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	Congenital Adrenal Hyperplasiaâ€"Current Insights in Pathophysiology, Diagnostics, and Management. Endocrine Reviews, 2022, 43, 91-159.	20.1	182
2	Prenatal dexamethasone treatment for classic 21-hydroxylase deficiency in Europe. European Journal of Endocrinology, 2022, 186, K17-K24.	3.7	7
3	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
4	More severe than CVID: Combined immunodeficiency due to a novel NFKB2 mutation. Pediatric Allergy and Immunology, 2021, 32, 793-797.	2.6	5
5	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
6	Biochemical testing for neuroblastoma using plasma free 3â€Oâ€methyldopa, 3â€methoxytyramine, and normetanephrine. Pediatric Blood and Cancer, 2020, 67, e28081.	1.5	14
7	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
8	Cancer Stem Cells in Pheochromocytoma and Paraganglioma. Frontiers in Endocrinology, 2020, 11, 79.	3.5	20
9	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
10	MON-175 Structural Instability as an Underlying Pathomechanism in Congenital Adrenal Hyperplasia. Journal of the Endocrine Society, 2020, 4, .	0.2	0
11	Homozygous deletion of the entire AAAS gene in a triple A syndrome patient. European Journal of Medical Genetics, 2019, 62, 103665.	1.3	3
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	ALADIN is required for the production of fertile mouse oocytes. Molecular Biology of the Cell, 2017, 28, 2470-2478.	2.1	13
14	Phenotypeâ€"genotype spectrum of AAA syndrome from Western India and systematic review of literature. Endocrine Connections, 2017, 6, 901-913.	1.9	28
15	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
16	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
17	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	2.6	69
18	A founder mutation in Anoctamin 5 is a major cause of limb girdle muscular dystrophy. Brain, 2011, 134, 171-182.	7.6	254

#	Article	IF	Citations
19	Nonclassic Lipoid Congenital Adrenal Hyperplasia Masquerading as Familial Glucocorticoid Deficiency. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 3865-3871.	3.6	138
20	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
21	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
22	Chromosomal fragility in patients with triple A syndrome. American Journal of Medical Genetics Part A, 2003, 117A, 30-36.	2.4	20
23	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
24	Tall stature in familial glucocorticoid deficiency. Clinical Endocrinology, 2000, 53, 423-430.	2.4	88