

# Jes s Jvd Del Valle

## List of Publications by Year in descending order

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Version: 2024-02-01

47  
papers

1,196  
citations

430874

18  
h-index

395702

33  
g-index

48  
all docs

48  
docs citations

48  
times ranked

3092  
citing authors

#	ARTICLE	IF	CITATIONS
1	Solving the enigma of POLD1 p.V295M as a potential cause of increased cancer risk. <i>European Journal of Human Genetics</i> , 2022, 30, 485-489.	2.8	2
2	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.	6.3	19
3	Variants of uncertain significance (VUS) in cancer predisposing genes: What are we learning from multigene panels?. <i>European Journal of Medical Genetics</i> , 2022, 65, 104400.	1.3	4
4	Mosaicism in <i>PTEN</i> —new case and comment on the literature. <i>European Journal of Human Genetics</i> , 2022, 30, 641-644.	2.8	6
5	Characteristics of Adrenocortical Carcinoma Associated With Lynch Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, 318-325.	3.6	20
6	<i>BARD1</i> Pathogenic Variants Are Associated with Triple-Negative Breast Cancer in a Spanish Hereditary Breast and Ovarian Cancer Cohort. <i>Genes</i> , 2021, 12, 150.	2.4	11
7	The Challenge of Diagnosing Constitutional Mismatch Repair Deficiency Syndrome in Brain Malignancies from Young Individuals. <i>International Journal of Molecular Sciences</i> , 2021, 22, 4629.	4.1	9
8	CNVfilter: an R/Bioconductor package to identify false positives produced by germline NGS CNV detection tools. <i>Bioinformatics</i> , 2021, 37, 4227-4229.	4.1	1
9	Response to letter entitled: Re: ERCC3 a new ovarian cancer susceptibility gene?. <i>European Journal of Cancer</i> , 2021, 150, 281-282.	2.8	0
10	Genetic Screening for TLR7 Variants in Young and Previously Healthy Men With Severe COVID-19. <i>Frontiers in Immunology</i> , 2021, 12, 719115.	4.8	76
11	Paired Somatic-Germline Testing of 15 Polyposis and Colorectal Cancer—Predisposing Genes Highlights the Role of APC Mosaicism in de Novo Familial Adenomatous Polyposis. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 1452-1459.	2.8	10
12	RNA assay identifies a previous misclassification of <i>BARD1</i> c.1977A>G variant. <i>Scientific Reports</i> , 2021, 11, 22948.	3.3	0
13	Comprehensive Constitutional Genetic and Epigenetic Characterization of Lynch-Like Individuals. <i>Cancers</i> , 2020, 12, 1799.	3.7	15
14	Screening of CNVs using NGS data improves mutation detection yield and decreases costs in genetic testing for hereditary cancer. <i>Journal of Medical Genetics</i> , 2020, , jmedgenet-2020-107366.	3.2	3
15	Role of <i>POLE</i> and <i>POLD1</i> in familial cancer. <i>Genetics in Medicine</i> , 2020, 22, 2089-2100.	2.4	76
16	ERCC3, a new ovarian cancer susceptibility gene?. <i>European Journal of Cancer</i> , 2020, 141, 1-8.	2.8	8
17	Comprehensive analysis and ACMG—based classification of <i>CHEK2</i> variants in hereditary cancer patients. <i>Human Mutation</i> , 2020, 41, 2128-2142.	2.5	10
18	Evaluation of CNV detection tools for NGS panel data in genetic diagnostics. <i>European Journal of Human Genetics</i> , 2020, 28, 1645-1655.	2.8	67

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19	The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. <i>Cancers</i> , 2020, 12, 292.	3.7	11
20	Exploring the Role of Mutations in Fanconi Anemia Genes in Hereditary Cancer Patients. <i>Cancers</i> , 2020, 12, 829.	3.7	41
21	Improving Genetic Testing in Hereditary Cancer by RNA Analysis. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1453-1468.	2.8	9
22	Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019, 40, 1557-1578.	2.5	102
23	Opportunistic testing of <i>BRCA1</i> , <i>BRCA2</i> and mismatch repair genes improves the yield of phenotype driven hereditary cancer gene panels. <i>International Journal of Cancer</i> , 2019, 145, 2682-2691.	5.1	30
24	Highly sensitive MLH1 methylation analysis in blood identifies a cancer patient with low-level mosaic MLH1 epimutation. <i>Clinical Epigenetics</i> , 2019, 11, 171.	4.1	7
25	Does multilocus inherited neoplasia alleles syndrome have severe clinical expression?. <i>Journal of Medical Genetics</i> , 2019, 56, 521-525.	3.2	11
26	Primary constitutional MLH1 epimutations: a focal epigenetic event. <i>British Journal of Cancer</i> , 2018, 119, 978-987.	6.4	22
27	Genetic Testing in Hereditary Colorectal Cancer. , 2018, , 209-232.		0
28	Elucidating the clinical significance of two PMS2 missense variants coexisting in a family fulfilling hereditary cancer criteria. <i>Familial Cancer</i> , 2017, 16, 501-507.	1.9	3
29	Elucidating the molecular basis of MSH2-deficient tumors by combined germline and somatic analysis. <i>International Journal of Cancer</i> , 2017, 141, 1365-1380.	5.1	26
30	Benchmarking of Whole Exome Sequencing and Ad Hoc Designed Panels for Genetic Testing of Hereditary Cancer. <i>Scientific Reports</i> , 2017, 7, 37984.	3.3	35
31	A comprehensive custom panel design for routine hereditary cancer testing: preserving control, improving diagnostics and revealing a complex variation landscape. <i>Scientific Reports</i> , 2017, 7, 39348.	3.3	45
32	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.	1.6	152
33	Identification of a founder <i>BRCA1</i> mutation in the Moroccan population. <i>Clinical Genetics</i> , 2016, 90, 361-365.	2.0	13
34	Investigating the effect of 28 <i>BRCA1</i> and <i>BRCA2</i> mutations on their related transcribed mRNA. <i>Breast Cancer Research and Treatment</i> , 2016, 155, 253-260.	2.5	6
35	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in <i>BRCA2</i> mutation carriers. <i>Breast Cancer Research</i> , 2015, 17, 61.	5.0	26
36	ICO Amplicon NGS Data Analysis: A Web Tool for Variant Detection in Common High-Risk Hereditary Cancer Genes Analyzed by Amplicon GS Junior Next-Generation Sequencing. <i>Human Mutation</i> , 2014, 35, 271-277.	2.5	2

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37	Identification of a founder EPCAM deletion in Spanish Lynch syndrome families. <i>Clinical Genetics</i> , 2014, 85, 260-266.	2.0	12
38	Refining the role of <i>pms2</i> in Lynch syndrome: germline mutational analysis improved by comprehensive assessment of variants. <i>Journal of Medical Genetics</i> , 2013, 50, 552-563.	3.2	47
39	Next-generation sequencing meets genetic diagnostics: development of a comprehensive workflow for the analysis of BRCA1 and BRCA2 genes. <i>European Journal of Human Genetics</i> , 2013, 21, 864-870.	2.8	94
40	Assessing the RNA effect of 26 DNA variants in the BRCA1 and BRCA2 genes. <i>Breast Cancer Research and Treatment</i> , 2012, 132, 979-992.	2.5	20
41	Analysis of SLX4/FANCP in non-BRCA1/2-mutated breast cancer families. <i>BMC Cancer</i> , 2012, 12, 84.	2.6	14
42	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2012, 14, R33.	5.0	78
43	Abstract 4445: Defining a pipeline to use next generation sequencing for genetic testing in hereditary cancer. , 2012, , .		0
44	Identification of a new complex rearrangement affecting exon 20 of BRCA1. <i>Breast Cancer Research and Treatment</i> , 2011, 130, 341-344.	2.5	3
45	Identification and comprehensive characterization of large genomic rearrangements in the BRCA1 and BRCA2 genes. <i>Breast Cancer Research and Treatment</i> , 2010, 122, 733-743.	2.5	34
46	Comments on: Sluiter MD and van Rensburg EJ, Large genomic rearrangements of the BRCA1 and BRCA2 genes: review of the literature and report of a novel BRCA1 mutation. <i>Breast Cancer Research and Treatment</i> , 2010, 124, 295-296.	2.5	1
47	Small supernumerary marker chromosome causing partial trisomy 6p in a child with craniosynostosis. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1108-1113.	1.2	15