

Hideaki Sawai

List of Publications by Year in descending order

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Version: 2024-02-01

62
papers

1,075
citations

471061

17
h-index

476904

29
g-index

65
all docs

65
docs citations

65
times ranked

1233
citing authors

#	ARTICLE	IF	CITATIONS
1	Statin treatment rescues FGFR3 skeletal dysplasia phenotypes. <i>Nature</i> , 2014, 513, 507-511.	13.7	186
2	Nationwide demonstration project of next-generation sequencing of cell-free DNA in maternal plasma in Japan: 1-year experience. <i>Prenatal Diagnosis</i> , 2015, 35, 331-336.	1.1	59
3	Fetal cell-free DNA fraction in maternal plasma is affected by fetal trisomy. <i>Journal of Human Genetics</i> , 2016, 61, 647-652.	1.1	59
4	Prevalence of c.1559delT in ALPL, a common mutation resulting in the perinatal (lethal) form of hypophosphatasia in Japanese and effects of the mutation on heterozygous carriers. <i>Journal of Human Genetics</i> , 2011, 56, 166-168.	1.1	57
5	Live births from isolated primary/early secondary follicles following a multistep culture without organ culture in mice. <i>Reproduction</i> , 2013, 146, 37-47.	1.1	45
6	Current status of non-invasive prenatal testing in Japan. <i>Journal of Obstetrics and Gynaecology Research</i> , 2017, 43, 1245-1255.	0.6	40
7	Prenatal diagnosis of thanatophoric dysplasia by mutational analysis of the fibroblast growth factor receptor 3 gene and a proposed correction of previously published PCR results. , 1999, 19, 21-24.		36
8	Modeling type II collagenopathy skeletal dysplasia by directed conversion and induced pluripotent stem cells. <i>Human Molecular Genetics</i> , 2015, 24, 299-313.	1.4	35
9	Cloning and expression of the rat class I MHC gene RT1.A l. <i>Immunogenetics</i> , 1994, 39, 447-447.	1.2	30
10	Low prevalence of genetic prenatal diagnosis in Japan. <i>Prenatal Diagnosis</i> , 2011, 31, 1007-1009.	1.1	29
11	Clinical Practice Guidelines for Hypophosphatasia*. <i>Clinical Pediatric Endocrinology</i> , 2020, 29, 9-24.	0.4	28
12	Classification of factors involved in nonreportable results of noninvasive prenatal testing (NIPT) and prediction of success rate of second NIPT. <i>Prenatal Diagnosis</i> , 2019, 39, 100-106.	1.1	27
13	Multiple TL-like loci in the grc-G/C region of the rat. <i>Immunogenetics</i> , 1994, 39, 301-315.	1.2	23
14	Fetal cell-free DNA fraction in maternal plasma for the prediction of hypertensive disorders of pregnancy. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2018, 224, 165-169.	0.5	22
15	Mutation analysis of <i>SOX9</i> and single copy number variant analysis of the upstream region in eight patients with campomelic dysplasia and acampomelic campomelic dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2882-2885.	0.7	20
16	Retrospective details of false-positive and false-negative results in non-invasive prenatal testing for fetal trisomies 21, 18 and 13. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2021, 256, 75-81.	0.5	20
17	Birth of healthy neonates after intracytoplasmic injection of ejaculated or testicular spermatozoa from men with nonmosaic Klinefelter's syndrome: a report of 2 cases. <i>Journal of reproductive medicine, The</i> , 2004, 49, 126-30.	0.2	20
18	A survey on awareness of genetic counseling for non-invasive prenatal testing: the first year experience in Japan. <i>Journal of Human Genetics</i> , 2016, 61, 995-1001.	1.1	19

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19	Prenatal Diagnosis of Thanatophoric Dysplasia by 3-D Helical Computed Tomography and Genetic Analysis. <i>Fetal Diagnosis and Therapy</i> , 2008, 24, 420-424.	0.6	18
20	Severe perinatal hypophosphatasia due to homozygous deletion of T at nucleotide 1559 in the tissue nonspecific alkaline phosphatase gene. <i>Prenatal Diagnosis</i> , 2003, 23, 743-746.	1.1	15
21	Chromosome abnormalities diagnosed in utero: a Japanese study of 28‰983 amniotic fluid specimens collected before 22 weeks gestations. <i>Journal of Human Genetics</i> , 2015, 60, 133-137.	1.1	15
22	Molecular analysis of the Y chromosome AZFc region in Japanese infertile males with spermatogenic defects. <i>Journal of Reproductive Immunology</i> , 2002, 53, 37-44.	0.8	14
23	Factors affecting parental decisions to terminate pregnancy in the presence of chromosome abnormalities: a Japanese multicenter study. <i>Prenatal Diagnosis</i> , 2016, 36, 1121-1126.	1.1	14
24	Bone Morphogenetic Protein-2 Counterregulates Interleukin-18 mRNA and Protein in MC3T3-E1 Mouse Osteoblastic Cells. <i>Connective Tissue Research</i> , 2006, 47, 124-132.	1.1	13
25	A compound heterozygote harboring novel and recurrentDTDST mutations with intermediate phenotype between atelosteogenesis type II and diastrophic dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1143-1147.	0.7	13
26	Prenatal diagnosis of Kniest dysplasia with three-dimensional helical computed tomography. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2011, 24, 1181-1184.	0.7	13
27	Prenatal diagnosis of short-rib polydactyly syndrome type 3 (Verma-Naumoff type) by three-dimensional helical computed tomography. <i>Journal of Obstetrics and Gynaecology Research</i> , 2011, 37, 151-155.	0.6	13
28	Nationwide radiation dose survey of computed tomography for fetal skeletal dysplasias. <i>Pediatric Radiology</i> , 2014, 44, 971-979.	1.1	13
29	A novel mutation Ser344Cys in <i>FGFR3</i> causes achondroplasia with severe platyspondyly. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2851-2854.	0.7	13
30	Maternal age-specific risk for trisomy 21 based on the clinical performance of NIPT and empirically derived NIPT age-specific positive and negative predictive values in Japan. <i>Journal of Human Genetics</i> , 2018, 63, 1035-1040.	1.1	13
31	Possible presence of O-linked carbohydrate in the human male reproductive tract CD52. <i>Journal of Reproductive Immunology</i> , 2004, 62, 91-100.	0.8	12
32	Novel missense mutation resulting in the substitution of tyrosine by cysteine at codon 597 of the type X collagen gene associated with Schmid metaphyseal chondrodysplasia. <i>Journal of Human Genetics</i> , 1998, 43, 259-261.	1.1	11
33	Radiographic and genetic diagnosis of sporadic hypochondroplasia early in the neonatal period. <i>Prenatal Diagnosis</i> , 2004, 24, 45-49.	1.1	11
34	Criteria for radiologic diagnosis of hypochondroplasia in neonates. <i>Pediatric Radiology</i> , 2016, 46, 513-518.	1.1	11
35	Qualitative investigation of the factors that generate ambivalent feelings in women who give birth after receiving negative results from non-invasive prenatal testing. <i>BMC Pregnancy and Childbirth</i> , 2020, 20, 112.	0.9	10
36	The Current State of Genetic Counseling Before and After Amniocentesis for Fetal Karyotyping in Japan: A Survey of Obstetric Hospital Clients of a Prenatal Testing Laboratory. <i>Journal of Genetic Counseling</i> , 2013, 22, 795-804.	0.9	9

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37	Feelings about pregnancy and mother-infant bonding as predictors of persistent psychological distress in the perinatal period: The Japan Environment and Children's Study. <i>Journal of Psychiatric Research</i> , 2021, 140, 132-140.	1.5	8
38	National survey of prevalence and prognosis of thanatophoric dysplasia in Japan. <i>Pediatrics International</i> , 2019, 61, 748-753.	0.2	7
39	Parental serum alkaline phosphatase activity as an auxiliary tool for prenatal diagnosis of hypophosphatasia. <i>Prenatal Diagnosis</i> , 2017, 37, 491-496.	1.1	6
40	The current status of umbilical cord blood collection in Japanese medical centers: Survey of obstetricians. <i>Transfusion and Apheresis Science</i> , 2011, 44, 263-268.	0.5	5
41	Paternal occupational exposure to chemicals and secondary sex ratio: results from the Japan Environment and Children's Study. <i>Lancet Planetary Health</i> , The, 2019, 3, e529-e538.	5.1	5
42	Direct Production of the Fab Fragment Derived From the Sperm Immobilizing Antibody Using Polymerase Chain Reaction and cDNA Expression Vectors. <i>American Journal of Reproductive Immunology</i> , 1995, 34, 26-34.	1.2	4
43	Pig zona pellucida 2 (pZP2) protein does not participate in zona pellucida formation in transgenic mice. <i>Reproduction</i> , 2006, 132, 455-464.	1.1	4
44	Platyspondylic lethal skeletal dysplasia San Diego type (thanatophoric dysplasia type 1) associated with trisomy 21 presenting with nuchal translucency: a case report. <i>Prenatal Diagnosis</i> , 2009, 29, 715-717.	1.1	4
45	A Case of Thanatophoric Dysplasia Type I with Fetal Hydrops in the First Trimester. <i>Case Reports in Obstetrics and Gynecology</i> , 2016, 2016, 1-4.	0.2	4
46	Survey of prenatal testing for genetic disorders in <scp>Japan</scp>: Recent report. <i>Journal of Obstetrics and Gynaecology Research</i> , 2016, 42, 375-379.	0.6	4
47	Nationwide survey for current clinical status of amniocentesis and maternal serum marker test in Japan. <i>Journal of Human Genetics</i> , 2016, 61, 879-884.	1.1	4
48	Association of ALPL variants with serum alkaline phosphatase and bone traits in the general Japanese population: The Nagahama Study. <i>Journal of Human Genetics</i> , 2020, 65, 337-343.	1.1	4
49	Follow-Up Study on Fetal CT Radiation Dose in Japan: Validating the Decrease in Radiation Dose. <i>American Journal of Roentgenology</i> , 2017, 208, 862-867.	1.0	3
50	Challenges in Managing Patients with Hereditary Cancer at Gynecological Services. <i>Obstetrics and Gynecology International</i> , 2019, 2019, 1-9.	0.5	3
51	Association between the serum insulin-like growth factor-1 concentration in the first trimester of pregnancy and postpartum depression. <i>Psychiatry and Clinical Neurosciences</i> , 2021, 75, 159-165.	1.0	3
52	Stable Production of Recombinant Human Sperm Immobilizing Antibody Using cDNA Expression Vectors. <i>American Journal of Reproductive Immunology</i> , 1993, 29, 100-108.	1.2	2
53	Isolation and characterization of a human sperm antigen gene h-Sp-1. <i>Journal of Developmental and Physical Disabilities</i> , 2003, 26, 226-235.	3.6	2
54	Recurrence of osteogenesis imperfecta due to maternal mosaicism of a novel <i>COL1A1</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2969-2971.	0.7	2

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55	A large seminoma occurring 20years after diagnosis of complete androgen insensitivity syndrome: A case report. <i>Gynecologic Oncology Case Reports</i> , 2013, 5, 16-18.	0.9	2
56	Evaluation of the clinical performance of noninvasive prenatal testing at a Japanese laboratory. <i>Journal of Obstetrics and Gynaecology Research</i> , 2021, 47, 3437-3446.	0.6	2
57	Development of individuals with thanatophoric dysplasia surviving beyond infancy. <i>Pediatrics International</i> , 2022, 64, .	0.2	2
58	Prenatal Diagnosis of Achondroplasia Using the Nested Polymerase Chain Reaction with Modified Primer Sets. <i>Fetal Diagnosis and Therapy</i> , 1996, 11, 407-413.	0.6	1
59	Reproductive success in patients with Hallermannâ€“Streiff syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2311-2313.	0.7	1
60	Development of an integrated support system for hereditary cancer and its impact on gynecologic services. <i>International Journal of Clinical Oncology</i> , 2014, 19, 1043-1051.	1.0	1
61	Cytogenetic Analysis of Spontaneous Miscarriages Using Long-Term Culturing of Chorionic Villi. <i>Journal of Fetal Medicine</i> , 2019, 6, 1-6.	0.1	1
62	A Case of a Pregnant Woman with Thrombosis in an Artificial Aortic Valve Resulting in Severe Cerebral Hemorrhage in the Newborn. <i>Case Reports in Obstetrics and Gynecology</i> , 2018, 2018, 1-3.	0.2	0