

# Osamu Ohara

## List of Publications by Year in descending order

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Version: 2024-02-01

487  
papers

31,509  
citations

6486

82  
h-index

7234

158  
g-index

505  
all docs

505  
docs citations

505  
times ranked

48874  
citing authors

#	ARTICLE	IF	CITATIONS
1	Absent Xâ€linked inhibitor of apoptosis protein expression in T cell blasts and causal mutations including nonâ€coding deletion. <i>Pediatrics International</i> , 2022, 64, .	0.2	0
2	Transcriptome Analysis of Peripheral Blood Mononuclear Cells in Pulmonary Sarcoidosis. <i>Frontiers in Medicine</i> , 2022, 9, 822094.	1.2	6
3	Establishment of mouse stem cells that can recapitulate the developmental potential of primitive endoderm. <i>Science</i> , 2022, 375, 574-578.	6.0	16
4	In-Depth Serum Proteomics by DIA-MS with <i>In Silico</i> Spectral Libraries Reveals Dynamics during the Active Phase of Systemic Juvenile Idiopathic Arthritis. <i>ACS Omega</i> , 2022, 7, 7012-7023.	1.6	7
5	Data-Independent Acquisition Mass Spectrometry-Based Deep Proteome Analysis for Hydrophobic Proteins from Dried Blood Spots Enriched by Sodium Carbonate Precipitation. <i>Methods in Molecular Biology</i> , 2022, 2420, 39-52.	0.4	4
6	Evaluation of the Suitability of Dried Saliva Spots for In-Depth Proteome Analyses for Clinical Applications. <i>Journal of Proteome Research</i> , 2022, 21, 1340-1348.	1.8	0
7	Trapping of CDC42 C-terminal variants in the Golgi drives pyrin inflammasome hyperactivation. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	18
8	Single-Shot 10K Proteome Approach: Over 10,000 Protein Identifications by Data-Independent Acquisition-Based Single-Shot Proteomics with Ion Mobility Spectrometry. <i>Journal of Proteome Research</i> , 2022, 21, 1418-1427.	1.8	37
9	Clinical phenotypes and genetic analyses for diagnosis of systemic autoinflammatory diseases in adult patients with unexplained fever. <i>Modern Rheumatology</i> , 2021, 31, 704-709.	0.9	7
10	IRAK4 Deficiency Presenting with Anti-NMDAR Encephalitis and HHV6 Reactivation. <i>Journal of Clinical Immunology</i> , 2021, 41, 125-135.	2.0	10
11	Hemophagocytic Lymphohistiocytosis in Children with Chronic Granulomatous Diseaseâ€Single-Center Experience from North India. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 771-782.e3.	2.0	7
12	TAS4464, a NEDD8-activating enzyme inhibitor, activates both intrinsic and extrinsic apoptotic pathways via c-Myc-mediated regulation in acute myeloid leukemia. <i>Oncogene</i> , 2021, 40, 1217-1230.	2.6	15
13	Clinical and Immunological Heterogeneity in Japanese Patients with Gain-of-Function Variants in STAT3. <i>Journal of Clinical Immunology</i> , 2021, 41, 780-790.	2.0	10
14	Targeting critical kinases and anti-apoptotic molecules overcomes steroid resistance in MLL-rearranged leukaemia. <i>EBioMedicine</i> , 2021, 64, 103235.	2.7	2
15	Clinical, Immunological, and Molecular Profile of Chronic Granulomatous Disease: A Multi-Centric Study of 236 Patients From India. <i>Frontiers in Immunology</i> , 2021, 12, 625320.	2.2	31
16	Genetic alterations in squamous cell lung cancer associated with idiopathic pulmonary fibrosis. <i>International Journal of Cancer</i> , 2021, 148, 3008-3018.	2.3	7
17	Inherited CARD9 Deficiency in a Child with Invasive Disease Due to <i>Exophiala dermatitidis</i> and Two Older but Asymptomatic Siblings. <i>Journal of Clinical Immunology</i> , 2021, 41, 975-986.	2.0	15
18	ETV6-related thrombocytopenia associated with a transient decrease in von Willebrand factor. <i>International Journal of Hematology</i> , 2021, 114, 297-300.	0.7	1

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19	Rapid Flow Cytometry-Based Assay for the Functional Classification of MEFV Variants. <i>Journal of Clinical Immunology</i> , 2021, 41, 1187-1197.	2.0	13
20	A Simple Method for In-Depth Proteome Analysis of Mammalian Cell Culture Conditioned Media Containing Fetal Bovine Serum. <i>International Journal of Molecular Sciences</i> , 2021, 22, 2565.	1.8	10
21	Combined inhibition of XIAP and BCL2 drives maximal therapeutic efficacy in genetically diverse aggressive acute myeloid leukemia. <i>Nature Cancer</i> , 2021, 2, 340-356.	5.7	11
22	Wiskott Aldrich Syndrome: A Multi-Institutional Experience From India. <i>Frontiers in Immunology</i> , 2021, 12, 627651.	2.2	16
23	Genomics analysis of leukaemia predisposition in X-linked agammaglobulinaemia. <i>British Journal of Haematology</i> , 2021, 193, 1277-1281.	1.2	1
24	<i>Staphylococcus cohnii</i> is a potentially biotherapeutic skin commensal alleviating skin inflammation. <i>Cell Reports</i> , 2021, 35, 109052.	2.9	26
25	Sjögren's syndrome-associated SNPs increase GTF2I expression in salivary gland cells to enhance inflammation development. <i>International Immunology</i> , 2021, 33, 423-434.	1.8	9
26	A variant in human AIOLOS impairs adaptive immunity by interfering with IKAROS. <i>Nature Immunology</i> , 2021, 22, 893-903.	7.0	33
27	SCD2-mediated monounsaturated fatty acid metabolism regulates cGAS-STING-dependent type I IFN responses in CD4+ T cells. <i>Communications Biology</i> , 2021, 4, 820.	2.0	21
28	Influenza virus infection expands the breadth of antibody responses through IL-4 signalling in B cells. <i>Nature Communications</i> , 2021, 12, 3789.	5.8	21
29	Multiomic technologies for analyses of inborn errors of immunity: from snapshot of the average cell to dynamic temporal picture at single-cell resolution. <i>Inflammation and Regeneration</i> , 2021, 41, 19.	1.5	0
30	Hematopoietic Cell Transplantation Rescues Inflammatory Bowel Disease and Dysbiosis of Gut Microbiota in XIAP Deficiency. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 3767-3780.	2.0	15
31	Acetate differentially regulates IgA reactivity to commensal bacteria. <i>Nature</i> , 2021, 595, 560-564.	13.7	104
32	Detailed analysis of Japanese patients with adenosine deaminase 2 deficiency reveals characteristic elevation of type II interferon signature and STAT1 hyperactivation. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 550-562.	1.5	30
33	Genetic subtype classification using a simplified algorithm and mutational characteristics of diffuse large B-cell lymphoma in a Japanese cohort. <i>British Journal of Haematology</i> , 2021, 195, 731-742.	1.2	8
34	Toward proteome-wide exploration of proteins in dried blood spots using liquid chromatography-coupled mass spectrometry. <i>Proteomics</i> , 2021, 21, e2100019.	1.3	9
35	Transplantation of Human Autologous Synovial Mesenchymal Stem Cells with Trisomy 7 into the Knee Joint and 5 Years of Follow-up. <i>Stem Cells Translational Medicine</i> , 2021, 10, 1530-1543.	1.6	16
36	Essential Role of STAT3 Signaling in Hair Follicle Homeostasis. <i>Frontiers in Immunology</i> , 2021, 12, 663177.	2.2	7

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37	ACC1-expressing pathogenic T helper 2 cell populations facilitate lung and skin inflammation in mice. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	16
38	Acsbg1-dependent mitochondrial fitness is a metabolic checkpoint for tissue Treg cell homeostasis. <i>Cell Reports</i> , 2021, 37, 109921.	2.9	15
39	Functional Roles for CD26/DPP4 in Mediating Inflammatory Responses of Pulmonary Vascular Endothelial Cells. <i>Cells</i> , 2021, 10, 3508.	1.8	9
40	Prognostic factors for survival of herpes simplex virus-associated hemophagocytic lymphohistiocytosis. <i>International Journal of Hematology</i> , 2020, 111, 131-136.	0.7	7
41	Cysteinyl leukotriene metabolism of human eosinophils in allergic disease. <i>Allergology International</i> , 2020, 69, 28-34.	1.4	24
42	Human gain-of-function <i>STAT1</i> mutation disturbs IL-17 immunity in mice. <i>International Immunology</i> , 2020, 32, 259-272.	1.8	20
43	HaloTag-based conjugation of proteins to barcoding-oligonucleotides. <i>Nucleic Acids Research</i> , 2020, 48, e8-e8.	6.5	14
44	Novel HADHB mutations in a patient with mitochondrial trifunctional protein deficiency. <i>Human Genome Variation</i> , 2020, 7, 10.	0.4	2
45	Discovery of Candidate Stool Biomarker Proteins for Biliary Atresia Using Proteome Analysis by Data-Independent Acquisition Mass Spectrometry. <i>Proteomes</i> , 2020, 8, 36.	1.7	2
46	PD-L1 Expression Affects Neoantigen Presentation. <i>IScience</i> , 2020, 23, 101238.	1.9	9
47	Dysregulated metabolism of polyunsaturated fatty acids in eosinophilic allergic diseases. <i>Prostaglandins and Other Lipid Mediators</i> , 2020, 150, 106477.	1.0	14
48	Generation and Profiling of 2,135 Human ESC Lines for the Systematic Analyses of Cell States Perturbed by Inducing Single Transcription Factors. <i>Cell Reports</i> , 2020, 31, 107655.	2.9	28
49	Whole-Exome Sequencing-Based Approach for Germline Mutations in Patients with Inborn Errors of Immunity. <i>Journal of Clinical Immunology</i> , 2020, 40, 729-740.	2.0	20
50	ISSAID/EMQN Best Practice Guidelines for the Genetic Diagnosis of Monogenic Autoinflammatory Diseases in the Next-Generation Sequencing Era. <i>Clinical Chemistry</i> , 2020, 66, 525-536.	1.5	43
51	Autosomal recessive complete <i>STAT1</i> deficiency caused by compound heterozygous intronic mutations. <i>International Immunology</i> , 2020, 32, 663-671.	1.8	26
52	Clinical characteristics and treatment of 50 cases of Blau syndrome in Japan confirmed by genetic analysis of the <i>NOD2</i> mutation. <i>Annals of the Rheumatic Diseases</i> , 2020, 79, 1492-1499.	0.5	47
53	Microfluidic Immunoassays for Time-Resolved Measurement of Protein Secretion from Single Cells. <i>Annual Review of Analytical Chemistry</i> , 2020, 13, 67-84.	2.8	10
54	Simple and Sensitive Analysis for Dried Blood Spot Proteins by Sodium Carbonate Precipitation for Clinical Proteomics. <i>Journal of Proteome Research</i> , 2020, 19, 2821-2827.	1.8	14

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55	Clinical, Immunological, and Molecular Features of Severe Combined Immune Deficiency: A Multi-Institutional Experience From India. <i>Frontiers in Immunology</i> , 2020, 11, 619146.	2.2	31
56	Clinical and Genetic Profile of X-Linked Agammaglobulinemia: A Multicenter Experience From India. <i>Frontiers in Immunology</i> , 2020, 11, 612323.	2.2	16
57	CXCR4-Expressing Anti-CD25 CAR T-Cells Effectively Eliminate Human AML Cells In Vivo. <i>Blood</i> , 2020, 136, 35-36.	0.6	11
58	Japanese patients with mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency: <i>in vitro</i> functional analysis of five novel HMGCS2 mutations. <i>Experimental and Therapeutic Medicine</i> , 2020, 20, 1-1.	0.8	3
59	Allergy and the rules of cell society: An approach by using omics measurements. <i>Nihon Shoni Arerugi Gakkaishi the Japanese Journal of Pediatric Allergy and Clinical Immunology</i> , 2020, 34, 32-36.	0.0	0
60	Genomic Analysis of Diffuse Large B Cell Lymphoma in a Japanese Cohort Using Targeted DNA Sequencing. <i>Blood</i> , 2020, 136, 14-15.	0.6	0
61	Proteogenomic Analyses of Cellular Lysates Using a Phenol-Guanidinium Thiocyanate Reagent. <i>Journal of Proteome Research</i> , 2019, 18, 301-308.	1.8	13
62	Hematopoietic stem cell transplantation for progressive combined immunodeficiency and lymphoproliferation in patients with activated phosphatidylinositol-3-OH kinase $\gamma$ syndrome type 1. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 266-275.	1.5	49
63	Antiviral activity of zinc sulfate against hepatitis A virus replication. <i>Future Virology</i> , 2019, 14, 399-406.	0.9	12
64	Autosomal dominant Hashimoto's thyroiditis with a mutation in <i>TNFAIP3</i> . <i>Clinical Pediatric Endocrinology</i> , 2019, 28, 91-96.	0.4	8
65	TGF- $\beta$ 2 signaling promotes tube-structure-forming growth in pancreatic duct adenocarcinoma. <i>Scientific Reports</i> , 2019, 9, 11247.	1.6	5
66	Mast cells play role in wound healing through the ZnT2/GPR39/IL-6 axis. <i>Scientific Reports</i> , 2019, 9, 10842.	1.6	28
67	Short DNA Probes Developed for Sample Tracking and Quality Assurance in Gene Panel Testing. <i>Journal of Molecular Diagnostics</i> , 2019, 21, 1079-1094.	1.2	3
68	A deep intronic mutation of c.1166-285T>G in SLC46A1 is shared by four unrelated Japanese patients with hereditary folate malabsorption (HFM). <i>Clinical Immunology</i> , 2019, 208, 108256.	1.4	10
69	Detection of MYD88 L265P mutation by next-generation deep sequencing in peripheral blood mononuclear cells of Waldenström's macroglobulinemia and IgM monoclonal gammopathy of undetermined significance. <i>PLoS ONE</i> , 2019, 14, e0221941.	1.1	15
70	PCR-free whole exome sequencing: Cost-effective and efficient in detecting rare mutations. <i>PLoS ONE</i> , 2019, 14, e0222562.	1.1	5
71	PLD4 is a genetic determinant to systemic lupus erythematosus and involved in murine autoimmune phenotypes. <i>Annals of the Rheumatic Diseases</i> , 2019, 78, 509-518.	0.5	36
72	Dysregulated fatty acid metabolism in nasal polyp-derived eosinophils from patients with chronic rhinosinusitis. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 2019, 74, 1113-1124.	2.7	52

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73	Inhibition of T cell activation and function by the adaptor protein CIN85. <i>Science Signaling</i> , 2019, 12, .	1.6	14
74	Low-frequency mosaicism in cryopyrin-associated periodic fever syndrome: mosaicism in systemic autoinflammatory diseases. <i>International Immunology</i> , 2019, 31, 649-655.	1.8	16
75	Eomes transcription factor is required for the development and differentiation of invariant NKT cells. <i>Communications Biology</i> , 2019, 2, 150.	2.0	31
76	Co-activation of macrophages and T cells contribute to chronic GVHD in human IL-6 transgenic humanised mouse model. <i>EBioMedicine</i> , 2019, 41, 584-596.	2.7	20
77	BTNL 2 germline variants may be involved in the pathogenesis of renal granuloma. <i>Pediatrics International</i> , 2019, 61, 834-836.	0.2	0
78	Optimization of Data-Independent Acquisition Mass Spectrometry for Deep and Highly Sensitive Proteomic Analysis. <i>International Journal of Molecular Sciences</i> , 2019, 20, 5932.	1.8	73
79	Recurrent Acute Abdomen as the Main Manifestation of Hereditary Angioedema. <i>Internal Medicine</i> , 2019, 58, 213-216.	0.3	5
80	Mosaicism of an ELANE Mutation in an Asymptomatic Mother. <i>Journal of Clinical Immunology</i> , 2019, 39, 106-111.	2.0	4
81	Genetic and transcriptional landscape of plasma cells in POEMS syndrome. <i>Leukemia</i> , 2019, 33, 1723-1735.	3.3	28
82	Biallelic GALM pathogenic variants cause a novel type of galactosemia. <i>Genetics in Medicine</i> , 2019, 21, 1286-1294.	1.1	40
83	National survey of Japanese patients with mevalonate kinase deficiency reveals distinctive genetic and clinical characteristics. <i>Modern Rheumatology</i> , 2019, 29, 181-187.	0.9	18
84	Human NK cell development in hIL-7 and hIL-15 knockin NOD/SCID/IL2rgKO mice. <i>Life Science Alliance</i> , 2019, 2, e201800195.	1.3	41
85	Whole-exome sequencing in unresectable pancreatic cancer patients with long-term survival.. <i>Journal of Clinical Oncology</i> , 2019, 37, 260-260.	0.8	0
86	Targeting Critical Kinases and Anti-Apoptotic Molecules Overcomes Steroid Resistance in Infant MLL-Rearranged Leukemia. <i>Blood</i> , 2019, 134, 3885-3885.	0.6	0
87	Identification of Protein Targets of 12/15-Lipoxygenase-Derived Lipid Electrophiles in Mouse Peritoneal Macrophages Using Omega-Alkynyl Fatty Acid. <i>ACS Chemical Biology</i> , 2018, 13, 887-893.	1.6	11
88	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. <i>Cell</i> , 2018, 172, 952-965.e18.	13.5	92
89	A novel truncating mutation in FLNA causes periventricular nodular heterotopia, Ehlers-Danlos-like collagenopathy and macrothrombocytopenia. <i>Brain and Development</i> , 2018, 40, 489-492.	0.6	17
90	Haploinsufficiency of A20 causes autoinflammatory and autoimmune disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 1485-1488.e11.	1.5	100

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91	NK cell and IFN signatures are positive prognostic biomarkers for resectable pancreatic cancer. <i>Biochemical and Biophysical Research Communications</i> , 2018, 495, 2058-2065.	1.0	33
92	Characterization of a large UNC13D gene duplication in a patient with familial hemophagocytic lymphohistiocytosis type 3. <i>Clinical Immunology</i> , 2018, 191, 63-66.	1.4	7
93	Deregulated Mucosal Immune Surveillance through Gut-Associated Regulatory T Cells and PD-1+ T Cells in Human Colorectal Cancer. <i>Journal of Immunology</i> , 2018, 200, 3291-3303.	0.4	28
94	Human CTL-based functional analysis shows the reliability of a munc13-4 protein expression assay for FHL3 diagnosis. <i>Blood</i> , 2018, 131, 2016-2025.	0.6	13
95	Improving the efficiency of gene insertion in a human artificial chromosome vector and its transfer in human-induced pluripotent stem cells. <i>Biology Methods and Protocols</i> , 2018, 3, bpy013.	1.0	11
96	Late-onset ornithine transcarbamylase deficiency caused by a somatic mosaic mutation. <i>Human Genome Variation</i> , 2018, 5, 22.	0.4	13
97	Generation of orthotopically functional salivary gland from embryonic stem cells. <i>Nature Communications</i> , 2018, 9, 4216.	5.8	97
98	Development of a NanoLC-MS/MS System Using a Nonporous Reverse Phase Column for Ultrasensitive Proteome Analysis. <i>Analytical Chemistry</i> , 2018, 90, 12334-12338.	3.2	17
99	Clinical and Immunological Characterization of ICF Syndrome in Japan. <i>Journal of Clinical Immunology</i> , 2018, 38, 927-937.	2.0	29
100	Obesity Drives STAT-1-Dependent NASH and STAT-3-Dependent HCC. <i>Cell</i> , 2018, 175, 1289-1306.e20.	13.5	252
101	Sox12 promotes T reg differentiation in the periphery during colitis. <i>Journal of Experimental Medicine</i> , 2018, 215, 2509-2519.	4.2	7
102	Variant PRC1 competes with retinoic acid-related signals to repress <i>Meis2</i> in distal forelimb bud. <i>Development (Cambridge)</i> , 2018, 145, .	1.2	15
103	Development of a molecular diagnostic test for Retinitis Pigmentosa in the Japanese population. <i>Japanese Journal of Ophthalmology</i> , 2018, 62, 451-457.	0.9	29
104	Comprehensive molecular diagnosis of Epstein-Barr virus-associated lymphoproliferative diseases using next-generation sequencing. <i>International Journal of Hematology</i> , 2018, 108, 319-328.	0.7	6
105	Assessing the Accuracy of Variant Detection in Cost-Effective Gene Panel Testing by Next-Generation Sequencing. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 572-582.	1.2	33
106	Challenges in Developing Protein Secretion Assays at a Single-Cell Level. <i>Methods in Molecular Biology</i> , 2018, 1808, 1-7.	0.4	4
107	Clinical and molecular features of X-linked hyper IgM syndrome – An experience from North India. <i>Clinical Immunology</i> , 2018, 195, 59-66.	1.4	16
108	Enhanced AKT Phosphorylation of Circulating B Cells in Patients With Activated PI3K $\gamma$ Syndrome. <i>Frontiers in Immunology</i> , 2018, 9, 568.	2.2	15

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109	Rescue of recurrent deep intronic mutation underlying cell type-dependent quantitative NEMO deficiency. <i>Journal of Clinical Investigation</i> , 2018, 129, 583-597.	3.9	38
110	IL-25 enhances follicular helper T-cell differentiation and function downstream of ASC12. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 288-291.e8.	1.5	11
111	Accurate clinical genetic testing for autoinflammatory diseases using the next-generation sequencing platform MiSeq. <i>Biochemistry and Biophysics Reports</i> , 2017, 9, 146-152.	0.7	17
112	Alternative pathway for the development of V $\alpha$ 14+ NKT cells directly from CD4 <sup>+</sup> CD8 <sup>+</sup> thymocytes that bypasses the CD4+CD8+ stage. <i>Nature Immunology</i> , 2017, 18, 274-282.	7.0	55
113	An Immunogram for the Cancer-Immunity Cycle: Towards Personalized Immunotherapy of Lung Cancer. <i>Journal of Thoracic Oncology</i> , 2017, 12, 791-803.	0.5	127
114	Prediction and prioritization of neoantigens: integration of RNA sequencing data with whole-exome sequencing. <i>Cancer Science</i> , 2017, 108, 170-177.	1.7	63
115	Molecular mechanism and structural basis of gain-of-function of STAT1 caused by pathogenic R274Q mutation. <i>Journal of Biological Chemistry</i> , 2017, 292, 6240-6254.	1.6	21
116	Critical involvement of ZEB2 in collagen fibrillogenesis: the molecular similarity between Mowat-Wilson syndrome and Ehlers-Danlos syndrome. <i>Scientific Reports</i> , 2017, 7, 46565.	1.6	14
117	Common Variable Immunodeficiency Caused by FANC Mutations. <i>Journal of Clinical Immunology</i> , 2017, 37, 434-444.	2.0	18
118	A CD57+ CTL Degranulation Assay Effectively Identifies Familial Hemophagocytic Lymphohistiocytosis Type 3 Patients. <i>Journal of Clinical Immunology</i> , 2017, 37, 92-99.	2.0	13
119	Mitochondrial Acid 5 (MA-5) Facilitates ATP Synthase Oligomerization and Cell Survival in Various Mitochondrial Diseases. <i>EBioMedicine</i> , 2017, 20, 27-38.	2.7	46
120	Infection Profile in Chronic Granulomatous Disease: a 23-Year Experience from a Tertiary Care Center in North India. <i>Journal of Clinical Immunology</i> , 2017, 37, 319-328.	2.0	41
121	Alanine-scanning mutagenesis of human signal transducer and activator of transcription 1 to estimate loss- or gain-of-function variants. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 232-241.	1.5	43
122	Overcoming mutational complexity in acute myeloid leukemia by inhibition of critical pathways. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	19
123	Requirement of zinc transporter ZIP10 for epidermal development: Implication of the ZIP10-p63 axis in epithelial homeostasis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, 12243-12248.	3.3	45
124	Immunodeficiency in Two Female Patients with Incontinentia Pigmenti with Heterozygous NEMO Mutation Diagnosed by LPS Unresponsiveness. <i>Journal of Clinical Immunology</i> , 2017, 37, 529-538.	2.0	12
125	Newborn screening for carnitine palmitoyltransferase II deficiency using (C16 + C18:1)/C2: Evaluation of additional indices for adequate sensitivity and lower false-positivity. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 67-75.	0.5	26
126	Heterozygous carriers of succinyl-CoA:3-oxoacid CoA transferase deficiency can develop severe ketoacidosis. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 845-852.	1.7	12



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127	EPC1/TIP60-Mediated Histone Acetylation Facilitates Spermiogenesis in Mice. <i>Molecular and Cellular Biology</i> , 2017, 37, .	1.1	33
128	A deletion in the intergenic region upstream of <i>Ednrb</i> causes head spot in the rat strain KFRS4/Kyo. <i>BMC Genetics</i> , 2017, 18, 29.	2.7	8
129	A novel <i>KCNQ1</i> nonsense variant in the isoform-specific first exon causes both jervell and Lange-Nielsen syndrome 1 and long QT syndrome 1: a case report. <i>BMC Medical Genetics</i> , 2017, 18, 66.	2.1	10
130	Identification of a High-Frequency Somatic <i>NLR4</i> Mutation as a Cause of Autoinflammation by Pluripotent Cell-Based Phenotype Dissection. <i>Arthritis and Rheumatology</i> , 2017, 69, 447-459.	2.9	106
131	A Japanese family case with juvenile onset Behçet's disease caused by <i>NFAIP3</i> mutation. <i>Allergology International</i> , 2017, 66, 146-148.	1.4	48
132	Generation of safe and therapeutically effective human induced pluripotent stem cell-derived hepatocyte-like cells for regenerative medicine. <i>Hepatology Communications</i> , 2017, 1, 1058-1069.	2.0	57
133	<i>Rbm10</i> regulates inflammation development via alternative splicing of <i>Dnmt3b</i> . <i>International Immunology</i> , 2017, 29, 581-591.	1.8	31
134	Time evolution of microenvironment around cells regulated by the secretion activity and culture density of the cells. , 2017, , .		0
135	Widespread <i>Molluscum Contagiosum</i> with Atopic Dermatitis-like Skin Manifestations. <i>Acta Dermato-Venereologica</i> , 2017, 97, 291-292.	0.6	1
136	Nationwide survey of Baller-Gerold syndrome in Japanese population. <i>Molecular Medicine Reports</i> , 2017, 15, 3222-3224.	1.1	10
137	<i>Pyoderma Gangrenosum</i> , Acne and Suppurative Hidradenitis Syndrome Treated with Granulocyte and Monocyte Adsorption Apheresis. <i>Acta Dermato-Venereologica</i> , 2017, 97, 275-276.	0.6	10
138	Mapping the Interactome of a Major Mammalian Endoplasmic Reticulum Heat Shock Protein 90. <i>PLoS ONE</i> , 2017, 12, e0169260.	1.1	20
139	<i>PCGF6-PRC1</i> suppresses premature differentiation of mouse embryonic stem cells by regulating germ cell-related genes. <i>ELife</i> , 2017, 6, .	2.8	99
140	AB076. Heterozygous carriers of succinyl-CoA:3-oxoacid CoA transferase deficiency can develop severe ketoacidosis. <i>Annals of Translational Medicine</i> , 2017, 5, AB076-AB076.	0.7	1
141	Design and application of a target capture sequencing of exons and conserved non-coding sequences for the rat. <i>BMC Genomics</i> , 2016, 17, 593.	1.2	12
142	Clinical and Genetic Features of Patients With <i>TNFRSF1A</i> Variants in Japan: Findings of a Nationwide Survey. <i>Arthritis and Rheumatology</i> , 2016, 68, 2760-2771.	2.9	21
143	Invasive Bacterial Infection in Patients with Interleukin-1 Receptor-associated Kinase 4 Deficiency. <i>Medicine (United States)</i> , 2016, 95, e2437.	0.4	12
144	Establishment of a new three-dimensional human epidermal model reconstructed from plucked hair follicle-derived keratinocytes. <i>Experimental Dermatology</i> , 2016, 25, 903-906.	1.4	9

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145	Successful reduced-intensity stem cell transplantation for <sc>GATA</sc>2 deficiency before progression of advanced <sc>MDS</sc>. <i>Pediatric Transplantation</i> , 2016, 20, 333-336.	0.5	20
146	Genetic, immunological, and clinical features of patients with bacterial and fungal infections due to inherited IL-17RA deficiency. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E8277-E8285.	3.3	137
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