

# 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	Secreted amyloid $\beta$ 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. <i>Nature Medicine</i> , 1996, 2, 864-870.	30.7	2,545
2	Candidate Gene for the Chromosome 1 Familial Alzheimer's Disease Locus. <i>Science</i> , 1995, 269, 973-977.	12.6	2,455
3	X-linked neonatal diabetes mellitus, enteropathy and endocrinopathy syndrome is the human equivalent of mouse scurfy. <i>Nature Genetics</i> , 2001, 27, 18-20.	21.4	1,648
4	A Familial Alzheimer's Disease Locus on Chromosome 1. <i>Science</i> , 1995, 269, 970-973.	12.6	768
5	Mutant Adenosine Deaminase 2 in a Polyarteritis Nodosa Vasculopathy. <i>New England Journal of Medicine</i> , 2014, 370, 921-931.	27.0	566
6	A dual role for interleukin-1 in hippocampal-dependent memory processes. <i>Psychoneuroendocrinology</i> , 2007, 32, 1106-1115.	2.7	408
7	Genome-wide survey reveals predisposing diabetes type 2-related DNA methylation variations in human peripheral blood. <i>Human Molecular Genetics</i> , 2012, 21, 371-383.	2.9	317
8	Gilbert syndrome and glucose-6-phosphate dehydrogenase deficiency: A dose-dependent genetic interaction crucial to neonatal hyperbilirubinemia. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1997, 94, 12128-12132.	7.1	316
9	Cancer risks among BRCA1 and BRCA2 mutation carriers. <i>British Journal of Cancer</i> , 2007, 96, 11-15.	6.4	296
10	Population-based screening for breast and ovarian cancer risk due to <i>BRCA1</i> and <i>BRCA2</i> . <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 14205-14210.	7.1	286
11	Population-Based Screening for <i>BRCA1</i> and <i>BRCA2</i> . <i>JAMA - Journal of the American Medical Association</i> , 2014, 312, 1091.	7.4	236
12	Mutations in the DBP-Deficiency Protein HSD17B4 Cause Ovarian Dysgenesis, Hearing Loss, and Ataxia of Perrault Syndrome. <i>American Journal of Human Genetics</i> , 2010, 87, 282-288.	6.2	231
13	New ISSCR Guidelines Underscore Major Principles for Responsible Translational Stem Cell Research. <i>Cell Stem Cell</i> , 2008, 3, 607-609.	11.1	218
14	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in middle eastern families. <i>Genome Biology</i> , 2011, 12, R89.	9.6	183
15	A single nucleotide polymorphism in the RAD51 gene modifies cancer risk in BRCA2 but not BRCA1 carriers. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2001, 98, 3232-3236.	7.1	178
16	Mutations in LARS2, Encoding Mitochondrial Leucyl-tRNA Synthetase, Lead to Premature Ovarian Failure and Hearing Loss in Perrault Syndrome. <i>American Journal of Human Genetics</i> , 2013, 92, 614-620.	6.2	176
17	Spinal Muscular Atrophy with Pontocerebellar Hypoplasia Is Caused by a Mutation in the VRK1 Gene. <i>American Journal of Human Genetics</i> , 2009, 85, 281-289.	6.2	162
18	Neonatal Hyperbilirubinemia in Glucose-6-Phosphate Dehydrogenase-deficient Heterozygotes. <i>Pediatrics</i> , 1999, 104, 68-74.	2.1	119

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19	Functional and genomic approaches reveal an ancient CHEK2 allele associated with breast cancer in the Ashkenazi Jewish population. <i>Human Molecular Genetics</i> , 2005, 14, 555-563.	2.9	109
20	XX Ovarian Dysgenesis Is Caused by a PSMC3IP/HOP2 Mutation that Abolishes Coactivation of Estrogen-Driven Transcription. <i>American Journal of Human Genetics</i> , 2011, 89, 572-579.	6.2	99
21	Minichromosome maintenance complex component 8 (MCM8) gene mutations result in primary gonadal failure. <i>Journal of Medical Genetics</i> , 2015, 52, 391-399.	3.2	97
22	Wide range in age of onset for chromosome 1-related familial Alzheimer's disease. <i>Annals of Neurology</i> , 1996, 40, 932-936.	5.3	87
23	Prevalence of glucocerebrosidase mutations in the Israeli Ashkenazi Jewish population. <i>Human Mutation</i> , 1998, 12, 240-244.	2.5	84
24	Copy number variations in cryptogenic cerebral palsy. <i>Neurology</i> , 2015, 84, 1660-1668.	1.1	82
25	Familial clustering of site-specific cancer risks associated with BRCA1 and BRCA2 mutations in the Ashkenazi Jewish population. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 3770-3774.	7.1	81
26	Genetic factors in Alzheimer's disease: A review of recent advances. <i>Annals of Neurology</i> , 1996, 40, 829-840.	5.3	80
27	High Frequency of BRCA1 and BRCA2 Germline Mutations in Ashkenazi Jewish Ovarian Cancer Patients, Regardless of Family History. <i>Gynecologic Oncology</i> , 1997, 67, 123-126.	1.4	79
28	Is Uterine Serous Papillary Carcinoma a BRCA1-Related Disease? Case Report and Review of the Literature. <i>Gynecologic Oncology</i> , 1999, 75, 300-304.	1.4	79
29	Essential Role of <i>BRCA2</i> in Ovarian Development and Function. <i>New England Journal of Medicine</i> , 2018, 379, 1042-1049.	27.0	79
30	A mutation in the nucleoporin-107 gene causes XX gonadal dysgenesis. <i>Journal of Clinical Investigation</i> , 2015, 125, 4295-4304.	8.2	77
31	Low-dose enzyme replacement therapy for Gaucher's disease: Effects of age, sex, genotype, and clinical features on response to treatment. <i>American Journal of Medicine</i> , 1994, 97, 3-13.	1.5	74
32	Neonatal diabetes mellitus, enteropathy, thrombocytopenia, and endocrinopathy: Further evidence for an X-linked lethal syndrome. <i>Journal of Pediatrics</i> , 2001, 138, 577-580.	1.8	74
33	Apolipoprotein E genotypes and age of onset in early-onset familial Alzheimer's disease. <i>Annals of Neurology</i> , 1995, 38, 678-680.	5.3	68
34	Carrier Screening for Gaucher Disease. <i>JAMA - Journal of the American Medical Association</i> , 2007, 298, 1281.	7.4	68
35	BRCA Mutation Carriers Do Not Have Compromised Ovarian Reserve. <i>International Journal of Gynecological Cancer</i> , 2014, 24, 233-237.	2.5	68
36	Genomic Structure and Expression of STM2, the Chromosome 1 Familial Alzheimer Disease Gene. <i>Genomics</i> , 1996, 34, 198-204.	2.9	67

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37	Familial communication and cascade testing among relatives of BRCA population screening participants. <i>Genetics in Medicine</i> , 2018, 20, 1446-1454.	2.4	67
38	FMR1 Epigenetic Silencing Commonly Occurs in Undifferentiated Fragile X-Affected Embryonic Stem Cells. <i>Stem Cell Reports</i> , 2014, 3, 699-706.	4.8	66
39	Gilbert's syndrome and hyperbilirubinaemia in ABO-incompatible neonates. <i>Lancet, The</i> , 2000, 356, 652-653.	13.7	65
40	Cost-effectiveness of population based BRCA testing with varying Ashkenazi Jewish ancestry. <i>American Journal of Obstetrics and Gynecology</i> , 2017, 217, 578.e1-578.e12.	1.3	63
41	A role for <i>TENM1</i> mutations in congenital general anosmia. <i>Clinical Genetics</i> , 2016, 90, 211-219.	2.0	59
42	Precision Medicine Meets Public Health: Population Screening for BRCA1 and BRCA2. <i>Journal of the National Cancer Institute</i> , 2014, 107, dju420-dju420.	6.3	57
43	Amyloid (A $\beta$ ) deposition in chromosome 1-linked Alzheimer's disease: The volga german families. <i>Annals of Neurology</i> , 1997, 41, 52-57.	5.3	54
44	Fanconi anemia and breast cancer susceptibility meet again. <i>Nature Genetics</i> , 2010, 42, 368-369.	21.4	54
45	Hemolysis and bilirubin conjugation in association with UDP-glucuronosyltransferase 1A1 promoter polymorphism. <i>Hepatology</i> , 2002, 35, 905-911.	7.3	50
46	Double Heterozygotes for the Ashkenazi Founder Mutations in BRCA1 and BRCA2 Genes. <i>American Journal of Human Genetics</i> , 1998, 63, 1224-1227.	6.2	47
47	Molecular study of WISP3 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. <i>American Journal of Medical Genetics, Part A</i> , 2005, 138A, 118-126.	1.2	47
48	Functional variant of KLOTHO: a breast cancer risk modifier among BRCA1 mutation carriers of Ashkenazi origin. <i>Oncogene</i> , 2010, 29, 26-33.	5.9	46
49	Egr-1 upregulates the Alzheimer's disease presenilin-2 gene in neuronal cells. <i>Gene</i> , 2003, 318, 113-124.	2.2	44
50	Neonatal outcome after preimplantation genetic diagnosis. <i>Fertility and Sterility</i> , 2014, 102, 1016-1021.	1.0	44
51	Population screening for BRCA1/BRCA2 founder mutations in Ashkenazi Jews: proactive recruitment compared with self-referral. <i>Genetics in Medicine</i> , 2017, 19, 754-762.	2.4	44
52	Clinical testing of BRCA1 and BRCA2: a worldwide snapshot of technological practices. <i>Npj Genomic Medicine</i> , 2018, 3, 7.	3.8	44
53	BRCA1 germline mutations in women with uterine serous papillary carcinoma. <i>Obstetrics and Gynecology</i> , 2000, 96, 28-32.	2.4	43
54	Recent Advances in the Genetics of Alzheimer's Disease. <i>Journal of Geriatric Psychiatry and Neurology</i> , 1998, 11, 42-54.	2.3	41

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55	Uncovering the Role of Hypermethylation by CTG Expansion in Myotonic Dystrophy Type 1 Using Mutant Human Embryonic Stem Cells. <i>Stem Cell Reports</i> , 2015, 5, 221-231.	4.8	40
56	Genomic analysis of inherited breast cancer among Palestinian women: Genetic heterogeneity and a founder mutation in TP53. <i>International Journal of Cancer</i> , 2017, 141, 750-756.	5.1	40
57	Low-dose high-frequency enzyme replacement therapy for very young children with severe Gaucher disease. <i>British Journal of Haematology</i> , 1993, 85, 783-786.	2.5	37
58	Substrate profiling of human vaccinia-related kinases identifies coilin, a Cajal body nuclear protein, as a phosphorylation target with neurological implications. <i>Journal of Proteomics</i> , 2011, 75, 548-560.	2.4	37
59	The Spinal Muscular Atrophy with Pontocerebellar Hypoplasia Gene <i>VRK1</i> Regulates Neuronal Migration through an Amyloid- $\beta$ Precursor Protein-Dependent Mechanism. <i>Journal of Neuroscience</i> , 2015, 35, 936-942.	3.6	36
60	The landscape of autosomal-recessive pathogenic variants in European populations reveals phenotype-specific effects. <i>American Journal of Human Genetics</i> , 2021, 108, 608-619.	6.2	36
61	Testicular differentiation factor SF-1 is required for human spleen development. <i>Journal of Clinical Investigation</i> , 2014, 124, 2071-2075.	8.2	36
62	(TA) <sub>n</sub> UGT 1A1 Promoter Polymorphism: A Crucial Factor in the Pathophysiology of Jaundice in G-6-PD Deficient Neonates. <i>Pediatric Research</i> , 2007, 61, 727-731.	2.3	35
63	Pulmonary hypoplasia, diaphragmatic hernia, anophthalmia, cardiac defect (PDAC) syndrome due to <i>STRA6</i> mutations: What are the minimal criteria?. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2457-2463.	1.2	35
64	TODRA, a lncRNA at the RAD51 Locus, Is Oppositely Regulated to RAD51, and Enhances RAD51-Dependent DSB (Double Strand Break) Repair. <i>PLoS ONE</i> , 2015, 10, e0134120.	2.5	35
65	Haploseek: a 24-hour all-in-one method for preimplantation genetic diagnosis (PGD) of monogenic disease and aneuploidy. <i>Genetics in Medicine</i> , 2019, 21, 1390-1399.	2.4	35
66	Features of Patients With Hereditary Mixed Polyposis Syndrome Caused by Duplication of GREM1 and Implications for Screening and Surveillance. <i>Gastroenterology</i> , 2017, 152, 1876-1880.e1.	1.3	34
67	VRK1 regulates Cajal body dynamics and protects coilin from proteasomal degradation in cell cycle. <i>Scientific Reports</i> , 2015, 5, 10543.	3.3	33
68	Differing Pathogenesis of Perinatal Bilirubinemia in Glucose-6-Phosphate Dehydrogenase-Deficient Versus-Normal Neonates. <i>Pediatric Research</i> , 2001, 50, 532-537.	2.3	32
69	Loss of function of PCDH12 underlies recessive microcephaly mimicking intrauterine infection. <i>Neurology</i> , 2016, 86, 2016-2024.	1.1	32
70	Parkin differently regulates presenilin-1 and presenilin-2 functions by direct control of their promoter transcription. <i>Journal of Molecular Cell Biology</i> , 2013, 5, 132-142.	3.3	31
71	Population Screening for Inherited Predisposition to Breast and Ovarian Cancer. <i>Annual Review of Genomics and Human Genetics</i> , 2020, 21, 373-412.	6.2	31
72	PGD on a recombinant allele: crossover between the TSC2 gene and "linked" markers impairs accurate diagnosis. <i>Prenatal Diagnosis</i> , 2008, 28, 929-933.	2.3	27

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73	Female Sex Bias in Human Embryonic Stem Cell Lines. <i>Stem Cells and Development</i> , 2012, 21, 363-372.	2.1	27
74	(TA)n UDP-Glucuronosyltransferase 1A1 Promoter Polymorphism in Nigerian Neonates. <i>Pediatric Research</i> , 2008, 63, 109-111.	2.3	26
75	Collaborative genomics for human health and cooperation in the Mediterranean region. <i>Nature Genetics</i> , 2010, 42, 641-645.	21.4	26
76	PGD for fragile X syndrome: ovarian function is the main determinant of success. <i>Human Reproduction</i> , 2010, 25, 2629-2636.	0.9	26
77	Combined mineralocorticoid and glucocorticoid deficiency is caused by a novel founder nicotinamide nucleotide transhydrogenase mutation that alters mitochondrial morphology and increases oxidative stress. <i>Journal of Medical Genetics</i> , 2015, 52, 636-641.	3.2	26
78	Mitochondrial <i>PITRM1</i> peptidase loss-of-function in childhood cerebellar atrophy. <i>Journal of Medical Genetics</i> , 2018, 55, 599-606.	3.2	26
79	Single-sperm analysis for haplotype construction of de-novo paternal mutations: application to PGD for neurofibromatosis type 1. <i>Human Reproduction</i> , 2006, 21, 2047-2051.	0.9	25
80	Simultaneous preimplantation genetic diagnosis for Tay-Sachs and Gaucher disease. <i>Reproductive BioMedicine Online</i> , 2007, 15, 83-88.	2.4	25
81	Vesicular acetylcholine transporter defect underlies devastating congenital myasthenia syndrome. <i>Neurology</i> , 2017, 88, 1021-1028.	1.1	25
82	Population screening for BRCA1/BRCA2 mutations: lessons from qualitative analysis of the screening experience. <i>Genetics in Medicine</i> , 2017, 19, 628-634.	2.4	25
83	Proof-of-principle rapid noninvasive prenatal diagnosis of autosomal recessive founder mutations. <i>Journal of Clinical Investigation</i> , 2015, 125, 3757-3765.	8.2	25
84	A single Mediterranean, possibly Jewish, origin for the Val59Gly CDKN2A mutation in four melanoma-prone families. <i>European Journal of Human Genetics</i> , 2003, 11, 288-296.	2.8	24
85	Preimplantation genetic diagnosis (PGD) for nonsyndromic deafness by polar body and blastomere biopsy. <i>Journal of Assisted Reproduction and Genetics</i> , 2009, 26, 391-397.	2.5	24
86	CANCER: Enhanced: A Risky Business—Assessing Breast Cancer Risk. <i>Science</i> , 2003, 302, 574-575.	12.6	22
87	Successful polar body-based preimplantation genetic diagnosis for achondroplasia. <i>Reproductive BioMedicine Online</i> , 2008, 16, 276-282.	2.4	22
88	p53-dependent control of transactivation of the Pen2 promoter by presenilins. <i>Journal of Cell Science</i> , 2009, 122, 4003-4008.	2.0	21
89	Human Embryo Editing: Opportunities and Importance of Transnational Cooperation. <i>Cell Stem Cell</i> , 2017, 21, 423-426.	11.1	21
90	High Frequency of Factor V Leiden in a Population of Israeli Arabs. <i>Thrombosis and Haemostasis</i> , 1999, 82, 1768-1768.	3.4	21

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91	Advantages of multiple markers and polar body analysis in preimplantation genetic diagnosis for Alagille disease. <i>Prenatal Diagnosis</i> , 2007, 27, 317-321.	2.3	20
92	Real-time reverse linkage using polar body analysis for preimplantation genetic diagnosis in female carriers of de novo mutations. <i>Human Reproduction</i> , 2009, 24, 3225-3229.	0.9	20
93	A novel severe N-terminal splice site KISS1R gene mutation causes hypogonadotropic hypogonadism but enables a normal development of neonatal external genitalia. <i>European Journal of Endocrinology</i> , 2012, 167, 209-216.	3.7	20
94	Expanding the phenotype of <i>CRB2</i> mutations—A new ciliopathy syndrome?. <i>Clinical Genetics</i> , 2016, 90, 540-544.	2.0	18
95	Preconception carrier screening yield: effect of variants of unknown significance in partners of carriers with clinically significant variants. <i>Genetics in Medicine</i> , 2020, 22, 646-653.	2.4	18
96	Presymptomatic Awareness of Germline Pathogenic <i>BRCA</i> Variants and Associated Outcomes in Women With Breast Cancer. <i>JAMA Oncology</i> , 2020, 6, 1460.	7.1	18
97	Monogenic determinants of familial Alzheimer's disease: presenilin-2 mutations. <i>Cellular and Molecular Life Sciences</i> , 1998, 54, 910-919.	5.4	17
98	Familial haplotyping and embryo analysis for Preimplantation Genetic Diagnosis (PGD) using DNA microarrays: a proof of principle study. <i>Journal of Assisted Reproduction and Genetics</i> , 2013, 30, 1595-1603.	2.5	17
99	Ethnic ancestry and increased paternal age are risk factors for breast cancer before the age of 40 years. <i>European Journal of Cancer Prevention</i> , 2007, 16, 549-554.	1.3	16
100	BRCA genetic testing of individuals from families with low prevalence of cancer: experiences of carriers and implications for population screening. <i>Genetics in Medicine</i> , 2012, 14, 688-694.	2.4	16
101	Hiding in Plain Sight—Somatic Mutation in Human Disease. <i>New England Journal of Medicine</i> , 2020, 383, 2680-2682.	27.0	16
102	Breast cancer risk and hormone replacement therapy among BRCA carriers after risk-reducing salpingo-oophorectomy. <i>European Journal of Cancer</i> , 2021, 148, 95-102.	2.8	16
103	Preimplantation genetic diagnosis (PGD)—prevention of the birth of children affected with endocrine diseases. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2011, 24, 543-8.	0.9	15
104	Brain calcifications and <i>PCDH12</i> variants. <i>Neurology: Genetics</i> , 2017, 3, e166.	1.9	15
105	Preventing mucopolysaccharidosis type II (Hunter syndrome): PGD and establishing a Hunter (46, XX) stem cell line. <i>Prenatal Diagnosis</i> , 2011, 31, 853-860.	2.3	14
106	A deleterious founder mutation in the <i>BMPER</i> gene causes diaphanospondylodysostosis (DSD). <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2801-2806.	1.2	14
107	Vrk1 partial Knockdown in Mice Results in Reduced Brain Weight and Mild Motor Dysfunction, and Indicates Neuronal VRK1 Target Pathways. <i>Scientific Reports</i> , 2018, 8, 11265.	3.3	14
108	Snord 3A: A Molecular Marker and Modulator of Prion Disease Progression. <i>PLoS ONE</i> , 2013, 8, e54433.	2.5	14

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109	Preimplantation genetic diagnosis (PGD) for a treatable disorder: Gaucher disease type 1 as a model. <i>Blood Cells, Molecules, and Diseases</i> , 2011, 46, 15-18.	1.4	13
110	Prevention of Lysosomal Storage Diseases and Derivation of Mutant Stem Cell Lines by Preimplantation Genetic Diagnosis. <i>Molecular Biology International</i> , 2012, 2012, 1-9.	1.7	13
111	10 Gaucher's disease: genetic counselling and population screening. <i>Best Practice and Research: Clinical Haematology</i> , 1997, 10, 779-792.	1.1	12
112	Incidence of thrombophilia in patients with Gaucher disease. <i>American Journal of Medical Genetics Part A</i> , 2000, 95, 429-431.	2.4	12
113	PGD for germline mosaicism. <i>Reproductive BioMedicine Online</i> , 2012, 25, 390-395.	2.4	12
114	Alpha-thalassemia intellectual disability: variable phenotypic expression among males with a recurrent nonsense mutation (p.R37X). <i>Clinical Genetics</i> , 2015, 87, 461-466.	2.0	12
115	Developmental neuropsychological assessment of 4- to 5-year-old children born following Preimplantation Genetic Diagnosis (PGD): A pilot study. <i>Child Neuropsychology</i> , 2016, 22, 458-471.	1.3	12
116	Garrod's fourth inborn error of metabolism solved by the identification of mutations causing pentosuria. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, 18313-18317.	7.1	11
117	High school Tay Sachs disease carrier screening: 5 to 11-year follow-up. <i>Journal of Community Genetics</i> , 2014, 5, 139-146.	1.2	11
118	<i>NKX2-2</i> Mutation Causes Congenital Diabetes and Infantile Obesity With Paradoxical Glucose-Induced Ghrelin Secretion. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 3486-3495.	3.6	11
119	Attitudes of couples identified through screening as carriers of Gaucher disease type 1. <i>Clinical Genetics</i> , 2008, 74, 566-570.	2.0	9
120	Transcriptional regulation of the murine Presenilin-2 gene reveals similarities and differences to its human orthologue. <i>Gene</i> , 2009, 446, 81-89.	2.2	9
121	Medical genetics in Israel's diverse population. <i>Lancet, The</i> , 2017, 389, 2453-2455.	13.7	9
122	Variable Features of Juvenile Polyposis Syndrome With Gastric Involvement Among Patients With a Large Genomic Deletion of BMPR1A. <i>Clinical and Translational Gastroenterology</i> , 2019, 10, e00054.	2.5	9
123	A defect in GPI synthesis as a suggested mechanism for the role of ARV1 in intellectual disability and seizures. <i>Neurogenetics</i> , 2020, 21, 259-267.	1.4	9
124	Preimplantation Genetic Diagnosis for Fetal Neonatal Alloimmune Thrombocytopenia Due to Antihuman Platelet Antigen Maternal Antibodies. <i>Obstetrics and Gynecology</i> , 2012, 119, 338-343.	2.4	8
125	Prenatal Isolated Ventricular Septal Defect May Not Be Associated with Trisomy 21. <i>Journal of Clinical Medicine</i> , 2014, 3, 432-439.	2.4	8
126	The Founder Mutations in the BRCA1, BRCA2, and ATM Genes in Moroccan Jewish Women with Breast Cancer. <i>Genetic Testing and Molecular Biomarkers</i> , 2000, 4, 403-407.	1.7	7



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127	Polar Body-Based Preimplantation Genetic Diagnosis for <i>N</i> -Acetylglutamate Synthase Deficiency. Fetal Diagnosis and Therapy, 2008, 24, 170-176.	1.4	7
128	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
129	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
130	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
131	Homozygous stop-gain variant in LRRC32, encoding a TGF $\beta$ 2 receptor, associated with cleft palate, proliferative retinopathy, and developmental delay. European Journal of Human Genetics, 2019, 27, 1315-1319.	2.8	7
132	Performance comparison: exome sequencing as a single test replacing Sanger sequencing. Molecular Genetics and Genomics, 2021, 296, 653-663.	2.1	7
133	Why should primary care physicians know about breast cancer genetics?. Western Journal of Medicine, 2001, 175, 168-173.	0.3	7
134	Population-based BRCA1/BRCA2 screening in Ashkenazi Jews: A call for evidence. Genetics in Medicine, 2009, 11, 620-621.	2.4	6
135	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
136	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
137	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
138	Establishment of Homozygote Mutant Human Embryonic Stem Cells by Parthenogenesis. PLoS ONE, 2015, 10, e0138893.	2.5	5
139	Israeli Position Paper. Chest, 2020, 158, 2278-2281.	0.8	5
140	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
141	Response to ACMG guideline: Carrier screening in individuals of Ashkenazi Jewish decent. Genetics in Medicine, 2008, 10, 462-462.	2.4	3
142	Mosaic compound heterozygosity of SHOX resulting in Leri's 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
143	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
144	Proposed Shift in Screening for Breast Cancer—Reply. JAMA - Journal of the American Medical Association, 2015, 313, 525.	7.4	3

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145	Salt-Losing 21-Hydroxylase Deficiency Caused by Double Homozygosity for Two "Mild" Mutations. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e680-e686.	3.6	3
146	BRCA1 Germline Mutations in Women With Uterine Serous Papillary Carcinoma. Obstetrics and Gynecology, 2000, 96, 28-32.	2.4	2
147	BRCA1 and BRCA2 mutation carriers as potential candidates for chemoprevention trials. Journal of Cellular Biochemistry, 2000, 77, 13-18.	2.6	2
148	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
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