

# John C Achermann

## List of Publications by Year in descending order

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137  
papers

12,581  
citations

25014

57  
h-index

24961

109  
g-index

143  
all docs

143  
docs citations

143  
times ranked

7546  
citing authors

#	ARTICLE	IF	CITATIONS
1	Consensus Statement on Management of Intersex Disorders. <i>Pediatrics</i> , 2006, 118, e488-e500.	1.0	1,378
2	Consensus statement on management of intersex disorders. <i>Archives of Disease in Childhood</i> , 2005, 91, 554-563.	1.0	900
3	A mutation in the gene encoding steroidogenic factor-1 causes XY sex reversal and adrenal failure in humans. <i>Nature Genetics</i> , 1999, 22, 125-126.	9.4	642
4	Consensus statement on management of intersex disorders. <i>Journal of Pediatric Urology</i> , 2006, 2, 148-162.	0.6	516
5	Mutations in <i>NR5A1</i> Associated with Ovarian Insufficiency. <i>New England Journal of Medicine</i> , 2009, 360, 1200-1210.	13.9	339
6	Mutations within <i>Sox2/SOX2</i> are associated with abnormalities in the hypothalamo-pituitary-gonadal axis in mice and humans. <i>Journal of Clinical Investigation</i> , 2006, 116, 2442-55.	3.9	285
7	Two Novel Missense Mutations in G Protein-Coupled Receptor 54 in a Patient with Hypogonadotropic Hypogonadism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005, 90, 1849-1855.	1.8	264
8	Gonadal Determination and Adrenal Development Are Regulated by the Orphan Nuclear Receptor Steroidogenic Factor-1, in a Dose-Dependent Manner. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 1829-1833.	1.8	251
9	Human Male Infertility Associated with Mutations in <i>NR5A1</i> Encoding Steroidogenic Factor 1. <i>American Journal of Human Genetics</i> , 2010, 87, 505-512.	2.6	210
10	Summary of Consensus Statement on Intersex Disorders and Their Management. <i>Pediatrics</i> , 2006, 118, 753-757.	1.0	200
11	Society for Endocrinology <sc>UK</sc> guidance on the initial evaluation of an infant or an adolescent with a suspected disorder of sex development (Revised 2015). <i>Clinical Endocrinology</i> , 2016, 84, 771-788.	1.2	196
12	Heterozygous Missense Mutations in Steroidogenic Factor 1 ( <i>SF1/Ad4BP, NR5A1</i> ) Are Associated with 46,XY Disorders of Sex Development with Normal Adrenal Function. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2007, 92, 991-999.	1.8	189
13	Analysis of <i>DAX1 (NROB1)</i> and Steroidogenic Factor-1 ( <i>NR5A1</i> ) in Children and Adults with Primary Adrenal Failure: Ten Years' Experience. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006, 91, 3048-3054.	1.8	183
14	<i>DAX-1 (NROB1)</i> and steroidogenic factor-1 ( <i>SF-1, NR5A1</i> ) in human disease. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2015, 29, 607-619.	2.2	183
15	A novel mutation in <i>DAX1</i> causes delayed-onset adrenal insufficiency and incomplete hypogonadotropic hypogonadism. <i>Journal of Clinical Investigation</i> , 2000, 105, 321-328.	3.9	171
16	Mutations in the PCNA-binding domain of <i>CDKN1C</i> cause <i>IMAGe</i> syndrome. <i>Nature Genetics</i> , 2012, 44, 788-792.	9.4	169
17	Nonclassic Congenital Lipoid Adrenal Hyperplasia: A New Disorder of the Steroidogenic Acute Regulatory Protein with Very Late Presentation and Normal Male Genitalia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006, 91, 4781-4785.	1.8	164
18	Steroidogenic Factor-1 (<i>SF-1, Ad4BP, NR5A1</i>) and Disorders of Testis Development. <i>Sexual Development</i> , 2008, 2, 200-209.	1.1	163

#	ARTICLE	IF	CITATIONS
19	X-Linked Adrenal Hypoplasia Congenita: A Mutation in DAX1 Expands the Phenotypic Spectrum in Males and Females. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 4501-4509.	1.8	157
20	SOX2 Plays a Critical Role in the Pituitary, Forebrain, and Eye during Human Embryonic Development. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008, 93, 1865-1873.	1.8	154
21	Phenotypic spectrum of mutations in DAX-1 and SF-1. <i>Molecular and Cellular Endocrinology</i> , 2001, 185, 17-25.	1.6	146
22	Mitochondrial disease and endocrine dysfunction. <i>Nature Reviews Endocrinology</i> , 2017, 13, 92-104.	4.3	146
23	Clinical and Functional Effects of Mutations in the DAX-1 Gene in Patients with Adrenal Hypoplasia Congenita. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 504-511.	1.8	143
24	Steroidogenic factor-1 (SF-1, NR5A1) and human disease. <i>Molecular and Cellular Endocrinology</i> , 2011, 336, 198-205.	1.6	143
25	Five novel mutations in steroidogenic factor 1 (SF1, NR5A1) in 46,XY patients with severe underandrogenization but without adrenal insufficiency. <i>Human Mutation</i> , 2008, 29, 59-64.	1.1	141
26	Sphingosine-1-phosphate lyase mutations cause primary adrenal insufficiency and steroid-resistant nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2017, 127, 942-953.	3.9	139
27	Nonclassic Lipoid Congenital Adrenal Hyperplasia Masquerading as Familial Glucocorticoid Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 3865-3871.	1.8	138
28	Mutational analysis of steroidogenic factor 1 (NR5a1) in 24 boys with bilateral anorchia: a French collaborative study. <i>Human Reproduction</i> , 2007, 22, 3255-3261.	0.4	131
29	Somatic mutations and progressive monosomy modify SAMD9-related phenotypes in humans. <i>Journal of Clinical Investigation</i> , 2017, 127, 1700-1713.	3.9	129
30	Rare Causes of Primary Adrenal Insufficiency: Genetic and Clinical Characterization of a Large Nationwide Cohort. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 284-292.	1.8	128
31	Severe Combined Adrenal and Gonadal Deficiency Caused by Novel Mutations in the Cholesterol Side Chain Cleavage Enzyme, P450scc. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008, 93, 696-702.	1.8	125
32	UK guidance on the initial evaluation of an infant or an adolescent with a suspected disorder of sex development. <i>Clinical Endocrinology</i> , 2011, 75, 12-26.	1.2	124
33	X-Linked Adrenal Hypoplasia Congenita: A Mutation in DAX1 Expands the Phenotypic Spectrum in Males and Females. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 4501-4509.	1.8	124
34	The spectrum of phenotypes associated with mutations in steroidogenic factor 1 (SF-1, NR5A1, Ad4BP) includes severe penoscrotal hypospadias in 46,XY males without adrenal insufficiency. <i>European Journal of Endocrinology</i> , 2009, 161, 237-242.	1.9	115
35	Homozygous Disruption of P450 Side-Chain Cleavage (CYP11A1) Is Associated with Prematurity, Complete 46,XY Sex Reversal, and Severe Adrenal Failure. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005, 90, 538-541.	1.8	112
36	Clinical and Functional Effects of Mutations in the DAX-1 Gene in Patients with Adrenal Hypoplasia Congenita. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 504-511.	1.8	110

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37	Variable Phenotypes Associated with Aromatase (CYP19) Insufficiency in Humans. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2007, 92, 982-990.	1.8	108
38	An Alternate Translation Initiation Site Circumvents an Amino-Terminal DAX1 Nonsense Mutation Leading to a Mild Form of X-Linked Adrenal Hypoplasia Congenita. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 417-423.	1.8	103
39	Holistic management of DSD. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2010, 24, 335-354.	2.2	101
40	A novel loss of function mutation in exon 10 of the FSH receptor gene causing hypergonadotrophic hypogonadism: clinical and molecular characteristics. <i>Human Reproduction</i> , 2003, 18, 251-256.	0.4	100
41	Hypogonadotropic Hypogonadism as a Presenting Feature of Late-Onset X-Linked Adrenal Hypoplasia Congenita. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 44-48.	1.8	94
42	A recurrent p.Arg92Trp variant in steroidogenic factor-1 (NR5A1) can act as a molecular switch in human sex development. <i>Human Molecular Genetics</i> , 2016, 25, 3446-3453.	1.4	90
43	Constitutional <i>SAMD9L</i> mutations cause familial myelodysplastic syndrome and transient monosomy 7. <i>Haematologica</i> , 2018, 103, 427-437.	1.7	83
44	Disorders of sex development: effect of molecular diagnostics. <i>Nature Reviews Endocrinology</i> , 2015, 11, 478-488.	4.3	81
45	Severe loss-of-function mutations in the adrenocorticotropin receptor (ACTHR, MC2R) can be found in patients diagnosed with salt-losing adrenal hypoplasia. <i>Clinical Endocrinology</i> , 2007, 66, 205-210.	1.2	79
46	Tumor to normal single-cell mRNA comparisons reveal a pan-neuroblastoma cancer cell. <i>Science Advances</i> , 2021, 7, .	4.7	78
47	Mutational Analysis of DAX1 in Patients with Hypogonadotropic Hypogonadism or Pubertal Delay1. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 4497-4500.	1.8	77
48	Inherited disorders of the gonadotropin hormones. <i>Molecular and Cellular Endocrinology</i> , 2001, 179, 89-96.	1.6	77
49	Wide spectrum of NR5A1-related phenotypes in 46,XY and 46,XX individuals. <i>Birth Defects Research Part C: Embryo Today Reviews</i> , 2016, 108, 309-320.	3.6	76
50	A Naturally Occurring Steroidogenic Factor-1 Mutation Exhibits Differential Binding and Activation of Target Genes. <i>Journal of Biological Chemistry</i> , 2000, 275, 31708-31714.	1.6	75
51	The role of SF1 in adrenal and reproductive function: insight from naturally occurring mutations in humans. <i>Molecular Genetics and Metabolism</i> , 2002, 76, 85-91.	0.5	73
52	Clinical, Genetic, and Functional Characterization of Four Patients Carrying Partial Loss-of-Function Mutations in the Steroidogenic Acute Regulatory Protein (StAR). <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, 3352-3359.	1.8	73
53	Genetic Causes of Human Reproductive Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 2447-2454.	1.8	70
54	Human RSPO1/R-spondin1 Is Expressed during Early Ovary Development and Augments $\beta$ -Catenin Signaling. <i>PLoS ONE</i> , 2011, 6, e16366.	1.1	70

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55	Missense Mutations Cluster within the Carboxyl-Terminal Region of DAX-1 and Impair Transcriptional Repression. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2001, 86, 3171-3175.	1.8	69
56	Steroidogenic Factor-1 and Human Disease. <i>Seminars in Reproductive Medicine</i> , 2012, 30, 374-381.	0.5	64
57	Loss of Function of the Nuclear Receptor NR2F2, Encoding COUP-TF2, Causes Testis Development and Cardiac Defects in 46,XX Children. <i>American Journal of Human Genetics</i> , 2018, 102, 487-493.	2.6	64
58	Mutations involving the SRY-related gene SOX8 are associated with a spectrum of human reproductive anomalies. <i>Human Molecular Genetics</i> , 2018, 27, 1228-1240.	1.4	64
59	A Homozygous R262Q Mutation in the Gonadotropin-Releasing Hormone Receptor Presenting as Constitutional Delay of Growth and Puberty with Subsequent Borderline Oligospermia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006, 91, 5117-5121.	1.8	62
60	Gonadal Determination and Adrenal Development Are Regulated by the Orphan Nuclear Receptor Steroidogenic Factor-1, in a Dose-Dependent Manner. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 1829-1833.	1.8	61
61	Presymptomatic diagnosis of X-linked adrenal hypoplasia congenita by analysis of DAX1. <i>Journal of Pediatrics</i> , 2000, 137, 878-881.	0.9	57
62	Clinical and gonadal features and early surgical management of 45,X/46,XY and 45,X/47,XYY chromosomal mosaicism presenting with genital anomalies. <i>Journal of Pediatric Urology</i> , 2013, 9, 139-144.	0.6	56
63	SF1 in the Development of the Adrenal Gland and Gonads. <i>Hormone Research in Paediatrics</i> , 2003, 59, 94-98.	0.8	55
64	A genomic atlas of human adrenal and gonad development. <i>Wellcome Open Research</i> , 2017, 2, 25.	0.9	55
65	Primary adrenal insufficiency: New genetic causes and their long-term consequences. <i>Clinical Endocrinology</i> , 2020, 92, 11-20.	1.2	54
66	New Technologies for the Identification of Novel Genetic Markers of Disorders of Sex Development (DSD). <i>Sexual Development</i> , 2010, 4, 213-224.	1.1	53
67	Fertility and Infertility: Genetic Contributions from the Hypothalamic-Pituitary- Gonadal Axis. <i>Molecular Endocrinology</i> , 1999, 13, 812-818.	3.7	51
68	A Novel Mutation L260P of the Steroidogenic Acute Regulatory Protein Gene in Three Unrelated Patients of Swiss Ancestry with Congenital Lipoid Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005, 90, 5304-5308.	1.8	47
69	Birth after TESE-ICSI in a man with hypogonadotropic hypogonadism and congenital adrenal hypoplasia linked to a DAX-1 (NR0B1) mutation. <i>Human Reproduction</i> , 2011, 26, 724-728.	0.4	47
70	Next-Generation Sequencing Reveals Novel Genetic Variants (SRY, DMRT1, NR5A1, DHH, DHX37) in Adults With 46,XY DSD. <i>Journal of the Endocrine Society</i> , 2019, 3, 2341-2360.	0.1	46
71	Sterol O-Acyltransferase 1 (SOAT1, ACAT) Is a Novel Target of Steroidogenic Factor-1 (SF-1, NR5A1,) Tj ETQq1 1 0.784314 rgBT / Over	1.8	45
72	Disorders of Adrenal Development. , 2008, 13, 19-32.		41

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73	Variable Presentation of X-linked Adrenal Hypoplasia Congenita. Journal of Pediatric Endocrinology and Metabolism, 2001, 14, 1093-6.	0.4	40
74	Skewed X inactivation is associated with phenotype in a female with adrenal hypoplasia congenita. Journal of Medical Genetics, 2008, 45, e1-e1.	1.5	40
75	Role of DAX-1 &lt;i>&lt;/i>(NR0B1)&lt;i>&lt;/i> and Steroidogenic Factor-1 &lt;i>&lt;/i>(NR5A1)&lt;i>&lt;/i> in Human Adrenal Function. Endocrine Development, 2011, 20, 38-46.	1.3	40
76	Testosterone production during puberty in two 46,XY patients with disorders of sex development and novel NR5A1 (SF-1) mutations. European Journal of Endocrinology, 2012, 167, 125-130.	1.9	40
77	A Novel Point Mutation in P450c17 (CYP17) Causing Combined 17 $\beta$ -Hydroxylase/17,20-Lyase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 2428-2431.	1.8	38
78	The Role of DAX-1 in Reproduction. Trends in Endocrinology and Metabolism, 1998, 9, 169-175.	3.1	36
79	CBP/p300-Interacting Transactivator, with Glu/Asp-Rich C-Terminal Domain, 2, and Pre-B-Cell Leukemia Transcription Factor 1 in Human Adrenal Development and Disease. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 678-683.	1.8	35
80	Dehydroepiandrosterone replacement for patients with adrenal insufficiency. Lancet, The, 2001, 357, 1381-1382.	6.3	34
81	Pathogenic variants in the DEAH-box RNA helicase DHX37 are a frequent cause of 46,XY gonadal dysgenesis and 46,XY testicular regression syndrome. Genetics in Medicine, 2020, 22, 150-159.	1.1	34
82	Genetic Analysis of Pediatric Primary Adrenal Insufficiency of Unknown Etiology: 25 Yearsâ€™ Experience in the UK. Journal of the Endocrine Society, 2021, 5, bvab086.	0.1	34
83	We used to call them hermaphrodites. Genetics in Medicine, 2007, 9, 65-66.	1.1	33
84	A genomic atlas of human adrenal and gonad development. Wellcome Open Research, 0, 2, 25.	0.9	33
85	Late-onset X-linked adrenal hypoplasia (DAX-1, NR0B1): two new adult-onset cases from a single center. Pituitary, 2017, 20, 585-593.	1.6	30
86	Progressive Onset of Adrenal Insufficiency and Hypogonadism of Pituitary Origin Caused by a Complex Genetic Rearrangement within DAX-1. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4094-4100.	1.8	29
87	Stress Response and Child Health <b>Meeting Information</b> : The European Society for Paediatric Endocrinology (ESPE) New Inroads to Child Health (NICHe) Conference on Stress Response and Child Health took place in Heraklion, Crete, Greece, 18 to 20 May 2012.. Science Signaling, 2012, 5, mr1.	1.6	29
88	Society for Endocrinology UK Guidance on the initial evaluation of a suspected difference or disorder of sex development (Revised 2021). Clinical Endocrinology, 2021, 95, 818-840.	1.2	29
89	Genetic disorders of nuclear receptors. Journal of Clinical Investigation, 2017, 127, 1181-1192.	3.9	28
90	The relationship between the growth hormone and insulin-like growth factor axis in long-term survivors of childhood brain tumours. Clinical Endocrinology, 1998, 49, 639-645.	1.2	27

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91	ChIP&#x2013;chip analysis reveals angiotensin 2 (Ang2, ANGPT2) as a novel target of steroidogenic factor&#x2013;1 (SF&#x2013;1, NR5A1) in the human adrenal gland. FASEB Journal, 2011, 25, 1166-1175.	0.2	27
92	Predicted Benign and Synonymous Variants in CYP11A1 Cause Primary Adrenal Insufficiency Through Missplicing. Journal of the Endocrine Society, 2019, 3, 201-221.	0.1	27
93	Single cell derived mRNA signals across human kidney tumors. Nature Communications, 2021, 12, 3896.	5.8	27
94	Molecular and Structural Analysis of Two Novel STAR Mutations in Patients with Lipoid Congenital Adrenal Hyperplasia. Molecular Genetics and Metabolism, 2001, 73, 354-357.	0.5	26
95	Inherited adrenal hypoplasia: not just for kids!. Clinical Endocrinology, 2004, 60, 529-537.	1.2	26
96	A novel NR 5 A 1 variant in an infant with elevated testosterone from an Australasian cohort of 46, XY patients with disorders of sex development. Clinical Endocrinology, 2013, 78, 545-550.	1.2	24
97	Current Insights Into Adrenal Insufficiency in the Newborn and Young Infant. Frontiers in Pediatrics, 2020, 8, 619041.	0.9	23
98	Disorders of Sex Development. , 2011, , 868-934.		23
99	Clearing up at 2.5 years. British Journal of Developmental Psychology, 1991, 9, 365-376.	0.9	21
100	Isolated 17,20-lyase (desmolase) deficiency in a 46,XX female presenting with delayed puberty. Fertility and Sterility, 2005, 83, 1548.e23-1548.e26.	0.5	21
101	Kisspeptin Is a Novel Regulator of Human Fetal Adrenocortical Development and Function: A Finding With Important Implications for the Human Fetoplacental Unit. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 3349-3359.	1.8	21
102	Puberty, stress, and sudden death. Lancet, The, 2010, 376, 1512.	6.3	19
103	The Adrenal Cortex and Its Disorders. , 2008, , 444-511.		19
104	A Novel Missense Mutation in Dax-1 with an Unusual Presentation of X-Linked Adrenal Hypoplasia Congenita. Hormone Research in Paediatrics, 2007, 68, 32-37.	0.8	18
105	The growth hormone receptor gene deleted for exon three (<i>GHRd3</i>) polymorphism is associated with birth and placental weight. Clinical Endocrinology, 2012, 76, 236-240.	1.2	17
106	A Patient with Proopiomelanocortin Deficiency: An Increasingly Important Diagnosis to Make. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018, 10, 68-73.	0.4	17
107	Battle of the sexes: new insights into genetic pathways of gonadal development. Transactions of the American Clinical and Climatological Association, 2003, 114, 51-63; discussion 64-5.	0.9	17
108	Birth Weight Influences the Initial Response to Growth Hormone Treatment in Growth Hormone-insufficient Children. Pediatrics, 1998, 102, 342-345.	1.0	16

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109	Fertility and Infertility: Genetic Contributions from the Hypothalamic-Pituitary- Gonadal Axis. <i>Molecular Endocrinology</i> , 1999, 13, 812-818.	3.7	16
110	Long-term outcome of partial P450 side-chain cleavage enzyme deficiency in three brothers: the importance of early diagnosis. <i>European Journal of Endocrinology</i> , 2020, 182, K15-K24.	1.9	16
111	Steroidogenic Factor-1 (SF-1, NR5A1) and 46,XX Ovotesticular Disorders of Sex Development: One Factor, Many Phenotypes. <i>Hormone Research in Paediatrics</i> , 2017, 87, 189-190.	0.8	15
112	Analysis of CDKN1C in fetal growth restriction and pregnancy loss. <i>F1000Research</i> , 2019, 8, 90.	0.8	15
113	X-Linked Adrenal Hypoplasia Congenita and DAX-1. , 2000, 10, 289-299.		13
114	Analysis of LIN28A in early human ovary development and as a candidate gene for primary ovarian insufficiency. <i>Molecular and Cellular Endocrinology</i> , 2012, 351, 264-268.	1.6	13
115	Human sex development: targeted technologies to improve diagnosis. <i>Genome Biology</i> , 2016, 17, 257.	3.8	13
116	<i>ZSWIM7</i> Is Associated With Human Female Meiosis and Familial Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, e254-e263.	1.8	13
117	IMAGe association and congenital adrenal hypoplasia: No disease-causing mutations found in the ACD gene. <i>Molecular Genetics and Metabolism</i> , 2006, 88, 66-70.	0.5	12
118	Pediatric Disorders of Sex Development. , 2016, , 893-963.		12
119	The Adrenal. <i>Hormone Research in Paediatrics</i> , 2004, 62, 22-29.	0.8	11
120	Regarding the Consensus Statement on 21-Hydroxylase Deficiency from the Lawson Wilkins Pediatric Endocrine Society and The European Society for Paediatric Endocrinology. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 3455-3455.	1.8	9
121	Analysis of CDKN1C in fetal growth restriction and pregnancy loss. <i>F1000Research</i> , 2019, 8, 90.	0.8	9
122	Steroidogenic factor-1 (SF-1) and its relevance to pediatric endocrinology. <i>Pediatric Endocrinology Reviews</i> , 2006, 3, 359-64.	1.2	9
123	Characterisation and Validation of Insertions and Deletions in 173 Patient Exomes. <i>PLoS ONE</i> , 2012, 7, e51292.	1.1	8
124	Pathogenic variants in the human m6A reader YTHDC2 are associated with primary ovarian insufficiency. <i>JCI Insight</i> , 2022, 7, .	2.3	8
125	Insights From Long-term Follow-up of a Girl With Adrenal Insufficiency and Sphingosine-1-Phosphate Lyase Deficiency. <i>Journal of the Endocrine Society</i> , 2022, 6, bvac020.	0.1	6
126	Genetic Disorders Involving Adrenal Development. , 2007, 11, 36-46.		5



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127	Pathogenic variants in RNPC3 are associated with hypopituitarism and primary ovarian insufficiency. <i>Genetics in Medicine</i> , 2022, 24, 384-397.	1.1	4
128	Can Digenic, Tri-Allelic Inheritance of Variants in STAR and CYP11A1 Give Rise to Primary Adrenal Insufficiency? A Case Report. <i>Frontiers in Endocrinology</i> , 2022, 13, 860055.	1.5	4
129	A retrospective analysis of endocrine disease in sphingosine-1-phosphate lyase insufficiency: case series and literature review. <i>Endocrine Connections</i> , 2022, 11, .	0.8	4
130	Mutations in NR5A1 Associated With Ovarian Insufficiency. <i>Obstetrical and Gynecological Survey</i> , 2009, 64, 665-666.	0.2	3
131	Co-Existence of Congenital Adrenal Hyperplasia and Bartter Syndrome due to Maternal Uniparental Isodisomy of <i>HSD3B2</i> and <i>CLCNKB</i> Mutations. <i>Hormone Research in Paediatrics</i> , 2020, 93, 137-142.	0.8	2
132	Missplicing due to a synonymous, T96= exonic substitution in the T-box transcription factor TBX19 resulting in isolated ACTH deficiency. <i>Endocrinology, Diabetes and Metabolism Case Reports</i> , 2021, .	0.2	2
133	Management of a Girl With Delayed Puberty and Elevated Gonadotropins. <i>Journal of the Endocrine Society</i> , 2022, 6, .	0.1	2
134	Disorders of sex development. , 0, , 15-34.		1
135	Adrenal Development. , 2014, , 5-27.		1
136	Reproductive Endocrinology. , 2005, , 89-106.		0
137	Adrenals. <i>Yearbook of Paediatric Endocrinology</i> , 2009, , 93-110.	0.0	0