Pascal Brouillard

List of Publications by Year
in descending order

38 papers	1,974 citations	361388 20 h-index	377849 34 g-index
39 all docs	39 docs citations	39 times ranked	2000 citing authors

#	Article	IF	Citations
1	Mutations in a Novel Factor, Glomulin, Are Responsible for Glomuvenous Malformations (â€œGlomangiomasâ€). American Journal of Human Genetics, 2002, 70, 866-874.	6.2	304
2	Genetics of lymphatic anomalies. Journal of Clinical Investigation, 2014, 124, 898-904.	8.2	271
3	Genetic causes of vascular malformations. Human Molecular Genetics, 2007, 16, R140-R149.	2.9	226
4	A Gene for Inherited Cutaneous Venous Anomalies (â€œGlomangiomasâ€) Localizes to Chromosome 1p21-22. American Journal of Human Genetics, 1999, 65, 125-133.	6.2	156
5	Vascular malformations: localized defects in vascular morphogenesis. Clinical Genetics, 2003, 63, 340-351.	2.0	103
6	Loss of ADAMTS3 activity causes Hennekam lymphangiectasiaâ€“lymphedema syndrome 3. Human Molecular Genetics, 2017, 26, 4095-4104.	2.9	95
7	Mutations in the VEGFR3 Signaling Pathway Explain 36% of Familial Lymphedema. Molecular Syndromology, 2013, 4, 257-266.	0.8	92
8	Somatic Uniparental Isodisomy Explains Multifocality of Glomuvenous Malformations. American Journal of Human Genetics, 2013, 92, 188-196.	6.2	71
9	Efficient activation of the lymphangiogenic growth factor VEGF-C requires the C-terminal domain of VEGF-C and the N-terminal domain of CCBE1. Scientific Reports, 2017, 7, 4916.	3.3	69
10	Blockade of VEGF-C signaling inhibits lymphatic malformations driven by oncogenic PIK3CA mutation. Nature Communications, 2020, 11, 2869.	12.8	59
11	Genotypes and Phenotypes of 162 Families with aGlomulinMutation. Molecular Syndromology, 2013, 4, 157-64.	0.8	55
12	Association of PDGFRB Mutations With Pediatric Myofibroma and Myofibromatosis. JAMA Dermatology, 2019, 155, 946.	4.1	43
13	High-Resolution Physical and Transcript Map of the Locus for Venous Malformations with Glomus Cells (VMGLOM) on Chromosome 1p21â€“p22. Genomics, 2000, 67, 96-101.	2.9	42
14	Linkage disequilibrium narrows locus for venous malformation with glomus cells (VMGLOM) to a single 1.48ÂMbp YAC. European Journal of Human Genetics, 2001, 9, 34-38.	2.8	42
15	Glomulin is predominantly expressed in vascular smooth muscle cells in the embryonic and adult mouse. Gene Expression Patterns, 2004, 4, 351-358.	0.8	39
16	RASA1mosaic mutations in patients with capillary malformation-arteriovenous malformation. Journal of Medical Genetics, 2020, 57, 48-52.	3.2	38
17	Primary lymphoedema. Nature Reviews Disease Primers, 2021, 7, 77.	30.5	33
18	Characterization of ANGPT2 mutations associated with primary lymphedema. Science Translational Medicine, 2020, 12, .	12.4	31

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19	KRAS-driven model of Gorham-Stout disease effectively treated with trametinib. JCI Insight, 2021, 6, .	5.0	27
20	Non-hotspot PIK3CA mutations are more frequent in CLOVES than in common or combined lymphatic malformations. Orphanet Journal of Rare Diseases, 2021, 16, 267.	2.7	26
21	Structure of the TSC2 GAP Domain: Mechanistic Insight into Catalysis and Pathogenic Mutations. Structure, 2020, 28, 933-942.e4.	3.3	20
22	Microcephaly, intellectual impairment, bilateral vesicoureteral reflux, distichiasis, and glomuvenous malformations associated with a 16q24.3 contiguous gene deletion and a Glomulin mutation. American Journal of Medical Genetics, Part A, 2012, 158A, 839-849.	1.2	18
23	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
24	Congenital Plaqueâ€Type Glomuvenous Malformations Associated with Fetal Pleural Effusion and Ascites. Pediatric Dermatology, 2011, 28, 528-531.	0.9	15
25	Pathogenic variants in MDFIC cause recessive central conducting lymphatic anomaly with lymphedema. Science Translational Medicine, 2022, 14, eabm4869.	12.4	14
26	Aberrant Membrane Composition and Biophysical Properties Impair Erythrocyte Morphology and Functionality in Elliptocytosis. Biomolecules, 2020, 10, 1120.	4.0	10
27	<scp>EPHB4</scp> mutation causes adult and adolescentâ€onset primary lymphedema. American Journal of Medical Genetics, Part A, 2021, 185, 3810-3813.	1.2	10
28	GATA2 null mutation associated with incomplete penetrance in a family with Emberger syndrome. Hematology, 2017, 22, 1-5.	1.5	8
29	Hypotrichosisâ€lymphedemaâ€telangiectasia syndrome: Report of ileal atresia associated with a <scp>SOX18</scp> de novo pathogenic variant and review of the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 2153-2159.	1.2	8
30	Angiosarcoma arising from congenital primary lymphedema. Pediatric Dermatology, 2018, 35, e382-e388.	0.9	7
31	Reconstructive surgery in the management of a patient with CLOVES syndrome. Journal of Plastic, Reconstructive and Aesthetic Surgery, 2013, 66, 1813-1815.	1.0	6
32	Tumor sequencing is useful to refine the analysis of germline variants in unexplained high-risk breast cancer families. Breast Cancer Research, 2020, 22, 36.	5.0	6
33	First Draft Genome of the Trypanosomatid Herpetomonas muscarum ingenoplastis through MinION Oxford Nanopore Technology and Illumina Sequencing. Tropical Medicine and Infectious Disease, 2020, 5, 25.	2.3	4
34	SOX18 and the Hypotrichosis-Lymphedema-Telangiectasia Syndrome. , 2016, , 867-869.		4
35	BiallelicANGPT2loss-of-function causes severe early-onset non-immune hydrops fetalis. Journal of Medical Genetics, 2023, 60, 57-64.	3.2	4
36	Molecular Genetics of Lymphatic and Complex Vascular Malformations. , 2018, , 753-763.		1

#	Article	IF	Citations
37	Disorders of the Venous System. , 2013, , 1-9.		0
38	Disorders of the Venous System. , 2020, , 251-260.		0