

# Juan Ignacio Arostegui

## List of Publications by Citations

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112  
papers

5,612  
citations

38  
h-index

74  
g-index

130  
ext. papers

6,738  
ext. citations

4.9  
avg, IF

4.84  
L-index

#	Paper	IF	Citations
112	Pyogenic bacterial infections in humans with MyD88 deficiency. <i>Science</i> , <b>2008</b> , 321, 691-6	33.3	608
111	The NLRP3 inflammasome is released as a particulate danger signal that amplifies the inflammatory response. <i>Nature Immunology</i> , <b>2014</b> , 15, 738-48	19.1	522
110	B cell-helper neutrophils stimulate the diversification and production of immunoglobulin in the marginal zone of the spleen. <i>Nature Immunology</i> , <b>2011</b> , 13, 170-80	19.1	501
109	Clinical features and outcome of patients with IRAK-4 and MyD88 deficiency. <i>Medicine (United States)</i> , <b>2010</b> , 89, 403-425	1.8	297
108	Innate lymphoid cells integrate stromal and immunological signals to enhance antibody production by splenic marginal zone B cells. <i>Nature Immunology</i> , <b>2014</b> , 15, 354-364	19.1	208
107	High incidence of NLRP3 somatic mosaicism in patients with chronic infantile neurologic, cutaneous, articular syndrome: results of an International Multicenter Collaborative Study. <i>Arthritis and Rheumatism</i> , <b>2011</b> , 63, 3625-32		200
106	NOD2 gene-associated pediatric granulomatous arthritis: clinical diversity, novel and recurrent mutations, and evidence of clinical improvement with interleukin-1 blockade in a Spanish cohort. <i>Arthritis and Rheumatism</i> , <b>2007</b> , 56, 3805-13		198
105	Classification criteria for autoinflammatory recurrent fevers. <i>Annals of the Rheumatic Diseases</i> , <b>2019</b> , 78, 1025-1032	2.4	159
104	MCC950 closes the active conformation of NLRP3 to an inactive state. <i>Nature Chemical Biology</i> , <b>2019</b> , 15, 560-564	11.7	156
103	Heterogeneity among patients with tumor necrosis factor receptor-associated periodic syndrome phenotypes. <i>Arthritis and Rheumatism</i> , <b>2003</b> , 48, 2632-44		143
102	Crohn's disease patients carrying Nod2/CARD15 gene variants have an increased and early need for first surgery due to stricturing disease and higher rate of surgical recurrence. <i>Annals of Surgery</i> , <b>2005</b> , 242, 693-700	7.8	141
101	Blau syndrome: cross-sectional data from a multicentre study of clinical, radiological and functional outcomes. <i>Rheumatology</i> , <b>2015</b> , 54, 1008-16	3.9	96
100	NOD2-associated pediatric granulomatous arthritis, an expanding phenotype: study of an international registry and a national cohort in Spain. <i>Arthritis and Rheumatism</i> , <b>2009</b> , 60, 1797-803		94
99	Somatic NLRP3 mosaicism in Muckle-Wells syndrome. A genetic mechanism shared by different phenotypes of cryopyrin-associated periodic syndromes. <i>Annals of the Rheumatic Diseases</i> , <b>2015</b> , 74, 603-10	2.4	83
98	Evaluating the Genetics of Common Variable Immunodeficiency: Monogenetic Model and Beyond. <i>Frontiers in Immunology</i> , <b>2018</b> , 9, 636	8.4	81
97	A novel Pyrin-Associated Autoinflammation with Neutrophilic Dermatitis mutation further defines 14-3-3 binding of pyrin and distinction to Familial Mediterranean Fever. <i>Annals of the Rheumatic Diseases</i> , <b>2017</b> , 76, 2085-2094	2.4	75
96	IL36RN mutations define a severe autoinflammatory phenotype of generalized pustular psoriasis. <i>Journal of Allergy and Clinical Immunology</i> , <b>2015</b> , 135, 1067-1070.e9	11.5	73

95	New workflow for classification of genetic variants Spathogenicity applied to hereditary recurrent fevers by the International Study Group for Systemic Autoinflammatory Diseases (INSAID). <i>Journal of Medical Genetics</i> , <b>2018</b> , 55, 530-537	5.8	73
94	Single amino acid charge switch defines clinically distinct proline-serine-threonine phosphatase-interacting protein 1 (PSTPIP1)-associated inflammatory diseases. <i>Journal of Allergy and Clinical Immunology</i> , <b>2015</b> , 136, 1337-45	11.5	73
93	Clinical and genetic heterogeneity among Spanish patients with recurrent autoinflammatory syndromes associated with the CIAS1/PYPAF1/NALP3 gene. <i>Arthritis and Rheumatism</i> , <b>2004</b> , 50, 4045-50		72
92	Late-Onset Cryopyrin-Associated Periodic Syndromes Caused by Somatic NLRP3 Mosaicism-UK Single Center Experience. <i>Frontiers in Immunology</i> , <b>2017</b> , 8, 1410	8.4	67
91	A somatic NLRP3 mutation as a cause of a sporadic case of chronic infantile neurologic, cutaneous, articular syndrome/neonatal-onset multisystem inflammatory disease: Novel evidence of the role of low-level mosaicism as the pathophysiologic mechanism underlying mendelian inherited diseases. <i>Arthritis and Rheumatism</i> , <b>2010</b> , 62, 1158-66		64
90	Familial CD8 deficiency due to a mutation in the CD8 alpha gene. <i>Journal of Clinical Investigation</i> , <b>2001</b> , 108, 117-23	15.9	63
89	Disparity for the minor histocompatibility antigen HA-1 is associated with an increased risk of acute graft-versus-host disease (GvHD) but it does not affect chronic GvHD incidence, disease-free survival or overall survival after allogeneic human leucocyte antigen-identical sibling donor transplantation. <i>British Journal of Haematology</i> , <b>2001</b> , 114, 931-6	4.5	60
88	IL-12R $\beta$ deficiency: mutation update and description of the IL12RB1 variation database. <i>Human Mutation</i> , <b>2013</b> , 34, 1329-39	4.7	56
87	Inherited biallelic CSF3R mutations in severe congenital neutropenia. <i>Blood</i> , <b>2014</b> , 123, 3811-7	2.2	55
86	LTBP2 and CYP1B1 mutations and associated ocular phenotypes in the Roma/Gypsy founder population. <i>European Journal of Human Genetics</i> , <b>2011</b> , 19, 326-33	5.3	50
85	Abnormal serum free light chain ratio in patients with multiple myeloma in complete remission has strong association with the presence of oligoclonal bands: implications for stringent complete remission definition. <i>Blood</i> , <b>2009</b> , 114, 4954-6	2.2	50
84	Clinical and genetic characterization of the autoinflammatory diseases diagnosed in an adult reference center. <i>Autoimmunity Reviews</i> , <b>2016</b> , 15, 9-15	13.6	49
83	Molecular genetic investigation, clinical features, and response to treatment in 21 patients with Schnitzler syndrome. <i>Blood</i> , <b>2018</b> , 131, 974-981	2.2	47
82	An unexpectedly high frequency of MEFV mutations in patients with anti-citrullinated protein antibody-negative palindromic rheumatism. <i>Arthritis and Rheumatism</i> , <b>2007</b> , 56, 2784-8		47
81	Effect of NOD2/CARD15 variants in T-cell depleted allogeneic stem cell transplantation. <i>Haematologica</i> , <b>2006</b> , 91, 1372-6	6.6	47
80	Brief Report: Late-Onset Cryopyrin-Associated Periodic Syndrome Due to Myeloid-Restricted Somatic NLRP3 Mosaicism. <i>Arthritis and Rheumatology</i> , <b>2016</b> , 68, 3035-3041	9.5	43
79	Somatic NOD2 mosaicism in Blau syndrome. <i>Journal of Allergy and Clinical Immunology</i> , <b>2015</b> , 136, 484-7.e25		42
78	DNA demethylation of inflammasome-associated genes is enhanced in patients with cryopyrin-associated periodic syndromes. <i>Journal of Allergy and Clinical Immunology</i> , <b>2017</b> , 139, 202-211.e6	11.5	40

77	Blau Syndrome-Associated Uveitis: Preliminary Results From an International Prospective Interventional Case Series. <i>American Journal of Ophthalmology</i> , <b>2018</b> , 187, 158-166	4.9	40
76	Open-Label, Phase II Study to Assess the Efficacy and Safety of Canakinumab Treatment in Active Hyperimmunoglobulinemia D With Periodic Fever Syndrome. <i>Arthritis and Rheumatology</i> , <b>2017</b> , 69, 1679-1688	9.5	38
75	Etanercept plus colchicine treatment in a child with tumour necrosis factor receptor-associated periodic syndrome abolishes auto-inflammatory episodes without normalising the subclinical acute phase response. <i>European Journal of Pediatrics</i> , <b>2005</b> , 164, 13-6	4.1	38
74	Complement factor H binding of monomeric C-reactive protein downregulates proinflammatory activity and is impaired with at risk polymorphic CFH variants. <i>Scientific Reports</i> , <b>2016</b> , 6, 22889	4.9	38
73	Brief Report: whole-exome sequencing revealing somatic NLRP3 mosaicism in a patient with chronic infantile neurologic, cutaneous, articular syndrome. <i>Arthritis and Rheumatology</i> , <b>2014</b> , 66, 197-202	9.5	37
72	Evolving M-protein pattern in patients with smoldering multiple myeloma: impact on early progression. <i>Leukemia</i> , <b>2018</b> , 32, 1427-1434	10.7	36
71	Brief Report: Association of Tumor Necrosis Factor Receptor-Associated Periodic Syndrome With Gonosomal Mosaicism of a Novel 24-Nucleotide TNFRSF1A Deletion. <i>Arthritis and Rheumatology</i> , <b>2016</b> , 68, 2044-9	9.5	35
70	The inflammasome pathway in stable COPD and acute exacerbations. <i>ERJ Open Research</i> , <b>2016</b> , 2,	3.5	35
69	Brief Report: First Identification of Intrafamilial Recurrence of Blau Syndrome due to Gonosomal NOD2 Mosaicism. <i>Arthritis and Rheumatology</i> , <b>2016</b> , 68, 1039-44	9.5	34
68	Emergence of oligoclonal bands in patients with multiple myeloma in complete remission after induction chemotherapy: association with the use of novel agents. <i>Haematologica</i> , <b>2011</b> , 96, 171-3	6.6	33
67	Natural history and prognostic impact of oligoclonal humoral response in patients with multiple myeloma after autologous stem cell transplantation: long-term results from a single institution. <i>Haematologica</i> , <b>2013</b> , 98, 1142-6	6.6	31
66	mTOR intersects antibody-inducing signals from TACI in marginal zone B cells. <i>Nature Communications</i> , <b>2017</b> , 8, 1462	17.4	31
65	Common variants in NLRP2 and NLRP3 genes are strong prognostic factors for the outcome of HLA-identical sibling allogeneic stem cell transplantation. <i>Blood</i> , <b>2008</b> , 112, 4337-42	2.2	30
64	Unexpected relevant role of gene mosaicism in patients with primary immunodeficiency diseases. <i>Journal of Allergy and Clinical Immunology</i> , <b>2019</b> , 143, 359-368	11.5	29
63	First report of vertical transmission of a somatic NLRP3 mutation in cryopyrin-associated periodic syndromes. <i>Annals of the Rheumatic Diseases</i> , <b>2013</b> , 72, 1109-10	2.4	27
62	A novel G6PC3 homozygous 1-bp deletion as a cause of severe congenital neutropenia. <i>Blood</i> , <b>2009</b> , 114, 1718-9	2.2	27
61	Role of TNFRSF13B variants in patients with common variable immunodeficiency. <i>Blood</i> , <b>2009</b> , 114, 2846-8	2.8	26
60	Clinical, genetic, and therapeutic diversity in 2 patients with severe mevalonate kinase deficiency. <i>Pediatrics</i> , <b>2012</b> , 129, e535-9	7.4	25

59	Prognostic impact of serum immunoglobulin heavy/light chain ratio in patients with multiple myeloma in complete remission after autologous stem cell transplantation. <i>Biology of Blood and Marrow Transplantation</i> , <b>2012</b> , 18, 1076-9	4.7	21
58	Predictive value of selected biomarkers, polymorphisms, and clinical features for oligoarticular juvenile idiopathic arthritis-associated uveitis. <i>Ocular Immunology and Inflammation</i> , <b>2014</b> , 22, 208-12	2.8	20
57	Cryopyrin-associated Periodic Syndromes in Italian Patients: Evaluation of the Rate of Somatic NLRP3 Mosaicism and Phenotypic Characterization. <i>Journal of Rheumatology</i> , <b>2017</b> , 44, 1667-1673	4.1	19
56	Optic nerve and retinal features in uveitis associated with juvenile systemic granulomatous disease (Blau syndrome). <i>Acta Ophthalmologica</i> , <b>2015</b> , 93, 253-7	3.7	18
55	Association of intermittent hydrarthrosis with MEFV gene mutations. <i>Arthritis and Rheumatism</i> , <b>2006</b> , 54, 2334-5		18
54	Deficiency of Adenosine Deaminase 2 in Adults and Children: Experience From India. <i>Arthritis and Rheumatology</i> , <b>2021</b> , 73, 276-285	9.5	18
53	Non-Hodgkin lymphoma in pediatric patients with common variable immunodeficiency. <i>European Journal of Pediatrics</i> , <b>2015</b> , 174, 1069-76	4.1	17
52	Prognostic Impact of Serum Heavy/Light Chain Pairs in Patients With Monoclonal Gammopathy of Undetermined Significance and Smoldering Myeloma: Long-Term Results From a Single Institution. <i>Clinical Lymphoma, Myeloma and Leukemia</i> , <b>2016</b> , 16, e71-7	2	15
51	Efficacy and safety of canakinumab in cryopyrin-associated periodic syndromes: results from a Spanish cohort. <i>Clinical and Experimental Rheumatology</i> , <b>2015</b> , 33, S67-71	2.2	15
50	Efficacy of anakinra in an adult patient with recurrent pericarditis and cardiac tamponade as initial manifestations of tumor necrosis factor receptor-associated periodic syndrome due to the R92Q TNFRSF1A variant. <i>International Journal of Rheumatic Diseases</i> , <b>2017</b> , 20, 510-514	2.3	14
49	Biallelic loss-of-function LACC1/FAMIN Mutations Presenting as Rheumatoid Factor-Negative Polyarticular Juvenile Idiopathic Arthritis. <i>Scientific Reports</i> , <b>2019</b> , 9, 4579	4.9	14
48	Severe Autoinflammatory Manifestations and Antibody Deficiency Due to Novel Hyperomorphic PLCG2 Mutations. <i>Journal of Clinical Immunology</i> , <b>2020</b> , 40, 987-1000	5.7	12
47	Acquired familial Mediterranean fever associated with a somatic MEFV mutation in a patient with JAK2 associated post-polycythemia myelofibrosis. <i>Orphanet Journal of Rare Diseases</i> , <b>2015</b> , 10, 86	4.2	10
46	Smoldering Multiple Myeloma: Impact of the Evolving Pattern on Early Progression. <i>Blood</i> , <b>2014</b> , 124, 3363-3363	2.2	10
45	Hyper-IgD and periodic fever syndrome: a new MVK mutation (p.R277G) associated with a severe phenotype. <i>Gene</i> , <b>2014</b> , 542, 217-20	3.8	9
44	Clinical and genetic features of hereditary periodic fever syndromes in Hispanic patients: the Chilean experience. <i>Clinical Rheumatology</i> , <b>2012</b> , 31, 829-34	3.9	9
43	Cerebrospinal fluid neopterin and cryopyrin-associated periodic syndrome. <i>Pediatric Neurology</i> , <b>2009</b> , 41, 448-50	2.9	9
42	Genetic diagnosis of autoinflammatory disease patients using clinical exome sequencing. <i>European Journal of Medical Genetics</i> , <b>2020</b> , 63, 103920	2.6	9

41	Massively parallel sequencing reveals maternal somatic IL2RG mosaicism in an X-linked severe combined immunodeficiency family. <i>Journal of Allergy and Clinical Immunology</i> , <b>2013</b> , 132, 741-743.e2	11.5	8
40	Inherited prion disease with 4-octapeptide repeat insertion linked to valine at codon 129. <i>Brain</i> , <b>2012</b> , 135, e212	11.2	7
39	I591T MEFV mutation in a Spanish kindred: is it a mild mutation, a benign polymorphism, or a variant influenced by another modifier?. <i>Human Mutation</i> , <b>2002</b> , 20, 148-50	4.7	7
38	Hidradenitis Suppurativa: Proposal of Classification in Two Endotypes with Two-Step Cluster Analysis. <i>Dermatology</i> , <b>2021</b> , 237, 365-371	4.4	7
37	Acute generalized exanthematous pustulosis and polyarthritis associated with a novel CARD14 mutation. <i>Australasian Journal of Dermatology</i> , <b>2018</b> , 59, e70-e73	1.3	6
36	Prognostic impact of immunoparesis at diagnosis and after treatment onset in patients with light-chain amyloidosis. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , <b>2017</b> , 24, 245-252	2.7	5
35	Differential humoral responses against heat-shock proteins after autologous stem cell transplantation in multiple myeloma. <i>Annals of Hematology</i> , <b>2014</b> , 93, 107-11	3	5
34	Multiple myeloma in serologic complete remission after autologous stem cell transplantation: impact of bone marrow plasma cell assessment by conventional morphology on disease progression. <i>Biology of Blood and Marrow Transplantation</i> , <b>2011</b> , 17, 1084-7	4.7	5
33	Newly Described Autoinflammatory Diseases in Pediatric Dermatology. <i>Pediatric Dermatology</i> , <b>2016</b> , 33, 602-614	1.9	4
32	Palindromic rheumatism: Evidence of four subtypes of palindromic-like arthritis based in either MEFV or rheumatoid factor/ACPA status. <i>Joint Bone Spine</i> , <b>2021</b> , 88, 105235	2.9	4
31	Impact of Autologous Stem Cell Transplantation on the Incidence and Outcome of Oligoclonal Bands in Patients with Light-Chain Amyloidosis. <i>Biology of Blood and Marrow Transplantation</i> , <b>2017</b> , 23, 1269-1275	4.7	3
30	Executive Summary of the Consensus Document on the Diagnosis and Management of Patients with Primary Immunodeficiencies. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , <b>2020</b> , 8, 3342-3347	5.4	3
29	A novel phenotype variant of severe congenital neutropenia caused by G6PC3 deficiency. <i>Pediatric Blood and Cancer</i> , <b>2013</b> , 60, E29-31	3	3
28	Pro-B acute lymphoblastic leukemia in a patient with severe congenital neutropenia: an unusual form of malignant evolution. <i>Leukemia and Lymphoma</i> , <b>2013</b> , 54, 2325-7	1.9	3
27	Excellent response to secukinumab in an infant with severe generalized pustular psoriasis. <i>Journal of Dermatology</i> , <b>2021</b> , 48, 907-910	1.6	3
26	Chronic urticaria in infants as the first manifestation of autoinflammatory disease. <i>Pediatric Dermatology</i> , <b>2018</b> , 35, e337-e340	1.9	3
25	Genetic Aspects of Investigating and Understanding Autoinflammation <b>2019</b> , 19-48		2
24	Short-term efficacy of adalimumab in a patient with pyrin-associated autoinflammation with neutrophilic dermatosis. <i>JDDG - Journal of the German Society of Dermatology</i> , <b>2018</b> , 16, 756-759	1.2	2

23	¿Qué es lo que hoy debo saber sobre los síndromes autoinflamatorios?. <i>Seminarios De La Fundación Española De Reumatología</i> , <b>2007</b> , 8, 34-44		2
22	Novel evidences of atypical manifestations in cryopyrin-associated periodic syndromes. <i>Clinical and Experimental Rheumatology</i> , <b>2017</b> , 35 Suppl 108, 27-31	2.2	2
21	Gene Mosaicism Screening Using Single-Molecule Molecular Inversion Probes in Routine Diagnostics for Systemic Autoinflammatory Diseases. <i>Journal of Molecular Diagnostics</i> , <b>2019</b> , 21, 943-950	5.1	1
20	CIAS1 and NOD2 genes in adult-onset Still's disease. <i>Journal of Rheumatology</i> , <b>2014</b> , 41, 1566-7	4.1	1
19	Manifestaciones cutáneas en las enfermedades autoinflamatorias sistémicas. <i>Piel</i> , <b>2009</b> , 24, 139-145	0.1	1
18	May some cases of intermittent hydrarthrosis represent an atypical form of calcium pyrophosphate dihydrate crystal deposition disease? Usefulness of mutational analysis of the MEFV gene. <i>Seminars in Arthritis and Rheumatism</i> , <b>2008</b> , 37, 269-70	5.3	1
17	A fatal Turkish case of CINCA-NOMID syndrome due to the novel Val-351-Leu CIAS1 gene mutation. <i>Rheumatology International</i> , <b>2008</b> , 28, 379-83	3.6	1
16	Familial Mediterranean fever in the pediatric population. <i>Allergologia Et Immunopathologia</i> , <b>2022</b> , 50, 25-30	1.9	1
15	Multiple sclerosis in a patient with cryopyrin-associated autoinflammatory syndrome: Evidence that autoinflammation is the common link. <i>Clinical Immunology</i> , <b>2021</b> , 227, 108750	9	1
14	Baseline correlations and prognostic impact of serum monoclonal proteins in follicular lymphoma. <i>British Journal of Haematology</i> , <b>2021</b> , 193, 299-306	4.5	1
13	Excess Serum Interleukin-18 Distinguishes Patients with Pathogenic Mutations in PSTPIP1		1
12	Excess Serum Interleukin-18 Distinguishes Patients with Pathogenic Mutations in PSTPIP1. <i>Arthritis and Rheumatology</i> , <b>2021</b> ,	9.5	1
11	Serum monoclonal component in chronic lymphocytic leukemia: baseline correlations and prognostic impact. <i>Haematologica</i> , <b>2021</b> , 106, 1754-1757	6.6	0
10	Assessment of the gene mosaicism burden in blood and its implications for immune disorders. <i>Scientific Reports</i> , <b>2021</b> , 11, 12940	4.9	0
9	A Novel Pathogenic Variant in a Mother and Daughter with Blau Syndrome. <i>Ophthalmic Genetics</i> , <b>2021</b> , 42, 753-764	1.2	0
8	Executive Summary of the Consensus Document on the Diagnosis and Management of Patients with Primary Immunodeficiencies. <i>Enfermedades Infecciosas Y Microbiología Clínica</i> , <b>2020</b> , 38, 438-443	0.9	
7	Hereditary systemic autoinflammatory diseases. <i>Reumatología Clínica (English Edition)</i> , <b>2011</b> , 7, 45-50	0.1	
6	Response: Free light chain assay and stringent complete remission in multiple myeloma: more questions than answers. <i>Blood</i> , <b>2010</b> , 115, 3414-3415	2.2	

- 5 Polymorphisms of NOD2/CARD15 Are Associated with Clinical Outcome in T-Cell Depleted HLA-Identical Sibling Allogeneic Stem Cell Transplantation.. *Blood*, **2005**, 106, 1408-1408 2.2
- 4 Genetic Polymorphisms in the Inflammasomes Are Associated with Relapse and Survival in HLA-Identical Sibling Donor Allogeneic Stem Cell Transplantation.. *Blood*, **2007**, 110, 1075-1075 2.2
- 3 Smoldering Multiple Myeloma: Usefulness of Serum Heavy/Light Chain Measurements for the Evaluation of Evolving Pattern. *Blood*, **2018**, 132, 4514-4514 2.2
- 2 Long-Term Survivors after Stem Cell Transplantation in Multiple Myeloma: Bone Marrow Minimal Residual Disease, PET/CT and Immunological Status. *Blood*, **2015**, 126, 4192-4192 2.2
- 1 Inherited Biallelic Loss-Of-Function Mutations In CSF3R Define a Novel Type Of Severe Congenital Neutropenia With Full Myeloid Cell Maturation and Refractoriness To RhG-CSF. *Blood*, **2013**, 122, 1025-1025 2.2