

Eva Klopocki

List of Publications by Year in descending order

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107
papers

6,541
citations

87888

38
h-index

71685

76
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113
all docs

113
docs citations

113
times ranked

11112
citing authors

#	ARTICLE	IF	CITATIONS
1	Generation of the induced pluripotent stem cell line UKWNLi005-A derived from a patient with the GLA mutation c.376A→G of unknown pathogenicity in Fabry disease. <i>Stem Cell Research</i> , 2022, 61, 102747.	0.7	1
2	Generation of induced pluripotent stem cell (iPSC) lines carrying a heterozygous (UKWMPi002-A-1) and null mutant knockout (UKWMPi002-A-2) of Cadherin 13 associated with neurodevelopmental disorders using CRISPR/Cas9. <i>Stem Cell Research</i> , 2021, 51, 102169.	0.7	3
3	The importance of TNAP/Tnap for dental development in human cell culture and in zebrafish. <i>Bone Reports</i> , 2021, 14, 101006.	0.4	0
4	CRISPR/Cas9-edited PKP2 knock-out (JMUi001-A-2) and DSG2 knock-out (JMUi001-A-3) iPSC lines as an isogenic human model system for arrhythmogenic cardiomyopathy (ACM). <i>Stem Cell Research</i> , 2021, 53, 102256.	0.7	1
5	Exploration of zebrafish larvae as an alternative whole-animal model for nephrotoxicity testing. <i>Toxicology Letters</i> , 2021, 344, 69-81.	0.8	9
6	Generation of multiple human iPSC lines from peripheral blood mononuclear cells of two SLC2A3 deletion and two SLC2A3 duplication carriers. <i>Stem Cell Research</i> , 2021, 56, 102526.	0.7	0
7	Novel variants in <i>FERMT3</i> and <i>RASGRP2</i> Genetic linkage in Glanzmann-like bleeding disorders. <i>Pediatric Blood and Cancer</i> , 2020, 67, e28078.	1.5	6
8	New Insights on Genetic Diagnostics in Cardiomyopathy and Arrhythmia Patients Gained by Stepwise Exome Data Analysis. <i>Journal of Clinical Medicine</i> , 2020, 9, 2168.	2.4	4
9	Investigation of <i>alpl</i> expression and Tnap-activity in zebrafish implies conserved functions during skeletal and neuronal development. <i>Scientific Reports</i> , 2020, 10, 13321.	3.3	10
10	Heterogeneous phenotypes in families with duplications of the paternal allele within the imprinting center 1 (<i>H19</i> / <i>IGF2</i>) (<i>TSS</i> Δ <i>DMR</i>) in 11p15.5. <i>Clinical Genetics</i> , 2020, 98, 418-419.	2.0	3
11	Tissue-Nonspecific Alkaline Phosphatase A Gatekeeper of Physiological Conditions in Health and a Modulator of Biological Environments in Disease. <i>Biomolecules</i> , 2020, 10, 1648.	4.0	19
12	Generation of two patient-derived iPSC lines from siblings (LIBUCi001-A and LIBUCi002-A) and a genetically modified iPSC line (JMUi001-A-1) to mimic dilated cardiomyopathy with ataxia (DCMA) caused by a homozygous <i>DNAJC19</i> mutation. <i>Stem Cell Research</i> , 2020, 46, 101856.	0.7	5
13	A Novel Locus and Candidate Gene for Familial Developmental Dyslexia on Chromosome 4q. <i>Zeitschrift für Kinder- Und Jugendpsychiatrie Und Psychotherapie</i> , 2020, 48, 478-489.	0.7	5
14	Atypical 22q11.2 Microduplication with "Typical" Signs and Overgrowth. <i>Cytogenetic and Genome Research</i> , 2020, 160, 659-663.	1.1	1
15	ECM alterations in <i>Fndc3a</i> (Fibronectin Domain Containing Protein 3A) deficient zebrafish cause temporal fin development and regeneration defects. <i>Scientific Reports</i> , 2019, 9, 13383.	3.3	8
16	On the traces of <i>tcf12</i> : Investigation of the gene expression pattern during development and cranial suture patterning in zebrafish (<i>Danio rerio</i>). <i>PLoS ONE</i> , 2019, 14, e0218286.	2.5	6
17	Characterization of six Merkel cell polyomavirus-positive Merkel cell carcinoma cell lines: Integration pattern suggest that large T antigen truncating events occur before or during integration. <i>International Journal of Cancer</i> , 2019, 145, 1020-1032.	5.1	44
18	Biallelic mutation in <i>MYH7</i> and <i>MYBPC3</i> leads to severe cardiomyopathy with left ventricular noncompaction phenotype. <i>Human Mutation</i> , 2019, 40, 1101-1114.	2.5	22

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19	Generation of two induced pluripotent stem cell lines from skin fibroblasts of sisters carrying a c.1094C>A variation in the SCN10A gene potentially associated with small fiber neuropathy. <i>Stem Cell Research</i> , 2019, 35, 101396.	0.7	3
20	Biallelic intragenic deletion in MASP1 in an adult female with 3MC syndrome. <i>European Journal of Medical Genetics</i> , 2018, 61, 363-368.	1.3	17
21	Noncoding copy-number variations are associated with congenital limb malformation. <i>Genetics in Medicine</i> , 2018, 20, 599-607.	2.4	42
22	Use of Targeted High-Throughput Sequencing for Genetic Classification of Patients with Bleeding Diathesis and Suspected Platelet Disorder. <i>TH Open</i> , 2018, 02, e445-e454.	1.4	18
23	Generation of the human induced pluripotent stem cell line UKWNLI002-A from dermal fibroblasts of a woman with a heterozygous c.608 C>T (p.Thr203Met) mutation in exon 3 of the nerve growth factor gene potentially associated with hereditary sensory and autonomic neuropathy type 5. <i>Stem Cell Research</i> , 2018, 33, 171-174.	0.7	4
24	Interstitial Deletion of 5q22.2q23.1 Including APC and TSSK1B in a Patient with Adenomatous Polyposis and Asthenoteratozoospermia. <i>Molecular Syndromology</i> , 2018, 9, 235-240.	0.8	4
25	Recessive grey platelet-like syndrome with unaffected erythropoiesis in the absence of the splice isoform GF11B-p37. <i>Haematologica</i> , 2017, 102, e375-e378.	3.5	16
26	A novel two-nucleotide deletion in <i>HPS6</i> affects mepacrine uptake and platelet dense granule secretion in a family with Hermansky-Pudlak syndrome. <i>Pediatric Blood and Cancer</i> , 2017, 64, e26320.	1.5	10
27	Composition and dosage of a multipartite enhancer cluster control developmental expression of <i>Ihh</i> (Indian hedgehog). <i>Nature Genetics</i> , 2017, 49, 1539-1545.	21.4	107
28	Mutation c.943G>T (p.Ala315Ser) in <i>FGFR2</i> Causing a Mild Phenotype of Crouzon Craniofacial Dysostosis in a Three-Generation Family. <i>Molecular Syndromology</i> , 2017, 8, 93-97.	0.8	8
29	Exome sequencing identifies a nonsense mutation in <i>Fam46a</i> associated with bone abnormalities in a new mouse model for skeletal dysplasia. <i>Mammalian Genome</i> , 2016, 27, 111-121.	2.2	27
30	An Autosomal-Recessive GF11B Mutation Defines the Splice Isoform p37 As Essential for Biogenesis of Functional Human Platelets, but Dispensable for Erythropoiesis. <i>Blood</i> , 2016, 128, 2644-2644.	1.4	1
31	<i>DFNB16</i> is a frequent cause of congenital hearing impairment: implementation of <i>STRC</i> mutation analysis in routine diagnostics. <i>Clinical Genetics</i> , 2015, 87, 49-55.	2.0	86
32	Microdeletions on 6p22.3 are associated with mesomelic dysplasia Savarirayan type. <i>Journal of Medical Genetics</i> , 2015, 52, 476-483.	3.2	27
33	Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. <i>Cell</i> , 2015, 161, 1012-1025.	28.9	1,725
34	Molecular mechanism of <i>CHRD1</i> -mediated X-linked megalocornea in humans and in <i>Xenopus</i> model. <i>Human Molecular Genetics</i> , 2015, 24, 3119-3132.	2.9	24
35	Homozygous missense and nonsense mutations in <i>BMP1B</i> cause acromesomelic chondrodysplasia-type Grebe. <i>European Journal of Human Genetics</i> , 2014, 22, 726-733.	2.8	23
36	Microduplications encompassing the Sonic hedgehog limb enhancer <i>ZRS</i> are associated with Haas-type polysyndactyly and Laurin-Sandrow syndrome. <i>Clinical Genetics</i> , 2014, 86, 318-325.	2.0	72

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37	<i>De novo</i> partial deletion in <i>GRID2</i> presenting with complicated spastic paraplegia. Muscle and Nerve, 2014, 49, 289-292.	2.2	33
38	Large homozygous RAB3GAP1 gene microdeletion causes Warburg Micro Syndrome 1. Orphanet Journal of Rare Diseases, 2014, 9, 113.	2.7	5
39	X-linked congenital ptosis and associated intellectual disability, short stature, microcephaly, cleft palate, digital and genital abnormalities define novel Xq25q26 duplication syndrome. Human Genetics, 2014, 133, 625-638.	3.8	17
40	Partial trisomy 1q41–qter and partial trisomy 9pter–q21.32 in a newborn infant: An array CGH analysis and review. American Journal of Medical Genetics, Part A, 2014, 164, 490-494.	1.2	6
41	Deletions of exons with regulatory activity at the DYNC111 locus are associated with split-hand/split-foot malformation: array CGH screening of 134 unrelated families. Orphanet Journal of Rare Diseases, 2014, 9, 108.	2.7	43
42	Interstitial 12p deletion involving more than 40 genes in a patient with postnatal microcephaly, psychomotor delay, optic nerve atrophy, and facial dysmorphism. Meta Gene, 2014, 2, 72-82.	0.6	4
43	CNVs of noncoding cis-regulatory elements in human disease. Current Opinion in Genetics and Development, 2013, 23, 249-256.	3.3	43
44	A case of paroxysmal nocturnal hemoglobinuria caused by a germline mutation and a somatic mutation in PIGT. Blood, 2013, 122, 1312-1315.	1.4	77
45	Proximal and distal 15q25.2 microdeletions–genotype–phenotype delineation of two neurodevelopmental susceptibility loci. American Journal of Medical Genetics, Part A, 2013, 161, 218-224.	1.2	10
46	Cerebellar and posterior fossa malformations in patients with autism–associated chromosome 22q13 terminal deletion. American Journal of Medical Genetics, Part A, 2013, 161, 131-136.	1.2	65
47	Fine Mapping of the 1p36 Deletion Syndrome Identifies Mutation of PRDM16 as a Cause of Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 67-77.	6.2	164
48	Novel mutations of the <i><sc>PRKAR1A</sc></i> gene in–patients with acrodysostosis. Clinical Genetics, 2013, 84, 531-538.	2.0	27
49	Phenotypic variant of Brachydactyly-mental retardation syndrome in a family with an inherited interstitial 2q37.3 microdeletion including HDAC4. European Journal of Human Genetics, 2013, 21, 743-748.	2.8	46
50	Complex genetics of radial ray deficiencies: screening of a cohort of 54 patients. Genetics in Medicine, 2013, 15, 195-202.	2.4	15
51	Whole exome sequencing identified a novel zinc-finger gene<i>ZNF141</i> associated with autosomal recessive postaxial polydactyly type A. Journal of Medical Genetics, 2013, 50, 47-53.	3.2	51
52	Phenotypic overlap in the contribution of individual genes to CNV pathogenicity revealed by cross-species computational analysis of single-gene mutations in humans, mice and zebrafish. DMM Disease Models and Mechanisms, 2013, 6, 358-72.	2.4	43
53	Proximal microdeletions and microduplications of 1q21.1 contribute to variable abnormal phenotypes. European Journal of Human Genetics, 2012, 20, 754-761.	2.8	84
54	Two patterns of thrombopoietin signaling suggest no coupling between platelet production and thrombopoietin reactivity in thrombocytopenia-absent radii syndrome. Haematologica, 2012, 97, 73-81.	3.5	24

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55	Microduplications upstream of <i>MSX2</i> are associated with a phenocopy of cleidocranial dysplasia. <i>Journal of Medical Genetics</i> , 2012, 49, 437-441.	3.2	12
56	Duplications of <i>BHLHA9</i> are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. <i>Journal of Medical Genetics</i> , 2012, 49, 119-125.	3.2	81
57	Heterozygote <i>FANCD2</i> mutations associated with childhood T Cell ALL and testicular seminoma. <i>Familial Cancer</i> , 2012, 11, 661-665.	1.9	21
58	Left ventricular hypertrabeculation/noncompaction with epilepsy, other heart defects, minor facial anomalies and new copy number variants. <i>BMC Medical Genetics</i> , 2012, 13, 60.	2.1	3
59	Deletions in <i>PITX1</i> cause a spectrum of lower-limb malformations including mirror-image polydactyly. <i>European Journal of Human Genetics</i> , 2012, 20, 705-708.	2.8	63
60	Homeotic Arm-to-Leg Transformation Associated with Genomic Rearrangements at the <i>PITX1</i> Locus. <i>American Journal of Human Genetics</i> , 2012, 91, 629-635.	6.2	111
61	Das 2q37-Deletionssyndrom. <i>Medizinische Genetik</i> , 2012, 24, 40-47.	0.2	0
62	Copy-Number Variations, Noncoding Sequences, and Human Phenotypes. <i>Annual Review of Genomics and Human Genetics</i> , 2011, 12, 53-72.	6.2	53
63	Microdeletions of chromosome 7p21, including <i>TWIST1</i> , associated with significant microcephaly, facial dysmorphism, and short stature. <i>European Journal of Medical Genetics</i> , 2011, 54, 256-261.	1.3	9
64	Homozygous deletion of chromosome 15q13.3 including <i>CHRNA7</i> causes severe mental retardation, seizures, muscular hypotonia, and the loss of <i>KLF13</i> and <i>TRPM1</i> potentially cause macrocytosis and congenital retinal dysfunction in siblings. <i>European Journal of Medical Genetics</i> , 2011, 54, e441-e445.	1.3	47
65	The core <i>FOXP1</i> syndrome phenotype consists of postnatal microcephaly, severe mental retardation, absent language, dyskinesia, and corpus callosum hypogenesis. <i>Journal of Medical Genetics</i> , 2011, 48, 396-406.	3.2	220
66	Copy-Number Variations Involving the <i>IHH</i> Locus Are Associated with Syndactyly and Craniosynostosis. <i>American Journal of Human Genetics</i> , 2011, 88, 70-75.	6.2	89
67	Negative enrichment by immunomagnetic nanobeads for unbiased characterization of circulating tumor cells from peripheral blood of cancer patients. <i>Journal of Translational Medicine</i> , 2011, 9, 70.	4.4	162
68	Madelung deformity in a girl with a novel and de novo mutation in the <i>GNAS</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2566-2570.	1.2	11
69	Combined partial trisomy 11q and partial monosomy 10p in a 19-year-old female patient: Phenotypic and genotypic findings. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 3075-3081.	1.2	3
70	A <i>BACH2-BCL2L1</i> fusion gene resulting from a t(6;20)(q15;q11.2) chromosomal translocation in the lymphoma cell line BLUE1. <i>Genes Chromosomes and Cancer</i> , 2011, 50, 389-396.	2.8	22
71	Deletion and Point Mutations of <i>PTHLH</i> Cause Brachydactyly Type E. <i>American Journal of Human Genetics</i> , 2010, 86, 434-439.	6.2	127
72	Deletions of the <i>RUNX2</i> gene are present in about 10% of individuals with cleidocranial dysplasia. <i>Human Mutation</i> , 2010, 31, E1587-E1593.	2.5	61

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73	Identification of FOXP1 deletions in three unrelated patients with mental retardation and significant speech and language deficits. <i>Human Mutation</i> , 2010, 31, E1851-E1860.	2.5	130
74	Four unrelated patients with lubs Xâ€linked mental retardation syndrome and different Xq28 duplications. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 305-312.	1.2	27
75	Mesomelic dysplasia Kantaputra type is associated with duplications of the HOXD locus on chromosome 2q. <i>European Journal of Human Genetics</i> , 2010, 18, 1310-1314.	2.8	32
76	Analysis of relative gene dosage and expression differences of the paralogs RABL2A and RABL2B by Pyrosequencing. <i>Gene</i> , 2010, 455, 1-7.	2.2	10
77	The Abrogated Thrombopoietin (TPO) Signal Transduction In Pediatric Patients Suffering From Thrombocytopenia-Absent Radii Syndrome Is Restored In Adult Patients, Suggesting An Additional, c-Mpl-Jak2-Independent Mechanism for Platelet Biogenesis.. <i>Blood</i> , 2010, 116, 1561-1561.	1.4	0
78	Duplications of noncoding elements 5â€² of SOX9 are associated with brachydactyly-anonychia. <i>Nature Genetics</i> , 2009, 41, 862-863.	21.4	105
79	Duplications Involving a Conserved Regulatory Element Downstream of BMP2 Are Associated with Brachydactyly Type A2. <i>American Journal of Human Genetics</i> , 2009, 84, 483-492.	6.2	139
80	De novo 9â€Mb deletion of 6q23.2q24.1 disrupting the gene EYA4 in a patient with sensorineural hearing loss, cardiac malformation, and mental retardation. <i>European Journal of Medical Genetics</i> , 2009, 52, 450-453.	1.3	11
81	Expanded Clinical Spectrum in Hepatocyte Nuclear Factor 1B-Maturity-Onset Diabetes of the Young. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 2658-2664.	3.6	77
82	Gomez-Lopez-Hernandez syndrome (cerebello-trigeminal-dermal dysplasia): description of an additional case and review of the literature. <i>European Journal of Pediatrics</i> , 2008, 167, 123-126.	2.7	23
83	A further case of the recurrent 15q24 microdeletion syndrome, detected by array CGH. <i>European Journal of Pediatrics</i> , 2008, 167, 903-908.	2.7	47
84	Expanding the phenotype of alopeciaâ€contracturesâ€dwarfism mental retardation syndrome (ACD) Tj ETQq0 0 0 rgBT /Overlock 10 T <i>Pediatrics</i> , 2008, 167, 1057-1062.	2.7	7
85	Agenesis and dysgenesis of the corpus callosum: Clinical, genetic and neuroimaging findings in a series of 41 patients. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2501-2511.	1.2	148
86	Mutations of CASK cause an X-linked brain malformation phenotype with microcephaly and hypoplasia of the brainstem and cerebellum. <i>Nature Genetics</i> , 2008, 40, 1065-1067.	21.4	252
87	Triangular tibia with fibular aplasia associated with a microdeletion on 2q11.2 encompassing <i>LAF4</i>. <i>Clinical Genetics</i> , 2008, 74, 560-565.	2.0	28
88	Catelâ€Manzke syndrome: Two new patients and a critical review of the literature. <i>European Journal of Medical Genetics</i> , 2008, 51, 452-465.	1.3	32
89	A cryptic unbalanced translocation t(2;9)(p25.2;q34.3) causes the phenotype of 9q subtelomeric deletion syndrome and additional exophthalmos and joint contractures. <i>European Journal of Medical Genetics</i> , 2008, 51, 615-621.	1.3	5
90	<i>HNF1B</i> Abnormality (Mature-Onset Diabetes of the Young 5) in Children and Adolescents. <i>Diabetes Care</i> , 2008, 31, e83-e83.	8.6	9

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91	A microduplication of the long range SHH limb regulator (ZRS) is associated with triphalangeal thumb-polysyndactyly syndrome. <i>Journal of Medical Genetics</i> , 2008, 45, 370-375.	3.2	118
92	Complex Inheritance Pattern Resembling Autosomal Recessive Inheritance Involving a Microdeletion in Thrombocytopeniaâ€‘Absent Radius Syndrome. <i>American Journal of Human Genetics</i> , 2007, 80, 232-240.	6.2	290
93	Impact of Array Comparative Genomic Hybridizationâ€‘Derived Information on Genetic Counseling Demonstrated by Prenatal Diagnosis of the TAR (Thrombocytopenia-Absent-Radius) Syndromeâ€‘Associated Microdeletion 1q21.1. <i>American Journal of Human Genetics</i> , 2007, 81, 866-868.	6.2	17
94	Amplification and translocation of 3q26 with overexpression ofEVI1 in Fanconi anemia-derived childhood acute myeloid leukemia with biallelicFANCD1/BRCA2 disruption. <i>Genes Chromosomes and Cancer</i> , 2007, 46, 359-372.	2.8	28
95	Tandem duplication ofDMD exon 18 associated with epilepsy, macroglossia, and endocrinologic abnormalities. <i>Muscle and Nerve</i> , 2007, 35, 396-401.	2.2	7
96	Molecular cytogenetic characterisation of an interstitial deletion 12p detected by prenatal diagnosis. <i>Prenatal Diagnosis</i> , 2007, 27, 475-478.	2.3	11
97	Frequent loss of SFRP1 expression in multiple human solid tumours: association with aberrant promoter methylation in renal cell carcinoma. <i>Oncogene</i> , 2007, 26, 5680-5691.	5.9	127
98	Ulnarâ€‘mammary syndrome with dysmorphic facies and mental retardation caused by a novel 1.28 Mb deletion encompassing the TBX3 gene. <i>European Journal of Human Genetics</i> , 2006, 14, 1274-1279.	2.8	41
99	A complex phenotype with cystic renal disease. <i>Kidney International</i> , 2006, 70, 1656-1660.	5.2	14
100	Aberrant methylation of the Wnt antagonist SFRP1 in breast cancer is associated with unfavourable prognosis. <i>Oncogene</i> , 2006, 25, 3479-3488.	5.9	234
101	A novel 8 Mb interstitial deletion of chromosome 8p12-p21.2. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 873-877.	1.2	21
102	Systematic identification and molecular characterization of genes differentially expressed in breast and ovarian cancer. <i>Journal of Pathology</i> , 2005, 205, 21-28.	4.5	52
103	P4: Ulnar-mammary syndrome with mental retardation caused by a novel 0.8 Mb deletion encompassing the TBX3 gene. <i>European Journal of Medical Genetics</i> , 2005, 48, 450.	1.3	0
104	Loss of SFRP1 is associated with breast cancer progression and poor prognosis in early stage tumors. <i>International Journal of Oncology</i> , 2004, 25, 641.	3.3	62
105	Deletions of chromosome 8p and loss of sFRP1 expression are progression markers of papillary bladder cancer. <i>Laboratory Investigation</i> , 2004, 84, 465-478.	3.7	134
106	ITIH5, a novel member of the inter-Î±-trypsin inhibitor heavy chain family is downregulated in breast cancer. <i>Cancer Letters</i> , 2004, 204, 69-77.	7.2	73
107	Distinct Secreted Frizzled Receptor Protein 1 Staining Pattern in Patients With Hyperplastic Polyposis Coli Syndrome. <i>Archives of Pathology and Laboratory Medicine</i> , 2004, 128, 967-973.	2.5	10