# JesÃºs Jvd Del Valle

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250. | 1.6 | 152 |
| 2 | Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578. | 2.5 | 102 |
| 3 | Next-generation sequencing meets genetic diagnostics: development of a comprehensive workflow for the analysis of BRCA1 and BRCA2 genes. European Journal of Human Genetics, 2013, 21, 864-870. | 2.8 | 94 |
| 4 | Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2mutation carriers. Breast Cancer Research, 2012, 14, R33. | 5.0 | 78 |
| 5 | Role of POLE and POLD1 in familial cancer. Genetics in Medicine, 2020, 22, 2089-2100. | 2.4 | 76 |
| 6 | Genetic Screening for TLR7 Variants in Young and Previously Healthy Men With Severe COVID-19. Frontiers in Immunology, 2021, 12, 719115. | 4.8 | 76 |
| 7 | Evaluation of CNV detection tools for NGS panel data in genetic diagnostics. European Journal of Human Genetics, 2020, 28, 1645-1655. | 2.8 | 67 |
| 8 | Refining the role ofpms2in Lynch syndrome: germline mutational analysis improved by comprehensive assessment of variants. Journal of Medical Genetics, 2013, 50, 552-563. | 3.2 | 47 |
| 9 | A comprehensive custom panel design for routine hereditary cancer testing: preserving control, improving diagnostics and revealing a complex variation landscape. Scientific Reports, 2017, 7, 39348. | 3.3 | 45 |
| 10 | Exploring the Role of Mutations in Fanconi Anemia Genes in Hereditary Cancer Patients. Cancers, 2020, 12, 829. | 3.7 | 41 |
| 11 | Benchmarking of Whole Exome Sequencing and Ad Hoc Designed Panels for Genetic Testing of Hereditary Cancer. Scientific Reports, 2017, 7, 37984. | 3.3 | 35 |
| 12 | Identification and comprehensive characterization of large genomic rearrangements in the BRCA1 and BRCA2 genes. Breast Cancer Research and Treatment, 2010, 122, 733-743. | 2.5 | 34 |
| 13 | Opportunistic testing of BRCA1, BRCA2 and mismatch repair genes improves the yield of phenotype driven hereditary cancer gene panels. International Journal of Cancer, 2019, 145, 2682-2691. | 5.1 | 30 |
| 14 | An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. Breast Cancer Research, 2015, 17, 61. | 5.0 | 26 |
| 15 | Elucidating the molecular basis of MSH2â€deficient tumors by combined germline and somatic analysis. International Journal of Cancer, 2017, 141, 1365-1380. | 5.1 | 26 |
| 16 | Primary constitutional MLH1 epimutations: a focal epigenetic event. British Journal of Cancer, 2018, 119, 978-987. | 6.4 | 22 |
| 17 | Assessing the RNA effect of 26 DNA variants in the BRCA1 and BRCA2 genes. Breast Cancer Research and Treatment, 2012, 132, 979-992. | 2.5 | 20 |
| 18 | Characteristics of Adrenocortical Carcinoma Associated With Lynch Syndrome. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 318-325. | 3.6 | 20 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122. | 6.3 | 19 |
| 20 | Small supernumerary marker chromosome causing partial trisomy 6p in a child with craniosynostosis. American Journal of Medical Genetics, Part A, 2007, 143A, 1108-1113. | 1.2 | 15 |
| 21 | Comprehensive Constitutional Genetic and Epigenetic Characterization of Lynch-Like Individuals. Cancers, 2020, 12, 1799. | 3.7 | 15 |
| 22 | Analysis of SLX4/FANCP in non-BRCA1/2-mutated breast cancer families. BMC Cancer, 2012, 12, 84. | 2.6 | 14 |
| 23 | Identification of a founder <scp>BRCA1</scp> mutation in the Moroccan population. Clinical Genetics, 2016, 90, 361-365. | 2.0 | 13 |
| 24 | Identification of a founder EPCAM deletion in Spanish Lynch syndrome families. Clinical Genetics, 2014, 85, 260-266. | 2.0 | 12 |
| 25 | Does multilocus inherited neoplasia alleles syndrome have severe clinical expression?. Journal of Medical Genetics, 2019, 56, 521-525. | 3.2 | 11 |
| 26 | The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292. | 3.7 | 11 |
| 27 | BARD1 Pathogenic Variants Are Associated with Triple-Negative Breast Cancer in a Spanish Hereditary Breast and Ovarian Cancer Cohort. Genes, 2021, 12, 150. | 2.4 | 11 |
| 28 | Comprehensive analysis and ACMGâ€based classification of CHEK2 variants in hereditary cancer patients. Human Mutation, 2020, 41, 2128-2142. | 2.5 | 10 |
| 29 | Paired Somatic-Germline Testing of 15 Polyposis and Colorectal Cancerâ€“Predisposing Genes Highlights the Role of APC Mosaicism in de Novo Familial Adenomatous Polyposis. Journal of Molecular Diagnostics, 2021, 23, 1452-1459. | 2.8 | 10 |
| 30 | The Challenge of Diagnosing Constitutional Mismatch Repair Deficiency Syndrome in Brain Malignancies from Young Individuals. International Journal of Molecular Sciences, 2021, 22, 4629. | 4.1 | 9 |
| 31 | Improving Genetic Testing in Hereditary Cancer by RNA Analysis. Journal of Molecular Diagnostics, 2020, 22, 1453-1468. | 2.8 | 9 |
| 32 | ERCC3, a new ovarian cancer susceptibility gene?. European Journal of Cancer, 2020, 141, 1-8. | 2.8 | 8 |
| 33 | Highly sensitive MLH1 methylation analysis in blood identifies a cancer patient with low-level mosaic MLH1 epimutation. Clinical Epigenetics, 2019, 11, 171. | 4.1 | 7 |
| 34 | Investigating the effect of 28 BRCA1 and BRCA2 mutations on their related transcribed mRNA. Breast Cancer Research and Treatment, 2016, 155, 253-260. | 2.5 | 6 |
| 35 | Mosaicism in PTENâ€”new case and comment on the literature. European Journal of Human Genetics, 2022, 30, 641-644. | 2.8 | 6 |
| 36 | Variants of uncertain significance (VUS) in cancer predisposing genes: What are we learning from multigene panels?. European Journal of Medical Genetics, 2022, 65, 104400. | 1.3 | 4 |

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|---|---|---|---|
| 37 | Identification of a new complex rearrangement affecting exon 20 of BRCA1. Breast Cancer Research and Treatment, 2011, 130, 341-344. | 2.5 | 3 |
| 38 | Elucidating the clinical significance of two PMS2 missense variants coexisting in a family fulfilling hereditary cancer criteria. Familial Cancer, 2017, 16, 501-507. | 1.9 | 3 |
| 39 | Screening of CNVs using NGS data improves mutation detection yield and decreases costs in genetic testing for hereditary cancer. Journal of Medical Genetics, 2020, , jmedgenet-2020-107366. | 3.2 | 3 |
| 40 | 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. Human Mutation, 2014, 35, 271-277. | 2.5 | 2 |
| 41 | Solving the enigma of POLD1 p.V295M as a potential cause of increased cancer risk. European Journal of Human Genetics, 2022, 30, 485-489. | 2.8 | 2 |
| 42 | 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. Breast Cancer Research and Treatment, 2010, 124, 295-296. | 2.5 | 1 |
| 43 | CNVfilteR: an R/Bioconductor package to identify false positives produced by germline NGS CNV detection tools. Bioinformatics, 2021, 37, 4227-4229. | 4.1 | 1 |
| 44 | Genetic Testing in Hereditary Colorectal Cancer. , 2018, , 209-232. | | 0 |
| 45 | Response to letter entitled: Re: ERCC3 a new ovarian cancer susceptibility gene?. European Journal of Cancer, 2021, 150, 281-282. | 2.8 | 0 |
| 46 | Abstract 4445: Defining a pipeline to use next generation sequencing for genetic testing in hereditary cancer. , 2012, , . | | 0 |
| 47 | RNA assay identifies a previous misclassification of BARD1 c.1977A>G variant. Scientific Reports, 2021, 11, 22948. | 3.3 | 0 |