

# Miguel Angel Pujana

## List of Publications by Year in descending order

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Version: 2024-02-01

142  
papers

9,081  
citations

57631

44  
h-index

48187

88  
g-index

154  
all docs

154  
docs citations

154  
times ranked

17334  
citing authors

| #  | ARTICLE  | IF   | CITATIONS |
|----|--|------|-----------|
| 1  | Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2022, 114, 109-122.           | 3.0  | 19        |
| 2  | Evidence for shared genetic risk factors between lymphangiomyomatosis and pulmonary function. <i>ERJ Open Research</i> , 2022, 8, 00375-2021.  | 1.1  | 0         |
| 3  | CDK5RAP3, a New <i>BRCA2</i> Partner That Regulates DNA Repair, Is Associated with Breast Cancer Survival. <i>Cancers</i> , 2022, 14, 353.   | 1.7  | 0         |
| 4  | Validation of Anticorrelated TGF $\beta$ 2 Signaling and Alternative End-Joining DNA Repair Signatures that Predict Response to Genotoxic Cancer Therapy. <i>Clinical Cancer Research</i> , 2022, 28, 1372-1382. | 3.2  | 6         |
| 5  | Modification of <i>BRCA1</i> -associated breast cancer risk by HMMR overexpression. <i>Nature Communications</i> , 2022, 13, 1895.   | 5.8  | 19        |
| 6  | Pathogenic <i>BRCA1</i> variants disrupt <i>PLK1</i> -regulation of mitotic spindle orientation. <i>Nature Communications</i> , 2022, 13, 2200.  | 5.8  | 3         |
| 7  | A High-Throughput Screening Platform Identifies Novel Combination Treatments for Malignant Peripheral Nerve Sheath Tumors. <i>Molecular Cancer Therapeutics</i> , 2022, 21, 1246-1258.                           | 1.9  | 2         |
| 8  | A case-only study to identify genetic modifiers of breast cancer risk for <i>BRCA1/BRCA2</i> mutation carriers. <i>Nature Communications</i> , 2021, 12, 1078.   | 5.8  | 19        |
| 9  | <i>RNF168</i> regulates R-loop resolution and genomic stability in <i>BRCA1/2</i> -deficient tumors. <i>Journal of Clinical Investigation</i> , 2021, 131, .   | 3.9  | 38        |
| 10 | Loss of TGF $\beta$ 2 signaling increases alternative end-joining DNA repair that sensitizes to genotoxic therapies across cancer types. <i>Science Translational Medicine</i> , 2021, 13, .                     | 5.8  | 33        |
| 11 | Mammary epithelial cells have lineage-rooted metabolic identities. <i>Nature Metabolism</i> , 2021, 3, 665-681.  | 5.1  | 24        |
| 12 | Long-term results of sirolimus treatment in lymphangiomyomatosis: a single referral centre experience. <i>Scientific Reports</i> , 2021, 11, 10171.  | 1.6  | 9         |
| 13 | Heterogeneity and Cancer-Related Features in Lymphangiomyomatosis Cells and Tissue. <i>Molecular Cancer Research</i> , 2021, 19, 1840-1853.  | 1.5  | 3         |
| 14 | Altered regulation of <i>BRCA1</i> exon 11 splicing is associated with breast cancer risk in carriers of <i>BRCA1</i> pathogenic variants. <i>Human Mutation</i> , 2021, 42, 1488-1502.                          | 1.1  | 7         |
| 15 | Histamine signaling and metabolism identify potential biomarkers and therapies for lymphangiomyomatosis. <i>EMBO Molecular Medicine</i> , 2021, 13, e13929.  | 3.3  | 6         |
| 16 | Tumour <i>DDR1</i> promotes collagen fibre alignment to instigate immune exclusion. <i>Nature</i> , 2021, 599, 673-678.  | 18.7 | 139       |
| 17 | Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020, 80, 624-638.  | 0.4  | 39        |
| 18 | Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73.   | 9.4  | 120       |

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|----|--|-----|-----------|
| 19 | Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1653-1666.   | 1.1 | 82        |
| 20 | Immune Cell Associations with Cancer Risk. <i>IScience</i> , 2020, 23, 101296.   | 1.9 | 6         |
| 21 | NEK10 tyrosine phosphorylates p53 and controls its transcriptional activity. <i>Oncogene</i> , 2020, 39, 5252-5266.  | 2.6 | 12        |
| 22 | Characterization of the Cancer Spectrum in Men With Germline <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>JAMA Oncology</i> , 2020, 6, 1218.  | 3.4 | 48        |
| 23 | Tumors defective in homologous recombination rely on oxidative metabolism: relevance to treatments with <i>PARP</i> inhibitors. <i>EMBO Molecular Medicine</i> , 2020, 12, e11217.   | 3.3 | 37        |
| 24 | EV11 as a Prognostic and Predictive Biomarker of Clear Cell Renal Cell Carcinoma. <i>Cancers</i> , 2020, 12, 300.  | 1.7 | 9         |
| 25 | Looking for a Better Characterization of Triple-Negative Breast Cancer by Means of Circulating Tumor Cells. <i>Journal of Clinical Medicine</i> , 2020, 9, 353.  | 1.0 | 17        |
| 26 | Abstract 1388: Loss of <i>TGF<math>\beta</math>2</i> signaling increases alternative end-joining and could sensitize high-grade serous ovarian cancer to <i>PARP</i> inhibitors. , 2020, , .   |     | 0         |
| 27 | Allergy in patients with lymphangiomyomatosis. , 2020, , .   |     | 0         |
| 28 | Abstract P4-10-17: Baseline and pharmacodynamic changes of circulating exosomal microRNAs predict early versus late progression to palbociclib plus endocrine therapy in patients with metastatic breast cancer. A sub-analysis of the PARSIFAL-1 trial. , 2020, , . |     | 0         |
| 29 | The <i>FANCM</i> :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019, 5, 38.   | 2.3 | 28        |
| 30 | Chromosome 12p Amplification in Triple-Negative/ <i>BRCA1</i> -Mutated Breast Cancer Associates with Emergence of Docetaxel Resistance and Carboplatin Sensitivity. <i>Cancer Research</i> , 2019, 79, 4258-4270.  | 0.4 | 17        |
| 31 | Risk of breast cancer in patients with lymphangiomyomatosis. <i>Cancer Epidemiology</i> , 2019, 61, 154-156.   | 0.8 | 2         |
| 32 | A genome-wide association study implicates <i>NR2F2</i> in lymphangiomyomatosis pathogenesis. <i>European Respiratory Journal</i> , 2019, 53, 1900329.   | 3.1 | 14        |
| 33 | Differential metabolic activity and discovery of therapeutic targets using summarized metabolic pathway models. <i>Npj Systems Biology and Applications</i> , 2019, 5, 7.  | 1.4 | 30        |
| 34 | AhR controls redox homeostasis and shapes the tumor microenvironment in <i>BRCA1</i> -associated breast cancer. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 3604-3613.                                       | 3.3 | 96        |
| 35 | Evolutionary Changes after Translational Challenges Imposed by Horizontal Gene Transfer. <i>Genome Biology and Evolution</i> , 2019, 11, 814-831.  | 1.1 | 23        |
| 36 | Reactive oxygen species modulate macrophage immunosuppressive phenotype through the up-regulation of PD-L1. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 4326-4335.   | 3.3 | 137       |

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|----|---|-----|-----------|
| 37 | Generalised mosaicism for TSC2 mutation in isolated lymphangioleiomyomatosis. <i>European Respiratory Journal</i> , 2019, 54, 1900938.  | 3.1 | 5         |
| 38 | Decapping protein EDC4 regulates DNA repair and phenocopies BRCA1. <i>Nature Communications</i> , 2018, 9, 967.   | 5.8 | 33        |
| 39 | Tumor xenograft modeling identifies TCF4/ITF2 loss associated with breast cancer chemoresistance. <i>DMM Disease Models and Mechanisms</i> , 2018, 11, .  | 1.2 | 15        |
| 40 | Orthoxenografts of Testicular Germ Cell Tumors Demonstrate Genomic Changes Associated with Cisplatin Resistance and Identify PDMP as a Resensitizing Agent. <i>Clinical Cancer Research</i> , 2018, 24, 3755-3766.                          | 3.2 | 17        |
| 41 | ALK1 Loss Results in Vascular Hyperplasia in Mice and Humans Through PI3K Activation. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2018, 38, 1216-1229.  | 1.1 | 75        |
| 42 | Cell Cycle-Dependent Tumor Engraftment and Migration Are Enabled by Aurora-A. <i>Molecular Cancer Research</i> , 2018, 16, 16-31.   | 1.5 | 27        |
| 43 | Disease networks identify specific conditions and pleiotropy influencing multimorbidity in the general population. <i>Scientific Reports</i> , 2018, 8, 15970.  | 1.6 | 22        |
| 44 | A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. <i>Cancer Research</i> , 2018, 78, 5419-5430.  | 0.4 | 54        |
| 45 | Gene Expression Integration into Pathway Modules Reveals a Pan-Cancer Metabolic Landscape. <i>Cancer Research</i> , 2018, 78, 6059-6072.  | 0.4 | 40        |
| 46 | Subjugation of TGF $\beta$ 2 Signaling by Human Papilloma Virus in Head and Neck Squamous Cell Carcinoma Shifts DNA Repair from Homologous Recombination to Alternative End Joining. <i>Clinical Cancer Research</i> , 2018, 24, 6001-6014. | 3.2 | 71        |
| 47 | Ubiquitin ligase RNF8 suppresses Notch signaling to regulate mammary development and tumorigenesis. <i>Journal of Clinical Investigation</i> , 2018, 128, 4525-4542.  | 3.9 | 31        |
| 48 | Abstract 2812: Status of TGFbeta signaling determines PARP inhibitor sensitivity in head and neck cancer. , 2018, , .   |     | 0         |
| 49 | AURKA Overexpression Is Driven by FOXM1 and MAPK/ERK Activation in Melanoma Cells Harboring BRAF or NRAS Mutations: Impact on Melanoma Prognosis and Therapy. <i>Journal of Investigative Dermatology</i> , 2017, 137, 1297-1310.           | 0.3 | 40        |
| 50 | Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017, 49, 680-691.   | 9.4 | 356       |
| 51 | Stem cell-like transcriptional reprogramming mediates metastatic resistance to mTOR inhibition. <i>Oncogene</i> , 2017, 36, 2737-2749.  | 2.6 | 34        |
| 52 | Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017, 49, 1767-1778.  | 9.4 | 289       |
| 53 | DNA Methylomes Reveal Biological Networks Involved in Human Eye Development, Functions and Associated Disorders. <i>Scientific Reports</i> , 2017, 7, 11762.  | 1.6 | 44        |
| 54 | Attenuation of RNA polymerase II pausing mitigates BRCA1-associated R-loop accumulation and tumorigenesis. <i>Nature Communications</i> , 2017, 8, 15908.   | 5.8 | 118       |

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|----|---|-----|-----------|
| 55 | Prediction of Breast and Prostate Cancer Risks in Male <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017, 35, 2240-2250.            | 0.8 | 152       |
| 56 | <i>BRCA1</i> controls the cell division axis and governs ploidy and phenotype in human mammary cells. <i>Oncotarget</i> , 2017, 8, 32461-32475.   | 0.8 | 14        |
| 57 | Radioresistance of mesenchymal glioblastoma initiating cells correlates with patient outcome and is associated with activation of inflammatory program. <i>Oncotarget</i> , 2017, 8, 73640-73653.         | 0.8 | 33        |
| 58 | Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>PLoS ONE</i> , 2016, 11, e0158801.               | 1.1 | 10        |
| 59 | The acetyltransferase Tip60 contributes to mammary tumorigenesis by modulating DNA repair. <i>Cell Death and Differentiation</i> , 2016, 23, 1198-1208.   | 5.0 | 62        |
| 60 | Large-scale analysis of genome and transcriptome alterations in multiple tumors unveils novel cancer-relevant splicing networks. <i>Genome Research</i> , 2016, 26, 732-744.                              | 2.4 | 225       |
| 61 | Male breast cancer in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers: pathology data from the Consortium of Investigators of Modifiers of <i>BRCA1/2</i> . <i>Breast Cancer Research</i> , 2016, 18, 15. | 2.2 | 88        |
| 62 | Cancer network activity associated with therapeutic response and synergism. <i>Genome Medicine</i> , 2016, 8, 88.   | 3.6 | 7         |
| 63 | RANK Signaling Blockade Reduces Breast Cancer Recurrence by Inducing Tumor Cell Differentiation. <i>Cancer Research</i> , 2016, 76, 5857-5869.  | 0.4 | 47        |
| 64 | RANKL/RANK control <i>Brca1</i> mutation-driven mammary tumors. <i>Cell Research</i> , 2016, 26, 761-774.   | 5.7 | 128       |
| 65 | Study of breast cancer incidence in patients of lymphangioleiomyomatosis. <i>Breast Cancer Research and Treatment</i> , 2016, 156, 195-201.   | 1.1 | 9         |
| 66 | Rankl Impairs Lactogenic Differentiation Through Inhibition of the Prolactin/Stat5 Pathway at Midgestation. <i>Stem Cells</i> , 2016, 34, 1027-1039.  | 1.4 | 26        |
| 67 | Breast cancer risk variants at 6q25 display different phenotype associations and regulate <i>ESR1</i> , <i>RMND1</i> and <i>CCDC170</i> . <i>Nature Genetics</i> , 2016, 48, 374-386.                     | 9.4 | 125       |
| 68 | Cancer Stem-like Cells Act via Distinct Signaling Pathways in Promoting Late Stages of Malignant Progression. <i>Cancer Research</i> , 2016, 76, 1245-1259.   | 0.4 | 21        |
| 69 | Analysis of Paired Primary-Metastatic Hormone-Receptor Positive Breast Tumors (HRPBC) Uncovers Potential Novel Drivers of Hormonal Resistance. <i>PLoS ONE</i> , 2016, 11, e0155840.                      | 1.1 | 20        |
| 70 | Gasdermin B expression predicts poor clinical outcome in HER2-positive breast cancer. <i>Oncotarget</i> , 2016, 7, 56295-56308.   | 0.8 | 83        |
| 71 | Comprehensive establishment and characterization of orthoxenograft mouse models of malignant peripheral nerve sheath tumors for personalized medicine. <i>EMBO Molecular Medicine</i> , 2015, 7, 608-627. | 3.3 | 36        |
| 72 | Assessing Associations between the <i>AURKA-HMMR-TPX2-TUBG1</i> Functional Module and Breast Cancer Risk in <i>BRCA1/2</i> Mutation Carriers. <i>PLoS ONE</i> , 2015, 10, e0120020.                       | 1.1 | 34        |

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|----|---|-----|-----------|
| 73 | Lymphangiomyomatosis Biomarkers Linked to Lung Metastatic Potential and Cell Stemness. PLoS ONE, 2015, 10, e0132546.  | 1.1 | 15        |
| 74 | Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.   | 9.4 | 221       |
| 75 | <i>FANCM</i> c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355. | 1.4 | 91        |
| 76 | Germline Mutations in FAN1 Cause Hereditary Colorectal Cancer by Impairing DNA Repair. Gastroenterology, 2015, 149, 563-566.  | 0.6 | 94        |
| 77 | A validated gene regulatory network and GWAS identifies early regulators of T cell-associated diseases. Science Translational Medicine, 2015, 7, 313ra178.  | 5.8 | 66        |
| 78 | PKA signaling drives mammary tumorigenesis through Src. Oncogene, 2015, 34, 1160-1173.  | 2.6 | 75        |
| 79 | Integrating gene expression and epidemiological data for the discovery of genetic interactions associated with cancer risk. Carcinogenesis, 2014, 35, 578-585.  | 1.3 | 1         |
| 80 | Modules, networks and systems medicine for understanding disease and aiding diagnosis. Genome Medicine, 2014, 6, 82.  | 3.6 | 169       |
| 81 | VAV3 mediates resistance to breast cancer endocrine therapy. Breast Cancer Research, 2014, 16, R53.   | 2.2 | 28        |
| 82 | Integrated genomic and prospective clinical studies show the importance of modular pleiotropy for disease susceptibility, diagnosis and treatment. Genome Medicine, 2014, 6, 17.                      | 3.6 | 27        |
| 83 | Tubers from patients with tuberous sclerosis complex are characterized by changes in microtubule biology through <i>ROCK2</i> signalling. Journal of Pathology, 2014, 233, 247-257.                   | 2.1 | 7         |
| 84 | Integrating germline and somatic data towards a personalized cancer medicine. Trends in Molecular Medicine, 2014, 20, 413-415.  | 3.5 | 9         |
| 85 | Linkage of DNA Methylation Quantitative Trait Loci to Human Cancer Risk. Cell Reports, 2014, 7, 331-338.  | 2.9 | 76        |
| 86 | Constitutive activation of RANK disrupts mammary cell fate leading to tumorigenesis. Stem Cells, 2013, 31, 1954-1965.   | 1.4 | 40        |
| 87 | Serum 25-hydroxyvitamin D3 levels and vitamin D receptor variants in melanoma patients from the Mediterranean area of Barcelona. BMC Medical Genetics, 2013, 14, 26.                                  | 2.1 | 24        |
| 88 | Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.                                      | 1.5 | 244       |
| 89 | Evaluation of <i>PAX3</i> genetic variants and nevus number. Pigment Cell and Melanoma Research, 2013, 26, 666-676.   | 1.5 | 7         |
| 90 | Functional and Structural Analysis of C-Terminal BRCA1 Missense Variants. PLoS ONE, 2013, 8, e61302.  | 1.1 | 16        |

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|-----|--|-----|-----------|
| 91  | Genomic imbalance of <i>HMMR/RHAMM</i> regulates the sensitivity and response of malignant peripheral nerve sheath tumour cells to aurora kinase inhibition. <i>Oncotarget</i> , 2013, 4, 80-93.             | 0.8 | 27        |
| 92  | Lurbinectedin (PM01183), a New DNA Minor Groove Binder, Inhibits Growth of Orthotopic Primary Graft of Cisplatin-Resistant Epithelial Ovarian Cancer. <i>Clinical Cancer Research</i> , 2012, 18, 5399-5411. | 3.2 | 86        |
| 93  | Cancer develops, progresses and responds to therapies through restricted perturbation of the protein-protein interaction network. <i>Integrative Biology (United Kingdom)</i> , 2012, 4, 1038.               | 0.6 | 10        |
| 94  | DNA Methylation Plasticity of Human Adipose-Derived Stem Cells in Lineage Commitment. <i>American Journal of Pathology</i> , 2012, 181, 2079-2093.   | 1.9 | 36        |
| 95  | Distinct DNA methylomes of newborns and centenarians. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 10522-10527.                                       | 3.3 | 687       |
| 96  | Analysis of SLX4/FANCP in non-BRCA1/2-mutated breast cancer families. <i>BMC Cancer</i> , 2012, 12, 84.  | 1.1 | 14        |
| 97  | Tools for protein-protein interaction network analysis in cancer research. <i>Clinical and Translational Oncology</i> , 2012, 14, 3-14.  | 1.2 | 35        |
| 98  | Abstract 2608: Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. , 2012, , .   |     | 0         |
| 99  | Exploring the link between MORF4L1 and risk of breast cancer. <i>Breast Cancer Research</i> , 2011, 13, R40.   | 2.2 | 23        |
| 100 | Evidence for a link between TNFRSF11A and risk of breast cancer. <i>Breast Cancer Research and Treatment</i> , 2011, 129, 947-954.   | 1.1 | 12        |
| 101 | Validation of a DNA methylation microarray for 450,000 CpG sites in the human genome. <i>Epigenetics</i> , 2011, 6, 692-702.   | 1.3 | 908       |
| 102 | Gene Expression Differences between Colon and Rectum Tumors. <i>Clinical Cancer Research</i> , 2011, 17, 7303-7312.  | 3.2 | 69        |
| 103 | Interplay between BRCA1 and RHAMM Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. <i>PLoS Biology</i> , 2011, 9, e1001199.   | 2.6 | 91        |
| 104 | Biological reprogramming in acquired resistance to endocrine therapy of breast cancer. <i>Oncogene</i> , 2010, 29, 6071-6083.  | 2.6 | 59        |
| 105 | Exploring the Link between Germline and Somatic Genetic Alterations in Breast Carcinogenesis. <i>PLoS ONE</i> , 2010, 5, e14078.   | 1.1 | 33        |
| 106 | TACC3-TSC2 maintains nuclear envelope structure and controls cell division. <i>Cell Cycle</i> , 2010, 9, 1143-1155.  | 1.3 | 46        |
| 107 | Gene set-based analysis of polymorphisms: finding pathways or biological processes associated to traits in genome-wide association studies. <i>Nucleic Acids Research</i> , 2009, 37, W340-W344.             | 6.5 | 64        |
| 108 | Gene expression profiling integrated into network modelling reveals heterogeneity in the mechanisms of BRCA1 tumorigenesis. <i>British Journal of Cancer</i> , 2009, 101, 1469-1480.                         | 2.9 | 13        |

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|-----|--|-----|-----------|
| 109 | Biological Convergence of Cancer Signatures. PLoS ONE, 2009, 4, e4544.   | 1.1 | 20        |
| 110 | Biological processes, properties and molecular wiring diagrams of candidate low-penetrance breast cancer susceptibility genes. BMC Medical Genomics, 2008, 1, 62.  | 0.7 | 13        |
| 111 | Genetic and genomic analysis modeling of germline c-MYC overexpression and cancer susceptibility. BMC Genomics, 2008, 9, 12.   | 1.2 | 27        |
| 112 | CLEAR-test: Combining inference for differential expression and variability in microarray data analysis. Journal of Biomedical Informatics, 2008, 41, 33-45.   | 2.5 | 8         |
| 113 | Genetic interactions: the missing links for a better understanding of cancer susceptibility, progression and treatment. Molecular Cancer, 2008, 7, 4.  | 7.9 | 10        |
| 114 | Genetic Variants in Apoptosis and Immunoregulation-Related Genes Are Associated with Risk of Chronic Lymphocytic Leukemia. Cancer Research, 2008, 68, 10178-10186.   | 0.4 | 67        |
| 115 | Fas-activated serine/threonine phosphoprotein (FAST) is a regulator of alternative splicing. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 11370-11375.  | 3.3 | 32        |
| 116 | Integrative analysis of a cancer somatic mutome. Molecular Cancer, 2007, 6, 13.  | 7.9 | 28        |
| 117 | Molecular characterization of a t(9;12)(p21;q13) balanced chromosome translocation in combination with integrative genomics analysis identifies C9orf14 as a candidate tumor-suppressor. Genes Chromosomes and Cancer, 2007, 46, 155-162.                | 1.5 | 10        |
| 118 | Network modeling links breast cancer susceptibility and centrosome dysfunction. Nature Genetics, 2007, 39, 1338-1349.  | 9.4 | 602       |
| 119 | Evidence for systems-level molecular mechanisms of tumorigenesis. BMC Genomics, 2007, 8, 185.  | 1.2 | 31        |
| 120 | Geminin is bound to chromatin in G2/M phase to promote proper cytokinesis. International Journal of Biochemistry and Cell Biology, 2006, 38, 1207-1220.  | 1.2 | 15        |
| 121 | Transgenic mice overexpressing the full-length neurotrophin receptor TrkC exhibit increased catecholaminergic neuron density in specific brain areas and increased anxiety-like behavior and panic reaction. Neurobiology of Disease, 2006, 24, 403-418. | 2.1 | 50        |
| 122 | Novel Pheochromocytoma Susceptibility Loci Identified by Integrative Genomics. Cancer Research, 2005, 65, 9651-9658.   | 0.4 | 88        |
| 123 | Enrichment of segmental duplications in regions of breaks of synteny between the human and mouse genomes suggest their involvement in evolutionary rearrangements. Human Molecular Genetics, 2003, 12, 2201-2208.  | 1.4 | 121       |
| 124 | Human Chromosome 7: DNA Sequence and Biology. Science, 2003, 300, 767-772.   | 6.0 | 185       |
| 125 | Genomic inversions of human chromosome 15q11-q13 in mothers of Angelman syndrome patients with class II (BP2/3) deletions. Human Molecular Genetics, 2003, 12, 849-858.  | 1.4 | 131       |
| 126 | Human chromosome 15q11-q14 regions of rearrangements contain clusters of LCR15 duplicons. European Journal of Human Genetics, 2002, 10, 26-35.   | 1.4 | 92        |



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|-----|--|------|-----------|
| 127 | Chromosomal regions containing high-density and ambiguously mapped putative single nucleotide polymorphisms (SNPs) correlate with segmental duplications in the human genome. <i>Human Molecular Genetics</i> , 2002, 11, 1987-1995. | 1.4  | 80        |
| 128 | Cloning of S4D-SRCRB, a new soluble member of the group B scavenger receptor cysteine-rich family (SRCR-SF) mapping to human Chromosome 7q11.23. <i>Immunogenetics</i> , 2002, 54, 621-634.  | 1.2  | 13        |
| 129 | 5â€² UTR-region SNP in the NTRK3 gene is associated with panic disorder. <i>Molecular Psychiatry</i> , 2002, 7, 928-930.   | 4.1  | 28        |
| 130 | A Polymorphic Genomic Duplication on Human Chromosome 15 Is a Susceptibility Factor for Panic and Phobic Disorders. <i>Cell</i> , 2001, 106, 367-379.  | 13.5 | 219       |
| 131 | Additional Complexity on Human Chromosome 15q: Identification of a Set of Newly Recognized Duplicons (LCR15) on 15q11-q13, 15q24, and 15q26. <i>Genome Research</i> , 2001, 11, 98-111.  | 2.4  | 60        |
| 132 | Isolation and characterisation of a novel human gene (C9orf11) on chromosome 9p21, a region frequently deleted in human cancer. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 2000, 1517, 128-134.               | 2.4  | 6         |
| 133 | Genomic organization of the human CD5 gene. <i>Immunogenetics</i> , 2000, 51, 993-1001.  | 1.2  | 40        |
| 134 | HMG20A and HMG20B map to human chromosomes 15q24 and 19p13.3 and constitute a distinct class of HMG-box genes with ubiquitous expression. <i>Cytogenetic and Genome Research</i> , 2000, 88, 62-67.                                  | 0.6  | 36        |
| 135 | Spinocerebellar ataxias in Spanish patients: genetic analysis of familial and sporadic cases. <i>Human Genetics</i> , 1999, 104, 516-522.  | 1.8  | 140       |
| 136 | Anticipation is not associated with CAG repeat expansion in parent-offspring pairs of patients affected with schizophrenia. , 1999, 88, 50-56.   |      | 13        |
| 137 | Uncloned expanded CAG/CTG repeat sequences in autosomal dominant cerebellar ataxia (ADCA) detected by the repeat expansion detection (RED) method.. <i>Journal of Medical Genetics</i> , 1998, 35, 99-102.                           | 1.5  | 6         |
| 138 | Large CAG/CTG repeat templates produced by PCR, usefulness for the DIRECT method of cloning genes with CAG/CTG repeat expansions. <i>Nucleic Acids Research</i> , 1998, 26, 1352-1353.   | 6.5  | 13        |
| 139 | Analysis of amino-acid and nucleotide variants in the spinocerebellar ataxia type 1 ( SCA1 ) gene in schizophrenic patients. <i>Human Genetics</i> , 1997, 99, 772-775.  | 1.8  | 6         |
| 140 | Polymorphisms at 13 expressed human sequences containing CAG/CTG repeats and analysis in autosomal dominant cerebellar ataxia (ADCA) patients. <i>Human Genetics</i> , 1997, 101, 18-21.   | 1.8  | 3         |
| 141 | The repeat expansion detection method in the analysis of diseases with CAG/CTG repeat expansion: Usefulness and limitations. <i>Human Mutation</i> , 1997, 10, 486-488.  | 1.1  | 9         |
| 142 | Cloning (CAG/GTC) <sub>n</sub> STSs by an Alu-(CAG/GTC) <sub>n</sub> PCR method: an approach to human chromosome 12 and spinocerebellar ataxia 2 (SCA2). <i>Nucleic Acids Research</i> , 1996, 24, 3651-3652.                        | 6.5  | 2         |