# Juan Ignacio Arostegui

## List of Publications by Year in Descending Order



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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 | Familial Mediterranean fever in the pediatric population. *Allergologia Et Immunopathologia*, **2022**, 50, 25-30 | 1.9 | 1 |
| 111 | Serum monoclonal component in chronic lymphocytic leukemia: baseline correlations and prognostic impact. *Haematologica*, **2021**, 106, 1754-1757 | 6.6 | 0 |
| 110 | Assessment of the gene mosaicism burden in blood and its implications for immune disorders. *Scientific Reports*, **2021**, 11, 12940 | 4.9 | 0 |
| 109 | Multiple sclerosis in a patient with cryopyrin-associated autoinflammatory syndrome: Evidence that autoinflammation is the common link. *Clinical Immunology*, **2021**, 227, 108750 | 9 | 1 |
| 108 | Hidradenitis Suppurativa: Proposal of Classification in Two Endotypes with Two-Step Cluster Analysis. *Dermatology*, **2021**, 237, 365-371 | 4.4 | 7 |
| 107 | Baseline correlations and prognostic impact of serum monoclonal proteins in follicular lymphoma. *British Journal of Haematology*, **2021**, 193, 299-306 | 4.5 | 1 |
| 106 | Deficiency of Adenosine Deaminase 2 in Adults and Children: Experience From India. *Arthritis and Rheumatology*, **2021**, 73, 276-285 | 9.5 | 18 |
| 105 | Excellent response to secukinumab in an infant with severe generalized pustular psoriasis. *Journal of Dermatology*, **2021**, 48, 907-910 | 1.6 | 3 |
| 104 | A Novel Pathogenic Variant in a Mother and Daughter with Blau Syndrome. *Ophthalmic Genetics*, **2021**, 42, 753-764 | 1.2 | 0 |
| 103 | Excess Serum Interleukin-18 Distinguishes Patients with Pathogenic Mutations in PSTPIP1. *Arthritis and Rheumatology*, **2021**, | 9.5 | 1 |
| 102 | Palindromic rheumatism: Evidence of four subtypes of palindromic-like arthritis based in either MEFV or rheumatoid factor/ACPA status. *Joint Bone Spine*, **2021**, 88, 105235 | 2.9 | 4 |
| 101 | Executive Summary of the Consensus Document on the Diagnosis and Management of Patients with Primary Immunodeficiencies. *Enfermedades Infecciosas Y Microbiología Clínica*, **2020**, 38, 438-443 | 0.9 | |
| 100 | Executive Summary of the Consensus Document on the Diagnosis and Management of Patients with Primary Immunodeficiencies. *Journal of Allergy and Clinical Immunology: in Practice*, **2020**, 8, 3342-3347 | 5.4 | 3 |
| 99 | Severe Autoinflammatory Manifestations and Antibody Deficiency Due to Novel Hypermorphic PLCG2 Mutations. *Journal of Clinical Immunology*, **2020**, 40, 987-1000 | 5.7 | 12 |
| 98 | Genetic diagnosis of autoinflammatory disease patients using clinical exome sequencing. *European Journal of Medical Genetics*, **2020**, 63, 103920 | 2.6 | 9 |
| 97 | MCC950 closes the active conformation of NLRP3 to an inactive state. *Nature Chemical Biology*, **2019**, 15, 560-564 | 11.7 | 156 |
| 96 | Classification criteria for autoinflammatory recurrent fevers. *Annals of the Rheumatic Diseases*, **2019**, 78, 1025-1032 | 2.4 | 159 |

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| 95 | Biallelic loss-of-function LACC1/FAMIN Mutations Presenting as Rheumatoid Factor-Negative Polyarticular Juvenile Idiopathic Arthritis. *Scientific Reports*, **2019**, 9, 4579 | 4.9 | 14 |
| 94 | Genetic Aspects of Investigating and Understanding Autoinflammation **2019**, 19-48 | | 2 |
| 93 | Gene Mosaicism Screening Using Single-Molecule Molecular Inversion Probes in Routine Diagnostics for Systemic Autoinflammatory Diseases. *Journal of Molecular Diagnostics*, **2019**, 21, 943-950 | 5.1 | 1 |
| 92 | Unexpected relevant role of gene mosaicism in patients with primary immunodeficiency diseases. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 359-368 | 11.5 | 29 |
| 91 | Evolving M-protein pattern in patients with smoldering multiple myeloma: impact on early progression. *Leukemia*, **2018**, 32, 1427-1434 | 10.7 | 36 |
| 90 | Molecular genetic investigation, clinical features, and response to treatment in 21 patients with Schnitzler syndrome. *Blood*, **2018**, 131, 974-981 | 2.2 | 47 |
| 89 | Short-term efficacy of adalimumab in a patient with pyrin-associated autoinflammation with neutrophilic dermatosis. *JDDG - Journal of the German Society of Dermatology*, **2018**, 16, 756-759 | 1.2 | 2 |
| 88 | New workflow for classification of genetic variantsSpathogenicity applied to hereditary recurrent fevers by the International Study Group for Systemic Autoinflammatory Diseases (INSAID). *Journal of Medical Genetics*, **2018**, 55, 530-537 | 5.8 | 73 |
| 87 | Blau Syndrome-Associated Uveitis: Preliminary Results From an International Prospective Interventional Case Series. *American Journal of Ophthalmology*, **2018**, 187, 158-166 | 4.9 | 40 |
| 86 | Evaluating the Genetics of Common Variable Immunodeficiency: Monogenetic Model and Beyond. *Frontiers in Immunology*, **2018**, 9, 636 | 8.4 | 81 |
| 85 | Smoldering Multiple Myeloma: Usefulness of Serum Heavy/Light Chain Measurements for the Evaluation of Evolving Pattern. *Blood*, **2018**, 132, 4514-4514 | 2.2 | |
| 84 | Acute generalized exanthematous pustulosis and polyarthritis associated with a novel CARD14 mutation. *Australasian Journal of Dermatology*, **2018**, 59, e70-e73 | 1.3 | 6 |
| 83 | Chronic urticaria in infants as the first manifestation of autoinflammatory disease. *Pediatric Dermatology*, **2018**, 35, e337-e340 | 1.9 | 3 |
| 82 | DNA demethylation of inflammasome-associated genes is enhanced in patients with cryopyrin-associated periodic syndromes. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 202-211.e6 | 11.5 | 40 |
| 81 | Open-Label, Phase II Study to Assess the Efficacy and Safety of Canakinumab Treatment in Active Hyperimmunoglobulinemia D With Periodic Fever Syndrome. *Arthritis and Rheumatology*, **2017**, 69, 1679-1688 | 9.5 | 38 |
| 80 | Impact of Autologous Stem Cell Transplantation on the Incidence and Outcome of Oligoclonal Bands in Patients with Light-Chain Amyloidosis. *Biology of Blood and Marrow Transplantation*, **2017**, 23, 1269-1275 | 4.7 | 3 |
| 79 | 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. *International Journal of Rheumatic Diseases*, **2017**, 20, 510-514 | 2.3 | 14 |
| 78 | Prognostic impact of immunoparesis at diagnosis and after treatment onset in patients with light-chain amyloidosis. *Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis*, **2017**, 24, 245-252 | 2.7 | 5 |

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| 77 | A novel Pyrin-Associated Autoinflammation with Neutrophilic Dermatosis mutation further defines 14-3-3 binding of pyrin and distinction to Familial Mediterranean Fever. *Annals of the Rheumatic Diseases*, **2017**, 76, 2085-2094 | 2.4 | 75 |
| 76 | Cryopyrin-associated Periodic Syndromes in Italian Patients: Evaluation of the Rate of Somatic NLRP3 Mosaicism and Phenotypic Characterization. *Journal of Rheumatology*, **2017**, 44, 1667-1673 | 4.1 | 19 |
| 75 | mTOR intersects antibody-inducing signals from TACI in marginal zone B cells. *Nature Communications*, **2017**, 8, 1462 | 17.4 | 31 |
| 74 | Late-Onset Cryopyrin-Associated Periodic Syndromes Caused by Somatic NLRP3 Mosaicism-UK Single Center Experience. *Frontiers in Immunology*, **2017**, 8, 1410 | 8.4 | 67 |
| 73 | Novel evidences of atypical manifestations in cryopyrin-associated periodic syndromes. *Clinical and Experimental Rheumatology*, **2017**, 35 Suppl 108, 27-31 | 2.2 | 2 |
| 72 | Clinical and genetic characterization of the autoinflammatory diseases diagnosed in an adult reference center. *Autoimmunity Reviews*, **2016**, 15, 9-15 | 13.6 | 49 |
| 71 | Brief Report: First Identification of Intrafamilial Recurrence of Blau Syndrome due to Gonosomal NOD2 Mosaicism. *Arthritis and Rheumatology*, **2016**, 68, 1039-44 | 9.5 | 34 |
| 70 | 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. *Clinical Lymphoma, Myeloma and Leukemia*, **2016**, 16, e71-7 | 2 | 15 |
| 69 | Brief Report: Late-Onset Cryopyrin-Associated Periodic Syndrome Due to Myeloid-Restricted Somatic NLRP3 Mosaicism. *Arthritis and Rheumatology*, **2016**, 68, 3035-3041 | 9.5 | 43 |
| 68 | Brief Report: Association of Tumor Necrosis Factor Receptor-Associated Periodic Syndrome With Gonosomal Mosaicism of a Novel 24-Nucleotide TNFRSF1A Deletion. *Arthritis and Rheumatology*, **2016**, 68, 2044-9 | 9.5 | 35 |
| 67 | Complement factor H binding of monomeric C-reactive protein downregulates proinflammatory activity and is impaired with at risk polymorphic CFH variants. *Scientific Reports*, **2016**, 6, 22889 | 4.9 | 38 |
| 66 | The inflammasome pathway in stable COPD and acute exacerbations. *ERJ Open Research*, **2016**, 2, | 3.5 | 35 |
| 65 | Newly Described Autoinflammatory Diseases in Pediatric Dermatology. *Pediatric Dermatology*, **2016**, 33, 602-614 | 1.9 | 4 |
| 64 | IL36RN mutations define a severe autoinflammatory phenotype of generalized pustular psoriasis. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1067-1070.e9 | 11.5 | 73 |
| 63 | Non-Hodgkin lymphoma in pediatric patients with common variable immunodeficiency. *European Journal of Pediatrics*, **2015**, 174, 1069-76 | 4.1 | 17 |
| 62 | Optic nerve and retinal features in uveitis associated with juvenile systemic granulomatous disease (Blau syndrome). *Acta Ophthalmologica*, **2015**, 93, 253-7 | 3.7 | 18 |
| 61 | Somatic NOD2 mosaicism in Blau syndrome. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 484-7.e2 | 11.5 | 42 |
| 60 | Blau syndrome: cross-sectional data from a multicentre study of clinical, radiological and functional outcomes. *Rheumatology*, **2015**, 54, 1008-16 | 3.9 | 96 |

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| 59 | Somatic NLRP3 mosaicism in Muckle-Wells syndrome. A genetic mechanism shared by different phenotypes of cryopyrin-associated periodic syndromes. *Annals of the Rheumatic Diseases*, **2015**, 74, 603-10 | 2.4 | 83 |
| 58 | Acquired familial Mediterranean fever associated with a somatic MEFV mutation in a patient with JAK2 associated post-polycythemia myelofibrosis. *Orphanet Journal of Rare Diseases*, **2015**, 10, 86 | 4.2 | 10 |
| 57 | Single amino acid charge switch defines clinically distinct proline-serine-threonine phosphatase-interacting protein 1 (PSTPIP1)-associated inflammatory diseases. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 1337-45 | 11.5 | 73 |
| 56 | 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 | |
| 55 | Efficacy and safety of canakinumab in cryopyrin-associated periodic syndromes: results from a Spanish cohort. *Clinical and Experimental Rheumatology*, **2015**, 33, S67-71 | 2.2 | 15 |
| 54 | Innate lymphoid cells integrate stromal and immunological signals to enhance antibody production by splenic marginal zone B cells. *Nature Immunology*, **2014**, 15, 354-364 | 19.1 | 208 |
| 53 | Differential humoral responses against heat-shock proteins after autologous stem cell transplantation in multiple myeloma. *Annals of Hematology*, **2014**, 93, 107-11 | 3 | 5 |
| 52 | The NLRP3 inflammasome is released as a particulate danger signal that amplifies the inflammatory response. *Nature Immunology*, **2014**, 15, 738-48 | 19.1 | 522 |
| 51 | Hyper-IgD and periodic fever syndrome: a new MVK mutation (p.R277G) associated with a severe phenotype. *Gene*, **2014**, 542, 217-20 | 3.8 | 9 |
| 50 | Predictive value of selected biomarkers, polymorphisms, and clinical features for oligoarticular juvenile idiopathic arthritis-associated uveitis. *Ocular Immunology and Inflammation*, **2014**, 22, 208-12 | 2.8 | 20 |
| 49 | CIAS1 and NOD2 genes in adult-onset Still's disease. *Journal of Rheumatology*, **2014**, 41, 1566-7 | 4.1 | 1 |
| 48 | Inherited biallelic CSF3R mutations in severe congenital neutropenia. *Blood*, **2014**, 123, 3811-7 | 2.2 | 55 |
| 47 | Brief Report: whole-exome sequencing revealing somatic NLRP3 mosaicism in a patient with chronic infantile neurologic, cutaneous, articular syndrome. *Arthritis and Rheumatology*, **2014**, 66, 197-202 | 9.5 | 37 |
| 46 | Smoldering Multiple Myeloma: Impact of the Evolving Pattern on Early Progression. *Blood*, **2014**, 124, 3363-3363 | 2.2 | 10 |
| 45 | IL-12Rβ1 deficiency: mutation update and description of the IL12RB1 variation database. *Human Mutation*, **2013**, 34, 1329-39 | 4.7 | 56 |
| 44 | 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. *Haematologica*, **2013**, 98, 1142-6 | 6.6 | 31 |
| 43 | Massively parallel sequencing reveals maternal somatic IL2RG mosaicism in an X-linked severe combined immunodeficiency family. *Journal of Allergy and Clinical Immunology*, **2013**, 132, 741-743.e2 | 11.5 | 8 |
| 42 | A novel phenotype variant of severe congenital neutropenia caused by G6PC3 deficiency. *Pediatric Blood and Cancer*, **2013**, 60, E29-31 | 3 | 3 |

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| 41 | Pro-B acute lymphoblastic leukemia in a patient with severe congenital neutropenia: an unusual form of malignant evolution. *Leukemia and Lymphoma*, **2013**, 54, 2325-7 | 1.9 | 3 |
| 40 | First report of vertical transmission of a somatic NLRP3 mutation in cryopyrin-associated periodic syndromes. *Annals of the Rheumatic Diseases*, **2013**, 72, 1109-10 | 2.4 | 27 |
| 39 | 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 | |
| 38 | Prognostic impact of serum immunoglobulin heavy/light chain ratio in patients with multiple myeloma in complete remission after autologous stem´cell transplantation. *Biology of Blood and Marrow Transplantation*, **2012**, 18, 1076-9 | 4.7 | 21 |
| 37 | Clinical and genetic features of hereditary periodic fever syndromes in Hispanic patients: the Chilean experience. *Clinical Rheumatology*, **2012**, 31, 829-34 | 3.9 | 9 |
| 36 | Clinical, genetic, and therapeutic diversity in 2 patients with severe mevalonate kinase deficiency. *Pediatrics*, **2012**, 129, e535-9 | 7.4 | 25 |
| 35 | Inherited prion disease with 4-octapeptide repeat insertion linked to valine at codon 129. *Brain*, **2012**, 135, e212 | 11.2 | 7 |
| 34 | B cell-helper neutrophils stimulate the diversification and production of immunoglobulin in the marginal zone of the spleen. *Nature Immunology*, **2011**, 13, 170-80 | 19.1 | 501 |
| 33 | Hereditary systemic autoinflammatory diseases. *Reumatología Clínica (English Edition)*, **2011**, 7, 45-50 | 0.1 | |
| 32 | Multiple myeloma in serologic complete remission after autologous stem cell transplantation: impact of bone marrow plasma cell assessment by conventional morphology on disease progression. *Biology of Blood and Marrow Transplantation*, **2011**, 17, 1084-7 | 4.7 | 5 |
| 31 | Emergence of oligoclonal bands in patients with multiple myeloma in complete remission after induction chemotherapy: association with the use of novel agents. *Haematologica*, **2011**, 96, 171-3 | 6.6 | 33 |
| 30 | LTBP2 and CYP1B1 mutations and associated ocular phenotypes in the Roma/Gypsy founder population. *European Journal of Human Genetics*, **2011**, 19, 326-33 | 5.3 | 50 |
| 29 | High incidence of NLRP3 somatic mosaicism in patients with chronic infantile neurologic, cutaneous, articular syndrome: results of an International Multicenter Collaborative Study. *Arthritis and Rheumatism*, **2011**, 63, 3625-32 | | 200 |
| 28 | Clinical features and outcome of patients with IRAK-4 and MyD88 deficiency. *Medicine (United States)*, **2010**, 89, 403-425 | 1.8 | 297 |
| 27 | Response: Free light chain assay and stringent complete remission in multiple myeloma: more questions than answers. *Blood*, **2010**, 115, 3414-3415 | 2.2 | |
| 26 | 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. *Arthritis and Rheumatism*, **2010**, 62, 1158-66 | | 64 |
| 25 | Manifestaciones cutáneas en las enfermedades autoinflamatorias sistémicas. *Piel*, **2009**, 24, 139-145 | 0.1 | 1 |
| 24 | NOD2-associated pediatric granulomatous arthritis, an expanding phenotype: study of an international registry and a national cohort in Spain. *Arthritis and Rheumatism*, **2009**, 60, 1797-803 | | 94 |

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| 23 | Cerebrospinal fluid neopterin and cryopyrin-associated periodic syndrome. *Pediatric Neurology*, **2009**, 41, 448-50 | 2.9 | 9 |
| 22 | 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. *Blood*, **2009**, 114, 4954-6 | 2.2 | 50 |
| 21 | A novel G6PC3 homozygous 1-bp deletion as a cause of severe congenital neutropenia. *Blood*, **2009**, 114, 1718-9 | 2.2 | 27 |
| 20 | Role of TNFRSF13B variants in patients with common variable immunodeficiency. *Blood*, **2009**, 114, 2846-8 | 2.2 | 26 |
| 19 | 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. *Seminars in Arthritis and Rheumatism*, **2008**, 37, 269-70 | 5.3 | 1 |
| 18 | Pyogenic bacterial infections in humans with MyD88 deficiency. *Science*, **2008**, 321, 691-6 | 33.3 | 608 |
| 17 | Common variants in NLRP2 and NLRP3 genes are strong prognostic factors for the outcome of HLA-identical sibling allogeneic stem cell transplantation. *Blood*, **2008**, 112, 4337-42 | 2.2 | 30 |
| 16 | A fatal Turkish case of CINCA-NOMID syndrome due to the novel Val-351-Leu CIAS1 gene mutation. *Rheumatology International*, **2008**, 28, 379-83 | 3.6 | 1 |
| 15 | ¿Qu□es lo que hoy debo saber sobre los s□dromes autoinflamatorios?. *Seminarios De La Fundaci□n Espa□ola De Reumatolog□a*, **2007**, 8, 34-44 | | 2 |
| 14 | An unexpectedly high frequency of MEFV mutations in patients with anti-citrullinated protein antibody-negative palindromic rheumatism. *Arthritis and Rheumatism*, **2007**, 56, 2784-8 | | 47 |
| 13 | 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. *Arthritis and Rheumatism*, **2007**, 56, 3805-13 | | 198 |
| 12 | 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 | |
| 11 | Association of intermittent hydrarthrosis with MEFV gene mutations. *Arthritis and Rheumatism*, **2006**, 54, 2334-5 | | 18 |
| 10 | Effect of NOD2/CARD15 variants in T-cell depleted allogeneic stem cell transplantation. *Haematologica*, **2006**, 91, 1372-6 | 6.6 | 47 |
| 9 | 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. *Annals of Surgery*, **2005**, 242, 693-700 | 7.8 | 141 |
| 8 | 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. *European Journal of Pediatrics*, **2005**, 164, 13-6 | 4.1 | 38 |
| 7 | 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 | |
| 6 | Clinical and genetic heterogeneity among Spanish patients with recurrent autoinflammatory syndromes associated with the CIAS1/PYPAF1/NALP3 gene. *Arthritis and Rheumatism*, **2004**, 50, 4045-50 | | 72 |

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| 5 | Heterogeneity among patients with tumor necrosis factor receptor-associated periodic syndrome phenotypes. *Arthritis and Rheumatism*, **2003**, 48, 2632-44 | | 143 |
| 4 | I591T MEFV mutation in a Spanish kindred: is it a mild mutation, a benign polymorphism, or a variant influenced by another modifier?. *Human Mutation*, **2002**, 20, 148-50 | 4.7 | 7 |
| 3 | 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. *British Journal of Haematology*, **2001**, 114, 931-6 | 4.5 | 60 |
| 2 | Familial CD8 deficiency due to a mutation in the CD8 alpha gene. *Journal of Clinical Investigation*, **2001**, 108, 117-23 | 15.9 | 63 |
| 1 | Excess Serum Interleukin-18 Distinguishes Patients with Pathogenic Mutations in PSTPIP1 | | 1 |