Paul Kuentz

List of Publications by Year
in descending order

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68 papers	1,810 citations	331670 21 h-index	315739 38 g-index
72 all docs	72 docs citations	72 times ranked	3966 citing authors

#	Article	IF	Citations
1	Clinical whole-exome sequencing for the diagnosis of rare disorders with congenital anomalies and/or intellectual disability: substantial interest of prospective annual reanalysis. Genetics in Medicine, 2018, 20, 645-654.	2.4	146
2	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	8.1	121
3	Globozoospermia is mainly due to DPY19L2 deletion via non-allelic homologous recombination involving two recombination hotspots. Human Molecular Genetics, 2012, 21, 3695-3702.	2.9	100
4	Lysosomal Signaling Licenses Embryonic Stem Cell Differentiation via Inactivation of Tfe3. Cell Stem Cell, 2019, 24, 257-270.e8.	11.1	97
5	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
6	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	2.4	90
7	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	2.5	70
8	Assisted oocyte activation overcomes fertilization failure in globozoospermic patients regardless of the DPY19L2 status. Human Reproduction, 2013, 28, 1054-1061.	0.9	67
9	Clinical reappraisal of <scp>SHORT</scp> syndrome with <scp>PIK3R1</scp> mutations: toward recommendation for molecular testing and management. Clinical Genetics, 2016, 89, 501-506.	2.0	66
10	NR2F1 regulates regional progenitor dynamics in the mouse neocortex and cortical gyrification in BBSOAS patients. EMBO Journal, 2020, 39, e104163.	7.8	49
11	Intragenic FMR1 disease-causing variants: a significant mutational mechanism leading to Fragile-X syndrome. European Journal of Human Genetics, 2017, 25, 423-431.	2.8	48
12	Dominant variants in the splicing factor PUF60 cause a recognizable syndrome with intellectual disability, heart defects and short stature. European Journal of Human Genetics, 2017, 25, 43-51.	2.8	44
13	OFIP/KIAA0753 forms a complex with OFD1 and FOR20 at pericentriolar satellites and centrosomes and is mutated in one individual with oral-facial-digital syndrome. Human Molecular Genetics, 2016, 25, 497-513.	2.9	42
14	PUF60 variants cause a syndrome of ID, short stature, microcephaly, coloboma, craniofacial, cardiac, renal and spinal features. European Journal of Human Genetics, 2017, 25, 552-559.	2.8	42
15	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. Genetics in Medicine, 2019, 21, 2025-2035.	2.4	40
16	A French multicenter study of over 700 patients with 22q11 deletions diagnosed using FISH or aCGH. European Journal of Human Genetics, 2016, 24, 844-851.	2.8	38
17	STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488.	3.2	35
18	Reverse Phenotyping in Patients with Skin Capillary Malformations and Mosaic GNAQ or GNA11 Mutations Defines a Clinical Spectrum with Genotype-Phenotype Correlation. Journal of Investigative Dermatology, 2020, 140, 1106-1110.e2.	0.7	30

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19	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La RÃ©union Island, in patients with Fryns syndrome. European Journal of Human Genetics, 2018, 26, 340-349.	2.8	27
20	MED13L-related intellectual disability: involvement of missense variants and delineation of the phenotype. Neurogenetics, 2018, 19, 93-103.	1.4	25
21	Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. Nature Genetics, 2019, 51, 1438-1441.	21.4	25
22	Mosaicâ€activating FGFR2 mutation in two fetuses with papillomatous pedunculated sebaceous naevus. British Journal of Dermatology, 2017, 176, 204-208.	1.5	23
23	Application of wholeâ€exome sequencing to unravel the molecular basis of undiagnosed syndromic congenital neutropenia with intellectual disability. American Journal of Medical Genetics, Part A, 2017, 173, 62-71.	1.2	23
24	<scp>AICA</scp>â€ribosiduria due to <scp>ATIC</scp> deficiency: Delineation of the phenotype with three novel cases, and longâ€term update on the first case. Journal of Inherited Metabolic Disease, 2020, 43, 1254-1264.	3.6	23
25	Reducing diagnostic turnaround times of exome sequencing for families requiring timely diagnoses. European Journal of Medical Genetics, 2017, 60, 595-604.	1.3	22
26	A new family with an SLC9A6 mutation expanding the phenotypic spectrum of Christianson syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2103-2110.	1.2	21
27	A standard of care for individuals with <scp>PIK3CA</scp>â€related disorders: An international expert consensus statement. Clinical Genetics, 2022, 101, 32-47.	2.0	21
28	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	2.4	21
29	Further delineation of a rare recessive encephalomyopathy linked to mutations in <scp>GFER</scp> thanks to data sharing of whole exome sequencing data. Clinical Genetics, 2017, 92, 188-198.	2.0	20
30	Chromosomal microarray analysis in fetuses with an isolated congenital heart defect: A retrospective, nationwide, multicenter study in France. Prenatal Diagnosis, 2019, 39, 464-470.	2.3	20
31	Homozygous FIBP nonsense variant responsible of syndromic overgrowth, with overgrowth, macrocephaly, retinal coloboma and learning disabilities. Clinical Genetics, 2016, 89, e1-4.	2.0	18
32	Heterozygous deletion of the LRFN2 gene is associated with working memory deficits. European Journal of Human Genetics, 2016, 24, 911-918.	2.8	18
33	Mosaicism for a KITLG Mutation in Linear and Whorled Nevoid Hypermelanosis. Journal of Investigative Dermatology, 2017, 137, 1575-1578.	0.7	18
34	Secondary actionable findings identified by exome sequencing: expected impact on the organisation of care from the study of 700 consecutive tests. European Journal of Human Genetics, 2019, 27, 1197-1214.	2.8	18
35	Genotype-first in a cohort of 95 fetuses with multiple congenital abnormalities: when exome sequencing reveals unexpected fetal phenotype-genotype correlations. Journal of Medical Genetics, 2021, 58, 400-413.	3.2	18
36	Postzygotic BRAF p.Lys601Asn Mutation in Phacomatosis Pigmentokeratotica with Woolly Hair Nevus and Focal Cortical Dysplasia. Journal of Investigative Dermatology, 2016, 136, 1060-1062.	0.7	15

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37	Autosomal recessive truncating <scp>MAB21L1</scp> mutation associated with a syndromic scrotal agenesis. Clinical Genetics, 2017, 91, 333-338.	2.0	15
38	Cardiomyopathy due to PRDM16 mutation: First description of a fetal presentation, with possible modifier genes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 129-135.	1.6	15
39	2.5 yearsâ€™ experience of GeneMatcher data-sharing: a powerful tool for identifying new genes responsible for rare diseases. Genetics in Medicine, 2019, 21, 1657-1661.	2.4	14
40	Clinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities. Genetics in Medicine, 2021, 23, 1484-1491.	2.4	14
41	9q33.3q34.11 microdeletion: new contiguous gene syndrome encompassing STXBP1, LMX1B and ENG genes assessed using reverse phenotyping. European Journal of Human Genetics, 2016, 24, 830-837.	2.8	13
42	Hearing impairment as an early sign of alphaâ€mannosidosis in children with a mild phenotype: Report of seven new cases. American Journal of Medical Genetics, Part A, 2019, 179, 1756-1763.	1.2	13
43	Genetic counselling difficulties and ethical implications of incidental findings from arrayâ€CGH: a 7â€year national survey. Clinical Genetics, 2016, 89, 630-635.	2.0	12
44	Novel KIAA1033 / WASHC4 mutations in three patients with syndromic intellectual disability and a review of the literature. American Journal of Medical Genetics, Part A, 2020, 182, 792-797.	1.2	12
45	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	3.2	11
46	TBL1XR1 mutations in Pierpont syndrome are not restricted to the recurrent p.Tyr446Cys mutation. American Journal of Medical Genetics, Part A, 2018, 176, 2813-2818.	1.2	10
47	Risk estimation of uniparental disomy of chromosome 14 or 15 in a fetus with a parent carrying a nonâ€homologous Robertsonian translocation. Should we still perform prenatal diagnosis?. Prenatal Diagnosis, 2019, 39, 986-992.	2.3	9
48	Incomplete Timothy syndrome secondary to a mosaic mutation of the CACNA1C gene diagnosed using nextâ€generation sequencing. American Journal of Medical Genetics, Part A, 2017, 173, 531-536.	1.2	8
49	New Insights into Potocki-Shaffer Syndrome: Report of Two Novel Cases and Literature Review. Brain Sciences, 2020, 10, 788.	2.3	7
50	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. Journal of Medical Genetics, 2020, 57, 466-474.	3.2	7
51	Cerebriform sebaceous nevus: a subtype of organoid nevus due to specific postzygotic FGFR2 mutations. Journal of the European Academy of Dermatology and Venereology, 2021, 35, 2085-2090.	2.4	6
52	Clinical and molecular data in cases of prenatal localized overgrowth disorder: major implication of genetic variants in <scp>PI3Kâ€AKTâ€mTOR</scp> signaling pathway. Ultrasound in Obstetrics and Gynecology, 2022, 59, 532-542.	1.7	6
53	A biâ€allelic lossâ€ofâ€function SARS1 variant in children with neurodevelopmental delay, deafness, cardiomyopathy, and decompensation during fever. Human Mutation, 2021, 42, 1576-1583.	2.5	6
54	Copy number variants calling from WES data through eXome hidden Markov model (XHMM) identifies additional 2.5% pathogenic genomic imbalances smaller than 30Âkb undetected by arrayâ€CGH. Annals of Human Genetics, 2022, 86, 171-180.	0.8	6

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55	Child with Beckwith-Wiedemann syndrome born after assisted reproductive techniques to an human immunodeficiency virus serodiscordant couple. Fertility and Sterility, 2011, 96, e35-e38.	1.0	5
56	Prenatal presentation of Aicardiâ€GoutiÃ¨res syndrome: Nonspecific phenotype necessitates exome sequencing for definitive diagnosis. Prenatal Diagnosis, 2019, 39, 806-810.	2.3	5
57	Primrose syndrome: a phenotypic comparison of patients with a ZBTB20 missense variant versus a 3q13.31 microdeletion including ZBTB20. European Journal of Human Genetics, 2020, 28, 1044-1055.	2.8	4
58	Severe gynaecological involvement in Proteus Syndrome. European Journal of Medical Genetics, 2019, 62, 270-272.	1.3	3
59	<scp>EPHA7</scp> haploinsufficiency is associated with a neurodevelopmental disorder. Clinical Genetics, 2021, 100, 396-404.	2.0	3
60	The diagnostic rate of inherited metabolic disorders by exome sequencing in a cohort of 547 individuals with developmental disorders. Molecular Genetics and Metabolism Reports, 2021, 29, 100812.	1.1	2
61	186 Mutational spectrum in PIK3CA -Related Overgrowth Spectrum (PROS) and recommendations for molecular testing. Journal of Investigative Dermatology, 2016, 136, S192.	0.7	1
62	Lossâ€ofâ€function variants in ARHGEF9 are associated with an Xâ€linked intellectual disability dominant disorder. Human Mutation, 2021, 42, 498-505.	2.5	1
63	Expanding the clinical spectrum of mosaic BRAF skin phenotypes. Journal of the European Academy of Dermatology and Venereology, 2021, 35, e690-e693.	2.4	1
64	The Largest Germline Heterozygous Deletion Encompassing Potockiâ€“Shaffer and WAGR Syndromes Loci to Date: A Case Report. Neuropediatrics, 2022, 53, 274-278.	0.6	1
65	Same performance of exome sequencing before and after fetal autopsy for congenital abnormalities: toward a paradigm shift in prenatal diagnosis?. European Journal of Human Genetics, 2022, , .	2.8	1
66	151 Postzygotic KITLG mutation in a congenital non-progressive linear nevoid hyperpigmentation. Journal of Investigative Dermatology, 2016, 136, S186.	0.7	0
67	616 Postzygotic mutations of RHOA cause a mosaic neuroectodermal syndrome. Journal of Investigative Dermatology, 2017, 137, S298.	0.7	0
68	Post-Essential Thrombocythemia Myelofibrosis and Multiple Isodicentric Y Chromosomes: A Unique Case among a Rare Association. Cytogenetic and Genome Research, 2020, 160, 18-21.	1.1	0