## Akira Oka

## List of Publications by Year in descending order

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516710 477307 1,066 68 16 29 h-index citations g-index papers 70 70 70 2390 all docs docs citations times ranked citing authors

| #  | Article  | IF   | Citations |
|----|--|------|-----------|
| 1  | Integrated genetic and epigenetic analysis defines novel molecular subgroups in rhabdomyosarcoma.<br>Nature Communications, 2015, 6, 7557.   | 12.8 | 149       |
| 2  | Recurrent SPI1 (PU.1) fusions in high-risk pediatric T cell acute lymphoblastic leukemia. Nature Genetics, 2017, 49, 1274-1281.  | 21.4 | 100       |
| 3  | Biallelic <i>DICER1</i> Mutations in Sporadic Pleuropulmonary Blastoma. Cancer Research, 2014, 74, 2742-2749.  | 0.9  | 67        |
| 4  | Autologous cord blood cell therapy for neonatal hypoxic-ischaemic encephalopathy: a pilot study for feasibility and safety. Scientific Reports, 2020, 10, 4603.  | 3.3  | 62        |
| 5  | Epidemiological changes of acute encephalopathy in Japan based on national surveillance for 2014–2017. Brain and Development, 2020, 42, 508-514.   | 1.1  | 41        |
| 6  | Long-term ventilator support in patients with Werdnig-Hoffmann disease. Pediatrics International, 2000, 42, 359-363.   | 0.5  | 34        |
| 7  | Integrated multiomics analysis of hepatoblastoma unravels its heterogeneity and provides novel druggable targets. Npj Precision Oncology, 2020, 4, 20.   | 5.4  | 30        |
| 8  | Contribution of gene mutations to Silver-Russell syndrome phenotype: multigene sequencing analysis in 92 etiology-unknown patients. Clinical Epigenetics, 2020, 12, 86.  | 4.1  | 29        |
| 9  | Amnionless-mediated glycosylation is crucial for cell surface targeting of cubilin in renal and intestinal cells. Scientific Reports, 2018, 8, 2351.   | 3.3  | 27        |
| 10 | <scp>NOTCH</scp> 1 pathway activating mutations and clonal evolution in pediatric Tâ€eell acute lymphoblastic leukemia. Cancer Science, 2019, 110, 784-794.  | 3.9  | 26        |
| 11 | Integrated Molecular Characterization of the Lethal Pediatric Cancer Pancreatoblastoma. Cancer Research, 2018, 78, 865-876.  | 0.9  | 25        |
| 12 | Association of the Emergence of Acyclovir-Resistant Herpes Simplex Virus Type 1 With Prognosis in Hematopoietic Stem Cell Transplantation Patients. Journal of Infectious Diseases, 2017, 215, 865-873.        | 4.0  | 23        |
| 13 | Molecular and clinical analyses of two patients with UPD(16)mat detected by screening 94 patients with Silver-Russell syndrome phenotype of unknown aetiology. Journal of Medical Genetics, 2019, 56, 413-418. | 3.2  | 23        |
| 14 | Identification of the genetic and clinical characteristics of neuroblastomas using genome-wide analysis. Oncotarget, 2017, 8, 107513-107529.   | 1.8  | 23        |
| 15 | Fever pattern and Câ€reactive protein predict response to rescue therapy in Kawasaki disease. Pediatrics International, 2016, 58, 180-184.   | 0.5  | 19        |
| 16 | Effect of i.v. immunoglobulin in the first 4 days of illness in Kawasaki disease. Pediatrics International, 2018, 60, 334-341.   | 0.5  | 19        |
| 17 | Epithelial protein lost in neoplasm modulates platelet-derived growth factor–mediated adhesion and motility of mesangial cells. Kidney International, 2014, 86, 548-557.                                       | 5.2  | 18        |
| 18 | Holoprosencephaly with cerebellar vermis hypoplasia in 13q deletion syndrome: Critical region for cerebellar dysgenesis within 13q32.2q34. Brain and Development, 2015, 37, 714-718.                           | 1.1  | 17        |

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|----|--|-----|-----------|
| 19 | LOX-1 Is a Novel Therapeutic Target in Neonatal Hypoxic-Ischemic Encephalopathy. American Journal of Pathology, 2014, 184, 1843-1852.  | 3.8 | 16        |
| 20 | Detection of Hereditary 1,25-Hydroxyvitamin D-Resistant Rickets Caused by Uniparental Disomy of Chromosome 12 Using Genome-Wide Single Nucleotide Polymorphism Array. PLoS ONE, 2015, 10, e0131157.                  | 2.5 | 15        |
| 21 | Integrated genetic and epigenetic analysis revealed heterogeneity of acute lymphoblastic leukemia in Down syndrome. Cancer Science, 2019, 110, 3358-3367.  | 3.9 | 15        |
| 22 | Functional analyses of a novel missense and other mutations of the vitamin D receptor in association with alopecia. Scientific Reports, 2017, 7, 5102.   | 3.3 | 14        |
| 23 | DNA methylation-based classification reveals difference between pediatric T-cell acute lymphoblastic leukemia and normal thymocytes. Leukemia, 2020, 34, 1163-1168.  | 7.2 | 14        |
| 24 | A Japanese patient with RAD51 â€associated Fanconi anemia. American Journal of Medical Genetics, Part A, 2019, 179, 900-902.   | 1.2 | 13        |
| 25 | Altered expression of Crb2 in podocytes expands a variation of CRB2 mutations in steroid-resistant nephrotic syndrome. Pediatric Nephrology, 2017, 32, 801-809.  | 1.7 | 12        |
| 26 | Accelerated Cardiomyocyte Proliferation in the Heart of a Neonate With LEOPARD Syndrome-Associated Fatal Cardiomyopathy. Circulation: Heart Failure, 2018, 11, e004660.  | 3.9 | 12        |
| 27 | A Qualitative Assessment of Adolescent Girls' Perception of Living with Congenital Heart Disease: Focusing on Future Pregnancies and Childbirth. Journal of Pediatric Nursing, 2018, 38, e12-e18.                    | 1.5 | 11        |
| 28 | A case of malignant rhabdoid tumor mimicking yolk sac tumor. Pediatric Blood and Cancer, 2019, 66, e27784.   | 1.5 | 11        |
| 29 | Case-control association study of rare nonsynonymous variants of SCN1A and KCNQ2 in acute encephalopathy with biphasic seizures and late reduced diffusion. Journal of the Neurological Sciences, 2020, 414, 116808. | 0.6 | 11        |
| 30 | Association of high-risk neuroblastoma classification based on expression profiles with differentiation and metabolism. PLoS ONE, 2021, 16, e0245526.  | 2.5 | 11        |
| 31 | Thermolabile polymorphism of carnitine palmitoyltransferase 2: A genetic risk factor of overall acute encephalopathy. Brain and Development, 2019, 41, 862-869.  | 1.1 | 10        |
| 32 | Leaky splicing variant in sepiapterin reductase deficiency. Neurology: Genetics, 2019, 5, e319.  | 1.9 | 10        |
| 33 | Direct hyperbilirubinemia in infants with congenital heart disease. Pediatrics International, 2018, 60, 179-182.   | 0.5 | 9         |
| 34 | Altered Expression of Astrocyte-Related Receptors and Channels Correlates With Epileptogenesis in Hippocampal Sclerosis. Pediatric and Developmental Pathology, 2019, 22, 532-539.                                   | 1.0 | 9         |
| 35 | Comprehensive genetic analysis of pediatric germ cell tumors identifies potential drug targets. Communications Biology, 2020, 3, 544.  | 4.4 | 9         |
| 36 | MLC1 mutations in Japanese patients with megalencephalic leukoencephalopathy with subcortical cysts. Human Genome Variation, 2014, 1, 14019.   | 0.7 | 8         |

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|----|--|-----|-----------|
| 37 | Systemic lupus erythematosus presenting with mixedâ€type fulminant autoimmune hemolytic anemia. Pediatrics International, 2016, 58, 527-530.   | 0.5 | 8         |
| 38 | Noonan syndromeâ€essociated biallelic <i>LZTR1</i> mutations cause cardiac hypertrophy and vascular malformations in zebrafish. Molecular Genetics & Enomic Medicine, 2020, 8, e1107.  | 1.2 | 8         |
| 39 | Association of human cytomegalovirus (HCMV) neutralizing antibodies with antibodies to the HCMV glycoprotein complexes. Virology Journal, 2020, 17, 120.   | 3.4 | 8         |
| 40 | Clinical and genetic characterization of nephropathy in patients with nail-patella syndrome. European Journal of Human Genetics, 2020, 28, 1414-1421.  | 2.8 | 8         |
| 41 | A Nationwide Epidemiological Survey of Adolescent Patients With Diverse Symptoms Similar to Those Following Human Papillomavirus Vaccination: Background Prevalence and Incidence for Considering Vaccine Safety in Japan. Journal of Epidemiology, 2022, 32, 34-43. | 2.4 | 8         |
| 42 | Afadin is localized at cell–cell contact sites in mesangial cells and regulates migratory polarity. Laboratory Investigation, 2016, 96, 49-59.   | 3.7 | 7         |
| 43 | Efficacy and safety of valganciclovir in patients with symptomatic congenital cytomegalovirus disease. Medicine (United States), 2020, 99, e19765.   | 1.0 | 7         |
| 44 | Medical, welfare, and educational challenges and psychological distress in parents caring for an individual with 22q11.2 deletion syndrome: A crossâ€sectional survey in Japan. American Journal of Medical Genetics, Part A, 2022, 188, 37-45.                      | 1.2 | 7         |
| 45 | Toward coâ€production of research in 22q11.2 deletion syndrome: Research needs from the caregiver's perspective. Psychiatry and Clinical Neurosciences, 2020, 74, 626-627.   | 1.8 | 7         |
| 46 | Novel DHCR7 mutation in a case of Smith–Lemli–Opitz syndrome showing 46,XY disorder of sex development. Human Genome Variation, 2017, 4, 17015.  | 0.7 | 6         |
| 47 | Nonosmotic secretion of arginine vasopressin and salt loss in hyponatremia in Kawasaki disease.<br>Pediatrics International, 2020, 62, 363-370.  | 0.5 | 6         |
| 48 | Correlation between fetal heart rate evolution patterns and magnetic resonance imaging findings in severe cerebral palsy: AÂlongitudinal study. BJOG: an International Journal of Obstetrics and Gynaecology, 2022, 129, 1574-1582.                                  | 2.3 | 6         |
| 49 | Deletion in the Cobalamin Synthetase W Domain–Containing Protein 1 Gene Is associated with Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2020, 31, 139-147.   | 6.1 | 5         |
| 50 | The cellular model of albumin endocytosis uncovers link between membrane and nuclear proteins. Journal of Cell Science, 2020, 133, .   | 2.0 | 5         |
| 51 | Relationship between post-IVIG IgG levels and clinical outcomes in Kawasaki disease patients: new insight into the mechanism of action of IVIG. Clinical Rheumatology, 2020, 39, 3747-3755.  | 2.2 | 5         |
| 52 | Brain edema with clasmatodendrosis complicating ataxia telangiectasia. Brain and Development, 2017, 39, 629-632.   | 1.1 | 4         |
| 53 | Incomplete cryptic splicing by an intronic mutation of OCRL in patients with partial phenotypes of Lowe syndrome. Journal of Human Genetics, 2020, 65, 831-839.  | 2.3 | 4         |
| 54 | Longitudinal Trends of Prevalence of Neutralizing Antibody against Human Cytomegalovirus over the Past 30 Years in Japanese Women. Japanese Journal of Infectious Diseases, 2022, 75, 496-503.   | 1.2 | 4         |

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|----|--|-----|-----------|
| 55 | Preferential involvement of U-fibers in human herpesvirus 6-associated acute encephalopathy. Annals of Neurology, 1999, 45, 684-684.   | 5.3 | 2         |
| 56 | <i>CBFA2T3â€GLIS2</i> àê€positive acute megakaryoblastic leukemia in a patient with Down syndrome.<br>Pediatric Blood and Cancer, 2020, 67, e28055.  | 1.5 | 2         |
| 57 | Rituximab-induced serum sickness in a 6-year-old boy with steroid-dependent nephrotic syndrome. CEN Case Reports, 2020, 9, 173-176.  | 0.9 | 2         |
| 58 | Effectiveness of Pediatric Teleconsultation to Prevent Skin Conditions in Infants and Reduce Parenting Stress in Mothers: Randomized Controlled Trial. JMIR Pediatrics and Parenting, 2022, 5, e27615. | 1.6 | 2         |
| 59 | Partial monosomy of 10p and duplication of another chromosome in two patients. Pediatrics International, 2017, 59, 99-102.   | 0.5 | 1         |
| 60 | Early-onset Marfan Syndrome Caused by a Splicing Mutation of <i>FBN1</i> Exon 29: A Case Report. Nihon Shoni Junkanki Gakkai Zasshi = Pediatric Cardiology and Cardiac Surgery, 2018, 34, 77-83.       | 0.0 | 1         |
| 61 | New ocular movement detector system as a communication tool in ventilatorâ€assisted<br>Werdnigâ€Hoffmann disease. Developmental Medicine and Child Neurology, 2000, 42, 61-64.                         | 2.1 | 0         |
| 62 | Genetic Landscapes Of Childhood T-Cell Acute Lymphoblastic Leukemia. Blood, 2013, 122, 3786-3786.  | 1.4 | 0         |
| 63 | TAL1 and MYB Abnormalities in Childhood T-Cell Acute Lymphoblastic Leukemia. Blood, 2015, 126, 2628-2628.  | 1.4 | 0         |
| 64 | Identifications of Highly Aggressive Phenotype with SPI1 Overexpression in Pediatric T Cell Acute Lymphoblastic Leukemia/Lymphoma. Blood, 2016, 128, 909-909.  | 1.4 | 0         |
| 65 | Gene Expression Profiles and Methylation Analysis in Down Syndrome Related Acute Lymphoblastic Leukemia. Blood, 2016, 128, 4084-4084.  | 1.4 | 0         |
| 66 | Distribution and Clinical Features of NOTCH1 Signaling Activating Alterations in Pediatric T-Cell Acute Lymphoblastic Leukemia (T-ALL). Blood, 2018, 132, 4089-4089.                                   | 1.4 | 0         |
| 67 | Comprehensive Genomic Analysis Identified Acute Lymphoblastic Leukemia in Down Syndrome Was Highly Heterogeneous with the High Prevalence of Ph-like Signature. Blood, 2018, 132, 2817-2817.           | 1.4 | 0         |
| 68 | Neonatal Enterovirus Myocarditis: A Case Report. Nihon Shoni Junkanki Gakkai Zasshi = Pediatric Cardiology and Cardiac Surgery, 2019, 35, 284-289.   | 0.0 | 0         |