

David Geneviève

List of Publications by Year in descending order

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Version: 2024-02-01

37
papers

1,043
citations

623734

14
h-index

454955

30
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39
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39
docs citations

39
times ranked

2210
citing authors

#	ARTICLE	IF	CITATIONS
1	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 356-370.	6.2	171
2	Mutation Update for Kabuki Syndrome Genes <i>KMT2D</i> and <i>KDM6A</i> and Further Delineation of X-Linked Kabuki Syndrome Subtype 2. <i>Human Mutation</i> , 2016, 37, 847-864.	2.5	134
3	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. <i>Genetics in Medicine</i> , 2016, 18, 49-56.	2.4	125
4	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. <i>Genetics in Medicine</i> , 2017, 19, 989-997.	2.4	90
5	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. <i>Genetics in Medicine</i> , 2014, 16, 720-724.	2.4	63
6	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. <i>Genetics in Medicine</i> , 2020, 22, 547-556.	2.4	63
7	The molecular and phenotypic spectrum of <i>IQSEC2</i> -related epilepsy. <i>Epilepsia</i> , 2016, 57, 1858-1869.	5.1	46
8	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100075.	1.7	42
9	The expanding spectrum of COL2A1 gene variants IN 136 patients with a skeletal dysplasia phenotype. <i>European Journal of Human Genetics</i> , 2016, 24, 992-1000.	2.8	39
10	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. <i>Genetics in Medicine</i> , 2020, 22, 181-188.	2.4	30
11	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. <i>American Journal of Human Genetics</i> , 2019, 105, 854-868.	6.2	29
12	Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. <i>Nature Genetics</i> , 2019, 51, 1438-1441.	21.4	25
13	Large deletions encompassing the TCOF1 and CAMK2A genes are responsible for Treacher Collins syndrome with intellectual disability. <i>European Journal of Human Genetics</i> , 2014, 22, 52-56.	2.8	22
14	Autosomal recessive Treacher Collins syndrome due to <i>POLR1C</i> mutations: Report of a new family and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1390-1394.	1.2	19
15	Developmental and epilepsy spectrum of <i>KCNB1</i> encephalopathy with long-term outcome. <i>Epilepsia</i> , 2020, 61, 2461-2473.	5.1	17
16	Clinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities. <i>Genetics in Medicine</i> , 2021, 23, 1484-1491.	2.4	14
17	Report on three additional patients and genotype-phenotype correlation in SLC25A22-related disorders group. <i>European Journal of Human Genetics</i> , 2019, 27, 1692-1700.	2.8	10
18	Clinical and Molecular Spectrum of Nonsyndromic Early-Onset Osteoarthritis. <i>Arthritis and Rheumatology</i> , 2020, 72, 1689-1693.	5.6	10

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19	Exclusion of the dymeclin and PAPSS2 genes in a novel form of spondyloepimetaphyseal dysplasia and mental retardation. <i>European Journal of Human Genetics</i> , 2005, 13, 541-546.	2.8	9
20	<i>TMEM187-IRAK1</i> Polymorphisms Associated with Rheumatoid Arthritis Susceptibility in Tunisian and French Female Populations: Influence of Geographic Origin. <i>Journal of Immunology Research</i> , 2017, 2017, 1-12.	2.2	9
21	CDK13-related disorder: Report of a series of 18 previously unpublished individuals and description of an epigenetic signature. <i>Genetics in Medicine</i> , 2022, 24, 1096-1107.	2.4	8
22	Clinical Utility of a Unique Genome-Wide DNA Methylation Signature for KMT2A-Related Syndrome. <i>International Journal of Molecular Sciences</i> , 2022, 23, 1815.	4.1	8
23	Growth charts in Kabuki syndrome 1. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 446-453.	1.2	7
24	Dysspondyloenchondromatosis without <i>COL2A1</i> mutation: Possible genetic heterogeneity. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 769-773.	1.2	6
25	The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2740-2750.	1.2	6
26	DNA methylation episignature in Gabriele-de Vries syndrome. <i>Genetics in Medicine</i> , 2022, 24, 905-914.	2.4	6
27	Associations between cognitive performance and the rehabilitation, medical care and social support provided to French children with Prader-Willi syndrome. <i>European Journal of Medical Genetics</i> , 2020, 63, 104064.	1.3	5
28	Genome Alert!: A standardized procedure for genomic variant reinterperation and automated gene phenotype reassessment in clinical routine. <i>Genetics in Medicine</i> , 2022, 24, 1316-1327.	2.4	5
29	Rapid exome sequencing in critically ill infants: implementation in routine care from French regional hospital's perspective. <i>European Journal of Human Genetics</i> , 2022, 30, 1076-1082.	2.8	5
30	Using deep-neural-network-driven facial recognition to identify distinct Kabuki syndrome 1 and 2 gestalt. <i>European Journal of Human Genetics</i> , 2022, 30, 682-686.	2.8	4
31	Extending the prenatal Noonan's phenotype by review of ultrasound and autopsy data. <i>Prenatal Diagnosis</i> , 2022, 42, 574-582.	2.3	4
32	Epiphyseal punctate calcifications (stippling) in complete trisomy 9. <i>Prenatal Diagnosis</i> , 2009, 29, 1085-1088.	2.3	3
33	Predominance of BRCA2 Mutation and Estrogen Receptor Positivity in Unselected Breast Cancer with BRCA1 or BRCA2 Mutation. <i>Cancers</i> , 2022, 14, 3266.	3.7	3
34	Treacher Collins Syndrome. <i>Plastic and Reconstructive Surgery</i> , 2016, 138, 374e-376e.	1.4	2
35	No Association of Early-Onset Breast or Ovarian Cancer with Early-Onset Cancer in Relatives in BRCA1 or BRCA2 Mutation Families. <i>Genes</i> , 2021, 12, 1100.	2.4	1
36	Toward clinical and molecular dissection of frontonasal dysplasia with facial skin polyps: From Pai syndrome to differential diagnosis through a series of 27 patients. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2036-2047.	1.2	1

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37	A second individual with rhizomelic spondyloepimetaphyseal dysplasia and homozygous variant in GNPAT1. European Journal of Medical Genetics, 2022, 65, 104495.	1.3	1