Jamie McDonald

List of Publications by Year in descending order

34 papers	2,668 citations	393982 19 h-index	377514 34 g-index
34 all docs	34 docs citations	34 times ranked	3028 citing authors

#	Article	IF	Citations
1	Hereditary hemorrhagic telangiectasia: genetics and molecular diagnostics in a new era. Frontiers in Genetics, 2015, 6, 1.	1.1	489
2	Hereditary hemorrhagic telangiectasia: An overview of diagnosis, management, and pathogenesis. Genetics in Medicine, 2011, 13, 607-616.	1.1	315
3	BMP9 Mutations Cause a Vascular-Anomaly Syndrome with Phenotypic Overlap with Hereditary Hemorrhagic Telangiectasia. American Journal of Human Genetics, 2013, 93, 530-537.	2.6	270
4	Second International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia. Annals of Internal Medicine, 2020, 173, 989-1001.	2.0	244
5	A fourth locus for hereditary hemorrhagic telangiectasia maps to chromosome 7. American Journal of Medical Genetics, Part A, 2006, 140A, 2155-2162.	0.7	205
6	Genotypeâ€“phenotype correlation in hereditary hemorrhagic telangiectasia: Mutations and manifestations. American Journal of Medical Genetics, Part A, 2006, 140A, 463-470.	0.7	188
7	Pathogenic variants that alter protein code often disrupt splicing. Nature Genetics, 2017, 49, 848-855.	9.4	176
8	Intracranial Hemorrhage in Infants and Children With Hereditary Hemorrhagic Telangiectasia (Osler-Weber-Rendu Syndrome). Pediatrics, 2002, 109, e12-e12.	1.0	113
9	Hereditary hemorrhagic telangiectasia: An overview of diagnosis and management in the molecular era for clinicians. Genetics in Medicine, 2004, 6, 175-191.	1.1	112
10	Appreciating the broad clinical features of SMAD4 mutation carriers: a multicenter chart review. Genetics in Medicine, 2014, 16, 588-593.	1.1	62
11	Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation. European Journal of Human Genetics, 2018, 26, 1521-1536.	1.4	42
12	Clinical and Analytical Sensitivities in Hereditary Hemorrhagic Telangiectasia Testing and a Report of de Novo Mutations. Journal of Molecular Diagnostics, 2007, 9, 258-265.	1.2	41
13	Phenotype of CM-AVM2 caused by variants in EPHB4: how much overlap with hereditary hemorrhagic telangiectasia (HHT)?. Genetics in Medicine, 2019, 21, 2007-2014.	1.1	38
14	RASA1 analysis: Clinical and molecular findings in a series of consecutive cases. European Journal of Medical Genetics, 2012, 55, 91-95.	0.7	37
15	Potential Second-Hits in Hereditary Hemorrhagic Telangiectasia. Journal of Clinical Medicine, 2020, 9, 3571.	1.0	37
16	CuraÃ§ao diagnostic criteria for hereditary hemorrhagic telangiectasia is highly predictive of a pathogenic variant in ENG or ACVRL1 (HHT1 and HHT2). Genetics in Medicine, 2020, 22, 1201-1205.	1.1	37
17	Mosaic ACVRL1 and ENG mutations in hereditary haemorrhagic telangiectasia patients. Journal of Medical Genetics, 2011, 48, 358-360.	1.5	32
18	Likelihood ratios to assess genetic evidence for clinical significance of uncertain variants: Hereditary hemorrhagic telangiectasia as a model. Experimental and Molecular Pathology, 2008, 85, 45-49.	0.9	28

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19	5'UTR mutations of ENG cause hereditary hemorrhagic telangiectasia. Orphanet Journal of Rare Diseases, 2011, 6, 85.	1.2	27
20	Inactivating mutations in Drosha mediate vascular abnormalities similar to hereditary hemorrhagic telangiectasia. Science Signaling, 2018, 11, .	1.6	23
21	Homozygous GDF2 nonsense mutations result in a loss of circulating BMP9 and BMP10 and are associated with either PAH or an â€œHHTâ€likeâ€•syndrome in children. Molecular Genetics & Genomic Medicine, 2021, 9, e1685.	0.6	19
22	Spinal arteriovenous fistulas in children with hereditary hemorrhagic telangiectasia. Journal of Neurosurgery: Pediatrics, 2012, 9, 654-659.	0.8	18
23	Epistaxis in children and adolescents with hereditary hemorrhagic telangiectasia. Laryngoscope, 2018, 128, 1714-1719.	1.1	17
24	Localization and age distribution of telangiectases in children and adolescents with hereditary hemorrhagic telangiectasia: A retrospective cohort study. Journal of the American Academy of Dermatology, 2019, 81, 950-955.	0.6	14
25	Genome sequencing reveals a deep intronic splicing ACVRL1 mutation hotspot in Hereditary Haemorrhagic Telangiectasia. Journal of Medical Genetics, 2018, 55, 824-830.	1.5	13
26	Genotypeâ€“Phenotype Correlations in Children with HHT. Journal of Clinical Medicine, 2020, 9, 2714.	1.0	12
27	Clinical presentation and treatment paradigms of brain arteriovenous malformations in patients with hereditary hemorrhagic telangiectasia. Journal of Clinical Neuroscience, 2018, 51, 22-28.	0.8	11
28	An evaluation of the severity and progression of epistaxis in hereditary hemorrhagic telangiectasia 1 versus hereditary hemorrhagic telangiectasia 2. Laryngoscope, 2016, 126, 786-790.	1.1	9
29	Life experiences of individuals with hereditary hemorrhagic telangiectasia and disclosing outside the family: a qualitative analysis. Journal of Community Genetics, 2016, 7, 81-89.	0.5	9
30	Tissueâ€specific mosaicism in hereditary hemorrhagic telangiectasia: Implications for genetic testing in families. American Journal of Medical Genetics, Part A, 2018, 176, 1618-1621.	0.7	9
31	The effects of nasal closure on quality of life in patients with hereditary hemorrhagic telangiectasia. Laryngoscope Investigative Otolaryngology, 2018, 3, 178-181.	0.6	8
32	Characterization of a family mutation in the 5â€™ untranslated region of the endoglin gene causative of hereditary hemorrhagic telangiectasia. Journal of Human Genetics, 2019, 64, 333-339.	1.1	7
33	Clinical presentation and treatment paradigms in patients with hereditary hemorrhagic telangiectasia and spinal vascular malformations. Journal of Clinical Neuroscience, 2018, 50, 51-57.	0.8	3
34	Vitamin D levels are associated with epistaxis severity and bleeding duration in hereditary hemorrhagic telangiectasia. Biomarkers in Medicine, 2018, 12, 365-371.	0.6	3