Eva Klopocki

List of Publications by Year
in descending order

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107	6,541	87888 38	71685 76
papers	citations	h-index	g-index

113	113	113	11112
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. Cell, 2015, 161, 1012-1025.	28.9	1,725
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	Mutations of CASK cause an X-linked brain malformation phenotype with microcephaly and hypoplasia of the brainstem and cerebellum. Nature Genetics, 2008, 40, 1065-1067.	21.4	252
4	Aberrant methylation of the Wnt antagonist SFRP1 in breast cancer is associated with unfavourable prognosis. Oncogene, 2006, 25, 3479-3488.	5.9	234
5	The core FOXG1 syndrome phenotype consists of postnatal microcephaly, severe mental retardation, absent language, dyskinesia, and corpus callosum hypogenesis. Journal of Medical Genetics, 2011, 48, 396-406.	3.2	220
6	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
7	Negative enrichment by immunomagnetic nanobeads for unbiased characterization of circulating tumor cells from peripheral blood of cancer patients. Journal of Translational Medicine, 2011, 9, 70.	4.4	162
8	Agenesis and dysgenesis of the corpus callosum: Clinical, genetic and neuroimaging findings in a series of 41 patients. American Journal of Medical Genetics, Part A, 2008, 146A, 2501-2511.	1.2	148
9	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
10	Deletions of chromosome 8p and loss of sFRP1 expression are progression markers of papillary bladder cancer. Laboratory Investigation, 2004, 84, 465-478.	3.7	134
11	Identification of FOXP1 deletions in three unrelated patients with mental retardation and significant speech and language deficits. Human Mutation, 2010, 31, E1851-E1860.	2.5	130
12	Frequent loss of SFRP1 expression in multiple human solid tumours: association with aberrant promoter methylation in renal cell carcinoma. Oncogene, 2007, 26, 5680-5691.	5.9	127
13	Deletion and Point Mutations of PTHLH Cause Brachydactyly Type E. American Journal of Human Genetics, 2010, 86, 434-439.	6.2	127
14	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
15	Homeotic Arm-to-Leg Transformation Associated with Genomic Rearrangements at the PITX1 Locus. American Journal of Human Genetics, 2012, 91, 629-635.	6.2	111
16	Composition and dosage of a multipartite enhancer cluster control developmental expression of Ihh (Indian hedgehog). Nature Genetics, 2017, 49, 1539-1545.	21.4	107
17	Duplications of noncoding elements 5â€² of SOX9 are associated with brachydactyly-anonychia. Nature Genetics, 2009, 41, 862-863.	21.4	105
18	Copy-Number Variations Involving the IHH Locus Are Associated with Syndactyly and Craniosynostosis. American Journal of Human Genetics, 2011, 88, 70-75.	6.2	89

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19	<scp>DFNB16</scp> is a frequent cause of congenital hearing impairment: implementation of <scp>STRC</scp> mutation analysis in routine diagnostics. Clinical Genetics, 2015, 87, 49-55.	2.0	86
20	Proximal microdeletions and microduplications of 1q21.1 contribute to variable abnormal phenotypes. European Journal of Human Genetics, 2012, 20, 754-761.	2.8	84
21	Duplications ofBHLHA9are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. Journal of Medical Genetics, 2012, 49, 119-125.	3.2	81
22	Expanded Clinical Spectrum in Hepatocyte Nuclear Factor 1B-Maturity-Onset Diabetes of the Young. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 2658-2664.	3.6	77
23	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
24	ITIH5, a novel member of the inter-Î±-trypsin inhibitor heavy chain family is downregulated in breast cancer. Cancer Letters, 2004, 204, 69-77.	7.2	73
25	Microduplications encompassing the Sonic hedgehog limb enhancer <scp>ZRS</scp> are associated with Haasâ€type polysyndactyly and Laurinâ€Sandrow syndrome. Clinical Genetics, 2014, 86, 318-325.	2.0	72
26	Cerebellar and posterior fossa malformations in patients with autismâ€associated chromosome 22q13 terminal deletion. American Journal of Medical Genetics, Part A, 2013, 161, 131-136.	1.2	65
27	Deletions in PITX1 cause a spectrum of lower-limb malformations including mirror-image polydactyly. European Journal of Human Genetics, 2012, 20, 705-708.	2.8	63
28	Loss of SFRP1 is associated with breast cancer progression and poor prognosis in early stage tumors. International Journal of Oncology, 2004, 25, 641.	3.3	62
29	Deletions of the RUNX2 gene are present in about 10% of individuals with cleidocranial dysplasia. Human Mutation, 2010, 31, E1587-E1593.	2.5	61
30	Copy-Number Variations, Noncoding Sequences, and Human Phenotypes. Annual Review of Genomics and Human Genetics, 2011, 12, 53-72.	6.2	53
31	Systematic identification and molecular characterization of genes differentially expressed in breast and ovarian cancer. Journal of Pathology, 2005, 205, 21-28.	4.5	52
32	Whole exome sequencing identified a novel zinc-finger geneZNF141associated with autosomal recessive postaxial polydactyly type A. Journal of Medical Genetics, 2013, 50, 47-53.	3.2	51
33	A further case of the recurrent 15q24 microdeletion syndrome, detected by array CGH. European Journal of Pediatrics, 2008, 167, 903-908.	2.7	47
34	Homozygous deletion of chromosome 15q13.3 including CHRNA7 causes severe mental retardation, seizures, muscular hypotonia, and the loss of KLF13 and TRPM1 potentially cause macrocytosis and congenital retinal dysfunction in siblings. European Journal of Medical Genetics, 2011, 54, e441-e445.	1.3	47
35	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
36	Characterization of six Merkel cell polyomavirusâ€positive Merkel cell carcinoma cell lines: Integration pattern suggest that large T antigen truncating events occur before or during integration. International Journal of Cancer, 2019, 145, 1020-1032.	5.1	44

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37	CNVs of noncoding cis-regulatory elements in human disease. Current Opinion in Genetics and Development, 2013, 23, 249-256.	3.3	43
38	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
39	Deletions of exons with regulatory activity at the DYNC1I1 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
40	Noncoding copy-number variations are associated with congenital limb malformation. Genetics in Medicine, 2018, 20, 599-607.	2.4	42
41	Ulnarâ€“mammary syndrome with dysmorphic facies and mental retardation caused by a novel 1.28 Mb deletion encompassing the TBX3 gene. European Journal of Human Genetics, 2006, 14, 1274-1279.	2.8	41
42	De novo partial deletion in GRID2 presenting with complicated spastic paraplegia. Muscle and Nerve, 2014, 49, 289-292.	2.2	33
43	Catelâ€“Manzke syndrome: Two new patients and a critical review of the literature. European Journal of Medical Genetics, 2008, 51, 452-465.	1.3	32
44	Mesomelic dysplasia Kantaputra type is associated with duplications of the HOXD locus on chromosome 2q. European Journal of Human Genetics, 2010, 18, 1310-1314.	2.8	32
45	Amplification and translocation of 3q26 with overexpression ofEVI1 in Fanconi anemia-derived childhood acute myeloid leukemia with biallelicFANCD1/BRCA2 disruption. Genes Chromosomes and Cancer, 2007, 46, 359-372.	2.8	28
46	Triangular tibia with fibular aplasia associated with a microdeletion on 2q11.2 encompassing LAF4. Clinical Genetics, 2008, 74, 560-565.	2.0	28
47	Four unrelated patients with lubs Xâ€linked mental retardation syndrome and different Xq28 duplications. American Journal of Medical Genetics, Part A, 2010, 152A, 305-312.	1.2	27
48	Novel mutations of the <scp>PRKAR1A</scp> gene inÂpatients with acrodysostosis. Clinical Genetics, 2013, 84, 531-538.	2.0	27
49	Microdeletions on 6p22.3 are associated with mesomelic dysplasia Savarirayan type. Journal of Medical Genetics, 2015, 52, 476-483.	3.2	27
50	Exome sequencing identifies a nonsense mutation in Fam46a associated with bone abnormalities in a new mouse model for skeletal dysplasia. Mammalian Genome, 2016, 27, 111-121.	2.2	27
51	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
52	Molecular mechanism of CHRDL1-mediated X-linked megalocornea in humans and in Xenopus model. Human Molecular Genetics, 2015, 24, 3119-3132.	2.9	24
53	Gomez-Lopez-Hernandez syndrome (cerebello-trigeminal-dermal dysplasia): description of an additional case and review of the literature. European Journal of Pediatrics, 2008, 167, 123-126.	2.7	23
54	Homozygous missense and nonsense mutations in BMPR1B cause acromesomelic chondrodysplasia-type Grebe. European Journal of Human Genetics, 2014, 22, 726-733.	2.8	23

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55	A BACH2â€BCL2L1 fusion gene resulting from a t(6;20)(q15;q11.2) chromosomal translocation in the lymphoma cell line BLUEâ€1. Genes Chromosomes and Cancer, 2011, 50, 389-396.	2.8	22
56	Biallelic mutation in MYH7 and MYBPC3 leads to severe cardiomyopathy with left ventricular noncompaction phenotype. Human Mutation, 2019, 40, 1101-1114.	2.5	22
57	A novel 8 Mb interstitial deletion of chromosome 8p12-p21.2. American Journal of Medical Genetics, Part A, 2006, 140A, 873-877.	1.2	21
58	Heterozygote FANCD2 mutations associated with childhood T Cell ALL and testicular seminoma. Familial Cancer, 2012, 11, 661-665.	1.9	21
59	Tissue-Nonspecific Alkaline Phosphataseâ€"A Gatekeeper of Physiological Conditions in Health and a Modulator of Biological Environments in Disease. Biomolecules, 2020, 10, 1648.	4.0	19
60	Use of Targeted High-Throughput Sequencing for Genetic Classification of Patients with Bleeding Diathesis and Suspected Platelet Disorder. TH Open, 2018, 02, e445-e454.	1.4	18
61	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. American Journal of Human Genetics, 2007, 81, 866-868.	6.2	17
62	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
63	Biallelic intragenic deletion in MASP1 in an adult female with 3MC syndrome. European Journal of Medical Genetics, 2018, 61, 363-368.	1.3	17
64	Recessive grey platelet-like syndrome with unaffected erythropoiesis in the absence of the splice isoform GFI1B-p37. Haematologica, 2017, 102, e375-e378.	3.5	16
65	Complex genetics of radial ray deficiencies: screening of a cohort of 54 patients. Genetics in Medicine, 2013, 15, 195-202.	2.4	15
66	A complex phenotype with cystic renal disease. Kidney International, 2006, 70, 1656-1660.	5.2	14
67	Microduplications upstream ofMSX2are associated with a phenocopy of cleidocranial dysplasia. Journal of Medical Genetics, 2012, 49, 437-441.	3.2	12
68	Molecular cytogenetic characterisation of an interstitial deletion 12p detected by prenatal diagnosis. Prenatal Diagnosis, 2007, 27, 475-478.	2.3	11
69	De novo 9ÂMb deletion of 6q23.2q24.1 disrupting the gene EYA4 in a patient with sensorineural hearing loss, cardiac malformation, and mental retardation. European Journal of Medical Genetics, 2009, 52, 450-453.	1.3	11
70	Madelung deformity in a girl with a novel and de novo mutation in the GNAS gene. American Journal of Medical Genetics, Part A, 2011, 155, 2566-2570.	1.2	11
71	Analysis of relative gene dosage and expression differences of the paralogs RABL2A and RABL2B by Pyrosequencing. Gene, 2010, 455, 1-7.	2.2	10
72	Proximal and distal 15q25.2 microdeletionsâ€"genotypeâ€"phenotype delineation of two neurodevelopmental susceptibility loci. American Journal of Medical Genetics, Part A, 2013, 161, 218-224.	1.2	10

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73	A novel twoâ€nucleotide deletion in HPS6 affects mepacrine uptake and platelet dense granule secretion in a family with Hermanskyâ€“Pudlak syndrome. Pediatric Blood and Cancer, 2017, 64, e26320.	1.5	10
74	Investigation of alpl expression and Tnap-activity in zebrafish implies conserved functions during skeletal and neuronal development. Scientific Reports, 2020, 10, 13321.	3.3	10
75	Distinct Secreted Frizzled Receptor Protein 1 Staining Pattern in Patients With Hyperplastic Polyposis Coli Syndrome. Archives of Pathology and Laboratory Medicine, 2004, 128, 967-973.	2.5	10
76	HNF1B Abnormality (Mature-Onset Diabetes of the Young 5) in Children and Adolescents. Diabetes Care, 2008, 31, e83-e83.	8.6	9
77	Microdeletions of chromosome 7p21, including TWIST1, associated with significant microcephaly, facial dysmorphism, and short stature. European Journal of Medical Genetics, 2011, 54, 256-261.	1.3	9
78	Exploration of zebrafish larvae as an alternative whole-animal model for nephrotoxicity testing. Toxicology Letters, 2021, 344, 69-81.	0.8	9
79	ECM alterations in Fndc3a (Fibronectin Domain Containing Protein 3A) deficient zebrafish cause temporal fin development and regeneration defects. Scientific Reports, 2019, 9, 13383.	3.3	8
80	Mutation c.943G>T (p.Ala315Ser) in FGFR2 Causing a Mild Phenotype of Crouzon Craniofacial Dysostosis in a Three-Generation Family. Molecular Syndromology, 2017, 8, 93-97.	0.8	8
81	Tandem duplication ofDMD exon 18 associated with epilepsy, macroglossia, and endocrinologic abnormalities. Muscle and Nerve, 2007, 35, 396-401.	2.2	7
82	Expanding the phenotype of alopeciaâ€“contracturesâ€“dwarfism mental retardation syndrome (ACD) Tj ETQq0 0 0 rgBT /Overlock 10 T Pediatrics, 2008, 167, 1057-1062.	2.7	7
83	Partial trisomy 1q41â€qter and partial trisomy 9pterâ€9q21.32 in a newborn infant: An array CGH analysis and review. American Journal of Medical Genetics, Part A, 2014, 164, 490-494.	1.2	6
84	On the traces of tcf12: Investigation of the gene expression pattern during development and cranial suture patterning in zebrafish (Danio rerio). PLoS ONE, 2019, 14, e0218286.	2.5	6
85	Novel variants in FERMT3 and RASGRP2â€”Genetic linkage in Glanzmannâ€like bleeding disorders. Pediatric Blood and Cancer, 2020, 67, e28078.	1.5	6
86	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. European Journal of Medical Genetics, 2008, 51, 615-621.	1.3	5
87	Large homozygous RAB3GAP1 gene microdeletion causes Warburg Micro Syndrome 1. Orphanet Journal of Rare Diseases, 2014, 9, 113.	2.7	5
88	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 DNAJC19 mutation. Stem Cell Research, 2020, 46, 101856.	0.7	5
89	A Novel Locus and Candidate Gene for Familial Developmental Dyslexia on Chromosome 4q. Zeitschrift FÃœr Kinder- Und Jugendpsychiatrie Und Psychotherapie, 2020, 48, 478-489.	0.7	5
90	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

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91	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. Stem Cell Research, 2018, 33, 171-174.	0.7	4
92	Interstitial Deletion of 5q22.2q23.1 Including APC and TSSK1B in a Patient with Adenomatous Polyposis and Asthenoteratozoospermia. Molecular Syndromology, 2018, 9, 235-240.	0.8	4
93	New Insights on Genetic Diagnostics in Cardiomyopathy and Arrhythmia Patients Gained by Stepwise Exome Data Analysis. Journal of Clinical Medicine, 2020, 9, 2168.	2.4	4
94	Combined partial trisomy 11q and partial monosomy 10p in a 19â€yearâ€old female patient: Phenotypic and genotypic findings. American Journal of Medical Genetics, Part A, 2011, 155, 3075-3081.	1.2	3
95	Left ventricular hypertrabeculation/noncompaction with epilepsy, other heart defects, minor facial anomalies and new copy number variants. BMC Medical Genetics, 2012, 13, 60.	2.1	3
96	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. Stem Cell Research, 2019, 35, 101396.	0.7	3
97	Heterogeneous phenotypes in families with duplications of the paternal allele within the imprinting center 1 (<scp>H19</scp>/<scp>IGF2</scp>:<scp>TSSâ€DMR</scp>) in 11p15.5. Clinical Genetics, 2020, 98, 418-419.	2.0	3
98	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. Stem Cell Research, 2021, 51, 102169.	0.7	3
99	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). Stem Cell Research, 2021, 53, 102256.	0.7	1
100	An Autosomal-Recessive GFI1B Mutation Defines the Splice Isoform p37 As Essential for Biogenesis of Functional Human Platelets, but Dispensable for Erythropoiesis. Blood, 2016, 128, 2644-2644.	1.4	1
101	Atypical 22q11.2 Microduplication with â€œTypicalâ€•Signs and Overgrowth. Cytogenetic and Genome Research, 2020, 160, 659-663.	1.1	1
102	Generation of the induced pluripotent stem cell line UKWNLi005-A derived from a patient with the GLA mutation c.376AÂ>ÂG of unknown pathogenicity in Fabry disease. Stem Cell Research, 2022, 61, 102747.	0.7	1
103	P4: Ulnar-mammary syndrome with mental retardation caused byÂaÂnovel 0.8ÂMb deletion encompassing theÂTBX3 gene. European Journal of Medical Genetics, 2005, 48, 450.	1.3	0
104	Das 2q37-Deletionssyndrom. Medizinische Genetik, 2012, 24, 40-47.	0.2	0
105	The importance of TNAP/Tnap for dental development in human cell culture and in zebrafish. Bone Reports, 2021, 14, 101006.	0.4	0
106	Generation of multiple human iPSC lines from peripheral blood mononuclear cells of two SLC2A3 deletion and two SLC2A3 duplication carriers. Stem Cell Research, 2021, 56, 102526.	0.7	0
107	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.. Blood, 2010, 116, 1561-1561.	1.4	0