# Susana Seixas

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5762971/publications.pdf

Version: 2024-02-01

| | | | |
|---|---|---|---|
| 60 papers | 1,320 citations | 394390 19 h-index | 377849 34 g-index |
| 60 all docs | 60 docs citations | 60 times ranked | 2204 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Microsatellite variation and evolution of human lactase persistence. Human Genetics, 2005, 117, 329-339. | 3.8 | 112 |
| 2 | Allele-specific CDH1 downregulation and hereditary diffuse gastric cancer. Human Molecular Genetics, 2010, 19, 943-952. | 2.9 | 100 |
| 3 | Characterization of microbiota in male infertility cases uncovers differences in seminal hyperviscosity and oligoasthenoteratozoospermia possibly correlated with increased prevalence of infectious bacteria. American Journal of Reproductive Immunology, 2018, 79, e12838. | 1.2 | 70 |
| 4 | A model to infer the pathogenic significance of CDH1 germline missense variants. Journal of Molecular Medicine, 2006, 84, 1023-1031. | 3.9 | 66 |
| 5 | Profiling of lung microbiota discloses differences in adenocarcinoma and squamous cell carcinoma. Scientific Reports, 2019, 9, 12838. | 3.3 | 64 |
| 6 | Known Mutations at the Cause of Alpha-1 Antitrypsin Deficiency an Updated Overview of SERPINA1 Variation Spectrum. The Application of Clinical Genetics, 2021, Volume 14, 173-194. | 3.0 | 57 |
| 7 | Patterns of haplotype diversity within the serpin gene cluster at 14q32.1: insights into the natural history of the Î±1-antitrypsin polymorphism. Human Genetics, 2001, 108, 20-30. | 3.8 | 53 |
| 8 | Alpha-1-antitrypsin (SERPINA1) mutation spectrum: Three novel variants and haplotype characterization of rare deficiency alleles identified in Portugal. Respiratory Medicine, 2016, 116, 8-18. | 2.9 | 53 |
| 9 | The Peopling of Sao Tome (Gulf of Guinea): Origins of Slave Settlers and Admixture with the Portuguese. Human Biology, 2002, 74, 397-411. | 0.2 | 48 |
| 10 | Bronchoalveolar Lavage Proteomics in Patients with Suspected Lung Cancer. Scientific Reports, 2017, 7, 42190. | 3.3 | 46 |
| 11 | Common coding variant in SERPINA1 increases the risk for large artery stroke. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 3613-3618. | 7.1 | 46 |
| 12 | A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154. | 12.8 | 38 |
| 13 | Hereditary Gastric and Breast Cancer Syndromes Related to CDH1 Germline Mutation: A Multidisciplinary Clinical Review. Cancers, 2020, 12, 1598. | 3.7 | 37 |
| 14 | Î±1-Antitrypsin null alleles: evidence for the recurrence of the L353fsX376 mutation and a novel Gâ†'A transition in position +1 of intron IC affecting normal mRNA splicing. Clinical Genetics, 2002, 62, 175-180. | 2.0 | 29 |
| 15 | Infection-associated FUT2 (Fucosyltransferase 2) genetic variation and impact on functionality assessed by in vivo studies. Glycoconjugate Journal, 2010, 27, 61-68. | 2.7 | 29 |
| 16 | Portuguese consensus document for the management of alpha-1-antitrypsin deficiency. Pulmonology, 2018, 24, 1-21. | 2.1 | 29 |
| 17 | Variants in GCNA, X-linked germ-cell genome integrity gene, identified in men with primary spermatogenic failure. Human Genetics, 2021, 140, 1169-1182. | 3.8 | 27 |
| 18 | Lobar Brain Hemorrhages and White Matter Changes: Clinical, Radiological and Laboratorial Profiles. Cerebrovascular Diseases, 2006, 22, 155-161. | 1.7 | 26 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Severe Î±â€1 antitrypsin deficiency caused by Q0<sub>OurÃ©m</sub> allele: clinical features, haplotype characterization and history. Clinical Genetics, 2012, 81, 462-469. | 2.0 | 26 |
| 20 | Microsatellite Variation and Evolution of the Human Duffy Blood Group Polymorphism. Molecular Biology and Evolution, 2002, 19, 1802-1806. | 8.9 | 24 |
| 21 | Geographical Distribution of E-cadherin Germline Mutations in the Context of Diffuse Gastric Cancer: A Systematic Review. Cancers, 2021, 13, 1269. | 3.7 | 21 |
| 22 | Influence of Apolipoprotein E Polymorphism on Cardiovascular Risk Factors in Obese Children. Annals of Nutrition and Metabolism, 2003, 47, 49-54. | 1.9 | 20 |
| 23 | Sequence Diversity at the Proximal 14q32.1 SERPIN Subcluster: Evidence for Natural Selection Favoring the Pseudogenization of SERPINA2. Molecular Biology and Evolution, 2006, 24, 587-598. | 8.9 | 20 |
| 24 | Alpha-1 Antitrypsin Deficiency Detection in a Portuguese Population. COPD: Journal of Chronic Obstructive Pulmonary Disease, 2018, 15, 4-9. | 1.6 | 19 |
| 25 | Loss and Gain of Function in SERPINB11: An Example of a Gene under Selection on Standing Variation, with Implications for Host-Pathogen Interactions. PLoS ONE, 2012, 7, e32518. | 2.5 | 18 |
| 26 | Evolutionary Constraints in the Î²-Globin Cluster: The Signature of Purifying Selection at the Î´-Globin (HBD) Locus and Its Role in Developmental Gene Regulation. Genome Biology and Evolution, 2013, 5, 559-571. | 2.5 | 18 |
| 27 | Genes from the TAS1R and TAS2R Families of Taste Receptors: Looking for Signatures of Their Adaptive Role in Human Evolution. Genome Biology and Evolution, 2018, 10, 1139-1152. | 2.5 | 18 |
| 28 | Reproduction and Immunity-Driven Natural Selection in the Human WFDC Locus. Molecular Biology and Evolution, 2013, 30, 938-950. | 8.9 | 17 |
| 29 | Rare double sex and mab-3-related transcription factor 1 regulatory variants in severe spermatogenic failure. Andrology, 2015, 3, 825-833. | 3.5 | 17 |
| 30 | Is the Proteome of Bronchoalveolar Lavage Extracellular Vesicles a Marker of Advanced Lung Cancer?. Cancers, 2020, 12, 3450. | 3.7 | 14 |
| 31 | Effect and in silico characterization of genetic variants associated with severe spermatogenic disorders in a large Iberian cohort. Andrology, 2021, 9, 1151-1165. | 3.5 | 12 |
| 32 | A novel alpha-1-antitrypsin R281del variant found in a population sample from the Basque country. Human Mutation, 2000, 15, 121-122. | 2.5 | 11 |
| 33 | Birth-and-Death of KLK3 and KLK2 in Primates: Evolution Driven by Reproductive Biology. Genome Biology and Evolution, 2012, 4, 1331-1338. | 2.5 | 11 |
| 34 | The Mutational Spectrum of WT1 in Male Infertility. Journal of Urology, 2015, 193, 1709-1715. | 0.4 | 11 |
| 35 | Sequence variation atKLKandWFDCclusters and its association to semen hyperviscosity and other male infertility phenotypes. Human Reproduction, 2016, 31, 2881-2891. | 0.9 | 11 |
| 36 | E-cadherin signal sequence disruption: a novel mechanism underlying hereditary cancer. Molecular Cancer, 2018, 17, 112. | 19.2 | 11 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Adaptive Evolution and Divergence of SERPINB3: A Young Duplicate in Great Apes. PLoS ONE, 2014, 9, e104935. | 2.5 | 10 |
| 38 | Evaluation of Male Fertility-Associated Loci in a European Population of Patients with Severe Spermatogenic Impairment. Journal of Personalized Medicine, 2021, 11, 22. | 2.5 | 10 |
| 39 | Intronic variation of the SOHLH2 gene confers risk to male reproductive impairment. Fertility and Sterility, 2020, 114, 398-406. | 1.0 | 9 |
| 40 | Differing Evolutionary Histories of WFDC8 (Short-Term Balancing) in Europeans and SPINT4 (Incomplete Selective Sweep) in Africans. Molecular Biology and Evolution, 2011, 28, 2811-2822. | 8.9 | 7 |
| 41 | The CDH1 c.1901C>T Variant: A Founder Variant in the Portuguese Population with Severe Impact in mRNA Splicing. Cancers, 2021, 13, 4464. | 3.7 | 7 |
| 42 | Distinctive Patterns of Evolution of the Î´-Globin Gene (HBD) in Primates. PLoS ONE, 2015, 10, e0123365. | 2.5 | 7 |
| 43 | SERPINA2 Is a Novel Gene with a Divergent Function from SERPINA1. PLoS ONE, 2013, 8, e66889. | 2.5 | 6 |
| 44 | Semen quality is affected by HLA class I alleles together with sexually transmitted diseases. Andrology, 2019, 7, 867-877. | 3.5 | 6 |
| 45 | Adaptive Evolution FavoringKLK4Downregulation in East Asians. Molecular Biology and Evolution, 2016, 33, 93-108. | 8.9 | 5 |
| 46 | Comparative analysis of the bronchoalveolar microbiome in Portuguese patients with different chronic lung disorders. Scientific Reports, 2021, 11, 15042. | 3.3 | 5 |
| 47 | Common genetic variation in KATNAL1 nonâ€coding regions is involved in the susceptibility to severe phenotypes of male infertility. Andrology, 2022, 10, 1339-1350. | 3.5 | 5 |
| 48 | A novel alpha-1-antitrypsin P362H variant found in a population sample from Sï¿½o Tomï¿½ e Prï¿½ncipe (Gulf of) Tj ETQq0 0 0 rgBT /O | 2.5 | 4 |
| 49 | Genetic Variation in a Compound Short Tandem Repeat/ Alu Haplotype System at the SB19.3 Locus: Properties and Interpretation. Human Biology, 2004, 76, 277-287. | 0.2 | 2 |
| 50 | Alpha-1 antitrypsin deficiency caused by a novel mutation (p.Leu263Pro): Pi*ZQ0gaia â€“ Q0gaia allele. Revista Portuguesa De Pneumologia, 2015, 21, 341-343. | 0.7 | 2 |
| 51 | GBA3: a polymorphic pseudogene in humans that experienced repeated gene loss during mammalian evolution. Scientific Reports, 2020, 10, 11565. | 3.3 | 2 |
| 52 | Pulmonary Emphysema in a Child With Alpha-1 Antitrypsin Deficiency: Evaluation of 2 Years of Intravenous Augmentation Therapy. Archivos De Bronconeumologia, 2019, 55, 502-504. | 0.8 | 2 |
| 53 | Sequence Diversity of Pan troglodytes Subspecies and the Impact of WFDC6 Selective Constraints in Reproductive Immunity. Genome Biology and Evolution, 2013, 5, 2512-2523. | 2.5 | 1 |
| 54 | A rare case of pulmonary disease combining alpha-1-antitrypsin deficiency and common variable immunodeficiency. Pulmonology, 2020, 26, 406-409. | 2.1 | 1 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | The Human SERPIN Repertoire and the Evolution of 14q32.1 and 18q21.3 Gene Clusters. , 2015, , 1-14. | | 0 |
| 56 | Pulmonary Emphysema in a Child With Alpha-1 Antitrypsin Deficiency: Evaluation of 2 Years of Intravenous Augmentation Therapy. Archivos De Bronconeumologia, 2019, 55, 502-504. | 0.8 | 0 |
| 57 | Detection of alpha1-antitrypsin deficiency in a European country without national registry yet. , 2016, , . | | 0 |
| 58 | Microbiota profile of Non-small Cell Lung Cancer (NSCLC): the study of a large cohort. , 2019, , . | | 0 |
| 59 | Unravelling the genetic landscape of early-onset emphysema by whole exome sequencing. , 2020, , . | | 0 |
| 60 | Common Variation in the PIN1 Locus Increases the Genetic Risk to Suffer from Sertoli Cell-Only Syndrome. Journal of Personalized Medicine, 2022, 12, 932. | 2.5 | 0 |