# Peter J Van Der Spek

List of Publications by Year
in descending order



| 71 | 3,673 | 186265 28 | 138484 58 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 73 | 73 | 73 | 6052 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Xeroderma Pigmentosum Group C Protein Complex Is the Initiator of Global Genome Nucleotide Excision Repair. Molecular Cell, 1998, 2, 223-232. | 9.7 | 796 |
| 2 | The Three Subfamilies of Leucine-Rich Repeat-Containing G Protein-Coupled Receptors (LGR): Identification of LGR6 and LGR7 and the Signaling Mechanism for LGR7. Molecular Endocrinology, 2000, 14, 1257-1271. | 3.7 | 329 |
| 3 | A new gene, encoding an anion transporter, is mutated in sialic acid storage diseases. Nature Genetics, 1999, 23, 462-465. | 21.4 | 252 |
| 4 | Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. Nature Genetics, 2018, 50, 834-848. | 21.4 | 239 |
| 5 | Rec8p, a Meiotic Recombination and Sister Chromatid Cohesion Phosphoprotein of the Rad21p Family Conserved from Fission Yeast to Humans. Molecular and Cellular Biology, 1999, 19, 3515-3528. | 2.3 | 235 |
| 6 | Diagnostic value of exome and whole genome sequencing in craniosynostosis. Journal of Medical Genetics, 2017, 54, 260-268. | 3.2 | 107 |
| 7 | A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS ONE, 2016, 11, e0162866. | 2.5 | 96 |
| 8 | Mutational analysis of the human nucleotide excision repair gene ERCC1. Nucleic Acids Research, 1996, 24, 3370-3380. | 14.5 | 93 |
| 9 | IL-7 Receptor Mutations and Steroid Resistance in Pediatric T cell Acute Lymphoblastic Leukemia: A Genome Sequencing Study. PLoS Medicine, 2016, 13, e1002200. | 8.4 | 89 |
| 10 | On the origin of proteins in human drusen: The meet, greet and stick hypothesis. Progress in Retinal and Eye Research, 2019, 70, 55-84. | 15.5 | 77 |
| 11 | A New Strategy to Identify and Annotate Human RPE-Specific Gene Expression. PLoS ONE, 2010, 5, e9341. | 2.5 | 72 |
| 12 | Cloning, Comparative Mapping, and RNA Expression of the Mouse Homologues of theSaccharomyces cerevisiaeNucleotide Excision Repair GeneRAD23. Genomics, 1996, 31, 20-27. | 2.9 | 66 |
| 13 | Human mutations in integrator complex subunits link transcriptome integrity to brain development. PLoS Genetics, 2017, 13, e1006809. | 3.5 | 66 |
| 14 | Gain-of-Function Mutations in ZIC1 Are Associated with Coronal Craniosynostosis and Learning Disability. American Journal of Human Genetics, 2015, 97, 378-388. | 6.2 | 56 |
| 15 | Functional annotation of the human retinal pigment epithelium transcriptome. BMC Genomics, 2009, 10, 164. | 2.8 | 52 |
| 16 | Early- and late-onset preeclampsia and the tissue-specific epigenome of the placenta and newborn. Placenta, 2017, 58, 122-132. | 1.5 | 52 |
| 17 | Gene Expression and Functional Annotation of the Human and Mouse Choroid Plexus Epithelium. PLoS ONE, 2013, 8, e83345. | 2.5 | 50 |
| 18 | ZC4H2, an XLID gene, is required for the generation of a specific subset of CNS interneurons. Human Molecular Genetics, 2015, 24, 4848-4861. | 2.9 | 48 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Comparison of Mouse and Human Retinal Pigment Epithelium Gene Expression Profiles: Potential Implications for Age-Related Macular Degeneration. PLoS ONE, 2015, 10, e0141597. | 2.5 | 47 |
| 20 | Bi-allelic TARS Mutations Are Associated with Brittle Hair Phenotype. American Journal of Human Genetics, 2019, 105, 434-440. | 6.2 | 42 |
| 21 | De Novo Missense Substitutions in the Gene Encoding CDK8, a Regulator of the Mediator Complex, Cause a Syndromic Developmental Disorder. American Journal of Human Genetics, 2019, 104, 709-720. | 6.2 | 41 |
| 22 | Molecular Binding Mechanism and Pharmacology Comparative Analysis of Noscapine for Repurposing against SARS-CoV-2 Protease. Journal of Proteome Research, 2020, 19, 4678-4689. | 3.7 | 41 |
| 23 | A Novel Heterozygous Mutation in the STAT1 SH2 Domain Causes Chronic Mucocutaneous Candidiasis, Atypically Diverse Infections, Autoimmunity, and Impaired Cytokine Regulation. Frontiers in Immunology, 2017, 8, 274. | 4.8 | 40 |
| 24 | De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203. | 6.2 | 37 |
| 25 | Studies of Nematode TFIIE Function Reveal a Link between Ser-5 Phosphorylation of RNA Polymerase II and the Transition from Transcription Initiation to Elongation. Molecular and Cellular Biology, 2001, 21, 1-15. | 2.3 | 35 |
| 26 | Automated Selection of Hotspots (ASH): enhanced automated segmentation and adaptive step finding for Ki67 hotspot detection in adrenal cortical cancer. Diagnostic Pathology, 2014, 9, 216. | 2.0 | 33 |
| 27 | Precursor lesions of vulvar squamous cell carcinoma â€“ histology and biomarkers: A systematic review. Critical Reviews in Oncology/Hematology, 2020, 147, 102866. | 4.4 | 32 |
| 28 | Guide-free Cas9 from pathogenic Campylobacter jejuni bacteria causes severe damage to DNA. Science Advances, 2020, 6, eaaz4849. | 10.3 | 31 |
| 29 | Characterization of a Novel Human Cell-Cycle-Regulated Homologue of Drosophila dlg1. Genomics, 2001, 77, 5-7. | 2.9 | 30 |
| 30 | ImmunoGlobulin galaxy (IGGalaxy) for simple determination and quantitation of immunoglobulin heavy chain rearrangements from NGS. BMC Immunology, 2014, 15, 59. | 2.2 | 30 |
| 31 | Increased serum levels of leptin and insulin in both schizophrenia and major depressive disorder: A cross-disorder proteomics analysis. European Neuropsychopharmacology, 2019, 29, 835-846. | 0.7 | 26 |
| 32 | TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. American Journal of Human Genetics, 2019, 105, 1126-1147. | 6.2 | 25 |
| 33 | A de novo substitution in BCL11B leads to loss of interaction with transcriptional complexes and craniosynostosis. Human Molecular Genetics, 2019, 28, 2501-2513. | 2.9 | 23 |
| 34 | Retinal pigment epithelial cells display specific transcriptional responses upon TNF-Î± stimulation. British Journal of Ophthalmology, 2015, 99, 700-704. | 3.9 | 21 |
| 35 | A point mutation in the pre-ZRS disrupts sonic hedgehog expression in the limb bud and results in triphalangeal thumbâ€“polysyndactyly syndrome. Genetics in Medicine, 2018, 20, 1405-1413. | 2.4 | 21 |
| 36 | Retinal Pigment Epithelial Cells Control Early Mycobacterium tuberculosis Infection via Interferon Signaling. , 2018, 59, 1384. | | 20 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | A novel definition and treatment of hyperinflammation in COVID-19 based on purinergic signalling. Purinergic Signalling, 2022, 18, 13-59. | 2.2 | 20 |
| 38 | Identification of Intragenic Exon Deletions and Duplication ofTCF12by Whole Genome or Targeted Sequencing as a Cause ofTCF12-Related Craniosynostosis. Human Mutation, 2016, 37, 732-736. | 2.5 | 19 |
| 39 | Fatigue in SjÃ¶gren's Syndrome: A Search for Biomarkers and Treatment Targets. Frontiers in Immunology, 2019, 10, 312. | 4.8 | 18 |
| 40 | Galaxy mothur Toolset (GmT): a user-friendly application for 16S rRNA gene sequencing analysis using mothur. GigaScience, 2019, 8, . | 6.4 | 17 |
| 41 | First trimester physiological development of the fetal foot position using threeâ€dimensional ultrasound in virtual reality. Journal of Obstetrics and Gynaecology Research, 2019, 45, 280-288. | 1.3 | 16 |
| 42 | A multidisciplinary review of triphalangeal thumb. Journal of Hand Surgery: European Volume, 2019, 44, 59-68. | 1.0 | 15 |
| 43 | A novel machine learning-based approach for the computational functional assessment of pharmacogenomic variants. Human Genomics, 2021, 15, 51. | 2.9 | 14 |
| 44 | The ethnogeographic variability of genetic factors underlying G6PD deficiency. Pharmacological Research, 2021, 173, 105904. | 7.1 | 14 |
| 45 | The tissue-specific aspect of genome-wide DNA methylation in newborn and placental tissues: implications for epigenetic epidemiologic studies. Journal of Developmental Origins of Health and Disease, 2021, 12, 113-123. | 1.4 | 13 |
| 46 | CXorf56, a dendritic neuronal protein, identified as a new candidate gene for X-linked intellectual disability. European Journal of Human Genetics, 2018, 26, 552-560. | 2.8 | 12 |
| 47 | Campylobacter jejuni Cas9 Modulates the Transcriptome in Caco-2 Intestinal Epithelial Cells. Genes, 2020, 11, 1193. | 2.4 | 12 |
| 48 | Early Pregnancy Placental Bed and Fetal Vascular Volume Measurements Using 3-D Virtual Reality. Ultrasound in Medicine and Biology, 2014, 40, 1796-1803. | 1.5 | 11 |
| 49 | Identification of Associated Genes and Diseases in Patients With Congenital Upper-Limb Anomalies: A Novel Application of the OMT Classification. Journal of Hand Surgery, 2017, 42, 533-545.e4. | 1.6 | 11 |
| 50 | Accuracy of fetal sex determination in the first trimester of pregnancy using 3D virtual reality ultrasound. Journal of Clinical Ultrasound, 2018, 46, 241-246. | 0.8 | 11 |
| 51 | Integrative Analysis of Proteomics and DNA Methylation in Orbital Fibroblasts From Gravesâ€™ Ophthalmopathy. Frontiers in Endocrinology, 2020, 11, 619989. | 3.5 | 11 |
| 52 | First-Trimester Detection of Surface Abnormalities: A Comparison of 2- and 3-Dimensional Ultrasound and 3-Dimensional Virtual Reality Ultrasound. Reproductive Sciences, 2014, 21, 993-999. | 2.5 | 10 |
| 53 | Genome-wide methylation analysis identifies novel CpG loci for perimembranous ventricular septal defects in human. Epigenomics, 2017, 9, 241-251. | 2.1 | 10 |
| 54 | Fusion transcripts and their genomic breakpoints in polyadenylated and ribosomal RNAâ€“minus RNA sequencing data. GigaScience, 2021, 10, . | 6.4 | 10 |

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|---|---|---|---|
| 55 | Evaluation of First-Trimester Physiological Midgut Herniation Using Three-Dimensional Ultrasound. Fetal Diagnosis and Therapy, 2019, 45, 332-338. | 1.4 | 9 |
| 56 | Evaluation of Immunohistochemical Markers, CK17 and SOX2, as Adjuncts to p53 for the Diagnosis of Differentiated Vulvar Intraepithelial Neoplasia (dVIN). Pharmaceuticals, 2021, 14, 324. | 3.8 | 9 |
| 57 | Endocrine Disorders Are Prominent Clinical Features in Patients With Primary Antibody Deficiencies. Frontiers in Immunology, 2019, 10, 2079. | 4.8 | 8 |
| 58 | Documentation of clinically relevant genomic biomarker allele frequencies in the nextâ€generation FINDbase worldwide database. Human Mutation, 2020, 41, 1112-1122. | 2.5 | 7 |
| 59 | Pollitt syndrome patients carry mutation in TTDN1. Meta Gene, 2014, 2, 616-618. | 0.6 | 6 |
| 60 | Type 1 interferon-inducible gene expression in QuantiFERON Gold TB-positive uveitis: A tool to stratify a high versus low risk of active tuberculosis?. PLoS ONE, 2018, 13, e0206073. | 2.5 | 6 |
| 61 | A Novel Text-Mining Approach for Retrieving Pharmacogenomics Associations From the Literature. Frontiers in Pharmacology, 2020, 11, 602030. | 3.5 | 5 |
| 62 | Identification of a novel variant of the ciliopathic gene FUZZY associated with craniosynostosis. European Journal of Human Genetics, 2022, 30, 282-290. | 2.8 | 5 |
| 63 | FuMa: reporting overlap in RNA-seq detected fusion genes. Bioinformatics, 2016, 32, 1226-1228. | 4.1 | 4 |
| 64 | Functional Analysis of Genetic Variation in the SECIS Element of Thyroid Hormone Activating Type 2 Deiodinase. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 1369-1377. | 3.6 | 4 |
| 65 | Exploring Differentially Methylated Genes in Vulvar Squamous Cell Carcinoma. Cancers, 2021, 13, 3580. | 3.7 | 4 |
| 66 | Circulating angiogenic cells in glioblastoma: toward defining crucial functional differences in CAC-induced neoplastic versus reactive neovascularization. Neuro-Oncology Advances, 2020, 2, vdaa040. | 0.7 | 3 |
| 67 | Molecular data show conserved DNA locations distinguishing lung cancer subtypes and regulation of immune genes. Lung Cancer, 2020, 146, 341-349. | 2.0 | 3 |
| 68 | Nuclear factorÂIB is downregulated in vulvar squamous cell carcinoma (VSCC): Unravelling differentially expressed genes in VSCC through gene expression dataset analysis. Oncology Letters, 2021, 21, 381. | 1.8 | 2 |
| 69 | Heart failure and promotion of physical activity before and after cardiac rehabilitation (HFâ€PProACH): a study protocol. ESC Heart Failure, 2021, 8, 3621-3627. | 3.1 | 2 |
| 70 | CRISPRs in the human genome are differentially expressed between malignant and normalÂadjacent to tumor tissue. Communications Biology, 2022, 5, 338. | 4.4 | 2 |
| 71 | A Single Microarray Assay for Simultaneous Diagnosis of t(15;17), t(8;21), Inv(16)/t(16;16), NPM1 Type A/B/D Mutation, CEBPA Double Mutation, and Aberrant Expression of BAALC or EVI1 in AML/APL Patients. Blood, 2011, 118, 4876-4876. | 1.4 | 0 |