Andrea L Sertie

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5962235/publications.pdf

Version: 2024-02-01

28	1,596	394421 19	501196 28
papers	citations	h-index	g-index

28	28	28	2121
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical spectrum of fibroblast growth factor receptor mutations. Human Mutation, 1999, 14, 115-125.	2.5	284
2	Collagen XVIII, containing an endogenous inhibitor of angiogenesis and tumor growth, plays a critical role in the maintenance of retinal structure and in neural tube closure (Knobloch syndrome). Human Molecular Genetics, 2000, 9, 2051-2058.	2.9	259
3	Neto1 Is a Novel CUB-Domain NMDA Receptorâ€“Interacting Protein Required for Synaptic Plasticity and Learning. PLoS Biology, 2009, 7, e1000041.	5.6	150
4	The Seventh Form of Autosomal Recessive Limb-Girdle Muscular Dystrophy Is Mapped to 17q11-12. American Journal of Human Genetics, 1997, 61, 151-159.	6.2	136
5	Molecular Analysis of Collagen XVIII Reveals Novel Mutations, Presence of a Third Isoform, and Possible Genetic Heterogeneity in Knobloch Syndrome. American Journal of Human Genetics, 2002, 71, 1320-1329.	6.2	128
6	A gene which causes severe ocular alterations and occipital encephalocele (Knobloch syndrome) is mapped to 21q22.3. Human Molecular Genetics, 1996, 5, 843-847.	2.9	76
7	Decreased cellular uptake and metabolism in Allan-Herndon-Dudley syndrome (AHDS) due to a novel mutation in the MCT8 thyroid hormone transporter. Journal of Medical Genetics, 2005, 43, 457-460.	3.2	59
8	New SMS mutation leads to a striking reduction in spermine synthase protein function and a severe form of Snyder-Robinson X-linked recessive mental retardation syndrome. Journal of Medical Genetics, 2008, 45, 539-543.	3.2	53
9	Mutations in collagen 18A1 (COL18A1) and their relevance to the human phenotype. Anais Da Academia Brasileira De Ciencias, 2006, 78, 123-131.	0.8	50
10	Dysfunctional mTORC1 Signaling: A Convergent Mechanism between Syndromic and Nonsyndromic Forms of Autism Spectrum Disorder?. International Journal of Molecular Sciences, 2017, 18, 659.	4.1	47
11	Description of a new mutation and characterization ofFGFR1, FGFR2, andFGFR3 mutations among Brazilian patients with syndromic craniosynostoses. American Journal of Medical Genetics Part A, 1998, 78, 237-241.	2.4	41
12	HighOCT4and Low<mml:math xmlns:mml="http://www.w3.org/1998/Math/MathML" id="M1"><mml:mrow><mml:msup><mml:mrow><mml:mtext>p16</mml:mtext></mml:mrow><mml:mrow><mml:mtext>INK4A</mml:mtext></mml:mrow></mml:msup></mml:mrow></mml:math>DetermineIn VitroLifespan of Mesenchymal Stem Cells. Stem Cells International, 2015, 2015, 1-11.	2.1	34
13	Linkage Analysis in a Large Brazilian Family with van der Woude Syndrome Suggests the Existence of a Susceptibility Locus for Cleft Palate at 17p11.2-11.1. American Journal of Human Genetics, 1999, 65, 433-440.	6.2	33
14	Collybistin binds and inhibits mTORC1 signaling: a potential novel mechanism contributing to intellectual disability and autism. European Journal of Human Genetics, 2016, 24, 59-65.	2.8	31
15	Apert p.Ser252Trp Mutation in FGFR2 Alters Osteogenic Potential and Gene Expression of Cranial Periosteal Cells. Molecular Medicine, 2007, 13, 422-442.	4.4	28
16	RareRELNvariants affect Reelin-DAB1 signal transduction in autism spectrum disorder. Human Mutation, 2018, 39, 1372-1383.	2.5	28
17	Pfeiffer mutation in an apert patient: How wide is the spectrum of variability due to mutations in the FGFR2 gene?. American Journal of Medical Genetics Part A, 1997, 71, 243-245.	2.4	27
18	Effects of antipsychotics with different weight gain liabilities on human in vitro models of adipose tissue differentiation and metabolism. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2011, 35, 1884-1890.	4.8	26

#	Article	IF	Citations
19	COL18A1 is highly expressed during human adipocyte differentiation and the SNP c.1136C > T in its "frizzled" motif is associated with obesity in diabetes type 2 patients. Anais Da Academia Brasileira De Ciencias, 2008, 80, 167-177.	0.8	21
20	Altered mTORC1 signaling in multipotent stem cells from nearly 25% of patients with nonsyndromic autism spectrum disorders. Molecular Psychiatry, 2015, 20, 551-552.	7.9	17
21	Presence of the Apert canonical S252W FGFR2 mutation in a patient without severe syndactyly.. Journal of Medical Genetics, 1998, 35, 677-679.	3.2	13
22	Crouzon syndrome: Association with absent pulmonary valve syndrome and severe tracheobronchomalacia. Pediatric Pulmonology, 2002, 34, 478-481.	2.0	12
23	Different gene expression profiles in iPSC-derived motor neurons from ALS8 patients with variable clinical courses suggest mitigating pathways for neurodegeneration. Human Molecular Genetics, 2020, 29, 1465-1475.	2.9	10
24	Collybistin and gephyrin are novel components of the eukaryotic translation initiation factor 3 complex. BMC Research Notes, 2010, 3, 242.	1.4	9
25	10q23.31 microduplication encompassing PTEN decreases mTOR signalling activity and is associated with autosomal dominant primary microcephaly. Journal of Medical Genetics, 2019, 56, 543-547.	3.2	9
26	How pathogenic is the p.D104N/endostatin polymorphic allele ofCOL18A1in Knobloch syndrome?. Human Mutation, 2005, 25, 314-315.	2.5	5
27	<scp>MS</scp>â€Ðriven Metabolic Alterations Are Recapitulated in <scp>iPSC</scp>â€Ðerived Astrocytes. Annals of Neurology, 2022, 91, 652-669.	5.3	5
28	Craniosynostosis associated with ocular and distal limb defects is very likely caused by mutations in a gene different fromFGFR,TWIST, andMSX2. American Journal of Medical Genetics Part A, 2002, 113, 200-206.	2.4	4