Nicole Revencu

List of Publications by Year
in descending order

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46 papers	2,610 citations	279798 23 h-index	206112 48 g-index
49 all docs	49 docs citations	49 times ranked	4634 citing authors

#	Article	IF	Citations
1	Pathogenic variants in MDFIC cause recessive central conducting lymphatic anomaly with lymphedema. Science Translational Medicine, 2022, 14, eabm4869.	12.4	14
2	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Human Mutation, 2022, 43, 582-594.	2.5	6
3	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	6.2	13
4	Somatic TEK variant with intraarticular venous malformation and knee hemarthrosis treated with rapamycin. Molecular Genetics & Genomic Medicine, 2022, , e1931.	1.2	2
5	Arteriovenous Cerebral High Flow Shunts in Children: From Genotype to Phenotype. Frontiers in Pediatrics, 2022, 10, 871565.	1.9	2
6	<scp>GNA11</scp>â€mutated <scp>Sturgeâ€“Weber</scp> syndrome has distinct neurological and dermatological features. European Journal of Neurology, 2022, 29, 3061-3070.	3.3	10
7	Aberrant sialylation in a patient with a HNF1Î± variant and liver adenomatosis. IScience, 2021, 24, 102323.	4.1	4
8	A human importin-Î²-related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8. American Journal of Human Genetics, 2021, 108, 1115-1125.	6.2	10
9	A novel RAD21 mutation in a boy with mild Cornelia de Lange presentation: Further delineation of the phenotype. European Journal of Medical Genetics, 2020, 63, 103620.	1.3	6
10	RASA1mosaic mutations in patients with capillary malformation-arteriovenous malformation. Journal of Medical Genetics, 2020, 57, 48-52.	3.2	38
11	Microdeletion of the entire IRF6 gene in a Subsaharian Africanâ€™s family with Van der Woude syndrome. Clinical Dysmorphology, 2020, 29, 24-27.	0.3	2
12	Characterization of ANGPT2 mutations associated with primary lymphedema. Science Translational Medicine, 2020, 12, .	12.4	31
13	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
14	Likely Pathogenic Variants in One Third of Non-Syndromic Discontinuous Cleft Lip and Palate Patients. Genes, 2019, 10, 833.	2.4	6
15	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
16	De Novo Mutations Affecting the Catalytic CÎ± Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 104, 139-156.	6.2	39
17	Cervical artery dissection: fibromuscular dysplasia versus vascular Ehlersâ€“Danlos syndrome. Blood Pressure, 2019, 28, 139-143.	1.5	5
18	Whole exome sequencing identifies mutations in 10% of patients with familial non-syndromic cleft lip and/or palate in genes mutated in well-known syndromes. Journal of Medical Genetics, 2018, 55, 449-458.	3.2	82

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19	CHARGE syndrome: a recurrent hotspot of mutations in CHD7 IVS25 analyzed by bioinformatic tools and minigene assays. European Journal of Human Genetics, 2018, 26, 287-292.	2.8	7
20	Unmasking familial CPX by WES and identification of novel clinical signs. American Journal of Medical Genetics, Part A, 2018, 176, 2661-2667.	1.2	3
21	Angiosarcoma arising from congenital primary lymphedema. Pediatric Dermatology, 2018, 35, e382-e388.	0.9	7
22	STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488.	3.2	35
23	Identification of a de novo variant in CHUK in a patient with an EEC/AEC syndromeâ€like phenotype and hypogammaglobulinemia. American Journal of Medical Genetics, Part A, 2017, 173, 1813-1820.	1.2	8
24	FOXP1-related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	3.2	48
25	Germline Loss-of-Function Mutations in EPHB4 Cause a Second Form of Capillary Malformation-Arteriovenous Malformation (CM-AVM2) Deregulating RAS-MAPK Signaling. Circulation, 2017, 136, 1037-1048.	1.6	204
26	Exploring the genetic basis of 3MC syndrome: Findings in 12 further families. American Journal of Medical Genetics, Part A, 2016, 170, 1216-1224.	1.2	25
27	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
28	Homozygous familial hypercholesterolemia in childhood: Genotype-phenotype description, established therapies and perspectives. Atherosclerosis, 2016, 247, 97-104.	0.8	14
29	No evidence of locus heterogeneity in familial microcephaly with or without chorioretinopathy, lymphedema, or mental retardation syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 52.	2.7	16
30	Retargeting of bile salt export pump and favorable outcome in children with progressive familial intrahepatic cholestasis type 2. Hepatology, 2015, 62, 198-206.	7.3	25
31	Heredity of port-wine stains: Investigation of families without a RASA1 mutation. Journal of Cosmetic and Laser Therapy, 2015, 17, 204-208.	0.9	7
32	Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. European Journal of Medical Genetics, 2014, 57, 151-156.	1.3	91
33	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	2.9	120
34	Clinical variability in neurohepatic syndrome due to combined mitochondrial DNA depletion and Gaucher disease. Molecular Genetics and Metabolism Reports, 2014, 1, 223-231.	1.1	2
35	Misdiagnosis as asphyxiating thoracic dystrophy and CMV-associated haemophagocytic lymphohistiocytosis in Shwachman-Diamond syndrome. European Journal of Pediatrics, 2013, 172, 613-622.	2.7	16
36	RASA1Mutations and Associated Phenotypes in 68 Families with Capillary Malformation-Arteriovenous Malformation. Human Mutation, 2013, 34, 1632-1641.	2.5	221

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37	Multifocal Capillary Malformations Due to RASA1 Mutation Misdiagnosed as Cutaneous Mastocytosis. Archives of Dermatology, 2012, 148, 1334.	1.4	5
38	How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. European Journal of Human Genetics, 2012, 20, 381-388.	2.8	142
39	Neonatal Ichthyosis and Sclerosing Cholangitis Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2011, 53, 350-354.	1.8	37
40	FAF1, a Gene that Is Disrupted in Cleft Palate and Has Conserved Function in Zebrafish. American Journal of Human Genetics, 2011, 88, 150-161.	6.2	57
41	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. Nature Genetics, 2010, 42, 483-485.	21.4	417
42	Prevalence and nonrandom distribution of exonic mutations in interferon regulatory factor 6 in 307 families with Van der Woude syndrome and 37 families with popliteal pterygium syndrome. Genetics in Medicine, 2009, 11, 241-247.	2.4	110
43	Sporadic In Utero Generalized Edema Caused by Mutations in the Lymphangiogenic Genes VEGFR3 and FOXC2. Journal of Pediatrics, 2009, 155, 90-93.	1.8	45
44	Parkes Weber syndrome, vein of Galen aneurysmal malformation, and other fast-flow vascular anomalies are caused byRASA1 mutations. Human Mutation, 2008, 29, 959-965.	2.5	382
45	Interferon regulatory factor-6: a gene predisposing to isolated cleft lip with or without cleft palate in the Belgian population. European Journal of Human Genetics, 2005, 13, 1239-1242.	2.8	81
46	Congenital diaphragmatic eventration and bilateral uretero-hydronephrosis in a patient with neonatal Marfan syndrome caused by a mutation in exon 25 of the FBN1 gene and review of the literature. European Journal of Pediatrics, 2004, 163, 33-37.	2.7	35