Claus-Eric Ott

List of Publications by Year
in descending order

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41	2,422	304701	233409
papers	citations	22 h-index	45 g-index

47	47	47	5214
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical Diagnostics in Human Genetics with Semantic Similarity Searches in Ontologies. American Journal of Human Genetics, 2009, 85, 457-464.	6.2	444
2	Complex Inheritance Pattern Resembling Autosomal Recessive Inheritance Involving a Microdeletion in Thrombocytopeniaâ€“Absent Radius Syndrome. American Journal of Human Genetics, 2007, 80, 232-240.	6.2	290
3	Deletions of chromosomal regulatory boundaries are associated with congenital disease. Genome Biology, 2014, 15, 423.	8.8	144
4	Duplications Involving a Conserved Regulatory Element Downstream of BMP2 Are Associated with Brachydactyly Type A2. American Journal of Human Genetics, 2009, 84, 483-492.	6.2	139
5	Acetylcholine Receptor Pathway Mutations Explain Various Fetal Akinesia Deformation Sequence Disorders. American Journal of Human Genetics, 2008, 82, 464-476.	6.2	124
6	miR-181a promotes osteoblastic differentiation through repression of TGF-Î² signaling molecules. International Journal of Biochemistry and Cell Biology, 2013, 45, 696-705.	2.8	120
7	A microduplication of the long range SHH limb regulator (ZRS) is associated with triphalangeal thumb-polysyndactyly syndrome. Journal of Medical Genetics, 2008, 45, 370-375.	3.2	118
8	MicroRNAs Differentially Expressed in Postnatal Aortic Development Downregulate Elastin via 3â€² UTR and Coding-Sequence Binding Sites. PLoS ONE, 2011, 6, e16250.	2.5	100
9	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
10	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	2.4	82
11	MiR-497â^¼195 Cluster MicroRNAs Regulate Osteoblast Differentiation by Targeting BMP Signaling. Journal of Bone and Mineral Research, 2015, 30, 796-808.	2.8	65
12	Composite transcriptome assembly of RNA-seq data in a sheep model for delayed bone healing. BMC Genomics, 2011, 12, 158.	2.8	63
13	Deletions of the RUNX2 gene are present in about 10% of individuals with cleidocranial dysplasia. Human Mutation, 2010, 31, E1587-E1593.	2.5	61
14	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
15	BMPs as new insulin sensitizers: enhanced glucose uptake in mature 3T3-L1 adipocytes via PPARÎ³ and GLUT4 upregulation. Scientific Reports, 2017, 7, 17192.	3.3	43
16	Severe congenital cutis laxa with cardiovascular manifestations due to homozygous deletions in ALDH18A1. Molecular Genetics and Metabolism, 2014, 112, 310-316.	1.1	41
17	Biaxial cell stimulation: A mechanical validation. Journal of Biomechanics, 2009, 42, 1692-1696.	2.1	39
18	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39

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19	The fibrillin-1 hypomorphic mgR/mgR murine model of Marfan syndrome shows severe elastolysis in all segments of the aorta. Journal of Vascular Surgery, 2013, 57, 1628-1636.e3.	1.1	36
20	Impaired proteoglycan glycosylation, elevated TGF-Î² signaling, and abnormal osteoblast differentiation as the basis for bone fragility in a mouse model for gerodermia osteodysplastica. PLoS Genetics, 2018, 14, e1007242.	3.5	36
21	Antagonism of GxxPG fragments ameliorates manifestations of aortic disease in Marfan syndrome mice. Human Molecular Genetics, 2013, 22, 433-443.	2.9	33
22	Promiscuous and Depolarization-Induced Immediate-Early Response Genes Are Induced by Mechanical Strain of Osteoblasts. Journal of Bone and Mineral Research, 2009, 24, 1247-1262.	2.8	26
23	Efficiency of Computer-Aided Facial Phenotyping (DeepGestalt) in Individuals With and Without a Genetic Syndrome: Diagnostic Accuracy Study. Journal of Medical Internet Research, 2020, 22, e19263.	4.3	26
24	Severe neuronopathic autosomal recessive osteopetrosis due to homozygous deletions affecting OSTM1. Bone, 2013, 55, 292-297.	2.9	22
25	Robinow syndrome: Phenotypic variability in a family with a novel intragenic ROR2 mutation. American Journal of Medical Genetics, Part A, 2008, 146A, 2804-2809.	1.2	19
26	Evaluation of the role of STAP1 in Familial Hypercholesterolemia. Scientific Reports, 2019, 9, 11995.	3.3	17
27	Novel variants in TECRL cause recessive inherited CPVT type 3 with severe and variable clinical symptoms. Journal of Cardiovascular Electrophysiology, 2020, 31, 1527-1535.	1.7	16
28	Indomethacin Prevents the Progression of Thoracic Aortic Aneurysm in Marfan Syndrome Mice. Aorta, 2013, 1, 5-12.	0.5	14
29	Transcriptional profiling of murine osteoblast differentiation based on RNA-seq expression analyses. Bone, 2018, 113, 29-40.	2.9	13
30	A novel mutation in CDH11, encoding cadherinâ€11, cause Branchioskeletogenital (Elsahyâ€Waters) syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2028-2033.	1.2	13
31	Microduplications upstream ofMSX2are associated with a phenocopy of cleidocranial dysplasia. Journal of Medical Genetics, 2012, 49, 437-441.	3.2	12
32	Improved bone defect healing by a superagonistic GDF5 variant derived from a patient with multiple synostoses syndrome. Bone, 2015, 73, 111-119.	2.9	12
33	Quantification and significance of fluid shear stress field in biaxial cell stretching device. Biomechanics and Modeling in Mechanobiology, 2011, 10, 559-564.	2.8	11
34	Differential effect of cataract-associated mutations in MAF on transactivation of MAF target genes. Molecular and Cellular Biochemistry, 2014, 396, 137-145.	3.1	11
35	Double NF1 Inactivation Affects Adrenocortical Function in NF1Prx1 Mice and a Human Patient. PLoS ONE, 2015, 10, e0119030.	2.5	10
36	The Interaction of BMP2â€Induced Defect Healing in Rat and Fixator Stiffness Modulates Matrix Alignment and Contraction. JBMR Plus, 2018, 2, 174-186.	2.7	7

#	Article	IF	Citations
37	On microRNA-214 suppressing osteogenic differentiation of C2C12 myoblast cells by targeting Osterix. Bone, 2013, 57, 325-327.	2.9	6
38	Prognostic significance of prenatal ultrasound in fetal arthrogryposis multiplex congenita. Archives of Gynecology and Obstetrics, 2021, 303, 943-953.	1.7	5
39	A machine learning-based screening tool for genetic syndromes in children. The Lancet Digital Health, 2022, 4, e295.	12.3	5
40	Brachydactyly Type C patient with compound heterozygosity for p.Gly319Val and p.Ile358Thr variants in the GDF5 proregion: benign variants or mutations?. Journal of Human Genetics, 2015, 60, 419-425.	2.3	4
41	Uncovering the Contribution of Moderate-Penetrance Susceptibility Genes to Breast Cancer by Whole-Exome Sequencing and Targeted Enrichment Sequencing of Candidate Genes in Women of European Ancestry. Cancers, 2022, 14, 3363.	3.7	2