical Genetics, 2015, 87, 461-466.	2.0	12
58	Proof-of-principle rapid noninvasive prenatal diagnosis of autosomal recessive founder mutations. Journal of Clinical Investigation, 2015, 125, 3757-3765.	8.2	25
59	A mutation in the nucleoporin-107 gene causes XX gonadal dysgenesis. Journal of Clinical Investigation, 2015, 125, 4295-4304.	8.2	77
60	Precision Medicine Meets Public Health: Population Screening for BRCA1 and BRCA2. Journal of the National Cancer Institute, 2014, 107, dju420-dju420.	6.3	57
61	Mutant Adenosine Deaminase 2 in a Polyarteritis Nodosa Vasculopathy. New England Journal of Medicine, 2014, 370, 921-931.	27.0	566
62	FMR1 Epigenetic Silencing Commonly Occurs in Undifferentiated Fragile X-Affected Embryonic Stem Cells. Stem Cell Reports, 2014, 3, 699-706.	4.8	66
63	BRCA Mutation Carriers Do Not Have Compromised Ovarian Reserve. International Journal of Gynecological Cancer, 2014, 24, 233-237.	2.5	68
64	Population-Based Screening forBRCA1andBRCA2. JAMA - Journal of the American Medical Association, 2014, 312, 1091.	7.4	236
65	High school Tayâ€“Sachs disease carrier screening: 5 to 11-year follow-up. Journal of Community Genetics, 2014, 5, 139-146.	1.2	11
66	Neonatal outcome after preimplantation genetic diagnosis. Fertility and Sterility, 2014, 102, 1016-1021.	1.0	44
67	Population-based screening for breast and ovarian cancer risk due to BRCA1 and BRCA2. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 14205-14210.	7.1	286
68	Prenatal Isolated Ventricular Septal Defect May Not Be Associated with Trisomy 21. Journal of Clinical Medicine, 2014, 3, 432-439.	2.4	8
69	Testicular differentiation factor SF-1 is required for human spleen development. Journal of Clinical Investigation, 2014, 124, 2071-2075.	8.2	36
70	Familial haplotyping and embryo analysis for Preimplantation Genetic Diagnosis (PGD) using DNA microarrays: a proof of principle study. Journal of Assisted Reproduction and Genetics, 2013, 30, 1595-1603.	2.5	17
71	Parkin acts as a transcription factor modulating presenilin-1 and presenilin-2 promoter transactivations. Molecular Neurodegeneration, 2013, 8, P56.	10.8	0
72	Mutations in LARS2, Encoding Mitochondrial Leucyl-tRNA Synthetase, Lead to Premature Ovarian Failure and Hearing Loss in Perrault Syndrome. American Journal of Human Genetics, 2013, 92, 614-620.	6.2	176

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73	Preimplantation Genetic Diagnosis in Genomic Regions with Duplications and Pseudogenes: Long-Range PCR in the Single-Cell Assay. Human Mutation, 2013, 34, 792-799.	2.5	7
74	Parkin differently regulates presenilin-1 and presenilin-2 functions by direct control of their promoter transcription. Journal of Molecular Cell Biology, 2013, 5, 132-142.	3.3	31
75	Snord 3A: A Molecular Marker and Modulator of Prion Disease Progression. PLoS ONE, 2013, 8, e54433.	2.5	14
76	A novel severe N-terminal splice site KISS1R gene mutation causes hypogonadotropic hypogonadism but enables a normal development of neonatal external genitalia. European Journal of Endocrinology, 2012, 167, 209-216.	3.7	20
77	Prevention of Lysosomal Storage Diseases and Derivation of Mutant Stem Cell Lines by Preimplantation Genetic Diagnosis. Molecular Biology International, 2012, 2012, 1-9.	1.7	13
78	Preimplantation Genetic Diagnosis for Fetal Neonatal Alloimmune Thrombocytopenia Due to Antihuman Platelet Antigen Maternal Antibodies. Obstetrics and Gynecology, 2012, 119, 338-343.	2.4	8
79	Genome-wide survey reveals predisposing diabetes type 2-related DNA methylation variations in human peripheral blood. Human Molecular Genetics, 2012, 21, 371-383.	2.9	317
80	PGD for germline mosaicism. Reproductive BioMedicine Online, 2012, 25, 390-395.	2.4	12
81	Female Sex Bias in Human Embryonic Stem Cell Lines. Stem Cells and Development, 2012, 21, 363-372.	2.1	27
82	BRCA genetic testing of individuals from families with low prevalence of cancer: experiences of carriers and implications for population screening. Genetics in Medicine, 2012, 14, 688-694.	2.4	16
83	Preimplantation genetic diagnosis (PGD) for a treatable disorder: Gaucher disease type 1 as a model. Blood Cells, Molecules, and Diseases, 2011, 46, 15-18.	1.4	13
84	Substrate profiling of human vaccinia-related kinases identifies coilin, a Cajal body nuclear protein, as a phosphorylation target with neurological implications. Journal of Proteomics, 2011, 75, 548-560.	2.4	37
85	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in middle eastern families. Genome Biology, 2011, 12, R89.	9.6	183
86	Preimplantation genetic diagnosis (PGD) â€“ prevention of the birth of children affected with endocrine diseases. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 543-8.	0.9	15
87	XX Ovarian Dysgenesis Is Caused by a PSMC3IP/HOP2 Mutation that Abolishes Coactivation of Estrogen-Driven Transcription. American Journal of Human Genetics, 2011, 89, 572-579.	6.2	99
88	Preimplantation genetic diagnosis (PGD) for SHOX-related haploinsufficiency in conjunction with trisomy 21 detection by molecular analysis. Journal of Assisted Reproduction and Genetics, 2011, 28, 233-238.	2.5	1
89	Preventing mucopolysaccharidosis type II (Hunter syndrome): PGD and establishing a Hunter (46, XX) stem cell line. Prenatal Diagnosis, 2011, 31, 853-860.	2.3	14
90	A non-pathogenic pseudoautosomal region 1 (PAR1) copy number variant downstream of SHOX. , 2011, 155, 938-939.		0

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91	Conflicts regarding genetic counseling for fragile X syndrome screening: A survey of clinical geneticists and genetic counselors in Israel. American Journal of Medical Genetics, Part A, 2011, 155, 2154-2160.	1.2	3
92	A deleterious founder mutation in the BMPER gene causes diaphanospondylodysostosis (DSD). American Journal of Medical Genetics, Part A, 2011, 155, 2801-2806.	1.2	14
93	Garrod's fourth inborn error of metabolism solved by the identification of mutations causing pentosuria. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 18313-18317.	7.1	11
94	Identification of BRCA1/BRCA2 carriers by screening in the healthy population and its implications.. Journal of Clinical Oncology, 2011, 29, 1513-1513.	1.6	6
95	Mutations in the DBP-Deficiency Protein HSD17B4 Cause Ovarian Dysgenesis, Hearing Loss, and Ataxia of Perrault Syndrome. American Journal of Human Genetics, 2010, 87, 282-288.	6.2	231
96	Mosaic compound heterozygosity of SHOX resulting in Leriâ€“Weill dyschondrosteosis with marked short stature: Implications for disease mechanisms and recurrence risks. American Journal of Medical Genetics, Part A, 2010, 152A, 2230-2235.	1.2	3
97	Functional variant of KLOTHO: a breast cancer risk modifier among BRCA1 mutation carriers of Ashkenazi origin. Oncogene, 2010, 29, 26-33.	5.9	46
98	Fanconi anemia and breast cancer susceptibility meet again. Nature Genetics, 2010, 42, 368-369.	21.4	54
99	Collaborative genomics for human health and cooperation in the Mediterranean region. Nature Genetics, 2010, 42, 641-645.	21.4	26
100	PGD for fragile X syndrome: ovarian function is the main determinant of success. Human Reproduction, 2010, 25, 2629-2636.	0.9	26
101	Frequencies of C282Y and H63D alleles in the HFE gene among various Jewish ethnic groups in Israel: A change of concept required. Genetics in Medicine, 2010, 12, 122-125.	2.4	6
102	Real-time reverse linkage using polar body analysis for preimplantation genetic diagnosis in female carriers of de novo mutations. Human Reproduction, 2009, 24, 3225-3229.	0.9	20
103	Population-based BRCA1/BRCA2 screening in Ashkenazi Jews: A call for evidence. Genetics in Medicine, 2009, 11, 620-621.	2.4	6
104	p53-dependent control of transactivation of the Pen2 promoter by presenilins. Journal of Cell Science, 2009, 122, 4003-4008.	2.0	21
105	Pulmonary hypoplasiaâ€“diaphragmatic herniaâ€“anophthalmiaâ€“cardiac defect (PDAC) syndrome due to STRA6 mutationsâ€”What are the minimal criteria?. American Journal of Medical Genetics, Part A, 2009, 149A, 2457-2463.	1.2	35
106	Preimplantation genetic diagnosis (PGD) for nonsyndromic deafness by polar body and blastomere biopsy. Journal of Assisted Reproduction and Genetics, 2009, 26, 391-397.	2.5	24
107	Spinal Muscular Atrophy with Pontocerebellar Hypoplasia Is Caused by a Mutation in the VRK1 Gene. American Journal of Human Genetics, 2009, 85, 281-289.	6.2	162
108	Transcriptional regulation of the murine Presenilin-2 gene reveals similarities and differences to its human orthologue. Gene, 2009, 446, 81-89.	2.2	9

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109	Non-invasive prenatal diagnosis using cell-free fetal DNA in maternal plasma from PGD pregnancies. Reproductive BioMedicine Online, 2009, 19, 714-720.	2.4	5
110	PGD on a recombinant allele: crossover between the TSC2 gene and â€˜linkedâ€™ markers impairs accurate diagnosis. Prenatal Diagnosis, 2008, 28, 929-933.	2.3	27
111	Attitudes of couples identified through screening as carriers of Gaucher disease type 1. Clinical Genetics, 2008, 74, 566-570.	2.0	9
112	Successful polar body-based preimplantation genetic diagnosis for achondroplasia. Reproductive BioMedicine Online, 2008, 16, 276-282.	2.4	22
113	New ISSCR Guidelines Underscore Major Principles for Responsible Translational Stem Cell Research. Cell Stem Cell, 2008, 3, 607-609.	11.1	218
114	Polar Body-Based Preimplantation Genetic Diagnosis for N-Acetylglutamate Synthase Deficiency. Fetal Diagnosis and Therapy, 2008, 24, 170-176.	1.4	7
115	(TA)n UDP-Glucuronosyltransferase 1A1 Promoter Polymorphism in Nigerian Neonates. Pediatric Research, 2008, 63, 109-111.	2.3	26
116	Response to ACMG guideline: Carrier screening in individuals of Ashkenazi Jewish decent. Genetics in Medicine, 2008, 10, 462-462.	2.4	3
117	Cancer risks among BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2007, 96, 11-15.	6.4	296
118	(TA)n UGT 1A1 Promoter Polymorphism: A Crucial Factor in the Pathophysiology of Jaundice in G-6-PD Deficient Neonates. Pediatric Research, 2007, 61, 727-731.	2.3	35
119	Carrier Screening for Gaucher Disease. JAMA - Journal of the American Medical Association, 2007, 298, 1281.	7.4	68
120	Ethnic ancestry and increased paternal age are risk factors for breast cancer before the age of 40 years. European Journal of Cancer Prevention, 2007, 16, 549-554.	1.3	16
121	Simultaneous preimplantation genetic diagnosis for Tayâ€“Sachs and Gaucher disease. Reproductive BioMedicine Online, 2007, 15, 83-88.	2.4	25
122	Advantages of multiple markers and polar body analysis in preimplantation genetic diagnosis for Alagille disease. Prenatal Diagnosis, 2007, 27, 317-321.	2.3	20
123	A dual role for interleukin-1 in hippocampal-dependent memory processes. Psychoneuroendocrinology, 2007, 32, 1106-1115.	2.7	408
124	Single-sperm analysis for haplotype construction of de-novo paternal mutations: application to PGD for neurofibromatosis type 1. Human Reproduction, 2006, 21, 2047-2051.	0.9	25
125	Familial clustering of site-specific cancer risks associated with BRCA1 and BRCA2 mutations in the Ashkenazi Jewish population. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 3770-3774.	7.1	81
126	Laparoscopic oophorectomy combined with breast surgery for breast cancer patients. Annals of Cancer Research and Therapy, 2006, 14, 19-22.	0.3	1

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127	Molecular study ofWISP3 in nine families originating from the Middle-East and presenting with progressive pseudorheumatoid dysplasia: Identification of two novel mutations, and description of a founder effect. American Journal of Medical Genetics, Part A, 2005, 138A, 118-126.	1.2	47
128	Functional and genomic approaches reveal an ancient CHEK2 allele associated with breast cancer in the Ashkenazi Jewish population. Human Molecular Genetics, 2005, 14, 555-563.	2.9	109
129	A single Mediterranean, possibly Jewish, origin for the Val59Gly CDKN2A mutation in four melanoma-prone families. European Journal of Human Genetics, 2003, 11, 288-296.	2.8	24
130	Egr-1 upregulates the Alzheimer's disease presenilin-2 gene in neuronal cells. Gene, 2003, 318, 113-124.	2.2	44
131	CANCER: Enhanced: A Risky Business–Assessing Breast Cancer Risk. Science, 2003, 302, 574-575.	12.6	22
132	Hemolysis and bilirubin conjugation in association with UDP-glucuronosyltransferase 1A1 promoter polymorphism. Hepatology, 2002, 35, 905-911.	7.3	50
133	Neonatal diabetes mellitus, enteropathy, thrombocytopenia, and endocrinopathy: Further evidence for an X-linked lethal syndrome. Journal of Pediatrics, 2001, 138, 577-580.	1.8	74
134	X-linked neonatal diabetes mellitus, enteropathy and endocrinopathy syndrome is the human equivalent of mouse scurfy. Nature Genetics, 2001, 27, 18-20.	21.4	1,648
135	A single nucleotide polymorphism in the RAD51 gene modifies cancer risk in BRCA2 but not BRCA1 carriers. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 3232-3236.	7.1	178
136	Differing Pathogenesis of Perinatal Bilirubinemia in Glucose-6-Phosphate Dehydrogenase-Deficient Versus-Normal Neonates. Pediatric Research, 2001, 50, 532-537.	2.3	32
137	Why should primary care physicians know about breast cancer genetics?. Western Journal of Medicine, 2001, 175, 168-173.	0.3	7
138	BRCA1 Germline Mutations in Women With Uterine Serous Papillary Carcinoma. Obstetrics and Gynecology, 2000, 96, 28-32.	2.4	2
139	BRCA1 and BRCA2 mutation carriers as potential candidates for chemoprevention trials. Journal of Cellular Biochemistry, 2000, 77, 13-18.	2.6	2
140	Incidence of thrombophilia in patients with Gaucher disease. American Journal of Medical Genetics Part A, 2000, 95, 429-431.	2.4	12
141	The Founder Mutations in theBRCA1,BRCA2, andATMGenes in Moroccan Jewish Women with Breast Cancer. Genetic Testing and Molecular Biomarkers, 2000, 4, 403-407.	1.7	7
142	BRCA1 germline mutations in women with uterine serous papillary carcinoma. Obstetrics and Gynecology, 2000, 96, 28-32.	2.4	43
143	Gilbert's syndrome and hyperbilirubinaemia in ABO-incompatible neonates. Lancet, The, 2000, 356, 652-653.	13.7	65
144	Is Uterine Serous Papillary Carcinoma a BRCA1-Related Disease? Case Report and Review of the Literature. Gynecologic Oncology, 1999, 75, 300-304.	1.4	79

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145	Neonatal Hyperbilirubinemia in Glucose-6-Phosphate Dehydrogenase-deficient Heterozygotes. Pediatrics, 1999, 104, 68-74.	2.1	119
146	High Frequency of Factor V Leiden in a Population of Israeli Arabs. Thrombosis and Haemostasis, 1999, 82, 1768-1768.	3.4	21
147	Prevalence of glucocerebrosidase mutations in the Israeli Ashkenazi Jewish population. Human Mutation, 1998, 12, 240-244.	2.5	84
148	Monogenic determinants of familial Alzheimer's disease: presenilin-2 mutations. Cellular and Molecular Life Sciences, 1998, 54, 910-919.	5.4	17
149	Double Heterozygotes for the Ashkenazi Founder Mutations in BRCA1 and BRCA2 Genes. American Journal of Human Genetics, 1998, 63, 1224-1227.	6.2	47
150	Recent Advances in the Genetics of Alzheimer's Disease. Journal of Geriatric Psychiatry and Neurology, 1998, 11, 42-54.	2.3	41
151	Gilbert syndrome and glucose-6-phosphate dehydrogenase deficiency: A dose-dependent genetic interaction crucial to neonatalâ€‰hyperbilirubinemia. Proceedings of the National Academy of Sciences of the United States of America, 1997, 94, 12128-12132.	7.1	316
152	10 Gaucher's disease: genetic counselling and population screening. Best Practice and Research: Clinical Haematology, 1997, 10, 779-792.	1.1	12
153	High Frequency of BRCA1 and BRCA2 Germline Mutations in Ashkenazi Jewish Ovarian Cancer Patients, Regardless of Family History. Gynecologic Oncology, 1997, 67, 123-126.	1.4	79
154	Amyloid (A?) deposition in chromosome 1-linked Alzheimer's disease: The volga german families. Annals of Neurology, 1997, 41, 52-57.	5.3	54
155	Genomic Structure and Expression of STM2, the Chromosome 1 Familial Alzheimer Disease Gene. Genomics, 1996, 34, 198-204.	2.9	67
156	Secreted amyloid Î²â€“protein similar to that in the senile plaques of Alzheimer's disease is increased in vivo by the presenilin 1 and 2 and APP mutations linked to familial Alzheimer's disease. Nature Medicine, 1996, 2, 864-870.	30.7	2,545
157	Genetic factors in Alzheimer's disease: A review of recent advances. Annals of Neurology, 1996, 40, 829-840.	5.3	80
158	Wide range in age of onset for chromosome 1-related familial Alzheimer's disease. Annals of Neurology, 1996, 40, 932-936.	5.3	87
159	Apolipoprotein E genotypes and age of onset in early-onset familial Alzheimer's disease. Annals of Neurology, 1995, 38, 678-680.	5.3	68
160	Candidate Gene for the Chromosome 1 Familial Alzheimer's Disease Locus. Science, 1995, 269, 973-977.	12.6	2,455
161	A Familial Alzheimer's Disease Locus on Chromosome 1. Science, 1995, 269, 970-973.	12.6	768
162	Low-dose enzyme replacement therapy for Gaucher's disease: Effects of age, sex, genotype, and clinical features on response to treatment. American Journal of Medicine, 1994, 97, 3-13.	1.5	74

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163	Low-dose high-frequency enzyme replacement therapy for very young children with severe Gaucher disease. British Journal of Haematology, 1993, 85, 783-786.	2.5	37