# Joris M Van Montfrans

List of Publications by Year
in descending order



| 40 | 1,752 | 394421 19 | 302126 39 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 41 | 41 | 41 | 3186 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Abnormal Results of Newborn Screening for SCID After Azathioprine Exposure In Utero: Benefit of TPMT Genotyping in Both Mother and Child. Journal of Clinical Immunology, 2022, 42, 199-202. | 3.8 | 6 |
| 2 | National external quality assessment for next-generation sequencing-based diagnostics of primary immunodeficiencies. European Journal of Human Genetics, 2021, 29, 20-28. | 2.8 | 5 |
| 3 | Immunoglobulin Replacement Therapy Versus Antibiotic Prophylaxis as Treatment for Incomplete Primary Antibody Deficiency. Journal of Clinical Immunology, 2021, 41, 382-392. | 3.8 | 7 |
| 4 | Parentsâ€™ Perspectives and Societal Acceptance of Implementation of Newborn Screening for SCID in the Netherlands. Journal of Clinical Immunology, 2021, 41, 99-108. | 3.8 | 25 |
| 5 | A Minimal Parameter Set Facilitating Early Decision-making in the Diagnosis of Hemophagocytic Lymphohistiocytosis. Journal of Clinical Immunology, 2021, 41, 1219-1228. | 3.8 | 8 |
| 6 | Severe Fatigue Is Common Among Pediatric Patients with Primary Immunodeficiency and Is Not Related to Disease Activity. Journal of Clinical Immunology, 2021, 41, 1198-1207. | 3.8 | 10 |
| 7 | New insights in phenotype and treatment of lung disease immuno-deficiency and chromosome breakage syndrome (LICS). Orphanet Journal of Rare Diseases, 2021, 16, 137. | 2.7 | 3 |
| 8 | Hematopoietic Cell Transplantation Cures Adenosine Deaminase 2 Deficiency: Report on 30 Patients. Journal of Clinical Immunology, 2021, 41, 1633-1647. | 3.8 | 43 |
| 9 | Internet and smartphone-based ecological momentary assessment and personalized advice (PROfeel) in adolescents with chronic conditions: A feasibility study. Internet Interventions, 2021, 25, 100395. | 2.7 | 12 |
| 10 | 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. Frontiers in Immunology, 2021, 12, 780134. | 4.8 | 12 |
| 11 | Long-term outcome of LRBA deficiency in 76 patients after various treatment modalities as evaluated by the immune deficiency and dysregulation activity (IDDA) score. Journal of Allergy and Clinical Immunology, 2020, 145, 1452-1463. | 2.9 | 112 |
| 12 | Managing Granulomatousâ€“Lymphocytic Interstitial Lung Disease in Common Variable Immunodeficiency Disorders: e-GLILDnet International Clinicians Survey. Frontiers in Immunology, 2020, 11, 606333. | 4.8 | 10 |
| 13 | Extended clinical and immunological phenotype and transplant outcome in CD27 and CD70 deficiency. Blood, 2020, 136, 2638-2655. | 1.4 | 64 |
| 14 | Cost and impact of early diagnosis in primary immunodeficiency disease: A literature review. Clinical Immunology, 2020, 213, 108359. | 3.2 | 25 |
| 15 | Diagnostic Yield of Next Generation Sequencing in Genetically Undiagnosed Patients with Primary Immunodeficiencies: a Systematic Review. Journal of Clinical Immunology, 2019, 39, 577-591. | 3.8 | 58 |
| 16 | The European Society for Immunodeficiencies (ESID) Registry Working Definitions for the ClinicalÂDiagnosis of Inborn Errors of Immunity. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 1763-1770. | 3.8 | 381 |
| 17 | Hematopoietic Stem Cell Transplantation as Treatment for Patients with DOCK8 Deficiency. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 848-855. | 3.8 | 67 |
| 18 | Phenotypic variability including BehÃ§et's disease-like manifestations in DADA2 patients due to a homozygous c.973-2A>G splice site mutation. Clinical and Experimental Rheumatology, 2019, 37 Suppl 121, 142-146. | 0.8 | 7 |

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|---|---|---|---|
| 19 | Proline-serine-threonine phosphatase interacting protein 1 (PSTPIP1) controls immune synapse stability in human T cells. Journal of Allergy and Clinical Immunology, 2018, 142, 1947-1955. | 2.9 | 17 |
| 20 | Exhaustion of the CD8+ T Cell Compartment in Patients with Mutations in Phosphoinositide 3-Kinase Delta. Frontiers in Immunology, 2018, 9, 446. | 4.8 | 52 |
| 21 | IgG trough levels and progression of pulmonary disease in pediatric and adult common variable immunodeficiency disorder patients. Journal of Allergy and Clinical Immunology, 2017, 140, 303-306.e4. | 2.9 | 16 |
| 22 | Differential Signalling and Kinetics of Neutrophil Extracellular Trap Release Revealed by Quantitative Live Imaging. Scientific Reports, 2017, 7, 6529. | 3.3 | 80 |
| 23 | Long-term outcomes of 176 patients with X-linked hyper-IgM syndrome treated with or without hematopoietic cell transplantation. Journal of Allergy and Clinical Immunology, 2017, 139, 1282-1292. | 2.9 | 107 |
| 24 | Embracing Complexity beyond Systems Medicine: A New Approach to Chronic Immune Disorders. Frontiers in Immunology, 2016, 7, 587. | 4.8 | 24 |
| 25 | Phenotypic variability in patients with ADA2 deficiency due to identical homozygous R169Q mutations. Rheumatology, 2016, 55, 902-910. | 1.9 | 116 |
| 26 | 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. Blood, 2016, 128, 366-366. | 1.4 | 2 |
| 27 | A novel human STAT3 mutation presents with autoimmunity involving Th17 hyperactivation. Oncotarget, 2015, 6, 20037-20042. | 1.8 | 30 |
| 28 | Antigen-specific IgA titres after 23-valent pneumococcal vaccine indicate transient antibody deficiency disease in children. Vaccine, 2015, 33, 6320-6326. | 3.8 | 8 |
| 29 | Viral PCR Positivity in Stool before Allogeneic Hematopoietic Cell Transplantation Is Strongly Associated with Acute Intestinal Graft-versus-Host Disease. Biology of Blood and Marrow Transplantation, 2015, 21, 772-774. | 2.0 | 16 |
| 30 | Overview of 15-year severe combined immunodeficiency in the Netherlands: towards newborn blood spot screening. European Journal of Pediatrics, 2015, 174, 1183-1188. | 2.7 | 16 |
| 31 | TREC Based Newborn Screening for Severe Combined Immunodeficiency Disease: A Systematic Review. Journal of Clinical Immunology, 2015, 35, 416-430. | 3.8 | 140 |
| 32 | Adenosine Deaminase 2 Deficiency As a Cause of Pure Red Cell Aplasia Mimicking Diamond Blackfan Anemia. Blood, 2015, 126, 3615-3615. | 1.4 | 9 |
| 33 | Dysfunctional BLK in common variable immunodeficiency perturbs B-cell proliferation and ability to elicit antigen-specific CD4+ T-cell help. Oncotarget, 2015, 6, 10759-10771. | 1.8 | 20 |
| 34 | Measurement of Pneumococcal Polysaccharide Vaccine Responses for Immunodeficiency Diagnostics: Combined IgG Responses Compared to Serotype Specific IgG Responses. Journal of Clinical Immunology, 2014, 34, 3-6. | 3.8 | 21 |
| 35 | B-cell development and functions and therapeutic options in adenosine deaminaseâ€“deficient patients. Journal of Allergy and Clinical Immunology, 2014, 133, 799-806.e10. | 2.9 | 30 |
| 36 | Targeted next-generation sequencing: AÂnovel diagnostic tool for primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2014, 133, 529-534.e1. | 2.9 | 143 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | A novel FcÎ³RIIa Q27W gene variant is associated with common variable immune deficiency through defective FcÎ³RIIa downstream signaling. Clinical Immunology, 2014, 155, 108-117. | 3.2 | 15 |
| 38 | 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. ISRN Immunology, 2013, 2013, 1-10. | 0.7 | 0 |
| 39 | The Role of Prolonged Viral Gastrointestinal Infections in the Development of Immunodeficiency-Related Enteropathy. Clinical Reviews in Allergy and Immunology, 2012, 42, 79-91. | 6.5 | 9 |
| 40 | Fatal hemophagocytic lymphohistiocytosis in Xâ€linked chronic granulomatous disease associated with a perforin gene variant. Pediatric Blood and Cancer, 2009, 52, 527-529. | 1.5 | 26 |