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	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
2	A novel definition and treatment of hyperinflammation in COVID-19 based on purinergic signalling. Purinergic Signalling, 2022, 18, 13-59.	2.2	20
3	CRISPRs in the human genome are differentially expressed between malignant and normalÂadjacent to tumor tissue. Communications Biology, 2022, 5, 338.	4.4	2
4	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
5	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
6	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
7	Exploring Differentially Methylated Genes in Vulvar Squamous Cell Carcinoma. Cancers, 2021, 13, 3580.	3.7	4
8	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
9	A novel machine learning-based approach for the computational functional assessment of pharmacogenomic variants. Human Genomics, 2021, 15, 51.	2.9	14
10	The ethnogeographic variability of genetic factors underlying G6PD deficiency. Pharmacological Research, 2021, 173, 105904.	7.1	14
11	Fusion transcripts and their genomic breakpoints in polyadenylated and ribosomal RNAâ€“minus RNA sequencing data. GigaScience, 2021, 10, .	6.4	10
12	A Novel Text-Mining Approach for Retrieving Pharmacogenomics Associations From the Literature. Frontiers in Pharmacology, 2020, 11, 602030.	3.5	5
13	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
14	Molecular data show conserved DNA locations distinguishing lung cancer subtypes and regulation of immune genes. Lung Cancer, 2020, 146, 341-349.	2.0	3
15	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
16	Campylobacter jejuni Cas9 Modulates the Transcriptome in Caco-2 Intestinal Epithelial Cells. Genes, 2020, 11, 1193.	2.4	12
17	Guide-free Cas9 from pathogenic Campylobacter jejuni bacteria causes severe damage to DNA. Science Advances, 2020, 6, eaaz4849.	10.3	31
18	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

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19	Documentation of clinically relevant genomic biomarker allele frequencies in the nextâ€generation FINDbase worldwide database. Human Mutation, 2020, 41, 1112-1122.	2.5	7
20	Integrative Analysis of Proteomics and DNA Methylation in Orbital Fibroblasts From Gravesâ€™ Ophthalmopathy. Frontiers in Endocrinology, 2020, 11, 619989.	3.5	11
21	Evaluation of First-Trimester Physiological Midgut Herniation Using Three-Dimensional Ultrasound. Fetal Diagnosis and Therapy, 2019, 45, 332-338.	1.4	9
22	Bi-allelic TARS Mutations Are Associated with Brittle Hair Phenotype. American Journal of Human Genetics, 2019, 105, 434-440.	6.2	42
23	Endocrine Disorders Are Prominent Clinical Features in Patients With Primary Antibody Deficiencies. Frontiers in Immunology, 2019, 10, 2079.	4.8	8
24	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
25	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
26	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
27	Fatigue in SjÃ¶gren's Syndrome: A Search for Biomarkers and Treatment Targets. Frontiers in Immunology, 2019, 10, 312.	4.8	18
28	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
29	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
30	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
31	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
32	Galaxy mothur Toolset (GmT): a user-friendly application for 16S rRNA gene sequencing analysis using mothur. GigaScience, 2019, 8, .	6.4	17
33	A multidisciplinary review of triphalangeal thumb. Journal of Hand Surgery: European Volume, 2019, 44, 59-68.	1.0	15
34	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
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	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

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37	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
38	Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. Nature Genetics, 2018, 50, 834-848.	21.4	239
39	Retinal Pigment Epithelial Cells Control Early Mycobacterium tuberculosis Infection via Interferon Signaling. , 2018, 59, 1384.		20
40	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
41	Genome-wide methylation analysis identifies novel CpG loci for perimembranous ventricular septal defects in human. Epigenomics, 2017, 9, 241-251.	2.1	10
42	Diagnostic value of exome and whole genome sequencing in craniosynostosis. Journal of Medical Genetics, 2017, 54, 260-268.	3.2	107
43	Early- and late-onset preeclampsia and the tissue-specific epigenome of the placenta and newborn. Placenta, 2017, 58, 122-132.	1.5	52
44	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
45	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
46	Human mutations in integrator complex subunits link transcriptome integrity to brain development. PLoS Genetics, 2017, 13, e1006809.	3.5	66
47	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS ONE, 2016, 11, e0162866.	2.5	96
48	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
49	FuMa: reporting overlap in RNA-seq detected fusion genes. Bioinformatics, 2016, 32, 1226-1228.	4.1	4
50	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
51	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
52	Retinal pigment epithelial cells display specific transcriptional responses upon TNF-Î± stimulation. British Journal of Ophthalmology, 2015, 99, 700-704.	3.9	21
53	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
54	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

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55	ImmunoGlobulin galaxy (IGGalaxy) for simple determination and quantitation of immunoglobulin heavy chain rearrangements from NGS. BMC Immunology, 2014, 15, 59.	2.2	30
56	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
57	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
58	Pollitt syndrome patients carry mutation in TTDN1. Meta Gene, 2014, 2, 616-618.	0.6	6
59	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
60	Gene Expression and Functional Annotation of the Human and Mouse Choroid Plexus Epithelium. PLoS ONE, 2013, 8, e83345.	2.5	50
61	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
62	A New Strategy to Identify and Annotate Human RPE-Specific Gene Expression. PLoS ONE, 2010, 5, e9341.	2.5	72
63	Functional annotation of the human retinal pigment epithelium transcriptome. BMC Genomics, 2009, 10, 164.	2.8	52
64	Characterization of a Novel Human Cell-Cycle-Regulated Homologue of Drosophila dlg1. Genomics, 2001, 77, 5-7.	2.9	30
65	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
66	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
67	A new gene, encoding an anion transporter, is mutated in sialic acid storage diseases. Nature Genetics, 1999, 23, 462-465.	21.4	252
68	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
69	Xeroderma Pigmentosum Group C Protein Complex Is the Initiator of Global Genome Nucleotide Excision Repair. Molecular Cell, 1998, 2, 223-232.	9.7	796
70	Cloning, Comparative Mapping, and RNA Expression of the Mouse Homologues of theSaccharomyces cerevisiaeNucleotide Excision Repair GeneRAD23. Genomics, 1996, 31, 20-27.	2.9	66
71	Mutational analysis of the human nucleotide excision repair gene ERCC1. Nucleic Acids Research, 1996, 24, 3370-3380.	14.5	93