David GeneviÃ¨ve

List of Publications by Year
in descending order

37 papers	1,043 citations	623734 14 h-index	454955 30 g-index
39 all docs	39 docs citations	39 times ranked	2210 citing authors

#	Article	IF	Citations
1	Using deep-neural-network-driven facial recognition to identify distinct Kabuki syndrome 1 and 2 gestalt. European Journal of Human Genetics, 2022, 30, 682-686.	2.8	4
2	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
3	DNA methylation episignature in Gabriele-de VriesÂsyndrome. Genetics in Medicine, 2022, 24, 905-914.	2.4	6
4	CDK13-related disorder: Report of a series of 18 previously unpublished individuals and description of an epigenetic signature.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.	2.4	8
5	Clinical Utility of a Unique Genome-Wide DNA Methylation Signature for KMT2A-Related Syndrome. International Journal of Molecular Sciences, 2022, 23, 1815.	4.1	8
6	Extending the prenatal Noonan's phenotype by review of ultrasound and autopsy data. Prenatal Diagnosis, 2022, 42, 574-582.	2.3	4
7	Genome Alert!: A standardized procedure for genomicÂvariant reinterpretation and automated geneâ€“phenotype reassessment in clinical routine. Genetics in Medicine, 2022, 24, 1316-1327.	2.4	5
8	Toward clinical and molecular dissection of frontonasal dysplasia with facial skin polyps: From Pai syndrome to differential diagnosis through a series of 27 patients. American Journal of Medical Genetics, Part A, 2022, 188, 2036-2047.	1.2	1
9	A second individual with rhizomelic spondyloepimetaphyseal dysplasia and homozygous variant in GNPNAT1. European Journal of Medical Genetics, 2022, 65, 104495.	1.3	1
10	Rapid exome sequencing in critically ill infants: implementation in routine care from French regional hospitalâ€™s perspective. European Journal of Human Genetics, 2022, 30, 1076-1082.	2.8	5
11	Predominance of BRCA2 Mutation and Estrogen Receptor Positivity in Unselected Breast Cancer with BRCA1 or BRCA2 Mutation. Cancers, 2022, 14, 3266.	3.7	3
12	Clinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities. Genetics in Medicine, 2021, 23, 1484-1491.	2.4	14
13	No Association of Early-Onset Breast or Ovarian Cancer with Early-Onset Cancer in Relatives in BRCA1 or BRCA2 Mutation Families. Genes, 2021, 12, 1100.	2.4	1
14	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. Genetics in Medicine, 2020, 22, 547-556.	2.4	63
15	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. Genetics in Medicine, 2020, 22, 181-188.	2.4	30
16	Growth charts in Kabuki syndrome 1. American Journal of Medical Genetics, Part A, 2020, 182, 446-453.	1.2	7
17	Associations between cognitive performance and the rehabilitation, medical care and social support provided to French children with Prader-Willi syndrome. European Journal of Medical Genetics, 2020, 63, 104064.	1.3	5
18	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17

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19	Clinical and Molecular Spectrum of Nonsyndromic Earlyâ€Onset Osteoarthritis. Arthritis and Rheumatology, 2020, 72, 1689-1693.	5.6	10
20	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	6.2	171
21	Report on three additional patients and genotypeâ€“phenotype correlation in SLC25A22-related disorders group. European Journal of Human Genetics, 2019, 27, 1692-1700.	2.8	10
22	Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. Nature Genetics, 2019, 51, 1438-1441.	21.4	25
23	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	6.2	29
24	Autosomal recessive Treacher Collins syndrome due to POLR1C mutations: Report of a new family and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1390-1394.	1.2	19
25	The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance. American Journal of Medical Genetics, Part A, 2018, 176, 2740-2750.	1.2	6
26	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
27	TMEM187-IRAK1Polymorphisms Associated with Rheumatoid Arthritis Susceptibility in Tunisian and French Female Populations: Influence of Geographic Origin. Journal of Immunology Research, 2017, 2017, 1-12.	2.2	9
28	Treacher Collins Syndrome. Plastic and Reconstructive Surgery, 2016, 138, 374e-376e.	1.4	2
29	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
30	The molecular and phenotypic spectrum of <scp>IQSEC</scp>2â€related epilepsy. Epilepsia, 2016, 57, 1858-1869.	5.1	46
31	The expanding spectrum of COL2A1 gene variants IN 136 patients with a skeletal dysplasia phenotype. European Journal of Human Genetics, 2016, 24, 992-1000.	2.8	39
32	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	2.4	125
33	Dysspondyloenchondromatosis without COL2A1 mutation: Possible genetic heterogeneity. American Journal of Medical Genetics, Part A, 2014, 164, 769-773.	1.2	6
34	Large deletions encompassing the TCOF1 and CAMK2A genes are responsible for Treacher Collins syndrome with intellectual disability. European Journal of Human Genetics, 2014, 22, 52-56.	2.8	22
35	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. Genetics in Medicine, 2014, 16, 720-724.	2.4	63
36	Epiphyseal punctate calcifications (stippling) in complete trisomy 9. Prenatal Diagnosis, 2009, 29, 1085-1088.	2.3	3

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37	Exclusion of the dymeclin and PAPSS2 genes in a novel form of spondyloepimetaphyseal dysplasia and mental retardation. European Journal of Human Genetics, 2005, 13, 541-546.	2.8	9