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List of Publications by Year in descending order

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Version: 2024-02-01

15
papers

1,026
citations

840119

11
h-index

996533

15
g-index

15
all docs

15
docs citations

15
times ranked

1846
citing authors

#	ARTICLE	IF	CITATIONS
1	Hereditary hemorrhagic telangiectasia: genetics and molecular diagnostics in a new era. <i>Frontiers in Genetics</i> , 2015, 6, 1.	1.1	489
2	BMP9 Mutations Cause a Vascular-Anomaly Syndrome with Phenotypic Overlap with Hereditary Hemorrhagic Telangiectasia. <i>American Journal of Human Genetics</i> , 2013, 93, 530-537.	2.6	270
3	Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation. <i>European Journal of Human Genetics</i> , 2018, 26, 1521-1536.	1.4	42
4	CuraÃ§ao diagnostic criteria for hereditary hemorrhagic telangiectasia is highly predictive of a pathogenic variant in ENG or ACVRL1 (HHT1 and HHT2). <i>Genetics in Medicine</i> , 2020, 22, 1201-1205.	1.1	37
5	Genetic Variants Associated with Port-Wine Stains. <i>PLoS ONE</i> , 2015, 10, e0133158.	1.1	35
6	5'UTR mutations of ENG cause hereditary hemorrhagic telangiectasia. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 85.	1.2	27
7	Executive summary of the 11th HHT international scientific conference. <i>Angiogenesis</i> , 2015, 18, 511-524.	3.7	24
8	Inactivating mutations in Drosha mediate vascular abnormalities similar to hereditary hemorrhagic telangiectasia. <i>Science Signaling</i> , 2018, 11, .	1.6	23
9	Homozygous <i>GDF2</i> nonsense mutations result in a loss of circulating BMP9 and BMP10 and are associated with either PAH or an "HHT-like" syndrome in children. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1685.	0.6	19
10	Variable expressivity and incomplete penetrance in a large family with non-classical Diamond-Blackfan anemia associated with <i>ribosomal protein L11</i> splicing variant. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2622-2627.	0.7	14
11	Genome sequencing reveals a deep intronic splicing <i>ACVRL1</i> mutation hotspot in Hereditary Haemorrhagic Telangiectasia. <i>Journal of Medical Genetics</i> , 2018, 55, 824-830.	1.5	13
12	Parkes Weber syndrome associated with two somatic pathogenic variants in <i>RASA1</i> . <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a005256.	0.5	13
13	Tissue-specific mosaicism in hereditary hemorrhagic telangiectasia: Implications for genetic testing in families. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1618-1621.	0.7	9
14	Characterization of a family mutation in the 5' untranslated region of the endoglin gene causative of hereditary hemorrhagic telangiectasia. <i>Journal of Human Genetics</i> , 2019, 64, 333-339.	1.1	7
15	Noncontinuously Binding Loop-Out Primers for Avoiding Problematic DNA Sequences in PCR and Sanger Sequencing. <i>Journal of Molecular Diagnostics</i> , 2014, 16, 477-480.	1.2	4