

Deborah P Merke

List of Publications by Year in descending order

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97
papers

8,833
citations

87401

40
h-index

51423

90
g-index

105
all docs

105
docs citations

105
times ranked

4978
citing authors

#	ARTICLE	IF	CITATIONS
1	Congenital Adrenal Hyperplasia—Current Insights in Pathophysiology, Diagnostics, and Management. <i>Endocrine Reviews</i> , 2022, 43, 91-159.	8.9	182
2	Management challenges and therapeutic advances in congenital adrenal hyperplasia. <i>Nature Reviews Endocrinology</i> , 2022, 18, 337-352.	4.3	34
3	Excess 11-Oxygenated Androgens in Women With Severe Insulin Resistance Are Mediated by Adrenal Insulin Receptor Signaling. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 2626-2635.	1.8	7
4	Adrenal disorders. , 2021, , 267-296.		0
5	Cardiovascular Disease Risk Factors and Metabolic Morbidity in a Longitudinal Study of Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e5247-e5257.	1.8	24
6	Letter to the Editor from Lao and Merke: “Ehlers—Danlos Syndrome: Molecular and Clinical Characterization of <i>CYP11A1/CYP11B1</i> Chimeras in Congenital Adrenal Hyperplasia.” <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e2835-e2836.	1.8	2
7	Molecular genetic testing of congenital adrenal hyperplasia due to 21-hydroxylase deficiency should include CAH-X chimeras. <i>European Journal of Human Genetics</i> , 2021, 29, 1047-1048.	1.4	9
8	Design of a Phase 1/2 Open-Label, Dose-Escalation Study of the Safety and Efficacy of Gene Therapy in Adults With Classic Congenital Adrenal Hyperplasia (CAH) Due to 21-hydroxylase Deficiency Through Administration of an Adeno-Associated Virus (AAV) Serotype 5-Based Recombinant Vector Encoding the Human <i>CYP21A2</i> Gene. <i>Journal of the Endocrine Society</i> , 2021, 5, A82-A82.	0.1	3
9	Tildacerfont in Adults With Classic Congenital Adrenal Hyperplasia: Results from Two Phase 2 Studies. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e4666-e4679.	1.8	21
10	Morphologic and Molecular Characterization of Adrenals and Adrenal Rest Affected by Congenital Adrenal Hyperplasia. <i>Frontiers in Endocrinology</i> , 2021, 12, 730947.	1.5	6
11	Modified-Release Hydrocortisone in Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e2063-e2077.	1.8	38
12	A <i>CYP11B1</i> splice donor site variant as a cause of hypermobility type Ehlers—Danlos syndrome in patients with congenital adrenal hyperplasia. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1556.	0.6	12
13	11-Oxygenated Androgens Useful in the Setting of Discrepant Conventional Biomarkers in 21-Hydroxylase Deficiency. <i>Journal of the Endocrine Society</i> , 2021, 5, bvaa192.	0.1	23
14	Younger age and early puberty are associated with cognitive function decline in children with Cushing disease. <i>Clinical Endocrinology</i> , 2021, , .	1.2	3
15	24-Hour Profiles of 11-Oxygenated C19 Steroids and 17 5-Steroid Sulfates during Oral and Continuous Subcutaneous Glucocorticoids in 21-Hydroxylase Deficiency. <i>Frontiers in Endocrinology</i> , 2021, 12, 751191.	1.5	10
16	Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency. <i>New England Journal of Medicine</i> , 2020, 383, 1248-1261.	13.9	155
17	A Phase 2, Multicenter Study of Nevanimibe for the Treatment of Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 2771-2778.	1.8	19
18	Multidimensional Aspects of Female Sexual Function in Congenital Adrenal Hyperplasia: A Case-Control Study. <i>Journal of the Endocrine Society</i> , 2020, 4, bvaa131.	0.1	10

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19	High-Throughput Screening for CYP21A1P-TNXA/TNXB Chimeric Genes Responsible for Ehlers-Danlos Syndrome in Patients with Congenital Adrenal Hyperplasia. <i>Journal of Molecular Diagnostics</i> , 2019, 21, 924-931.	1.2	22
20	Measurement of serum tenascin-X in patients with congenital adrenal hyperplasia at risk for Ehlers-Danlos contiguous gene deletion syndrome CAH-X. <i>BMC Research Notes</i> , 2019, 12, 711.	0.6	6
21	Adrenal morphology and associated comorbidities in congenital adrenal hyperplasia. <i>Clinical Endocrinology</i> , 2019, 91, 247-255.	1.2	13
22	Response to Letter to the Editor: "Congenital Adrenal Hyperplasia Due to Steroid 21-Hydroxylase Deficiency: An Endocrine Society Clinical Practice Guideline" <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 1928-1928.	1.8	1
23	Response to Letter to the Editor: "Characterization of the CYP11A1 Nonsynonymous Variant p.E314K in Children Presenting With Adrenal Insufficiency" <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 1415-1416.	1.8	0
24	Cover Image, Volume 91, Issue 2. <i>Clinical Endocrinology</i> , 2019, 91, i.	1.2	0
25	Characterization of the CYP11A1 Nonsynonymous Variant p.E314K in Children Presenting With Adrenal Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 269-276.	1.8	23
26	Revisiting the association of HLA alleles and haplotypes with CYP21A2 mutations in a large cohort of patients with congenital adrenal hyperplasia. <i>Gene</i> , 2019, 687, 30-34.	1.0	4
27	Complement component 4 variations may influence psychopathology risk in patients with congenital adrenal hyperplasia due to 21-hydroxylase deficiency. <i>Human Genetics</i> , 2018, 137, 955-960.	1.8	8
28	Congenital Adrenal Hyperplasia Due to Steroid 21-Hydroxylase Deficiency: An Endocrine Society* Clinical Practice Guideline. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 4043-4088.	1.8	667
29	Tenascin-X, Congenital Adrenal Hyperplasia, and the CAH-X Syndrome. <i>Hormone Research in Paediatrics</i> , 2018, 89, 352-361.	0.8	46
30	Long-term use of continuous subcutaneous hydrocortisone infusion therapy in patients with congenital adrenal hyperplasia. <i>Clinical Endocrinology</i> , 2018, 89, 399-407.	1.2	24
31	Longitudinal Assessment of Illnesses, Stress Dosing, and Illness Sequelae in Patients With Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 2336-2345.	1.8	51
32	Modified release and conventional glucocorticoids and diurnal androgen excretion in congenital adrenal hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, jc.2016-2855.	1.8	38
33	Genetics of Congenital Adrenal Hyperplasia. <i>Endocrinology and Metabolism Clinics of North America</i> , 2017, 46, 435-458.	1.2	56
34	11-Oxygenated Androgens Are Biomarkers of Adrenal Volume and Testicular Adrenal Rest Tumors in 21-Hydroxylase Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 2701-2710.	1.8	84
35	Revisiting the prevalence of nonclassic congenital adrenal hyperplasia in US Ashkenazi Jews and Caucasians. <i>Genetics in Medicine</i> , 2017, 19, 1276-1279.	1.1	90
36	Congenital adrenal hyperplasia. <i>Lancet</i> , The, 2017, 390, 2194-2210.	6.3	534

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37	Alterations in Hydrocortisone Pharmacokinetics in a Patient With Congenital Adrenal Hyperplasia Following Bariatric Surgery. <i>Journal of the Endocrine Society</i> , 2017, 1, 994-1001.	0.1	9
38	Ehlers-Danlos Syndrome Caused by Biallelic <i>TNXB</i> Variants in Patients with Congenital Adrenal Hyperplasia. <i>Human Mutation</i> , 2016, 37, 893-897.	1.1	36
39	A Phase 2 Study of Continuous Subcutaneous Hydrocortisone Infusion in Adults With Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 4690-4698.	1.8	68
40	Positive fertility outcomes in a female with classic congenital adrenal hyperplasia following bilateral adrenalectomy. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2016, 2016, 10.	1.6	7
41	Diagnosis and Treatment of Primary Adrenal Insufficiency: An Endocrine Society Clinical Practice Guideline. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 364-389.	1.8	1,166
42	Adrenal-derived 11-oxygenated 19-carbon steroids are the dominant androgens in classic 21-hydroxylase deficiency. <i>European Journal of Endocrinology</i> , 2016, 174, 601-609.	1.9	168
43	Cortical bone mineral density in patients with congenital adrenal hyperplasia due to 21-hydroxylase deficiency. <i>Clinical Endocrinology</i> , 2015, 82, 330-337.	1.2	46
44	A Phase 2 Study of Chronocort, a Modified-Release Formulation of Hydrocortisone, in the Treatment of Adults With Classic Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, 1137-1145.	1.8	124
45	Broadening the Spectrum of Ehlers Danlos Syndrome in Patients With Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E1143-E1152.	1.8	51
46	Hormonal circadian rhythms in patients with congenital adrenal hyperplasia: identifying optimal monitoring times and novel disease biomarkers. <i>European Journal of Endocrinology</i> , 2015, 173, 727-737.	1.9	39
47	Transforming growth factor- β (TGF- β) pathway abnormalities in tenascin-X deficiency associated with CAH-X syndrome. <i>European Journal of Medical Genetics</i> , 2014, 57, 95-102.	0.7	16
48	An oral multiparticulate, modified-release, hydrocortisone replacement therapy that provides physiological cortisol exposure. <i>Clinical Endocrinology</i> , 2014, 80, 554-561.	1.2	83
49	Tenascin-X gene defects and cardiovascular disease. <i>Medical Hypotheses</i> , 2014, 83, 844.	0.8	3
50	Comprehensive Mutation Analysis of the CYP21A2 Gene. <i>Journal of Molecular Diagnostics</i> , 2013, 15, 745-753.	1.2	38
51	Management of adolescents with congenital adrenal hyperplasia. <i>Lancet Diabetes and Endocrinology</i> , 2013, 1, 341-352.	5.5	90
52	Incentive processing in Congenital Adrenal Hyperplasia (CAH): A reward-based antisaccade study. <i>Psychoneuroendocrinology</i> , 2013, 38, 716-721.	1.3	9
53	Tenascin-X Haploinsufficiency Associated with Ehlers-Danlos Syndrome in Patients with Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013, 98, E379-E387.	1.8	59
54	Clinical Characteristics of a Cohort of 244 Patients with Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, 4429-4438.	1.8	242

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55	Use of PET/CT with Cosyntropin Stimulation to Identify and Localize Adrenal Rest Tissue following Adrenalectomy in a Woman with Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, E2084-E2089.	1.8	43
56	Junction Site Analysis of Chimeric CYP21A1P/CYP21A2 Genes in 21-Hydroxylase Deficiency. <i>Clinical Chemistry</i> , 2012, 58, 421-430.	1.5	60
57	Complement component 4 copy number variation and CYP21A2 genotype associations in patients with congenital adrenal hyperplasia due to 21-hydroxylase deficiency. <i>Human Genetics</i> , 2012, 131, 1889-1894.	1.8	11
58	Increased medial temporal lobe and striatal grey-matter volume in a rare disorder of androgen excess: a voxel-based morphometry (VBM) study. <i>International Journal of Neuropsychopharmacology</i> , 2011, 14, 445-457.	1.0	25
59	Phenotypic profiling of parents with cryptic nonclassic congenital adrenal hyperplasia: findings in 145 unrelated families. <i>European Journal of Endocrinology</i> , 2011, 164, 977-984.	1.9	69
60	Comprehensive Genetic Analysis of 182 Unrelated Families with Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011, 96, E161-E172.	1.8	154
61	Emotional Memory in Early Steroid Abnormalities: An fMRI Study of Adolescents With Congenital Adrenal Hyperplasia. <i>Developmental Neuropsychology</i> , 2011, 36, 473-492.	1.0	26
62	A pharmacokinetic and pharmacodynamic study of delayed and extended release hydrocortisone (Chronocort TM) vs. conventional hydrocortisone (Cortef TM) in the treatment of congenital adrenal hyperplasia. <i>Clinical Endocrinology</i> , 2010, 72, 441-447.	1.2	120
63	Guidelines for the Development of Comprehensive Care Centers for Congenital Adrenal Hyperplasia: Guidance from the CARES Foundation Initiative. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2010, 2010, 1-17.	1.6	35
64	Psychiatric characterization of children with genetic causes of hyperandrogenism. <i>European Journal of Endocrinology</i> , 2010, 163, 801-810.	1.9	69
65	Adrenomedullary Function in Patients with Nonclassic Congenital Adrenal Hyperplasia. <i>Hormone and Metabolic Research</i> , 2010, 42, 607-612.	0.7	19
66	Congenital Adrenal Hyperplasia Due to Steroid 21-Hydroxylase Deficiency: An Endocrine Society Clinical Practice Guideline. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, 4133-4160.	1.8	1,117
67	Early Hyperandrogenism Affects the Development of Hippocampal Function: Preliminary Evidence from a Functional Magnetic Resonance Imaging Study of Boys with Familial Male Precocious Puberty. <i>Journal of Child and Adolescent Psychopharmacology</i> , 2009, 19, 41-50.	0.7	28
68	Cardiovascular Disease Risk in Adult Women with Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency. <i>Seminars in Reproductive Medicine</i> , 2009, 27, 316-321.	0.5	37
69	Modified-Release Hydrocortisone to Provide Circadian Cortisol Profiles. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 1548-1554.	1.8	265
70	The phenotypic spectrum of contiguous deletion of CYP21A2 and tenascin XB: Quadricuspid aortic valve and other midline defects. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2803-2808.	0.7	26
71	Quality of life in children and adolescents 1 year after cure of Cushing syndrome: a prospective study. <i>Clinical Endocrinology</i> , 2009, 71, 326-333.	1.2	51
72	Steroid abnormalities and the developing brain: Declarative memory for emotionally arousing and neutral material in children with congenital adrenal hyperplasia. <i>Psychoneuroendocrinology</i> , 2008, 33, 238-245.	1.3	24

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73	Altered amygdala and hippocampus function in adolescents with hypercortisolemia: A functional magnetic resonance imaging study of Cushing syndrome. <i>Development and Psychopathology</i> , 2008, 20, 1177-1189.	1.4	62
74	Body Image in Adolescents with Disorders of Steroidogenesis. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2008, 21, 771-80.	0.4	6
75	Approach to the Adult with Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008, 93, 653-660.	1.8	103
76	Amygdala function in adolescents with congenital adrenal hyperplasia: A model for the study of early steroid abnormalities. <i>Neuropsychologia</i> , 2007, 45, 2104-2113.	0.7	70
77	Maternal 21-hydroxylase deficiency and uniparental isodisomy of chromosome 6 and X results in a child with 21-hydroxylase deficiency and Klinefelter syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 2236-2240.	0.7	25
78	The Adrenal Life Cycle: The Fetal and Adult Cortex and the Remaining Questions. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2006, 19, 1299-302.	0.4	8
79	Classic Congenital Adrenal Hyperplasia. , 2005, , 101-113.		1
80	Children Experience Cognitive Decline Despite Reversal of Brain Atrophy One Year After Resolution of Cushing Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005, 90, 2531-2536.	1.8	113
81	Congenital adrenal hyperplasia. <i>Lancet, The</i> , 2005, 365, 2125-2136.	6.3	615
82	Endocrinologic and Psychologic Evaluation of 21-Hydroxylase Deficiency Carriers and Matched Normal Subjects: Evidence for Physical and/or Psychologic Vulnerability to Stress. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2004, 89, 2228-2236.	1.8	35
83	Stress Dose of Hydrocortisone Is Not Beneficial in Patients with Classic Congenital Adrenal Hyperplasia Undergoing Short-Term, High-Intensity Exercise. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2004, 89, 3679-3684.	1.8	64
84	Patients with Classic Congenital Adrenal Hyperplasia Have Decreased Epinephrine Reserve and Defective Glucose Elevation in Response to High-Intensity Exercise. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2004, 89, 591-597.	1.8	64
85	Children with Classic Congenital Adrenal Hyperplasia Have Decreased Amygdala Volume: Potential Prenatal and Postnatal Hormonal Effects. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 1760-1765.	1.8	123
86	Adrenomedullary Function May Predict Phenotype and Genotype in Classic 21-Hydroxylase Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 3031-3037.	1.8	59
87	Children with Classic Congenital Adrenal Hyperplasia Have Elevated Serum Leptin Concentrations and Insulin Resistance: Potential Clinical Implications. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 2114-2120.	1.8	136
88	Flutamide Decreases Cortisol Clearance in Patients with Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 3197-3200.	1.8	12
89	NEW IDEAS FOR MEDICAL TREATMENT OF CONGENITAL ADRENAL HYPERPLASIA. <i>Endocrinology and Metabolism Clinics of North America</i> , 2001, 30, 121-135.	1.2	40
90	Congenital Adrenal Hyperplasia. <i>Paediatric Drugs</i> , 2001, 3, 599-611.	1.3	21

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91	Novel basic and clinical aspects of congenital adrenal hyperplasia. , 2001, 2, 289-296.		5
92	Hydrocortisone Suspension and Hydrocortisone Tablets Are Not Bioequivalent in the Treatment of Children with Congenital Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 441-445.	1.8	55
93	Adrenomedullary Dysplasia and Hypofunction in Patients with Classic 21-Hydroxylase Deficiency. New England Journal of Medicine, 2000, 343, 1362-1368.	13.9	229
94	Flutamide, Testolactone, and Reduced Hydrocortisone Dose Maintain Normal Growth Velocity and Bone Maturation Despite Elevated Androgen Levels in Children with Congenital Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 1114-1120.	1.8	157
95	Adrenal Lymphocytic Infiltration and Adrenocortical Tumors in a Patient with 21-Hydroxylase Deficiency. New England Journal of Medicine, 1999, 340, 1121-1122.	13.9	31
96	New Approaches to the Treatment of Congenital Adrenal Hyperplasia. JAMA - Journal of the American Medical Association, 1997, 277, 1073.	3.8	26
97	Adrenomedullary Function May Predict Phenotype and Genotype in Classic 21-Hydroxylase Deficiency. , 0, .		13