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List of Publications by Year in descending order

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840119 996533 15 1,026 11 15 citations h-index g-index papers 15 15 15 1846 docs citations times ranked all docs citing authors

#	Article	IF	CITATIONS
1	Homozygous <i>GDF2</i> nonsense mutations result in a loss of circulating BMP9 and BMP10 and are associated with either PAH or an "HHTâ€like―syndrome in children. Molecular Genetics & Genomic Medicine, 2021, 9, e1685.	0.6	19
2	Parkes Weber syndrome associated with two somatic pathogenic variants in <i>RASA1</i> . Journal of Physical Education and Sports Management, 2020, 6, a005256.	0.5	13
3	Curaçao 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
4	Characterization of a family mutation in the 5' untranslated region of the endoglin gene causative of hereditary hemorrhagic telangiectasia. Journal of Human Genetics, 2019, 64, 333-339.	1.1	7
5	Inactivating mutations in Drosha mediate vascular abnormalities similar to hereditary hemorrhagic telangiectasia. Science Signaling, 2018, 11, .	1.6	23
6	Genome sequencing reveals a deep intronic splicing <i>ACVRL1</i> mutation hotspot in Hereditary Haemorrhagic Telangiectasia. Journal of Medical Genetics, 2018, 55, 824-830.	1.5	13
7	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
8	Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation. European Journal of Human Genetics, 2018, 26, 1521-1536.	1.4	42
9	Variable expressivity and incomplete penetrance in a large family with nonâ€classical Diamondâ€Blackfan anemia associated with <i>ribosomal protein L11</i> splicing variant. American Journal of Medical Genetics, Part A, 2017, 173, 2622-2627.	0.7	14
10	Genetic Variants Associated with Port-Wine Stains. PLoS ONE, 2015, 10, e0133158.	1.1	35
11	Executive summary of the 11th HHT international scientific conference. Angiogenesis, 2015, 18, 511-524.	3.7	24
12	Hereditary hemorrhagic telangiectasia: genetics and molecular diagnostics in a new era. Frontiers in Genetics, 2015 , 6 , 1 .	1.1	489
13	Noncontinuously Binding Loop-Out Primers for Avoiding Problematic DNA Sequences in PCR and Sanger Sequencing. Journal of Molecular Diagnostics, 2014, 16, 477-480.	1.2	4
14	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
15	5'UTR mutations of ENG cause hereditary hemorrhagic telangiectasia. Orphanet Journal of Rare Diseases, 2011, 6, 85.	1.2	27