Miikka Vikkula

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/5380388/publications.pdf

Version: 2024-02-01

243 papers	21,282 citations	11908 72 h-index	11608 140 g-index
258 all docs	258 docs citations	258 times ranked	19051 citing authors

#	Article	IF	Citations
1	BiallelicANGPT2loss-of-function causes severe early-onset non-immune hydrops fetalis. Journal of Medical Genetics, 2023, 60, 57-64.	1.5	4
2	Increased Collagen Turnover Is a Feature of Fibromuscular Dysplasia and Associated With Hypertrophic Radial Remodeling: A Pilot, Urine Proteomic Study. Hypertension, 2022, 79, 93-103.	1.3	4
3	Abstract P1-05-03: Integrating spatial transcriptomics and high-resolution morphological annotation to investigate tumor heterogeneity and PAM50 molecular subtyping in lobular breast cancer. Cancer Research, 2022, 82, P1-05-03-P1-05-03.	0.4	0
4	Pathogenic variants in MDFIC cause recessive central conducting lymphatic anomaly with lymphedema. Science Translational Medicine, 2022, 14, eabm4869.	5.8	14
5	Recalibrating vascular malformations and mechanotransduction by pharmacological intervention. Journal of Clinical Investigation, 2022, 132, .	3.9	4
6	Arteriovenous Cerebral High Flow Shunts in Children: From Genotype to Phenotype. Frontiers in Pediatrics, 2022, 10, 871565.	0.9	2
7	The VASCERN-VASCA working group diagnostic and management pathways for severe and/or rare infantile hemangiomas. European Journal of Medical Genetics, 2022, 65, 104517.	0.7	1
8	Molecular pathways and possible therapies for head and neck vascular anomalies. Journal of Oral Pathology and Medicine, 2022, 51, 878-887.	1.4	3
9	<scp>GNA11</scp>â€mutated <scp>Sturgeâ€“Weber</scp> syndrome has distinct neurological and dermatological features. European Journal of Neurology, 2022, 29, 3061-3070.	1.7	10
10	Case report study of thalidomide therapy in 18 patients with severe arteriovenous malformationï»¿s. , 2022, 1, 562-567.		11
11	The European/International Fibromuscular Dysplasia Registry and Initiative (FEIRI)â€”clinical phenotypes and their predictors based on a cohort of 1000 patients. Cardiovascular Research, 2021, 117, 950-959.	1.8	33
12	PTGIR, a susceptibility gene for fibromuscular dysplasia?. Cardiovascular Research, 2021, 117, 990-992.	1.8	1
13	Dysregulation of Rho GTPases in orofacial cleft patients-derived primary cells leads to impaired cell migration, a potential cause of cleft/lip palate development. Cells and Development, 2021, 165, 203656.	0.7	6
14	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.	0.7	8
15	Genetic Basis and Therapies for Vascular Anomalies. Circulation Research, 2021, 129, 155-173.	2.0	85
16	Lymphatic Malformations: Genetics, Mechanisms and Therapeutic Strategies. Circulation Research, 2021, 129, 136-154.	2.0	88
17	KRAS-driven model of Gorham-Stout disease effectively treated with trametinib. JCI Insight, 2021, 6, .	2.3	27
18	Non-hotspot PIK3CA mutations are more frequent in CLOVES than in common or combined lymphatic malformations. Orphanet Journal of Rare Diseases, 2021, 16, 267.	1.2	26

#	Article	IF	Citations
19	A review of mechanisms of disease across PIK3CA-related disorders with vascular manifestations. Orphanet Journal of Rare Diseases, 2021, 16, 306.	1.2	62
20	<scp>EPHB4</scp> mutation causes adult and adolescentâ€onset primary lymphedema. American Journal of Medical Genetics, Part A, 2021, 185, 3810-3813.	0.7	10
21	Efficacy of Sirolimus in Patients Requiring Tracheostomy for Life-Threatening Lymphatic Malformation of the Head and Neck: A Report From the European Reference Network. Frontiers in Pediatrics, 2021, 9, 697960.	0.9	8
22	Primary lymphoedema. Nature Reviews Disease Primers, 2021, 7, 77.	18.1	33
23	Genetic investigation of fibromuscular dysplasia identifies risk loci and shared genetics with common cardiovascular diseases. Nature Communications, 2021, 12, 6031.	5.8	34
24	Human chromosome map of lymphedema-lymphangiogenesis genes: Template for current and future discovery.. Lymphology, 2021, 54, 167-169.	0.1	0
25	Letter: Is Developmental Venous Anomaly an Imaging Biomarker of PIK3CA Mutated Gliomas?. Neurosurgery, 2020, 86, E93-E93.	0.6	4
26	A Clinical Feasibility Study to Image Angiogenesis in Patients with Arteriovenous Malformations Using ⁶⁸Ga-RGD PET/CT. Journal of Nuclear Medicine, 2020, 61, 270-275.	2.8	7
27	RASA1mosaic mutations in patients with capillary malformation-arteriovenous malformation. Journal of Medical Genetics, 2020, 57, 48-52.	1.5	38
28	Disorders of the Venous System. , 2020, , 251-260.		0
29	Capillary Malformation/Arteriovenous Malformation. , 2020, , 261-266.		0
30	Genetics of vascular anomalies. Seminars in Pediatric Surgery, 2020, 29, 150967.	0.5	21
31	Structure of the TSC2 GAP Domain: Mechanistic Insight into Catalysis and Pathogenic Mutations. Structure, 2020, 28, 933-942.e4.	1.6	20
32	Guidance for the Management of Patients with Vascular Disease or Cardiovascular Risk Factors and COVID-19: Position Paper from VAS-European Independent Foundation in Angiology/Vascular Medicine. Thrombosis and Haemostasis, 2020, 120, 1597-1628.	1.8	131
33	Characterization of ANGPT2 mutations associated with primary lymphedema. Science Translational Medicine, 2020, 12, .	5.8	31
34	Enrichment of Rare Variants in Loeysâ€“Dietz Syndrome Genes in Spontaneous Coronary Artery Dissection but Not in Severe Fibromuscular Dysplasia. Circulation, 2020, 142, 1021-1024.	1.6	30
35	Aberrant Membrane Composition and Biophysical Properties Impair Erythrocyte Morphology and Functionality in Elliptocytosis. Biomolecules, 2020, 10, 1120.	1.8	10
36	Lack of FAM20A, Ectopic Gingival Mineralization and Chondro/Osteogenic Modifications in Enamel Renal Syndrome. Frontiers in Cell and Developmental Biology, 2020, 8, 605084.	1.8	9

#	Article	IF	Citations
37	New and Emerging Targeted Therapies for Vascular Malformations. American Journal of Clinical Dermatology, 2020, 21, 657-668.	3.3	59
38	Blockade of VEGF-C signaling inhibits lymphatic malformations driven by oncogenic PIK3CA mutation. Nature Communications, 2020, 11, 2869.	5.8	59
39	Liquid biopsy for mutational profiling of locoregional recurrent and/or metastatic head and neck squamous cell carcinoma. Oral Oncology, 2020, 104, 104631.	0.8	42
40	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.	0.9	4
41	Theranostic Advances in Vascular Malformations. Journal of Investigative Dermatology, 2020, 140, 756-763.	0.3	41
42	Tumor sequencing is useful to refine the analysis of germline variants in unexplained high-risk breast cancer families. Breast Cancer Research, 2020, 22, 36.	2.2	6
43	The Genetic Basis of Vascular Anomalies. , 2020, , 17-29.		3
44	Analysing ambiguities in trypanosomatids taxonomy by barcoding. Memorias Do Instituto Oswaldo Cruz, 2020, 115, e200504.	0.8	4
45	Abstract P5-03-04: Co-segregation of rare possibly-damaging variants in cancer-related genes correlates with phenotypic homogeneity in familial breast cancer. , 2020, , .		0
46	Abstract P5-03-03: Tumor sequencing is useful to reclassify germline variants in unexplained high-risk breast cancer families. , 2020, , .		0
47	Liquid biopsy for mutational profiling of locoregional recurrent and/or metastatic squamous cell carcinoma of the head and neck. Annals of Oncology, 2019, 30, v468-v469.	0.6	0
48	Likely Pathogenic Variants in One Third of Non-Syndromic Discontinuous Cleft Lip and Palate Patients. Genes, 2019, 10, 833.	1.0	6
49	Association of PDGFRB Mutations With Pediatric Myofibroma and Myofibromatosis. JAMA Dermatology, 2019, 155, 946.	2.0	43
50	DNA alterationâ€based classification of uveal melanoma gives better prognostic stratification than immune infiltration, which has a neutral effect in highâ€risk group. Cancer Medicine, 2019, 8, 3036-3046.	1.3	13
51	Arterial Tortuosity. Hypertension, 2019, 73, 951-960.	1.3	110
52	Rapamycin and treatment of venous malformations. Current Opinion in Hematology, 2019, 26, 185-192.	1.2	44
53	Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation. Neuron, 2019, 101, 429-443.e4.	3.8	56
54	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.	1.5	82

#	Article	IF	Citations
55	Venous Malformations of the Head and Neck. Otolaryngologic Clinics of North America, 2018, 51, 173-184.	0.5	51
56	Etiology and Genetics of Congenital Vascular Lesions. Otolaryngologic Clinics of North America, 2018, 51, 41-53.	0.5	57
57	Molecular Genetics of Lymphatic and Complex Vascular Malformations. , 2018, , 753-763.		1
58	Spliceâ€site mutations in VEGFC cause loss of function and Nonneâ€Milroyâ€like primary lymphedema. Clinical Genetics, 2018, 94, 179-181.	1.0	16
59	Ageâ€related heterogeneity of Burkitt lymphoma: response to Mbulaiteye and Anderson. British Journal of Haematology, 2018, 180, 155-156.	1.2	0
60	Expression of Contactin 4 Is Associated With Malignant Behavior in Pheochromocytomas and Paragangliomas. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 46-55.	1.8	19
61	Unmasking familial CPX by WES and identification of novel clinical signs. American Journal of Medical Genetics, Part A, 2018, 176, 2661-2667.	0.7	3
62	Prevalence of pathogenic variants and variants of unknown significance in patients at high risk of breast cancer: A systematic review and meta-analysis of gene-panel data. Critical Reviews in Oncology/Hematology, 2018, 132, 138-144.	2.0	26
63	Sirolimus is efficacious in treatment for extensive and/or complex slow-flow vascular malformations: a monocentric prospective phase II study. Orphanet Journal of Rare Diseases, 2018, 13, 191.	1.2	154
64	Angiosarcoma arising from congenital primary lymphedema. Pediatric Dermatology, 2018, 35, e382-e388.	0.5	7
65	Incidence of Cellulitis among Children with Primary Lymphedema. New England Journal of Medicine, 2018, 378, 2047-2048.	13.9	13
66	Genomics of Fibromuscular Dysplasia. International Journal of Molecular Sciences, 2018, 19, 1526.	1.8	35
67	GATA2 null mutation associated with incomplete penetrance in a family with Emberger syndrome. Hematology, 2017, 22, 1-5.	0.7	8
68	PDGFRB gain-of-function mutations in sporadic infantile myofibromatosis. Human Molecular Genetics, 2017, 26, 1801-1810.	1.4	77
69	KIF1B and NF1 are the most frequently mutated genes in paraganglioma and pheochromocytoma tumors. Endocrine-Related Cancer, 2017, 24, L57-L61.	1.6	16
70	Loss of ADAMTS3 activity causes Hennekam lymphangiectasiaâ€“lymphedema syndrome 3. Human Molecular Genetics, 2017, 26, 4095-4104.	1.4	95
71	Vascular Anomalies Caused by Abnormal Signaling within Endothelial Cells: Targets for Novel Therapies. Seminars in Interventional Radiology, 2017, 34, 233-238.	0.3	48
72	Comment on â€œIncreased Identification of Candidates for High-Risk Breast Cancer Screening Through Expanded Genetic Testingâ€. Journal of the American College of Radiology, 2017, 14, 582.	0.9	8

#	Article	IF	Citations
73	Un traitement mÃ©dicamenteux pour les malformations vasculaires. JMV-Journal De Medecine Vasculaire, 2017, 42, 95.	0.1	0
74	[OP.6B.03] KIF1B AND NF1 ARE THE MOST FREQUENTLY MUTATED GENES IN PARAGANGLIOMAS AND PHEOCHROMOCYTOMAS TUMOURS FROM A BELGIAN MULTICENTRIC COHORT. Journal of Hypertension, 2017, 35, e58.	0.3	0
75	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.	1.6	69
76	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
77	Blue Rubber Bleb Nevus (BRBN) Syndrome Is Caused by Somatic TEK (TIE2) Mutations. Journal of Investigative Dermatology, 2017, 137, 207-216.	0.3	148
78	Genetics of Arteriovenous Malformations. , 2017, , 23-36.		2
79	Abstract 534:PDGFRBgain-of-function mutations in multifocal infantile myofibromatosis: Implications for diagnosis & therapy. , 2017, , .		0
80	STK39 and WNK1 Are Potential Hypertension Susceptibility Genes in the BELHYPGEN Cohort. Medicine (United States), 2016, 95, e2968.	0.4	14
81	Genetic differences between paediatric and adult Burkitt lymphomas. British Journal of Haematology, 2016, 173, 137-144.	1.2	26
82	Cystathionine Î²-synthase genetic variant rs2124459 is associated with a reduced risk of cleft palate in French and Belgian populations. Journal of Medical Genetics, 2016, 53, 828-834.	1.5	5
83	The peculiar 11q-gain/loss aberration reported in a subset of MYC-negative high-grade B-cell lymphomas can also occur in a MYC-rearranged lymphoma. Cancer Genetics, 2016, 209, 117-118.	0.2	21
84	Five int22h homologous copies at the Xq28 locus identified in intron22 inversion type 3 of the Factor VIII gene. Thrombosis Research, 2016, 137, 224-227.	0.8	3
85	Inheritance Patterns of Infantile Hemangioma. Pediatrics, 2016, 138, .	1.0	33
86	Routine use of gene panel testing in hereditary breast cancer should be performed with caution. Critical Reviews in Oncology/Hematology, 2016, 108, 33-39.	2.0	17
87	Isolated bilateral transverse agenesis of the distal segments of the lower limbs at the level of the knee joint in a human fetus. American Journal of Medical Genetics, Part A, 2016, 170, 523-530.	0.7	1
88	RASA1 and Capillary Malformationâ€“Arteriovenous Malformation. , 2016, , 633-637.		3
89	SOX18 and the Hypotrichosis-Lymphedema-Telangiectasia Syndrome. , 2016, , 867-869.		4
90	GLMN and Glomuvenous Malformation. , 2016, , 1423-1427.		2

#	Article	IF	Citations
91	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.	1.2	16
92	Tandem inversion duplication within F8 Intron 1 associated with mild haemophilia A. Haemophilia, 2015, 21, 516-522.	1.0	5
93	Rapamycin as Novel Treatment for Refractory-to-Standard-Care Slow-Flow Vascular Malformations. Plastic and Reconstructive Surgery, 2015, 136, 38.	0.7	6
94	9B.09. Journal of Hypertension, 2015, 33, e122.	0.3	3
95	6C.05. Journal of Hypertension, 2015, 33, e80.	0.3	2
96	Blue bleb rubber nevus syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2015, 132, 223-230.	1.0	19
97	Rapamycin improves TIE2-mutated venous malformation in murine model and human subjects. Journal of Clinical Investigation, 2015, 125, 3491-3504.	3.9	167
98	SDHB/SDHA immunohistochemistry in pheochromocytomas and paragangliomas: a multicenter interobserver variation analysis using virtual microscopy: a Multinational Study of the European Network for the Study of Adrenal Tumors (ENS@T). Modern Pathology, 2015, 28, 807-821.	2.9	176
99	Vascular Anomalies Classification: Recommendations From the International Society for the Study of Vascular Anomalies. Pediatrics, 2015, 136, e203-e214.	1.0	1,026
100	Somatic Activating PIK3CA Mutations Cause Venous Malformation. American Journal of Human Genetics, 2015, 97, 914-921.	2.6	244
101	Common and specific effects of TIE2 mutations causing venous malformations. Human Molecular Genetics, 2015, 24, 6374-6389.	1.4	81
102	Overrepresentation of missense mutations in mild hemophilia A patients from Belgium: founder effect or independent occurrence?. Thrombosis Research, 2015, 135, 1057-1063.	0.8	6
103	Hypotrichosisâ€lymphedemaâ€telangiectasiaâ€renal defect associated with a truncating mutation in the <scp>SOX18</scp> gene. Clinical Genetics, 2015, 87, 378-382.	1.0	33
104	Antenatal presentation of hereditary lymphedema type I. European Journal of Medical Genetics, 2015, 58, 329-331.	0.7	9
105	Heredity of port-wine stains: Investigation of families without a RASA1 mutation. Journal of Cosmetic and Laser Therapy, 2015, 17, 204-208.	0.3	7
106	Molecular and Genetic Aspects of Hemangiomas and Vascular Malformations. , 2015, , 21-38.		3
107	Capillary Malformation/Arteriovenous Malformation. , 2014, , .		0
108	Pathognomonic oral profile of Enamel Renal Syndrome (ERS) caused by recessive FAM20A mutations. Orphanet Journal of Rare Diseases, 2014, 9, 84.	1.2	63

#	Article	IF	Citations
109	Genetics of vascular malformations. Seminars in Pediatric Surgery, 2014, 23, 221-226.	0.5	39
110	Common Somatic Alterations Identified in Maffucci Syndrome by Molecular Karyotyping. Molecular Syndromology, 2014, 5, 259-267.	0.3	20
111	Genetics of lymphatic anomalies. Journal of Clinical Investigation, 2014, 124, 898-904.	3.9	271
112	Disorders of the Venous System. , 2014, , .		0
113	Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations. Nephron Physiology, 2013, 122, 1-6.	1.5	84
114	Evaluation of Clinical Manifestations in Patients with Severe Lymphedema with and without CCBE1 Mutations. Molecular Syndromology, 2013, 4, 107-113.	0.3	45
115	Capillary Malformation/Arteriovenous Malformation. , 2013, , 1-7.		2
116	Disorders of the Venous System. , 2013, , 1-9.		0
117	Somatic Uniparental Isodisomy Explains Multifocality of Glomuvenous Malformations. American Journal of Human Genetics, 2013, 92, 188-196.	2.6	71
118	Intron 22 homologous regions are implicated in exons 1â€“22 duplications of the F8 gene. European Journal of Human Genetics, 2013, 21, 970-976.	1.4	20
119	Giant cell tumor occurring in familial Paget's disease of bone: Report of clinical characteristics and linkage analysis of a large pedigree. Journal of Bone and Mineral Research, 2013, 28, 341-350.	3.1	19
120	Capillary Malformation-Arteriovenous Malformation Syndrome: A Report of 2 Cases, Diagnostic Criteria, and Management. Actas Dermo-sifiliogrÃ¡ficas, 2013, 104, 710-713.	0.2	5
121	Reconstructive surgery in the management of a patient with CLOVES syndrome. Journal of Plastic, Reconstructive and Aesthetic Surgery, 2013, 66, 1813-1815.	0.5	6
122	Venous malformation-causative TIE2 mutations mediate an AKT-dependent decrease in PDGFB. Human Molecular Genetics, 2013, 22, 3438-3448.	1.4	82
123	Variable Somatic <i>TIE2</i> Mutations in Half of Sporadic Venous Malformations. Molecular Syndromology, 2013, 4, 179-183.	0.3	134
124	Genotypes and Phenotypes of 162 Families with aGlomulinMutation. Molecular Syndromology, 2013, 4, 157-64.	0.3	55
125	Micrometric segregation of fluorescent membrane lipids: relevance for endogenous lipids and biogenesis in erythrocytes. Journal of Lipid Research, 2013, 54, 1066-1076.	2.0	39
126	RASA1Mutations and Associated Phenotypes in 68 Families with Capillary Malformation-Arteriovenous Malformation. Human Mutation, 2013, 34, 1632-1641.	1.1	221

#	Article	IF	Citations
127	Germline Mutations in RASA1 Are Not Found in Patients with Klippel-Trenaunay Syndrome or Capillary Malformation with Limb Overgrowth. Molecular Syndromology, 2013, 4, 173-178.	0.3	36
128	Mutations in the VEGFR3 Signaling Pathway Explain 36% of Familial Lymphedema. Molecular Syndromology, 2013, 4, 257-266.	0.3	92
129	Ligand oligomerization state controls Tie2 receptor trafficking and Angiopoietin-2 ligand-specific responses. Journal of Cell Science, 2012, 125, 2212-23.	1.2	24
130	Vascular Anomalies: From Genetics toward Models for Therapeutic Trials. Cold Spring Harbor Perspectives in Medicine, 2012, 2, a009688-a009688.	2.9	48
131	Prevalence and Spectrum of SDHx Mutations in Pheochromocytoma and Paraganglioma in Patients from Belgium: An Update. Hormone and Metabolic Research, 2012, 44, 349-353.	0.7	9
132	Chromosome Instability Is Common in Human Cleavage-Stage Embryos. Obstetrical and Gynecological Survey, 2012, 67, 787-788.	0.2	1
133	Classification et gÃ©nÃ©tique des malformations vasculaires. Archives De Pediatrie, 2012, 19, H109-H110.	0.4	0
134	TMEM165 Deficiency Causes a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2012, 91, 15-26.	2.6	162
135	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.	0.7	18
136	Mutations in KIF11 Cause Autosomal-Dominant Microcephaly Variably Associated with Congenital Lymphedema and Chorioretinopathy. American Journal of Human Genetics, 2012, 90, 356-362.	2.6	138
137	Somatic mosaic IDH1 and IDH2 mutations are associated with enchondroma and spindle cell hemangioma in Ollier disease and Maffucci syndrome. Nature Genetics, 2011, 43, 1256-1261.	9.4	488
138	Pathogenesis of Vascular Anomalies. Clinics in Plastic Surgery, 2011, 38, 7-19.	0.7	96
139	A genome-wide association study-derived candidate gene seeks replication: STK39. Journal of Hypertension, 2011, 29, 434-436.	0.3	3
140	Congenital Plaqueâ€Type Glomuvenous Malformations Associated with Fetal Pleural Effusion and Ascites. Pediatric Dermatology, 2011, 28, 528-531.	0.5	15
141	Radio-opaque ethylcellulose-ethanol is a safe and efficient sclerosing agent for venous malformations. European Radiology, 2011, 21, 2647-2656.	2.3	45
142	Cerebrovascular disorders: molecular insights and therapeutic opportunities. Nature Neuroscience, 2011, 14, 1390-1397.	7.1	82
143	FAF1, a Gene that Is Disrupted in Cleft Palate and Has Conserved Function in Zebrafish. American Journal of Human Genetics, 2011, 88, 150-161.	2.6	57
144	5q14.3 neurocutaneous syndrome: A novel continguous gene syndrome caused by simultaneous deletion of RASA1 and MEF2C. American Journal of Medical Genetics, Part A, 2011, 155, 1640-1645.	0.7	28

#	Article	IF	Citations
145	KITLG Mutations Cause Familial Progressive Hyper- and Hypopigmentation. Journal of Investigative Dermatology, 2011, 131, 1234-1239.	0.3	54
146	Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. PLoS Genetics, 2011, 7, e1002050.	1.5	104
147	Refinement of 1p36 Alterations Not Involving PRDM16 in Myeloid and Lymphoid Malignancies. PLoS ONE, 2011, 6, e26311.	1.1	17
148	Molecular identification of aspartate N-acetyltransferase and its mutation in hypoacetylaspartia. Biochemical Journal, 2010, 425, 127-139.	1.7	144
149	Severe congenital lymphoedema not caused by mutations in known lymphoedema genes. British Journal of Dermatology, 2010, 163, 1358-1360.	1.4	2
150	Hereditary cutaneomucosal venous malformations are caused by TIE2 mutations with widely variable hyper-phosphorylating effects. European Journal of Human Genetics, 2010, 18, 414-420.	1.4	152
151	A Novel Association betweenRASA1Mutations and Spinal Arteriovenous Anomalies. American Journal of Neuroradiology, 2010, 31, 775-779.	1.2	91
152	Disruption of ST5 is associated with mental retardation and multiple congenital anomalies. Journal of Medical Genetics, 2010, 47, 91-98.	1.5	12
153	Venous malformation: update on aetiopathogenesis, diagnosis and management. Phlebology, 2010, 25, 224-235.	0.6	250
154	<i>IRF6</i> Screening of Syndromic and a priori Non-Syndromic Cleft Lip and Palate Patients: Identification of a New Type of Minor VWS Sign. Molecular Syndromology, 2010, 1, 67-74.	0.3	28
155	Spectrum and Prevalence of FP/TMEM127 Gene Mutations in Pheochromocytomas and Paragangliomas. JAMA - Journal of the American Medical Association, 2010, 304, 2611.	3.8	174
156	Cutaneous Venous Malformations in Familial Cerebral Cavernomatosis Caused by <i>KRIT1</i> Gene Mutations. Dermatology, 2009, 218, 307-313.	0.9	39
157	Elevated D-dimer Level in the Differential Diagnosis of Venous Malformations. Archives of Dermatology, 2009, 145, 1239-44.	1.7	134
158	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.	1.1	110
159	From germline towards somatic mutations in the pathophysiology of vascular anomalies. Human Molecular Genetics, 2009, 18, R65-R74.	1.4	104
160	Oral Contraceptive and D-Dimer Levelâ€"Reply. Archives of Dermatology, 2009, 145, .	1.7	3
161	Sporadic In Utero Generalized Edema Caused by Mutations in the Lymphangiogenic Genes VEGFR3 and FOXC2. Journal of Pediatrics, 2009, 155, 90-93.	0.9	45
162	Expression profiling of ependymomas unravels localization and tumor gradeâ€specific tumorigenesis. Cancer, 2009, 115, 3955-3968.	2.0	70

#	Article	IF	Citations
163	Somatic mutations in angiopoietin receptor gene TEK cause solitary and multiple sporadic venous malformations. Nature Genetics, 2009, 41, 118-124.	9.4	379
164	Mutations in CCBE1 cause generalized lymph vessel dysplasia in humans. Nature Genetics, 2009, 41, 1272-1274.	9.4	269
165	Recessive primary congenital lymphoedema caused by a VEGFR3 mutation. Journal of Medical Genetics, 2009, 46, 399-404.	1.5	60
166	Chromosome instability is common in human cleavage-stage embryos. Nature Medicine, 2009, 15, 577-583.	15.2	710
167	From Blue Jeans to Blue Genes. Journal of Craniofacial Surgery, 2009, 20, 703-706.	0.3	2
168	Rare presentation of familial paraganglioma without evidence of mutation in the SDH, RET and VHL genes: towards further genetic heterogeneity. Journal of Hypertension, 2009, 27, 76-82.	0.3	2
169	Genetic Aspects of Vascular Malformations. , 2009, , 99-108.		3
170	Parkes Weber syndrome, vein of Galen aneurysmal malformation, and other fast-flow vascular anomalies are caused byRASA1 mutations. Human Mutation, 2008, 29, 959-965.	1.1	382
171	Suppressed NFAT-dependent VEGFR1 expression and constitutive VEGFR2 signaling in infantile hemangioma. Nature Medicine, 2008, 14, 1236-1246.	15.2	325
172	Autosomal-Dominant Microtia Linked to Five Tandem Copies of a Copy-Number-Variable Region at Chromosome 4p16. American Journal of Human Genetics, 2008, 82, 181-187.	2.6	42
173	NEUROSERPIN MUTATION CAUSES ELECTRICAL STATUS EPILEPTICUS OF SLOW-WAVE SLEEP. Neurology, 2008, 71, 64-66.	1.5	60
174	Association of Localized Intravascular Coagulopathy With Venous Malformations. Archives of Dermatology, 2008, 144, 873-7.	1.7	257
175	PTHR1 mutations associated with Ollier disease result in receptor loss of function. Human Molecular Genetics, 2008, 17, 2766-2775.	1.4	103
176	Alpha-cardiac actin mutations produce atrial septal defects. Human Molecular Genetics, 2008, 17, 256-265.	1.4	128
177	High prevalence of SDHB mutations in head and neck paraganglioma in Belgium. Journal of Hypertension, 2008, 26, 1395-1401.	0.3	16
178	Arteriovenous Malformation in Mice and Men. , 2008, , 363-374.		2
179	Genetic causes of vascular malformations. Human Molecular Genetics, 2007, 16, R140-R149.	1.4	226
180	Trisomy 19 ependymoma, a newly recognized genetico-histological association, including clear cell ependymoma. Molecular Cancer, 2007, 6, 47.	7.9	38

#	Article	IF	Citations
181	Vascular Pathologies: Angiogenomics: towards a genetic nosology and understanding of vascular anomalies. European Journal of Human Genetics, 2007, 15, 821-822.	1.4	13
182	Vascular anomalies. Experimental Dermatology, 2007, 16, 877-879.	1.4	1
183	The first de novo mutation in the neuroserpin gene. FASEB Journal, 2007, 21, A30.	0.2	0
184	The first de novo mutation in the neuroserpin gene. Journal of Neuropathology and Experimental Neurology, 2007, 66, 430-431.	0.9	0
185	Genome-Wide Analysis of 30 Burkitt Lymphomas Using High-Density SNP Arrays Reveals Frequent Cryptic Alterations Including Amplification of the microRNA 17-92 Locus.. Blood, 2007, 110, 994-994.	0.6	8
186	Hereditary lymphedema type I associated with VEGFR3 mutation: the first de novo case and atypical presentations. Clinical Genetics, 2006, 70, 330-335.	1.0	75
187	DMP1 mutations in autosomal recessive hypophosphatemia implicate a bone matrix protein in the regulation of phosphate homeostasis. Nature Genetics, 2006, 38, 1248-1250.	9.4	487
188	Localization of candidate regions for a novel gene for Kartagener syndrome. European Journal of Human Genetics, 2006, 14, 809-815.	1.4	6
189	A novel CSX/NKX2-5 mutation causes autosomal-dominant AV block: are atrial fibrillation and syncopes part of the phenotype?. European Journal of Human Genetics, 2006, 14, 1313-1316.	1.4	69
190	Wide clinical spectrum in a family with hereditary lymphedema type I due to a novel missense mutation in VEGFR3. Journal of Human Genetics, 2006, 51, 846-850.	1.1	22
191	A novel mutation in the MSX2 homeobox gene of a family with foramina parietalia permagna, headache and vascular anomaly. European Journal of Pediatrics, 2006, 165, 734-735.	1.3	4
192	Congenital Plaque-Type Glomuvenous Malformations Presenting in Childhood. Archives of Dermatology, 2006, 142, 892-6.	1.7	60
193	Molecular Basis of Vascular Birthmarks. Seminars in Plastic Surgery, 2006, 20, 149-156.	0.8	3
194	Cerebral cavernous malformation: new molecular and clinical insights. Journal of Medical Genetics, 2006, 43, 716-721.	1.5	160
195	Orofacial clefting: update on the role of genetics. , 2006, 2 Suppl 4, 20-4.		5
196	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.	1.4	81
197	Hydrops fetalis: an unusual prenatal presentation of hereditary congenital lymphedema. Prenatal Diagnosis, 2005, 25, 1015-1018.	1.1	44
198	Four common glomulin mutations cause two thirds of glomuvenous malformations ("familial) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 62 T	1.5	126

#	Article	IF	Citations
199	SDH Genes: From Glomic Tumours to Pheochromocytomas. Current Hypertension Reviews, 2005, 1, 267-273.	0.5	1
200	RASA1: variable phenotype with capillary and arteriovenous malformations. Current Opinion in Genetics and Development, 2005, 15, 265-269.	1.5	240
201	Glomuvenous Malformation (Glomangioma) and Venous Malformation. Archives of Dermatology, 2004, 140, 971-6.	1.7	278
202	Six families with van der Woude and/or popliteal pterygium syndrome: all with a mutation in the IRF6 gene. Journal of Medical Genetics, 2004, 41, 15e-15.	1.5	48
203	Glomulin is predominantly expressed in vascular smooth muscle cells in the embryonic and adult mouse. Gene Expression Patterns, 2004, 4, 351-358.	0.3	39
204	A gene encoding a putative FAD-dependent L-2-hydroxyglutarate dehydrogenase is mutated in L-2-hydroxyglutaric aciduria. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 16849-16854.	3.3	166
205	A novel mutation in theSDHD gene in a family with inherited paragangliomas?implications of genetic diagnosis for follow up and treatment. Head and Neck, 2003, 25, 146-151.	0.9	36
206	Vascular malformations: localized defects in vascular morphogenesis. Clinical Genetics, 2003, 63, 340-351.	1.0	103
207	CDKN2A , CDKN2B and p14 ARF are frequently and differentially methylated in ependymal tumours. Neuropathology and Applied Neurobiology, 2003, 29, 574-583.	1.8	55
208	Tumour necrosis and microvascular proliferation are associated with 9p deletion and CDKN2A alterations in 1p/19q -deleted oligodendrogliomas. Neuropathology and Applied Neurobiology, 2003, 29, 462-471.	1.8	31
209	Mutations in the Transcription Factor Gene SOX18 Underlie Recessive and Dominant Forms of Hypotrichosis-Lymphedema-Telangiectasia. American Journal of Human Genetics, 2003, 72, 1470-1478.	2.6	370
210	Capillary Malformationâ€“Arteriovenous Malformation, a New Clinical and Genetic Disorder Caused by RASA1 Mutations. American Journal of Human Genetics, 2003, 73, 1240-1249.	2.6	647
211	Progressive Hyperpigmentation and Generalized Lentiginosis without Associated Systemic Symptoms: a Rare Hereditary Pigmentation Disorder in South-East Germany. Acta Dermato-Venereologica, 2003, 84, 57-60.	0.6	28
212	Mutations in a Novel Factor, Glomulin, Are Responsible for Glomuvenous Malformations (â€œGlomangiomasâ€). American Journal of Human Genetics, 2002, 70, 866-874.	2.6	304
213	Progressive AV-block and anomalous venous return among cardiac anomalies associated with two novel missense mutations in theCSX/NKX2-5 Gene. Human Mutation, 2002, 20, 75-76.	1.1	76
214	Locus for susceptibility for familial capillary malformation (â€˜port-wine stainâ€™) maps to 5q. European Journal of Human Genetics, 2002, 10, 375-380.	1.4	89
215	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	13.5	2,055
216	Molecular genetics of vascular malformations. Matrix Biology, 2001, 20, 327-335.	1.5	182

#	Article	IF	Citations
217	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.	1.4	42
218	Identification of eight novel 5â€²-exons in cerebral capillary malformation gene-1 (CCM1) encoding KRIT1. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2001, 1517, 464-467.	2.4	39
219	KRIT1 is mutated in hyperkeratotic cutaneous capillary-venous malformation associated with cerebral capillary malformation. Human Molecular Genetics, 2000, 9, 1351-1355.	1.4	134
220	High Frequency of Autologous Anti-Melanoma CTL Directed Against an Antigen Generated by a Point Mutation in a New Helicase Gene. Journal of Immunology, 2000, 164, 6057-6066.	0.4	85
221	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.	1.3	42
222	Congenital Hereditary Lymphedema Caused by a Mutation That Inactivates VEGFR3 Tyrosine Kinase. American Journal of Human Genetics, 2000, 67, 295-301.	2.6	380
223	The putative glucose 6-phosphate translocase gene is mutated in essentially all cases of glycogen storage disease type I non-a. European Journal of Human Genetics, 1999, 7, 717-723.	1.4	97
224	Endothelial receptor tyrosine kinases activate the STAT signaling pathway: mutant Tie-2 causing venous malformations signals a distinct STAT activation response. Oncogene, 1999, 18, 1-8.	2.6	132
225	A Gene for Inherited Cutaneous Venous Anomalies (â€œGlomangiomasâ€) Localizes to Chromosome 1p21-22. American Journal of Human Genetics, 1999, 65, 125-133.	2.6	156
226	Splicing Mutations of 54-bp Exons in the COL11A1 Gene Cause Marshall Syndrome, but Other Mutations Cause Overlapping Marshall/Stickler Phenotypes. American Journal of Human Genetics, 1999, 65, 974-983.	2.6	235
227	Molecular Basis of Vascular Anomalies. Trends in Cardiovascular Medicine, 1998, 8, 281-292.	2.3	109
228	A Gene on Chromosome 11q23 Coding for a Putative Glucose- 6-Phosphate Translocase Is Mutated in Glycogen-Storage Disease Types Ib and Ic. American Journal of Human Genetics, 1998, 63, 976-983.	2.6	116
229	The gene responsible for pseudohypoparathyroidism type Ib is paternally imprinted and maps in four unrelated kindreds to chromosome 20q13.3. Proceedings of the National Academy of Sciences of the United States of America, 1998, 95, 11798-11803.	3.3	184
230	Vascular Dysmorphogenesis Caused by an Activating Mutation in the Receptor Tyrosine Kinase TIE2. Cell, 1996, 87, 1181-1190.	13.5	734
231	Unravelling the Molecular Genetics of Osteoarthrosis. Annals of Medicine, 1996, 28, 301-304.	1.5	18
232	Osteoporosis-pseudoglioma syndrome, a disorder affecting skeletal strength and vision, is assigned to chromosome region 11q12-13. American Journal of Human Genetics, 1996, 59, 146-51.	2.6	174
233	Identification of col2a1 gene mutations in patients with chondrodysplasias and familial osteoarthritis. Arthritis and Rheumatism, 1995, 38, 999-1004.	6.7	66
234	A gene for familial venous malformations maps to chromosome 9p in a second large kindred.. Journal of Medical Genetics, 1995, 32, 197-199.	1.5	139

#	Article	IF	Citations
235	Autosomal dominant and recessive osteochondrodysplasias associated with the COL11A2 locus. Cell, 1995, 80, 431-437.	13.5	390
236	Assignment of a locus for dominantly inherited venous malformations to chromosome 9p. Human Molecular Genetics, 1994, 3, 1583-1587.	1.4	208
237	Type II Collagen Mutations in Rare and Common. Annals of Medicine, 1994, 26, 107-114.	1.5	86
238	Early-onset osteoarthritis linked to the type ii procollagen gene. detailed clinical phenotype and further analyses of the gene. Arthritis and Rheumatism, 1993, 36, 401-409.	6.7	62
239	A Mutation in the Amino-Terminal End of the Triple Helix of Type II Collagen Causing Severe Osteochondrodysplasia. Genomics, 1993, 16, 282-285.	1.3	31
240	Multiallelic polymorphism of the cartilage collagen gene: no association with osteoarthrosis.. Annals of the Rheumatic Diseases, 1993, 52, 762-764.	0.5	24
241	Structural analysis of the regulatory elements of the type-II procollagen gene. Conservation of promoter and first intron sequences between human and mouse. Biochemical Journal, 1992, 285, 287-294.	1.7	31
242	PREDISPOSITION TO FAMILIAL OSTEOARTHROSIS LINKED TO TYPE II COLLAGEN GENE. Lancet, The, 1989, 333, 924-927.	6.3	141
243	Structural analyses of the polymorphic area in type II collagen gene. FEBS Letters, 1989, 250, 171-174.	1.3	14