## Patrick Concannon

List of Publications by Year in descending order

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200 papers

23,579 citations

68 h-index 7950 149 g-index

206 all docs

206 docs citations

206 times ranked 26435 citing authors

| #  | Article  | lF   | Citations |
|----|--|------|-----------|
| 1  | Smoking, Radiation Therapy, and Contralateral Breast Cancer Risk in Young Women. Journal of the National Cancer Institute, 2022, 114, 631-634.   | 6.3  | 6         |
| 2  | Genetic Control of Splicing at <i>SIRPG</i> Modulates Risk of Type 1 Diabetes. Diabetes, 2022, 71, 350-358.  | 0.6  | 2         |
| 3  | Integrative analyses of TEDDY Omics data reveal lipid metabolism abnormalities, increased intracellular ROS and heightened inflammation prior to autoimmunity for type $1$ diabetes. Genome Biology, 2021, 22, 39.     | 8.8  | 22        |
| 4  | Fine-mapping, trans-ancestral and genomic analyses identify causal variants, cells, genes and drug targets for type $1$ diabetes. Nature Genetics, $2021$ , $53$ , $962$ - $971$ .                                     | 21.4 | 133       |
| 5  | Genes affecting ionizing radiation survival identified through combined exome sequencing and functional screening. Human Mutation, 2021, 42, 1124-1138.  | 2.5  | 0         |
| 6  | Cancer Risks Associated With Germline <i>PALB2</i> Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.   | 1.6  | 270       |
| 7  | A case-control study of the joint effect of reproductive factors and radiation treatment for first breast cancer and risk of contralateral breast cancer in the WECARE study. Breast, 2020, 54, 62-69.                 | 2.2  | 3         |
| 8  | Genome-Wide Association Study of Cryptosporidiosis in Infants Implicates <i>PRKCA</i> . MBio, 2020, 11,  | 4.1  | 20        |
| 9  | Radiation Treatment, <i>ATM</i> , <i>BRCA1/2</i> , and <i>CHEK2</i> *1100delC Pathogenic Variants and Risk of Contralateral Breast Cancer. Journal of the National Cancer Institute, 2020, 112, 1275-1279.             | 6.3  | 21        |
| 10 | Machine learning on genome-wide association studies to predict the risk of radiation-associated contralateral breast cancer in the WECARE Study. PLoS ONE, 2020, 15, e0226157.   | 2.5  | 22        |
| 11 | Genetics of Type 1 Diabetes Comes of Age. Diabetes Care, 2020, 43, 16-18.  | 8.6  | 11        |
| 12 | UBASH3A Regulates the Synthesis and Dynamics of TCR–CD3 Complexes. Journal of Immunology, 2019, 203, 2827-2836.  | 0.8  | 32        |
| 13 | Association of a Pathway-Specific Genetic Risk Score With Risk of Radiation-Associated Contralateral Breast Cancer. JAMA Network Open, 2019, 2, e1912259.  | 5.9  | 5         |
| 14 | Type 1 Diabetes Risk in African-Ancestry Participants and Utility of an Ancestry-Specific Genetic Risk Score. Diabetes Care, 2019, 42, 406-415.  | 8.6  | 62        |
| 15 | Molecular-genetic characterization of common, noncoding UBASH3A variants associated with type 1 diabetes. European Journal of Human Genetics, 2018, 26, 1060-1064.   | 2.8  | 23        |
| 16 | Agreement between self-reported and register-based cardiovascular events among Danish breast cancer survivors. Journal of Cancer Survivorship, 2018, 12, 95-100.   | 2.9  | 7         |
| 17 | Identification of ATIC as a Novel Target for Chemoradiosensitization. International Journal of Radiation Oncology Biology Physics, 2018, 100, 162-173.   | 0.8  | 22        |
| 18 | Breast Cancer Family History and Contralateral Breast Cancer Risk in Young Women: An Update From the Women's Environmental Cancer and Radiation Epidemiology Study. Journal of Clinical Oncology, 2018, 36, 1513-1520. | 1.6  | 44        |

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|----|---|------|-----------|
| 19 | Genome-Wide Association Study Reveals Genetic Link between Diarrhea-Associated Entamoeba histolytica Infection and Inflammatory Bowel Disease. MBio, 2018, 9, .   | 4.1  | 23        |
| 20 | Event Analysis: Using Transcript Events To Improve Estimates of Abundance in RNA-seq Data. G3: Genes, Genomes, Genetics, 2018, 8, 2923-2940.  | 1.8  | 11        |
| 21 | Genome-wide Analysis in Brazilians Reveals Highly Differentiated Native American Genome Regions.<br>Molecular Biology and Evolution, 2017, 34, msw249.  | 8.9  | 21        |
| 22 | ChIP Technique to Study Protein Dynamics at Defined DNA Double Strand Breaks. Methods in Molecular Biology, 2017, 1599, 245-262.  | 0.9  | 0         |
| 23 | The A946T variant of the RNA sensor IFIH1 mediates an interferon program that limits viral infection but increases the risk for autoimmunity. Nature Immunology, 2017, 18, 744-752.                             | 14.5 | 119       |
| 24 | UBASH3A Mediates Risk for Type 1 Diabetes Through Inhibition of T-Cell Receptor–Induced NF-κB Signaling. Diabetes, 2017, 66, 2033-2043.   | 0.6  | 54        |
| 25 | Disease-specific biases in alternative splicing and tissue-specific dysregulation revealed by multitissue profiling of lymphocyte gene expression in type 1 diabetes. Genome Research, 2017, 27, 1807-1815.     | 5.5  | 29        |
| 26 | <i>ATM</i> , radiation, and the risk of second primary breast cancer. International Journal of Radiation Biology, 2017, 93, 1121-1127.  | 1.8  | 34        |
| 27 | Hormone receptor status of a first primary breast cancer predicts contralateral breast cancer risk in the WECARE study population. Breast Cancer Research, 2017, 19, 83.  | 5.0  | 27        |
| 28 | Body mass index, weight change, and risk of second primary breast cancer in the <scp>WECARE</scp> study: influence of estrogen receptor status of the first breast cancer. Cancer Medicine, 2016, 5, 3282-3291. | 2.8  | 22        |
| 29 | Targeted Deep Sequencing in Multiple-Affected Sibships of European Ancestry Identifies Rare Deleterious Variants in <i>PTPN22</i> That Confer Risk for Type 1 Diabetes. Diabetes, 2016, 65, 794-802.            | 0.6  | 24        |
| 30 | Systematic Evaluation of Genes and Genetic Variants Associated with Type 1 Diabetes Susceptibility. Journal of Immunology, 2016, 196, 3043-3053.  | 0.8  | 47        |
| 31 | Fine mapping of type 1 diabetes susceptibility loci and evidence for colocalization of causal variants with lymphoid gene enhancers. Nature Genetics, 2015, 47, 381-386.  | 21.4 | 589       |
| 32 | Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.   | 21,4 | 221       |
| 33 | Novel Association Between Immune-Mediated Susceptibility Loci and Persistent Autoantibody Positivity in Type 1 Diabetes. Diabetes, 2015, 64, 3017-3027.   | 0.6  | 20        |
| 34 | Cytomegalovirus infection enhances the immune response to influenza. Science Translational Medicine, 2015, 7, 281ra43.  | 12.4 | 277       |
| 35 | Summary of the Type 1 Diabetes Genetics Consortium Autoantibody Workshop. Diabetes Care, 2015, 38, S45-S48.   | 8.6  | 2         |
| 36 | ATM-dependent phosphorylation of MRE11 controls extent of resection during homology directed repair by signalling through Exonuclease 1. Nucleic Acids Research, 2015, 43, 8352-8367.                           | 14.5 | 54        |

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|----|---|------|-----------|
| 37 | Effects of Type 1 Diabetes-Associated IFIH1 Polymorphisms on MDA5 Function and Expression. Current Diabetes Reports, 2015, 15, 96.  | 4.2  | 47        |
| 38 | Common variants at the <i>CHEK2 </i> gene locus and risk of epithelial ovarian cancer. Carcinogenesis, 2015, 36, 1341-1353.   | 2.8  | 24        |
| 39 | Role of Type 1 Diabetes–Associated SNPs on Autoantibody Positivity in the Type 1 Diabetes Genetics<br>Consortium: Overview. Diabetes Care, 2015, 38, S1-S3.   | 8.6  | 488       |
| 40 | Genetic and epigenetic variation in the lineage specification of regulatory T cells. ELife, 2015, 4, e07571.  | 6.0  | 49        |
| 41 | <i>CTSH</i> regulates $\hat{I}^2$ -cell function and disease progression in newly diagnosed type 1 diabetes patients. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 10305-10310.              | 7.1  | 81        |
| 42 | Breast-Cancer Risk in Families With Mutations in PALB2. Obstetrical and Gynecological Survey, 2014, 69, 659-660.  | 0.4  | 1         |
| 43 | Role of Leptin-Mediated Colonic Inflammation in Defense against Clostridium difficile Colitis.<br>Infection and Immunity, 2014, 82, 341-349.  | 2.2  | 46        |
| 44 | A Method for Geneâ€Based Pathway Analysis Using Genomewide Association Study Summary Statistics Reveals Nine New Type 1 Diabetes Associations. Genetic Epidemiology, 2014, 38, 661-670.   | 1.3  | 54        |
| 45 | Fine Mapping and Functional Studies of Risk Variants for Type 1 Diabetes at Chromosome 16p13.13. Diabetes, 2014, 63, 4360-4368.   | 0.6  | 17        |
| 46 | Breast-Cancer Risk in Families with Mutations in <i>PALB2</i> li>. New England Journal of Medicine, 2014, 371, 497-506.   | 27.0 | 745       |
| 47 | A hybrid qPCR/SNP array approach allows cost efficient assessment of KIR gene copy numbers in large samples. BMC Genomics, 2014, 15, 274.   | 2.8  | 12        |
| 48 | HLA-DRB1*07:01 is associated with a higher risk of asparaginase allergies. Blood, 2014, 124, 1266-1276.   | 1.4  | 84        |
| 49 | Intensity modulated radiotherapy for sinonasal malignancies with a focus on optic pathway preservation. Journal of Hematology and Oncology, 2013, 6, 4.   | 17.0 | 17        |
| 50 | Common variants in genes coding for chemotherapy metabolizing enzymes, transporters, and targets: a case $\hat{a}$ e"control study of contralateral breast cancer risk in the WECARE Study. Cancer Causes and Control, 2013, 24, 1605-1614. | 1.8  | 6         |
| 51 | Contralateral breast cancer after radiotherapy among BRCA1 and BRCA2 mutation carriers: A WECARE Study Report. European Journal of Cancer, 2013, 49, 2979-2985.   | 2.8  | 72        |
| 52 | Dense genotyping of immune-related disease regions identifies 14 new susceptibility loci for juvenile idiopathic arthritis. Nature Genetics, 2013, 45, 664-669.   | 21.4 | 337       |
| 53 | NBN Phosphorylation regulates the accumulation of MRN and ATM at sites of DNA double-strand breaks. Oncogene, 2013, 32, 4448-4456.  | 5.9  | 18        |
| 54 | Gut Microbiomes of Malawian Twin Pairs Discordant for Kwashiorkor. Science, 2013, 339, 548-554.   | 12.6 | 1,012     |

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|----|--|------|-----------|
| 55 | ImmunoChip Study Implicates Antigen Presentation to T Cells in Narcolepsy. PLoS Genetics, 2013, 9, e1003270.   | 3.5  | 206       |
| 56 | Risk of Asynchronous Contralateral Breast Cancer in Noncarriers of <i>BRCA1</i> and <i>BRCA2</i> Mutations With a Family History of Breast Cancer: A Report From the Women's Environmental Cancer and Radiation Epidemiology Study. Journal of Clinical Oncology, 2013, 31, 433-439. | 1.6  | 101       |
| 57 | Imputing Amino Acid Polymorphisms in Human Leukocyte Antigens. PLoS ONE, 2013, 8, e64683.  | 2.5  | 538       |
| 58 | HLA-DRB1*07:01 Is Associated With Asparaginase Allergies In Children With Acute Lymphoblastic Leukemia. Blood, 2013, 122, 60-60.   | 1.4  | 1         |
| 59 | Variants in tamoxifen metabolizing genes: a case-control study of contralateral breast cancer risk in the WECARE study. International Journal of Molecular Epidemiology and Genetics, 2013, 4, 35-48.  | 0.4  | 6         |
| 60 | Rare and functional SIAE variants are not associated with autoimmune disease risk in up to 66,924 individuals of European ancestry. Nature Genetics, 2012, 44, 3-5.  | 21.4 | 44        |
| 61 | Evidence for two independent associations with type 1 diabetes at the 12q13 locus. Genes and Immunity, 2012, 13, 66-70.  | 4.1  | 22        |
| 62 | Variation in Genes Related to Obesity, Weight, and Weight Change and Risk of Contralateral Breast Cancer in the WECARE Study Population. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 2261-2267.   | 2.5  | 11        |
| 63 | Evidence of Gene-Gene Interaction and Age-at-Diagnosis Effects in Type 1 Diabetes. Diabetes, 2012, 61, 3012-3017.  | 0.6  | 60        |
| 64 | Variants in activators and downstream targets of ATM, radiation exposure, and contralateral breast cancer risk in the WECARE study. Human Mutation, 2012, 33, 158-164.   | 2.5  | 23        |
| 65 | Rare germline mutations inPALB2and breast cancer risk: A population-based study. Human Mutation, 2012, 33, 674-680.  | 2.5  | 74        |
| 66 | Confirmation of novel type 1 diabetes risk loci in families. Diabetologia, 2012, 55, 996-1000.   | 6.3  | 50        |
| 67 | Single nucleotide polymorphisms associated with risk for contralateral breast cancer in the Women's Environment, Cancer, and Radiation Epidemiology (WECARE) Study. Breast Cancer Research, 2011, 13, R114.  | 5.0  | 33        |
| 68 | Risk of contralateral breast cancer associated with common variants in BRCA1 and BRCA2: potential modifying effect of BRCA1/BRCA2 mutation carrier status. Breast Cancer Research and Treatment, 2011, 127, 819-829.   | 2.5  | 11        |
| 69 | Assessment of rare BRCA1 and BRCA2 variants of unknown significance using hierarchical modeling. Genetic Epidemiology, 2011, 35, 389-397.  | 1.3  | 15        |
| 70 | Comprehensive Profiling of Radiosensitive Human Cell Lines with DNA Damage Response Assays Identifies the Neutral Comet Assay as a Potential Surrogate for Clonogenic Survival. Radiation Research, 2011, 177, 176.  | 1.5  | 12        |
| 71 | Oral contraceptives and postmenopausal hormones and risk of contralateral breast cancer among BRCA1 and BRCA2 mutation carriers and noncarriers: the WECARE Study. Breast Cancer Research and Treatment, 2010, 120, 175-183.   | 2.5  | 22        |
| 72 | Adjuvant systemic therapy for breast cancer in BRCA1/BRCA2 mutation carriers in a population-based study of risk of contralateral breast cancer. Breast Cancer Research and Treatment, 2010, 123, 491-498.   | 2.5  | 57        |

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|----|---|------|-----------|
| 73 | Reproductive factors and risk of contralateral breast cancer by BRCA1 and BRCA2 mutation status: results from the WECARE study. Cancer Causes and Control, 2010, 21, 839-846.   | 1.8  | 12        |
| 74 | Characterization of <i>BRCA1 </i> and <i>BRCA2 </i> deleterious mutations and variants of unknown clinical significance in unilateral and bilateral breast cancer: the WECARE study. Human Mutation, 2010, 31, E1200-E1240.   | 2.5  | 103       |
| 75 | HLA Class I and Genetic Susceptibility to Type 1 Diabetes. Diabetes, 2010, 59, 2972-2979.   | 0.6  | 202       |
| 76 | Dual Functions of Nbs1 in the Repair of DNA Breaks and Proliferation Ensure Proper V(D)J Recombination and T-Cell Development. Molecular and Cellular Biology, 2010, 30, 5572-5581.   | 2.3  | 23        |
| 77 | Population-Based Study of the Risk of Second Primary Contralateral Breast Cancer Associated With Carrying a Mutation in <i>BRCA1</i> Second Primary Contralateral Breast Cancer Associated With Carrying a Mutation in <i brca1<="" i="">Second Primary Contralateral Breast Cancer Associated With Carrying a Mutation in<i brca1<="" i="">Second Primary Contralateral Breast Cancer Associated With Carrying a Mutation in<i brca1<="" i="">Second Primary Contralateral Breast Cancer Associated With Carrying a Mutation in<!--</td--><td>1.6</td><td>166</td></i></i></i> | 1.6  | 166       |
| 78 | Genetics of Type 1 Diabetes: What's Next?. Diabetes, 2010, 59, 1561-1571.   | 0.6  | 256       |
| 79 | Radiation Exposure, the ATM Gene, and Contralateral Breast Cancer in the Women's Environmental Cancer and Radiation Epidemiology Study. Journal of the National Cancer Institute, 2010, 102, 475-483.   | 6.3  | 121       |
| 80 | Genetics of Type 1A Diabetes. New England Journal of Medicine, 2009, 360, 1646-1654.  | 27.0 | 437       |
| 81 | Nuclear Export of NBN Is Required for Normal Cellular Responses to Radiation. Molecular and Cellular Biology, 2009, 29, 1000-1006.  | 2.3  | 18        |
| 82 | Recent Progress in the Genetics of Diabetes. Hormone Research in Paediatrics, 2009, 71, 17-23.  | 1.8  | 7         |
| 83 | Genome-Wide Scan for Linkage to Type 1 Diabetes in 2,496 Multiplex Families From the Type 1 Diabetes Genetics Consortium. Diabetes, 2009, 58, 1018-1022.  | 0.6  | 87        |
| 84 | Functional and computational assessment of missense variants in the ataxia-telangiectasia mutated (ATM) gene: mutations with increased cancer risk. Human Mutation, 2009, 30, 12-21.  | 2.5  | 72        |
| 85 | Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. Nature Genetics, 2009, 41, 703-707.  | 21.4 | 1,513     |
| 86 | Endogenous hSNM1B/Apollo interacts with TRF2 and stimulates ATM in response to ionizing radiation. DNA Repair, 2008, 7, 1192-1201.  | 2.8  | 37        |
| 87 | Rapid screen for truncating ATM mutations by PTT-ELISA. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2008, 640, 139-144.  | 1.0  | 15        |
| 88 | Variants in the ATM Gene Associated with a Reduced Risk of Contralateral Breast Cancer. Cancer Research, 2008, 68, 6486-6491.   | 0.9  | 43        |
| 89 | A Human Type 1 Diabetes Susceptibility Locus Maps to Chromosome 21q22.3. Diabetes, 2008, 57, 2858-2861.   | 0.6  | 103       |
| 90 | Risk for contralateral breast cancer among carriers of the CHEK2*1100delC mutation in the WECARE Study. British Journal of Cancer, 2008, 98, 728-733.   | 6.4  | 42        |

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|-----|---|-----|-----------|
| 91  | Variation of Breast Cancer Risk Among BRCA1/2 Carriers. JAMA - Journal of the American Medical Association, 2008, 299, 194-201.   | 7.4 | 244       |
| 92  | Replication independent ATR signalling leads to G2/M arrest requiring Nbs1, 53BP1 and MDC1. Human Molecular Genetics, 2008, 17, 3247-3253.  | 2.9 | 33        |
| 93  | HLA DR-DQ Haplotypes and Genotypes and Type 1 Diabetes Risk. Diabetes, 2008, 57, 1084-1092.   | 0.6 | 631       |
| 94  | Genetic Variation in PTPN22 Corresponds to Altered Function of T and B Lymphocytes. Journal of Immunology, 2007, 179, 4704-4710.  | 0.8 | 295       |
| 95  | ATR-dependent phosphorylation and activation of ATM in response to UV treatment or replication fork stalling. EMBO Journal, 2006, 25, 5775-5782.  | 7.8 | 319       |
| 96  | Recent advances in the immunogenetics of human type $1$ diabetes. Current Opinion in Immunology, 2006, $18,634-638$ .   | 5.5 | 21        |
| 97  | The Type 1 Diabetes Genetics Consortium. Annals of the New York Academy of Sciences, 2006, 1079, 1-8.   | 3.8 | 116       |
| 98  | Population-based estimates of breast cancer risks associated with ATM gene variants c.7271T>G and c.1066-6T>G (IVS10-6T>G) from the Breast Cancer Family Registry. Human Mutation, 2006, 27, 1122-1128. | 2.5 | 88        |
| 99  | On the proposed association of the ATM variants 5557G>A and IVS38-8T>C and bilateral breast cancer. International Journal of Cancer, 2006, 119, 724-725.  | 5.1 | 13        |
| 100 | The CHEK2*1100delC Allelic Variant and Risk of Breast Cancer: Screening Results from the Breast Cancer Family Registry. Cancer Epidemiology Biomarkers and Prevention, 2006, 15, 348-352.               | 2.5 | 46        |
| 101 | Active Role for Nibrin in the Kinetics of Atm Activation. Molecular and Cellular Biology, 2006, 26, 1691-1699.  | 2.3 | 77        |
| 102 | A Haplotype-Based Analysis of the <i>PTPN22</i> Locus in Type 1 Diabetes. Diabetes, 2006, 55, 2883-2889.  | 0.6 | 53        |
| 103 | ATM Gene Founder Haplotypes and Associated Mutations in Polish Families with Ataxia-Telangiectasia. Annals of Human Genetics, 2005, 69, 657-664.  | 0.8 | 37        |
| 104 | Extended DR3-D6S273-HLA-B haplotypes are associated with increased susceptibility to type 1 diabetes in US Caucasians. Tissue Antigens, 2005, 65, 115-119.  | 1.0 | 27        |
| 105 | Functional variants in SUMO4, TAB2, and NFÎ $^{\circ}$ B and the risk of type 1 diabetes. Genes and Immunity, 2005, 6, 231-235.   | 4.1 | 41        |
| 106 | A patient with mutations in DNA Ligase IV: Clinical features and overlap with Nijmegen breakage syndrome. American Journal of Medical Genetics, Part A, 2005, 137A, 283-287.                            | 1.2 | 96        |
| 107 | DNA-dependent Protein Kinase and XRCC4-DNA Ligase IV Mobilization in the Cell in Response to DNA Double Strand Breaks. Journal of Biological Chemistry, 2005, 280, 7060-7069.                           | 3.4 | 129       |
| 108 | Type 1 Diabetes. Diabetes, 2005, 54, 2995-3001.   | 0.6 | 221       |

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|-----|---|-----|-----------|
| 109 | The genetics of type 1 diabetes: Lessons learned and future challenges. Journal of Autoimmunity, 2005, 25, 34-39.   | 6.5 | 19        |
| 110 | Independent Roles for Nibrin and Mre11-Rad50 in the Activation and Function of Atm. Journal of Biological Chemistry, 2004, 279, 38813-38819.  | 3.4 | 69        |
| 111 | Polymorphic variation in the CBLB gene in human type 1 diabetes. Genes and Immunity, 2004, 5, 232-235.  | 4.1 | 18        |
| 112 | A functional polymorphism (1858C/T) in the PTPN22 gene is linked and associated with type I diabetes in multiplex families. Genes and Immunity, 2004, 5, 678-680.                                 | 4.1 | 120       |
| 113 | Human SNM1B is required for normal cellular response to both DNA interstrand crosslink-inducing agents and ionizing radiation. Oncogene, 2004, 23, 8611-8618.                                     | 5.9 | 84        |
| 114 | Remapping the Insulin Gene/IDDM2 Locus in Type 1 Diabetes. Diabetes, 2004, 53, 1884-1889.   | 0.6 | 198       |
| 115 | Study design: Evaluating gene–environment interactions in the etiology of breast cancer – the WECARE study. Breast Cancer Research, 2004, 6, R199-214.  | 5.0 | 106       |
| 116 | An overview of three new disorders associated with genetic instability: LIG4 syndrome, RS-SCID and ATR-Seckel syndrome. DNA Repair, 2004, 3, 1227-1235.   | 2.8 | 174       |
| 117 | A functional variant of IRS1 is associated with type 1 diabetes in families from the US and UK. Molecular Genetics and Metabolism, 2004, 81, 291-294.   | 1.1 | 11        |
| 118 | Designing and implementing quality control for multi-center screening of mutations in the ATM gene among women with breast cancer. Human Mutation, 2003, 21, 542-550.                             | 2.5 | 56        |
| 119 | Functional delivery of large genomic DNA to human cells with a peptide-lipid vector. Journal of Gene Medicine, 2003, 5, 883-892.  | 2.8 | 29        |
| 120 | Improved diagnostic testing for ataxia–telangiectasia by immunoblotting of nuclear lysates for ATM protein expression. Molecular Genetics and Metabolism, 2003, 80, 437-443.                      | 1.1 | 78        |
| 121 | ATM variants 7271T>G and IVS10-6T>G among women with unilateral and bilateral breast cancer. British Journal of Cancer, 2003, 89, 1513-1516.  | 6.4 | 45        |
| 122 | Nibrin Forkhead-associated Domain and Breast Cancer C-terminal Domain Are Both Required for Nuclear Focus Formation and Phosphorylation. Journal of Biological Chemistry, 2003, 278, 21944-21951. | 3.4 | 63        |
| 123 | Medulloblastoma With Adverse Reaction to Radiation Therapy in Nijmegen Breakage Syndrome. Journal of Pediatric Hematology/Oncology, 2003, 25, 248-251.  | 0.6 | 50        |
| 124 | Challenges and Strategies for Investigating the Genetic Complexity of Common Human Diseases. Diabetes, 2002, 51, S288-S294.   | 0.6 | 34        |
| 125 | Complementarity-Determining Region 1 Sequence Requirements Drive Limited Vα Usage in Response to Influenza Hemagglutinin 307–319 Peptide. Journal of Immunology, 2002, 168, 3894-3901.            | 0.8 | 5         |
| 126 | Nijmegen breakage syndrome: Clinical characteristics and mutation analysis in eight unrelated Russian families. Journal of Pediatrics, 2002, 140, 355-361.  | 1.8 | 77        |

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|-----|---|------|-----------|
| 127 | Mutations and molecular variants of the NBS1 gene in non-Hodgkin lymphoma. Genes Chromosomes and Cancer, 2002, 35, 282-286.   | 2.8  | 43        |
| 128 | Mapping genes for autoimmunity in humans: type 1 diabetes as a model. Immunological Reviews, 2002, 190, 182-194.  | 6.0  | 51        |
| 129 | ATM heterozygosity and cancer risk. Nature Genetics, 2002, 32, 89-90.   | 21.4 | 55        |
| 130 | Parameters for reliable results in genetic association studies in common disease. Nature Genetics, 2002, 30, 149-150.   | 21.4 | 224       |
| 131 | Linkage and Association With Type 1 Diabetes on Chromosome 1q42. Diabetes, 2002, 51, 3318-3325.   | 0.6  | 15        |
| 132 | Etoposide and Adriamycin but Not Genistein Can Activate the Checkpoint Kinase Chk2 Independently of ATM/ATR. Biochemical and Biophysical Research Communications, 2001, 289, 1199-1204. | 2.1  | 28        |
| 133 | Linkage Studies of SOX13, the ICA12 Autoantigen Gene, in Families with Type 1 Diabetes. Molecular Genetics and Metabolism, 2001, 72, 356-359.   | 1.1  | 1         |
| 134 | Seven Regions of the Genome Show Evidence of Linkage to Type 1 Diabetes in a Consensus Analysis of 767 Multiplex Families. American Journal of Human Genetics, 2001, 69, 820-830.       | 6.2  | 245       |
| 135 | DNA Ligase IV Mutations Identified in Patients Exhibiting Developmental Delay and Immunodeficiency. Molecular Cell, 2001, 8, 1175-1185.   | 9.7  | 497       |
| 136 | Immune diversity and genomic stability: opposite goals but similar paths. Journal of Photochemistry and Photobiology B: Biology, 2001, 65, 88-96.                                       | 3.8  | 14        |
| 137 | Increased frequency of ATM mutations in breast carcinoma patients with early onset disease and positive family history. Cancer, 2001, 92, 479-487.                                      | 4.1  | 105       |
| 138 | Chk2 Activation Dependence on Nbs1 after DNA Damage. Molecular and Cellular Biology, 2001, 21, 5214-5222.   | 2.3  | 198       |
| 139 | Distinct Functional Domains of Nibrin Mediate Mre11 Binding, Focus Formation, and Nuclear Localization. Molecular and Cellular Biology, 2001, 21, 2184-2191.                            | 2.3  | 161       |
| 140 | ATM-dependent phosphorylation of nibrin in response to radiation exposure. Nature Genetics, 2000, 25, 115-119.  | 21.4 | 446       |
| 141 | Genetic variation in the gene encoding calpain-10 is associated with type 2 diabetes mellitus. Nature Genetics, 2000, 26, 163-175.  | 21.4 | 1,403     |
| 142 | Nijmegen breakage syndrome. Archives of Disease in Childhood, 2000, 82, 400-406.  | 1.9  | 253       |
| 143 | V(D)J rearrangement in Nijmegen breakage syndrome. Molecular Immunology, 2000, 37, 1131-1139.   | 2.2  | 46        |
| 144 | The role of X-chromosome inactivation in female predisposition to autoimmunity. Arthritis Research, 2000, 2, 399.   | 2.0  | 85        |

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|-----|---|------|-----------|
| 145 | Regulated Genomic Instability and Neoplasia in the Lymphoid Lineage. Blood, 1999, 94, 3997-4010.  | 1.4  | 117       |
| 146 | Loci on chromosomes 2 (NIDDM1) and 15 interact to increase susceptibility to diabetes in Mexican Americans. Nature Genetics, 1999, 21, 213-215.   | 21.4 | 374       |
| 147 | Splicing Defects in the Ataxia-Telangiectasia Gene, ATM: Underlying Mutations and Consequences. American Journal of Human Genetics, 1999, 64, 1617-1631.  | 6.2  | 290       |
| 148 | Cancer Risk in ATM Heterozygotes: A Model of Phenotypic and Mechanistic Differences between Missense and Truncating Mutations. Molecular Genetics and Metabolism, 1999, 68, 419-423.  | 1.1  | 182       |
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