AndrÃ©e Delahaye

List of Publications by Year
in descending order

41 papers	1,700 citations	236833 25 h-index	302012 39 g-index
45 all docs	45 docs citations	45 times ranked	3675 citing authors

#	Article	IF	Citations
1	CDK13-related disorder: Report of a series of 18 previously unpublished individuals and description of an epigenetic signature. Genetics in Medicine, 2022, 24, 1096-1107.	1.1	8
2	Dysfunction of AMPA receptor GluA3 is associated with aggressive behavior in human. Molecular Psychiatry, 2022, 27, 4092-4102.	4.1	7
3	Targeting Microglial Disturbances to Protect the Brain From Neurodevelopmental Disorders Associated With Prematurity. Journal of Neuropathology and Experimental Neurology, 2021, 80, 634-648.	0.9	3
4	Impact of Fetal Growth Restriction on the Neonatal Microglial Proteome in the Rat. Nutrients, 2021, 13, 3719.	1.7	4
5	16p13.11 microduplication in 45 new patients: refined clinical significance and genotypeâ€“phenotype correlations. Journal of Medical Genetics, 2020, 57, 301-307.	1.5	44
6	New evidence that biallelic loss of function in EEF1B2 gene leads to intellectual disability. Clinical Genetics, 2020, 97, 639-643.	1.0	5
7	Machine learning applications in drug development. Computational and Structural Biotechnology Journal, 2020, 18, 241-252.	1.9	135
8	Pontocerebellar hypoplasia with rhombencephalosynapsis and microlissencephaly expands the spectrum of PCH type 1B. European Journal of Medical Genetics, 2020, 63, 103814.	0.7	3
9	Cell Metabolic Alterations due to Mcph1 Mutation in Microcephaly. Cell Reports, 2020, 31, 107506.	2.9	23
10	Integrated systemsâ€genetic analyses reveal a network target for delaying glioma progression. Annals of Clinical and Translational Neurology, 2019, 6, 1616-1638.	1.7	8
11	Decreased microglial Wnt/Î²-catenin signalling drives microglial pro-inflammatory activation in the developing brain. Brain, 2019, 142, 3806-3833.	3.7	97
12	A systems-level framework for drug discovery identifies Csf1R as an anti-epileptic drug target. Nature Communications, 2018, 9, 3561.	5.8	75
13	Genome-wide analysis of differential RNA editing in epilepsy. Genome Research, 2017, 27, 440-450.	2.4	73
14	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. European Journal of Human Genetics, 2017, 25, 694-701.	1.4	33
15	Rare and common epilepsies converge on a shared gene regulatory network providing opportunities for novel antiepileptic drug discovery. Genome Biology, 2016, 17, 245.	3.8	75
16	Clinical and molecular findings in 39 patients with KBG syndrome caused by deletion or mutation of ANKRD11. American Journal of Medical Genetics, Part A, 2016, 170, 2847-2859.	0.7	62
17	Systems genetics identifies a convergent gene network for cognition and neurodevelopmental disease. Nature Neuroscience, 2016, 19, 223-232.	7.1	131
18	Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. Molecular Autism, 2015, 6, 19.	2.6	29

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19	WWOX-related encephalopathies: delineation of the phenotypical spectrum and emerging genotype-phenotype correlation. Journal of Medical Genetics, 2015, 52, 61-70.	1.5	74
20	Incomplete penetrance and phenotypic variability of 6q16 deletions including SIM1. European Journal of Human Genetics, 2015, 23, 1010-1018.	1.4	35
21	Molecular findings and clinical data in a cohort of 150 patients with anophthalmia/microphthalmia. Clinical Genetics, 2014, 86, 326-334.	1.0	88
22	Cerebral small-vessel disease associated with COL4A1 and COL4A2 gene duplications. Neurology, 2014, 83, 1029-1031.	1.5	24
23	Mutations in the C-terminus of CDKL5: proceed with caution. European Journal of Human Genetics, 2014, 22, 270-272.	1.4	12
24	Genomic imbalances detected by array-CGH in patients with syndromal ocular developmental anomalies. European Journal of Human Genetics, 2012, 20, 527-533.	1.4	19
25	Chromosomal microarray analysis in ocular developmental anomalies. Expert Review of Molecular Diagnostics, 2012, 12, 425-427.	1.5	2
26	HOXC4 homeoprotein efficiently expands human hematopoietic stem cells and triggers similar molecular alterations as HOXB4. Haematologica, 2012, 97, 168-178.	1.7	26
27	Preâ€•and postnatal phenotype of 6p25 deletions involving the FOXC1 gene. American Journal of Medical Genetics, Part A, 2012, 158A, 2430-2438.	0.7	30
28	Recurrent mutations in the CDKL5 gene: Genotypeâ€“phenotype relationships. American Journal of Medical Genetics, Part A, 2012, 158A, 1612-1619.	0.7	53
29	Haploinsufficiency ofSOX5at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. Human Mutation, 2012, 33, 728-740.	1.1	85
30	Molecular characterization of a de novo 6q24.2q25.3 duplication interrupting UTRN in a patient with arthrogryposis. American Journal of Medical Genetics, Part A, 2010, 152A, 1781-1788.	0.7	13
31	Identification of gene copy number variations in patients with mental retardation using array-CGH: Novel syndromes in a large French series. European Journal of Medical Genetics, 2010, 53, 66-75.	0.7	29
32	Further delineation of the 17p13.3 microdeletion involving YWHAE but distal to PAFAH1B1: Four additional patients. European Journal of Medical Genetics, 2010, 53, 303-308.	0.7	44
33	2q23.1 microdeletion identified by array comparative genomic hybridisation: an emerging phenotype with Angelman-like features?. Journal of Medical Genetics, 2009, 46, 847-855.	1.5	43
34	Chromosome 22q13.3 deletion syndrome with a de novo interstitial 22q13.3 cryptic deletion disrupting SHANK3. European Journal of Medical Genetics, 2009, 52, 328-332.	0.7	51
35	First cryptic balanced reciprocal translocation mosaicism and familial transmission. American Journal of Medical Genetics, Part A, 2008, 146A, 2971-2974.	0.7	3
36	Familial CHARGE syndrome because of CHD7 mutation: clinical intraâ€•and interfamilial variability. Clinical Genetics, 2007, 72, 112-121.	1.0	76

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37	Retrospective Diagnosis of Pallister-Killian Syndrome by CGH Array. Fetal Diagnosis and Therapy, 2006, 21, 485-488.	0.6	7
38	MEFV analysis is of particularly weak diagnostic value for recurrent fevers in Western European Caucasian patients. Arthritis and Rheumatism, 2005, 52, 3603-3605.	6.7	56
39	The E148QMEFV allele is not implicated in the development of familial Mediterranean fever. Human Mutation, 2003, 22, 339-340.	1.1	88
40	Clinical Evaluation of a Reverse Hybridization Assay for the Molecular Detection of Twelve MEFV Gene Mutations. Clinical Chemistry, 2003, 49, 1942-1945.	1.5	27
41	Reply to Pubpeer anonymous contributors: incomplete penetrance and phenotypic variability of 6q16 deletions including SIM1. European Journal of Human Genetics, 0, , .	1.4	0