# Bianca Tesi

## List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/6344873/publications.pdf

Version: 2024-02-01

| 43 | 1,967 | 394421 19 | 265206 42 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 45 | 45 | 45 | 3847 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Rubella vaccineâ€“induced granulomas are a novel phenotype with incomplete penetrance of genetic defects in cytotoxicity. Journal of Allergy and Clinical Immunology, 2022, 149, 388-399.e4. | 2.9 | 11 |
| 2 | Detection of germline mosaicism in fathers of children with intellectual disability syndromes caused by de novo variants. Molecular Genetics & Genomic Medicine, 2022, 10, e1880. | 1.2 | 10 |
| 3 | Childhood Kaposi sarcoma related to hypomorphic severe combined immunodeficiency caused by a novel CORO1A mutation. Pediatric Blood and Cancer, 2022, 69, e29487. | 1.5 | 0 |
| 4 | Patients with both Langerhans cell histiocytosis and Crohnâ€™s disease highlight a common role of interleukinâ€23. Acta Paediatrica, International Journal of Paediatrics, 2021, 110, 1315-1321. | 1.5 | 8 |
| 5 | Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40. | 8.2 | 116 |
| 6 | Clinical and laboratory signs of haemophagocytic lymphohistiocytosis associated with pandemic influenza A (H1N1) infection in patients needing extracorporeal membrane oxygenation. European Journal of Anaesthesiology, 2021, 38, 692-701. | 1.7 | 1 |
| 7 | Molecular Genetics Diversity of Primary Hemophagocytic Lymphohistiocytosis among Polish Pediatric Patients. Archivum Immunologiae Et Therapiae Experimentalis, 2021, 69, 31. | 2.3 | 4 |
| 8 | Adult-Onset Ataxia With Neuropathy and White Matter Abnormalities Due to a Novel SAMD9L Variant. Neurology: Genetics, 2021, 7, e628. | 1.9 | 1 |
| 9 | Whole Transcriptome Analysis Identifies Distinct Gene Expression Profiles between SF3B1mut and SF3B1 wt Myelodysplastic Syndrome with Ring Sideroblasts. Blood, 2021, 138, 3695-3695. | 1.4 | 0 |
| 10 | Multinational Study on the Clinical and Genetic Features of the ERCC6L2-Disease. Blood, 2021, 138, 864-864. | 1.4 | 2 |
| 11 | Alternative UNC13D Promoter Encodes a Functional Munc13-4 Isoform Predominantly Expressed in Lymphocytes and Platelets. Frontiers in Immunology, 2020, 11, 1154. | 4.8 | 2 |
| 12 | Diagnostic challenges for a novel SH2D1A mutation associated with Xâ€linked lymphoproliferative disease. Pediatric Blood and Cancer, 2020, 67, e28184. | 1.5 | 4 |
| 13 | Heterozygous variants in DCC. Neurology: Genetics, 2020, 6, e526. | 1.9 | 4 |
| 14 | Microdeletion of 7p12.1p13, including <scp>IKZF</scp>1, causes intellectual impairment, overgrowth, and susceptibility to leukaemia. British Journal of Haematology, 2019, 185, 354-357. | 2.5 | 4 |
| 15 | Human DEF6 deficiency underlies an immunodeficiency syndrome with systemic autoimmunity and aberrant CTLA-4 homeostasis. Nature Communications, 2019, 10, 3106. | 12.8 | 48 |
| 16 | Early activating somatic PIK3CA mutations promote ectopic muscle development and upper limb overgrowth. Clinical Genetics, 2019, 96, 118-125. | 2.0 | 14 |
| 17 | Haploinsufficiency of UNC13D increases the risk of lymphoma. Cancer, 2019, 125, 1848-1854. | 4.1 | 8 |
| 18 | Nordic Guidelines for Germline Predisposition to Myeloid Neoplasms in Adults: Recommendations for Genetic Diagnosis, Clinical Management and Follow-up. HemaSphere, 2019, 3, e321. | 2.7 | 51 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Fatal Central Nervous System Lymphocytic Vasculitis after Treatment for Burkitt Lymphoma in a Patient with a SH2D1A Mutation. Pediatric Infectious Disease Journal, 2019, 38, e29-e31. | 2.0 | 4 |
| 20 | Elevated ferritin and soluble CD25 in critically ill patients are associated with parameters of (hyper) inflammation and lymphocyte cytotoxicity. Minerva Anestesiologica, 2019, 85, 1289-1298. | 1.0 | 13 |
| 21 | A RAB27A 5â€² untranslated region structural variant associated with late-onset hemophagocytic lymphohistiocytosis and normal pigmentation. Journal of Allergy and Clinical Immunology, 2018, 142, 317-321.e8. | 2.9 | 22 |
| 22 | HLH: genomics illuminates pathophysiological diversity. Blood, 2018, 132, 5-7. | 1.4 | 11 |
| 23 | ARID5B regulates metabolic programming in human adaptive NK cells. Journal of Experimental Medicine, 2018, 215, 2379-2395. | 8.5 | 98 |
| 24 | Targeting SAMHD1 with the Vpx protein to improve cytarabine therapy for hematological malignancies. Nature Medicine, 2017, 23, 256-263. | 30.7 | 102 |
| 25 | Gain-of-function SAMD9L mutations cause a syndrome of cytopenia, immunodeficiency, MDS, and neurological symptoms. Blood, 2017, 129, 2266-2279. | 1.4 | 152 |
| 26 | A Case of Familial Hemophagocytic Lymphohistiocytosis Type 4 With Involvement of the Central Nervous System Complicated With Infarct. Journal of Pediatric Hematology/Oncology, 2017, 39, e321-e324. | 0.6 | 4 |
| 27 | Differences in Granule Morphology yet Equally Impaired Exocytosis among Cytotoxic T Cells and NK Cells from Chediakâ€“Higashi Syndrome Patients. Frontiers in Immunology, 2017, 8, 426. | 4.8 | 26 |
| 28 | Unperturbed Cytotoxic Lymphocyte Phenotype and Function in Myalgic Encephalomyelitis/Chronic Fatigue Syndrome Patients. Frontiers in Immunology, 2017, 8, 723. | 4.8 | 24 |
| 29 | Epigenetic Regulation of Adaptive NK Cell Diversification. Trends in Immunology, 2016, 37, 451-461. | 6.8 | 60 |
| 30 | Successful Hematopoietic Stem Cell Transplantation in a Patient with LPS-Responsive Beige-Like Anchor (LRBA) Gene Mutation. Journal of Clinical Immunology, 2016, 36, 480-489. | 3.8 | 30 |
| 31 | Cytopenia, Predisposition to Myelodysplastic Syndrome, Immunodeficiency, and Neurological Disease Caused By Gain-of-Function SAMD9L Mutations Is Frequently Ameliorated By Hematopoietic Revertant Mosaicism. Blood, 2016, 128, 4299-4299. | 1.4 | 1 |
| 32 | A Hemophagocytic Lymphohistiocytosis Case with Newly Defined UNC13D (c.175G>C; p.Ala59Pro) Mutation and a Rare Complication. Turkish Journal of Haematology, 2016, 32, 355-358. | 0.5 | 4 |
| 33 | Targeted high-throughput sequencing for genetic diagnostics of hemophagocytic lymphohistiocytosis. Genome Medicine, 2015, 7, 130. | 8.2 | 37 |
| 34 | The syndrome of hemophagocytic lymphohistiocytosis in primary immunodeficiencies: implications for differential diagnosis and pathogenesis. Haematologica, 2015, 100, 978-988. | 3.5 | 161 |
| 35 | Spectrum of Atypical Clinical Presentations in Patients with Biallelic PRF1 Missense Mutations. Pediatric Blood and Cancer, 2015, 62, 2094-2100. | 1.5 | 38 |
| 36 | Hemophagocytic lymphohistiocytosis in 2 patients with underlying IFN-Î³ receptor deficiency. Journal of Allergy and Clinical Immunology, 2015, 135, 1638-1641.e5. | 2.9 | 69 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Cytomegalovirus Infection Drives Adaptive Epigenetic Diversification of NK Cells with Altered Signaling and Effector Function. Immunity, 2015, 42, 443-456. | 14.3 | 650 |
| 38 | Diversification and Functional Specialization of Human NK Cell Subsets. Current Topics in Microbiology and Immunology, 2015, 395, 63-93. | 1.1 | 56 |
| 39 | An N-Terminal Missense Mutation in STX11 Causative of FHL4 Abrogates Syntaxin-11 Binding to Munc18-2. Frontiers in Immunology, 2014, 4, 515. | 4.8 | 20 |
| 40 | Pathophysiology and spectrum of diseases caused by defects in lymphocyte cytotoxicity. Experimental Cell Research, 2014, 325, 10-17. | 2.6 | 38 |
| 41 | Immunomodulatory activity of commonly used drugs on Fc-receptor-mediated human natural killer cell activation. Cancer Immunology, Immunotherapy, 2014, 63, 627-641. | 4.2 | 33 |
| 42 | Novel STAT3 Mutation Causing Hyper-IgE Syndrome: Studies of the Clinical Course and Immunopathology. Journal of Clinical Immunology, 2014, 34, 469-477. | 3.8 | 11 |
| 43 | An Indian boy with griscelli syndrome type 2: Case report and review of literature. Indian Journal of Dermatology, 2014, 59, 394. | 0.3 | 9 |