

Joris M Van Montfrans

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/844069/publications.pdf>

Version: 2024-02-01

40
papers

1,752
citations

394421

19
h-index

302126

39
g-index

41
all docs

41
docs citations

41
times ranked

3186
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|---|-----|-----------|
| 1 | The European Society for Immunodeficiencies (ESID) Registry Working Definitions for the Clinical Diagnosis of Inborn Errors of Immunity. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 1763-1770. | 3.8 | 381 |
| 2 | Targeted next-generation sequencing: A novel diagnostic tool for primary immunodeficiencies. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 529-534.e1. | 2.9 | 143 |
| 3 | TREC Based Newborn Screening for Severe Combined Immunodeficiency Disease: A Systematic Review. <i>Journal of Clinical Immunology</i> , 2015, 35, 416-430. | 3.8 | 140 |
| 4 | Phenotypic variability in patients with ADA2 deficiency due to identical homozygous R169Q mutations. <i>Rheumatology</i> , 2016, 55, 902-910. | 1.9 | 116 |
| 5 | Long-term outcome of LRBA deficiency in 76 patients after various treatment modalities as evaluated by the immune deficiency and dysregulation activity (IDDA) score. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 145, 1452-1463. | 2.9 | 112 |
| 6 | Long-term outcomes of 176 patients with X-linked hyper-IgM syndrome treated with or without hematopoietic cell transplantation. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 1282-1292. | 2.9 | 107 |
| 7 | Differential Signalling and Kinetics of Neutrophil Extracellular Trap Release Revealed by Quantitative Live Imaging. <i>Scientific Reports</i> , 2017, 7, 6529. | 3.3 | 80 |
| 8 | Hematopoietic Stem Cell Transplantation as Treatment for Patients with DOCK8 Deficiency. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 848-855. | 3.8 | 67 |
| 9 | Extended clinical and immunological phenotype and transplant outcome in CD27 and CD70 deficiency. <i>Blood</i> , 2020, 136, 2638-2655. | 1.4 | 64 |
| 10 | Diagnostic Yield of Next Generation Sequencing in Genetically Undiagnosed Patients with Primary Immunodeficiencies: a Systematic Review. <i>Journal of Clinical Immunology</i> , 2019, 39, 577-591. | 3.8 | 58 |
| 11 | Exhaustion of the CD8+ T Cell Compartment in Patients with Mutations in Phosphoinositide 3-Kinase Delta. <i>Frontiers in Immunology</i> , 2018, 9, 446. | 4.8 | 52 |
| 12 | Hematopoietic Cell Transplantation Cures Adenosine Deaminase 2 Deficiency: Report on 30 Patients. <i>Journal of Clinical Immunology</i> , 2021, 41, 1633-1647. | 3.8 | 43 |
| 13 | B-cell development and functions and therapeutic options in adenosine deaminase-deficient patients. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 799-806.e10. | 2.9 | 30 |
| 14 | A novel human STAT3 mutation presents with autoimmunity involving Th17 hyperactivation. <i>Oncotarget</i> , 2015, 6, 20037-20042. | 1.8 | 30 |
| 15 | Fatal hemophagocytic lymphohistiocytosis in X-linked chronic granulomatous disease associated with a perforin gene variant. <i>Pediatric Blood and Cancer</i> , 2009, 52, 527-529. | 1.5 | 26 |
| 16 | Cost and impact of early diagnosis in primary immunodeficiency disease: A literature review. <i>Clinical Immunology</i> , 2020, 213, 108359. | 3.2 | 25 |
| 17 | Parents' Perspectives and Societal Acceptance of Implementation of Newborn Screening for SCID in the Netherlands. <i>Journal of Clinical Immunology</i> , 2021, 41, 99-108. | 3.8 | 25 |
| 18 | Embracing Complexity beyond Systems Medicine: A New Approach to Chronic Immune Disorders. <i>Frontiers in Immunology</i> , 2016, 7, 587. | 4.8 | 24 |

| # | ARTICLE | IF | CITATIONS |
|----|---|-----|-----------|
| 19 | Measurement of Pneumococcal Polysaccharide Vaccine Responses for Immunodeficiency Diagnostics: Combined IgG Responses Compared to Serotype Specific IgG Responses. <i>Journal of Clinical Immunology</i> , 2014, 34, 3-6. | 3.8 | 21 |
| 20 | Dysfunctional BLK in common variable immunodeficiency perturbs B-cell proliferation and ability to elicit antigen-specific CD4+ T-cell help. <i>Oncotarget</i> , 2015, 6, 10759-10771. | 1.8 | 20 |
| 21 | Proline-serine-threonine phosphatase interacting protein 1 (PSTPIP1) controls immune synapse stability in human T cells. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 1947-1955. | 2.9 | 17 |
| 22 | Viral PCR Positivity in Stool before Allogeneic Hematopoietic Cell Transplantation Is Strongly Associated with Acute Intestinal Graft-versus-Host Disease. <i>Biology of Blood and Marrow Transplantation</i> , 2015, 21, 772-774. | 2.0 | 16 |
| 23 | Overview of 15-year severe combined immunodeficiency in the Netherlands: towards newborn blood spot screening. <i>European Journal of Pediatrics</i> , 2015, 174, 1183-1188. | 2.7 | 16 |
| 24 | IgG trough levels and progression of pulmonary disease in pediatric and adult common variable immunodeficiency disorder patients. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 303-306.e4. | 2.9 | 16 |
| 25 | A novel Fcγ3RIIa Q27W gene variant is associated with common variable immune deficiency through defective Fcγ3RIIa downstream signaling. <i>Clinical Immunology</i> , 2014, 155, 108-117. | 3.2 | 15 |
| 26 | Internet and smartphone-based ecological momentary assessment and personalized advice (PROfeel) in adolescents with chronic conditions: A feasibility study. <i>Internet Interventions</i> , 2021, 25, 100395. | 2.7 | 12 |
| 27 | Implementation of Early Next-Generation Sequencing for Inborn Errors of Immunity: A Prospective Observational Cohort Study of Diagnostic Yield and Clinical Implications in Dutch Genome Diagnostic Centers. <i>Frontiers in Immunology</i> , 2021, 12, 780134. | 4.8 | 12 |
| 28 | Managing Granulomatous Lymphocytic Interstitial Lung Disease in Common Variable Immunodeficiency Disorders: e-GLILDnet International Clinicians Survey. <i>Frontiers in Immunology</i> , 2020, 11, 606333. | 4.8 | 10 |
| 29 | Severe Fatigue Is Common Among Pediatric Patients with Primary Immunodeficiency and Is Not Related to Disease Activity. <i>Journal of Clinical Immunology</i> , 2021, 41, 1198-1207. | 3.8 | 10 |
| 30 | The Role of Prolonged Viral Gastrointestinal Infections in the Development of Immunodeficiency-Related Enteropathy. <i>Clinical Reviews in Allergy and Immunology</i> , 2012, 42, 79-91. | 6.5 | 9 |
| 31 | Adenosine Deaminase 2 Deficiency As a Cause of Pure Red Cell Aplasia Mimicking Diamond Blackfan Anemia. <i>Blood</i> , 2015, 126, 3615-3615. | 1.4 | 9 |
| 32 | Antigen-specific IgA titres after 23-valent pneumococcal vaccine indicate transient antibody deficiency disease in children. <i>Vaccine</i> , 2015, 33, 6320-6326. | 3.8 | 8 |
| 33 | A Minimal Parameter Set Facilitating Early Decision-making in the Diagnosis of Hemophagocytic Lymphohistiocytosis. <i>Journal of Clinical Immunology</i> , 2021, 41, 1219-1228. | 3.8 | 8 |
| 34 | Immunoglobulin Replacement Therapy Versus Antibiotic Prophylaxis as Treatment for Incomplete Primary Antibody Deficiency. <i>Journal of Clinical Immunology</i> , 2021, 41, 382-392. | 3.8 | 7 |
| 35 | Phenotypic variability including Behçet's disease-like manifestations in DADA2 patients due to a homozygous c.973-2A>G splice site mutation. <i>Clinical and Experimental Rheumatology</i> , 2019, 37 Suppl 121, 142-146. | 0.8 | 7 |
| 36 | Abnormal Results of Newborn Screening for SCID After Azathioprine Exposure In Utero: Benefit of TPMT Genotyping in Both Mother and Child. <i>Journal of Clinical Immunology</i> , 2022, 42, 199-202. | 3.8 | 6 |

| # | ARTICLE | IF | CITATIONS |
|----|--|-----|-----------|
| 37 | National external quality assessment for next-generation sequencing-based diagnostics of primary immunodeficiencies. <i>European Journal of Human Genetics</i> , 2021, 29, 20-28. | 2.8 | 5 |
| 38 | New insights in phenotype and treatment of lung disease immuno-deficiency and chromosome breakage syndrome (LICS). <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 137. | 2.7 | 3 |
| 39 | Wiskott-Aldrich Syndrome: A Retrospective Study on 575 Patients Analyzing the Impact of Splenectomy, Stem Cell Transplantation, or No Definitive Treatment on Frequency of Disease-Related Complications and Physician-Perceived Quality of Life. <i>Blood</i> , 2016, 128, 366-366. | 1.4 | 2 |
| 40 | Synergy in B-Cell Activation between Toll-Like Receptor 9 and Transmembrane Activator and Calcium-Modulating Cyclophilin Ligand Interactor (TACI) in A181E/C104R Compound Heterozygous Siblings. <i>ISRN Immunology</i> , 2013, 2013, 1-10. | 0.7 | 0 |