AndrÃ© Schaller

List of Publications by Year
in descending order

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62 papers	2,006 citations	304743 22 h-index	254184 43 g-index
63 all docs	63 docs citations	63 times ranked	3344 citing authors

#	Article	IF	Citations
1	Mutations in the Tight-Junction Gene Claudin 19 (CLDN19) Are Associated with Renal Magnesium Wasting, Renal Failure, and Severe Ocular Involvement. American Journal of Human Genetics, 2006, 79, 949-957.	6.2	446
2	The gene for histone RNA hairpin binding protein is located on human chromosome 4and encodes a novel type of RNA binding protein. EMBO Journal, 1997, 16, 769-778.	7.8	142
3	Loss of insulin-induced activation of TRPM6 magnesium channels results in impaired glucose tolerance during pregnancy. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 11324-11329.	7.1	122
4	Neutrophil extracellular trap formation requires OPA1-dependent glycolytic ATP production. Nature Communications, 2018, 9, 2958.	12.8	121
5	CLDN16 Genotype Predicts Renal Decline in Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis. Journal of the American Society of Nephrology: JASN, 2008, 19, 171-181.	6.1	111
6	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	3.7	90
7	Mutations inSDHDlead to autosomal recessive encephalomyopathy and isolated mitochondrial complex II deficiency. Journal of Medical Genetics, 2014, 51, 170-175.	3.2	75
8	The stem-loop binding protein stimulates histone translation at an early step in the initiation pathway. Rna, 2005, 11, 1030-1042.	3.5	59
9	The role of common single-nucleotide polymorphisms on exon 9 and exon 12 skipping in nonmutatedCFTR Alleles. Human Mutation, 2004, 24, 120-129.	2.5	50
10	Evaluation of genome-wide loci of iron metabolism in hereditary hemochromatosis identifies PCSK7 as a host risk factor of liver cirrhosis. Human Molecular Genetics, 2014, 23, 3883-3890.	2.9	50
11	The Vacuolar H+-ATPase B1 Subunit Polymorphism p.E161K Associates with Impaired Urinary Acidification in Recurrent Stone Formers. Journal of the American Society of Nephrology: JASN, 2016, 27, 1544-1554.	6.1	48
12	Broad phenotypes in heterozygous NR5A1 46,XY patients with a disorder of sex development: an oligogenic origin?. European Journal of Human Genetics, 2018, 26, 1329-1338.	2.8	47
13	Mitochondrial neurogastrointestinal encephalomyopathy in three siblings. Journal of Neurology, 2007, 254, 146-153.	3.6	36
14	A variant inMRPS14(uS14m) causes perinatal hypertrophic cardiomyopathy with neonatal lactic acidosis, growth retardation, dysmorphic features and neurological involvement. Human Molecular Genetics, 2019, 28, 639-649.	2.9	33
15	Molecular and biochemical characterisation of a novel mutation in POLGassociated with Alpers syndrome. BMC Neurology, 2011, 11, 4.	1.8	31
16	Rapid typing ofMoraxella catarrhalis subpopulations based on outer membrane proteins using mass spectrometry. Proteomics, 2006, 6, 172-180.	2.2	30
17	Usefulness of Genetic Testing in Sudden Cardiac Arrest Survivors With or Without Previous Clinical Evidence of Heart Disease. American Journal of Cardiology, 2019, 123, 2031-2038.	1.6	30
18	SDHA mutation with dominant transmission results in complex II deficiency with ocular, cardiac, and neurologic involvement. American Journal of Medical Genetics, Part A, 2017, 173, 225-230.	1.2	26

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19	Multiple clinical profiles of families with the short QT syndrome. Europace, 2018, 20, f113-f121.	1.7	26
20	A novel mitochondrial ATP6 frameshift mutation causing isolated complex V deficiency, ataxia and encephalomyopathy. European Journal of Medical Genetics, 2017, 60, 345-351.	1.3	25
21	Quantitative 1-Step DNA Methylation Analysis with Native Genomic DNA as Template. Clinical Chemistry, 2010, 56, 1098-1106.	3.2	24
22	Sensitive and rapid detection of ganciclovir resistance by PCR based MALDI-TOF analysis. Journal of Clinical Virology, 2012, 54, 359-363.	3.1	23
23	Diagnosis of adult-onset MELAS syndrome in a 63-year-old patient with suspected recurrent strokes â€“ a case report. BMC Neurology, 2019, 19, 91.	1.8	23
24	qPCR-based mitochondrial DNA quantification: Influence of template DNA fragmentation on accuracy. Biochemical and Biophysical Research Communications, 2012, 423, 441-447.	2.1	21
25	Movement disorders in genetically confirmed mitochondrial disease and the putative role of the cerebellum. Movement Disorders, 2018, 33, 146-155.	3.9	21
26	Functional importance of conserved nucleotides at the histone RNA 3' processing site. Rna, 1998, 4, 246-56.	3.5	21
27	Identification of SNPs in the cystic fibrosis interactome influencing pulmonary progression in cystic fibrosis. European Journal of Human Genetics, 2013, 21, 397-403.	2.8	20
28	Impairment of mitochondrial tRNAIle processing by a novel mutation associated with chronic progressive external ophthalmoplegia. Mitochondrion, 2011, 11, 488-496.	3.4	18
29	Phenotypic Spectrum of HCN4 Mutations. Circulation Genomic and Precision Medicine, 2018, 11, e002033.	3.6	18
30	Physiologic cold shock of Moraxella catarrhalis affects the expression of genes involved in the iron acquisition, serum resistance and immune evasion. BMC Microbiology, 2011, 11, 182.	3.3	17
31	Handling mammalian mitochondrial tRNAs and aminoacyl-tRNA synthetases for functional and structural characterization. Methods, 2008, 44, 176-189.	3.8	15
32	Effects of endurance training on skeletal muscle mitochondrial function in Huntington disease patients. Orphanet Journal of Rare Diseases, 2017, 12, 184.	2.7	15
33	Age-dependent suppression of SERCA2a mRNA in pediatric atrial myocardium. Biochemical and Biophysical Research Communications, 2005, 326, 344-348.	2.1	14
34	A novel mutation in BCS1L associated with deafness, tubulopathy, growth retardation and microcephaly. European Journal of Pediatrics, 2016, 175, 517-525.	2.7	14
35	Outer membrane porin M35 of Moraxella catarrhalis mediates susceptibility to aminopenicillins. BMC Microbiology, 2009, 9, 188.	3.3	13
36	Contiguous â^¼16â€‰Mb 1p36 deletion: Dominant features of classical distal 1p36 monosomy with haploâ€‹lethality. American Journal of Medical Genetics, Part A, 2011, 155, 1964-1968.	1.2	12

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37	Novel mitochondrial tRNAIle.4282A>G gene mutation leads to chronic progressive external ophthalmoplegia plus phenotype. British Journal of Ophthalmology, 2014, 98, 1453-1459.	3.9	11
38	Mitochondrial tRNA^{Leu(UUR)} mutation m.3302Aâ€‰>â€‰G presenting as childhoodâ€onset severe myopathy: threshold determination through segregation study. Journal of Inherited Metabolic Disease, 2010, 33, 219-226.	3.6	10
39	Simultaneous quantitative detection of relevant biomarkers in breast cancer by quantitative real-time PCR. International Journal of Biological Markers, 2006, 21, 30-39.	1.8	10
40	Characterization of the Calf Thymus Hairpin-binding Factor Involved in Histone Pre-mRNA 3â€² End Processing. Journal of Biological Chemistry, 1997, 272, 10435-10441.	3.4	8
41	Gender Modulates the Expression of Calcium-Regulating Proteins in Pediatric Atrial Myocardium. Experimental Biology and Medicine, 2005, 230, 853-859.	2.4	8
42	Clinical and molecular characterization of the potential CF disease modifier syntaxin 1A. European Journal of Human Genetics, 2013, 21, 1462-1466.	2.8	8
43	Mitochondrial leucine tRNA level and PTCD1 are regulated in response to leucine starvation. Amino Acids, 2014, 46, 1775-1783.	2.7	8
44	The CFTR frameshift mutation 3905insT and its effect at transcript and protein level. European Journal of Human Genetics, 2010, 18, 212-217.	2.8	7
45	Heterologous expression from the human D-Loop in organello. Mitochondrion, 2014, 17, 67-75.	3.4	7
46	A Novel Van der Woude Syndrome-Causing IRF6 Variant Is Subject to Incomplete Non-sense-Mediated mRNA Decay Affecting the Phenotype of Keratinocytes. Frontiers in Cell and Developmental Biology, 2020, 8, 583115.	3.7	7
47	Unexplained cardiac arrest: a tale of conflicting interpretations of KCNQ1 genetic test results. Clinical Research in Cardiology, 2018, 107, 670-678.	3.3	6
48	Characterization of two novel intronic OPA1 mutations resulting in aberrant pre-mRNA splicing. BMC Medical Genetics, 2017, 18, 22.	2.1	5
49	Triheptanoin â€“ Novel therapeutic approach for the ultra-rare disease mitochondrial malate dehydrogenase deficiency. Molecular Genetics and Metabolism Reports, 2021, 29, 100814.	1.1	5
50	Analysis of Inherited Optic Neuropathies. Klinische Monatsblatter Fur Augenheilkunde, 2019, 236, 451-461.	0.5	4
51	Longitudinal case study and phenotypic multimodal characterization of McArdle disease-linked retinopathy: insight into pathomechanisms. Ophthalmic Genetics, 2020, 41, 73-78.	1.2	4
52	Rare Case of Transcutaneous Oxygen Desaturation in a Cancer Patient. A&A Practice, 2019, 12, 96-98.	0.4	3
53	Sodium Pump Reduction Correlates with Aortic Clamp Time in Pediatric Heart Surgery. Experimental Biology and Medicine, 2006, 231, 1300-1305.	2.4	2
54	Cardiopulmonary bypass reduces atrial Na+â€“K+-ATPase expression in children. Biochemical and Biophysical Research Communications, 2005, 335, 700-704.	2.1	1

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55	Reduced atrial connexin43 expression after pediatric heart surgery. Biochemical and Biophysical Research Communications, 2006, 342, 310-315.	2.1	1
56	Rapid and reliable genotyping of polymorphic loci modifying correct splicing of CFTR pre-mRNA using mass spectrometry. European Journal of Human Genetics, 2007, 15, 53-61.	2.8	1
57	Mitochondrial cytopathy with common MELAS mutation presenting as multiple system atrophy mimic. Neurology: Genetics, 2016, 2, e121.	1.9	1
58	Lateâ€onset severe long QT syndrome. Annals of Noninvasive Electrocardiology, 2018, 23, e12517.	1.1	1
59	Earlyâ€onset leukoencephalomyelopathy due to a biallelic <scp>NDUFV1</scp> variant in a midâ€forties patient. Annals of Clinical and Translational Neurology, 2022, 9, 888-892.	3.7	1
60	Mitochondrial Encephalopathy with CADASIL-Like MRI. European Neurology, 2007, 58, 185-188.	1.4	0
61	Erleichterung und Sorge, Hoffnung und Angst â€“ der Weg zur Diagnosestellung einer SLC16A2-Mutation. Paediatrica, 2021, 32, .	0.1	0
62	Absence of Genotype/Phenotype Correlations Requires Molecular Diagnostic to Ascertain Stargardt and Stargardt-Like Swiss Patients. Genes, 2021, 12, 812.	2.4	0