Angela Huebner

List of Publications by Year
in descending order

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24	1,681	567281 15	642732 23
papers	citations	h-index	g-index

24	24	24	1927
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in MRAP, encoding a new interacting partner of the ACTH receptor, cause familial glucocorticoid deficiency type 2. Nature Genetics, 2005, 37, 166-170.	21.4	388
2	A founder mutation in Anoctamin 5 is a major cause of limb girdle muscular dystrophy. Brain, 2011, 134, 171-182.	7.6	254
3	Triple A syndrome is caused by mutations in AAAS, a new WD-repeat protein gene. Human Molecular Genetics, 2001, 10, 283-290.	2.9	231
4	Congenital Adrenal Hyperplasiaâ€"Current Insights in Pathophysiology, Diagnostics, and Management. Endocrine Reviews, 2022, 43, 91-159.	20.1	182
5	Nonclassic Lipoid Congenital Adrenal Hyperplasia Masquerading as Familial Glucocorticoid Deficiency. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 3865-3871.	3.6	138
6	Tall stature in familial glucocorticoid deficiency. Clinical Endocrinology, 2000, 53, 423-430.	2.4	88
7	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	2.6	69
8	Salbutamol-responsive limb-girdle congenital myasthenic syndrome due to a novel missense mutation and heteroallelic deletion in MUSK. Neuromuscular Disorders, 2014, 24, 31-35.	0.6	49
9	The nucleoporin ALADIN regulates Aurora A localization to ensure robust mitotic spindle formation. Molecular Biology of the Cell, 2015, 26, 3424-3438.	2.1	45
10	Mice Lacking the Nuclear Pore Complex Protein ALADIN Show Female Infertility but Fail To Develop a Phenotype Resembling Human Triple A Syndrome. Molecular and Cellular Biology, 2006, 26, 1879-1887.	2.3	41
11	Genetic Analysis of Pediatric Primary Adrenal Insufficiency of Unknown Etiology: 25 Yearsâ€™ Experience in the UK. Journal of the Endocrine Society, 2021, 5, bvab086.	0.2	34
12	Age-specific pediatric reference intervals for plasma free normetanephrine, metanephrine, 3-methoxytyramine and 3-O-methyldopa: Particular importance for early infancy. Clinica Chimica Acta, 2019, 494, 100-105.	1.1	29
13	Phenotypeâ€"genotype spectrum of AAA syndrome from Western India and systematic review of literature. Endocrine Connections, 2017, 6, 901-913.	1.9	28
14	Chromosomal fragility in patients with triple A syndrome. American Journal of Medical Genetics Part A, 2003, 117A, 30-36.	2.4	20
15	Cancer Stem Cells in Pheochromocytoma and Paraganglioma. Frontiers in Endocrinology, 2020, 11, 79.	3.5	20
16	Effects of androgen excess and glucocorticoid exposure on bone health in adult patients with 21-hydroxylase deficiency. Journal of Steroid Biochemistry and Molecular Biology, 2020, 204, 105734.	2.5	19
17	Biochemical testing for neuroblastoma using plasma free 3â€Oâ€methyldopa, 3â€methoxytyramine, and normetanephrine. Pediatric Blood and Cancer, 2020, 67, e28081.	1.5	14
18	ALADIN is required for the production of fertile mouse oocytes. Molecular Biology of the Cell, 2017, 28, 2470-2478.	2.1	13

#	Article	IF	Citations
19	Prenatal dexamethasone treatment for classic 21-hydroxylase deficiency in Europe. European Journal of Endocrinology, 2022, 186, K17-K24.	3.7	7
20	More severe than CVID: Combined immunodeficiency due to a novel NFKB2 mutation. Pediatric Allergy and Immunology, 2021, 32, 793-797.	2.6	5
21	Homozygous deletion of the entire AAAS gene in a triple A syndrome patient. European Journal of Medical Genetics, 2019, 62, 103665.	1.3	3
22	Homozygous mutation in murine retrovirus integration site 1 gene associated with a nonâ€syndromic form of isolated familial achalasia. Neurogastroenterology and Motility, 2020, 32, e13923.	3.0	2
23	CYP21A2 gene expression in a humanized 21-hydroxylase mouse model does not affect adrenocortical morphology and function. Journal of the Endocrine Society, 2022, 6, bvac062.	0.2	2
24	MON-175 Structural Instability as an Underlying Pathomechanism in Congenital Adrenal Hyperplasia. Journal of the Endocrine Society, 2020, 4, .	0.2	0