# John C Achermann

List of Publications by Year
in descending order



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|---|---|---|---|
| 135 papers | 12,581 citations | 28736 57 h-index | 29333 108 g-index |
| 143 all docs | 143 docs citations | 143 times ranked | 8084 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | ZSWIM7Is Associated With Human Female Meiosis and Familial Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e254-e263. | 1.8 | 13 |
| 2 | Pathogenic variants in RNPC3 are associated with hypopituitarism and primary ovarian insufficiency. Genetics in Medicine, 2022, 24, 384-397. | 1.1 | 4 |
| 3 | Insights From Long-term Follow-up of a Girl With Adrenal Insufficiency and Sphingosine-1-Phosphate Lyase Deficiency. Journal of the Endocrine Society, 2022, 6, bvac020. | 0.1 | 6 |
| 4 | Pathogenic variants in the human m6A reader YTHDC2 are associated with primary ovarian insufficiency. JCI Insight, 2022, 7, . | 2.3 | 8 |
| 5 | Can Digenic, Tri-Allelic Inheritance of Variants in STAR and CYP11A1 Give Rise to Primary Adrenal Insufficiency? A Case Report. Frontiers in Endocrinology, 2022, 13, 860055. | 1.5 | 4 |
| 6 | A retrospective analysis of endocrine disease in sphingosine-1-phosphate lyase insufficiency: case series and literature review. Endocrine Connections, 2022, 11, . | 0.8 | 4 |
| 7 | Management of a Girl With Delayed Puberty and Elevated Gonadotropins. Journal of the Endocrine Society, 2022, 6, . | 0.1 | 2 |
| 8 | Tumor to normal single-cell mRNA comparisons reveal a pan-neuroblastoma cancer cell. Science Advances, 2021, 7, . | 4.7 | 78 |
| 9 | 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 |
| 10 | Single cell derived mRNA signals across human kidney tumors. Nature Communications, 2021, 12, 3896. | 5.8 | 27 |
| 11 | 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 |
| 12 | Missplicing due to a synonymous, T96= exonic substitution in the T-box transcription factor TBX19 resulting in isolated ACTH deficiency. Endocrinology, Diabetes and Metabolism Case Reports, 2021, 2021, . | 0.2 | 2 |
| 13 | 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 |
| 14 | Primary adrenal insufficiency: New genetic causes and their longâ€term consequences. Clinical Endocrinology, 2020, 92, 11-20. | 1.2 | 54 |
| 15 | Current Insights Into Adrenal Insufficiency in the Newborn and Young Infant. Frontiers in Pediatrics, 2020, 8, 619041. | 0.9 | 23 |
| 16 | Co-Existence of Congenital Adrenal Hyperplasia and Bartter Syndrome due to Maternal Uniparental Isodisomy of HSD3B2 and CLCNKB Mutations. Hormone Research in Paediatrics, 2020, 93, 137-142. | 0.8 | 2 |
| 17 | Long-term outcome of partial P450 side-chain cleavage enzyme deficiency in three brothers: the importance of early diagnosis. European Journal of Endocrinology, 2020, 182, K15-K24. | 1.9 | 16 |
| 18 | Next-Generation Sequencing Reveals Novel Genetic Variants (SRY, DMRT1, NR5A1, DHH, DHX37) in Adults With 46,XY DSD. Journal of the Endocrine Society, 2019, 3, 2341-2360. | 0.1 | 46 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 20 | Analysis of CDKN1C in fetal growth restriction and pregnancy loss. F1000Research, 2019, 8, 90. | 0.8 | 15 |
| 21 | Analysis of CDKN1C in fetal growth restriction and pregnancy loss. F1000Research, 2019, 8, 90. | 0.8 | 9 |
| 22 | Loss of Function of the Nuclear Receptor NR2F2, Encoding COUP-TF2, Causes Testis Development and Cardiac Defects in 46,XX Children. American Journal of Human Genetics, 2018, 102, 487-493. | 2.6 | 64 |
| 23 | Mutations involving the SRY-related gene SOX8 are associated with a spectrum of human reproductive anomalies. Human Molecular Genetics, 2018, 27, 1228-1240. | 1.4 | 64 |
| 24 | Constitutional SAMD9L mutations cause familial myelodysplastic syndrome and transient monosomy 7. Haematologica, 2018, 103, 427-437. | 1.7 | 83 |
| 25 | 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 |
| 26 | Steroidogenic Factor-1 (SF-1, NR5A1) andâ€˜46,XX Ovotesticular Disorders of Sex Development:â€˜One Factor, Many Phenotypes. Hormone Research in Paediatrics, 2017, 87, 189-190. | 0.8 | 15 |
| 27 | 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 |
| 28 | 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 |
| 29 | Mitochondrial disease and endocrine dysfunction. Nature Reviews Endocrinology, 2017, 13, 92-104. | 4.3 | 146 |
| 30 | Somatic mutations and progressive monosomy modify SAMD9-related phenotypes in humans. Journal of Clinical Investigation, 2017, 127, 1700-1713. | 3.9 | 129 |
| 31 | Sphingosine-1-phosphate lyase mutations cause primary adrenal insufficiency and steroid-resistant nephrotic syndrome. Journal of Clinical Investigation, 2017, 127, 942-953. | 3.9 | 139 |
| 32 | Genetic disorders of nuclear receptors. Journal of Clinical Investigation, 2017, 127, 1181-1192. | 3.9 | 28 |
| 33 | A genomic atlas of human adrenal and gonad development. Wellcome Open Research, 2017, 2, 25. | 0.9 | 55 |
| 34 | Pediatric Disorders of Sex Development. , 2016, , 893-963. | | 12 |
| 35 | A recurrent p.Arg92Trp variant in steroidogenic factor-1 (NR5A1) can act as a molecular switch in human sex development. Human Molecular Genetics, 2016, 25, 3446-3453. | 1.4 | 90 |
| 36 | Human sex development: targeted technologies to improve diagnosis. Genome Biology, 2016, 17, 257. | 3.8 | 13 |

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|---|---|---|---|
| 37 | Society for Endocrinology <scp>UK</scp> guidance on the initial evaluation of an infant or an adolescent with a suspected disorder of sex development (Revised 2015). Clinical Endocrinology, 2016, 84, 771-788. | 1.2 | 196 |
| 38 | Rare Causes of Primary Adrenal Insufficiency: Genetic and Clinical Characterization of a Large Nationwide Cohort. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 284-292. | 1.8 | 128 |
| 39 | Wide spectrum of NR5A1â€related phenotypes in 46,XY and 46,XX individuals. Birth Defects Research Part C: Embryo Today Reviews, 2016, 108, 309-320. | 3.6 | 76 |
| 40 | DAX-1 (NR0B1) and steroidogenic factor-1 (SF-1, NR5A1) in human disease. Best Practice and Research in Clinical Endocrinology and Metabolism, 2015, 29, 607-619. | 2.2 | 183 |
| 41 | Disorders of sex development: effect of molecular diagnostics. Nature Reviews Endocrinology, 2015, 11, 478-488. | 4.3 | 81 |
| 42 | Adrenal Development. , 2014, , 5-27. | | 1 |
| 43 | Clinical and gonadal features and early surgical management of 45,X/46,XY and 45,X/47,XYY chromosomal mosaicism presenting with genital anomalies. Journal of Pediatric Urology, 2013, 9, 139-144. | 0.6 | 56 |
| 44 | A novel NR 5 A 1 variant in an infant with elevated testosterone from an A ustralasian cohort of 46, XY patients with disorders of sex development. Clinical Endocrinology, 2013, 78, 545-550. | 1.2 | 24 |
| 45 | 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 |
| 46 | Steroidogenic Factor-1 and Human Disease. Seminars in Reproductive Medicine, 2012, 30, 374-381. | 0.5 | 64 |
| 47 | Mutations in the PCNA-binding domain of CDKN1C cause IMAGe syndrome. Nature Genetics, 2012, 44, 788-792. | 9.4 | 169 |
| 48 | Stress Response and Child Health Meeting Information : 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 |
| 49 | Characterisation and Validation of Insertions and Deletions in 173 Patient Exomes. PLoS ONE, 2012, 7, e51292. | 1.1 | 8 |
| 50 | The growth hormone receptor gene deleted for exon three (GHRd3) polymorphism is associated with birth and placental weight. Clinical Endocrinology, 2012, 76, 236-240. | 1.2 | 17 |
| 51 | Analysis of LIN28A in early human ovary development and as a candidate gene for primary ovarian insufficiency. Molecular and Cellular Endocrinology, 2012, 351, 264-268. | 1.6 | 13 |
| 52 | Role of DAX-1 <i>(NR0B1)</i> and Steroidogenic Factor-1 <i>(NR5A1)</i> in Human Adrenal Function. Endocrine Development, 2011, 20, 38-46. | 1.3 | 40 |
| 53 | Steroidogenic factor-1 (SF-1, NR5A1) and human disease. Molecular and Cellular Endocrinology, 2011, 336, 198-205. | 1.6 | 143 |
| 54 | Human RSPO1/R-spondin1 Is Expressed during Early Ovary Development and Augments Î²-Catenin Signaling. PLoS ONE, 2011, 6, e16366. | 1.1 | 70 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | UK guidance on the initial evaluation of an infant or an adolescent with a suspected disorder of sex development. Clinical Endocrinology, 2011, 75, 12-26. | 1.2 | 124 |
| 56 | Birth after TESE-ICSI in a man with hypogonadotropic hypogonadism and congenital adrenal hypoplasia linked to a DAX-1 (NR0B1) mutation. Human Reproduction, 2011, 26, 724-728. | 0.4 | 47 |
| 57 | ChIPâ€onâ€chip analysis reveals angiopoietin 2 (Ang2, ANGPT2) as a novel target of steroidogenic factorâ€1 (SFâ€1, NR5A1) in the human adrenal gland. FASEB Journal, 2011, 25, 1166-1175. | 0.2 | 27 |
| 58 | Sterol O-Acyltransferase 1 (SOAT1, ACAT) Is a Novel Target of Steroidogenic Factor-1 (SF-1, NR5A1,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5 | 1.8 | 45 |
| 59 | Disorders of Sex Development. , 2011, , 868-934. | | 23 |
| 60 | Human Male Infertility Associated with Mutations in NR5A1 Encoding Steroidogenic Factor 1. American Journal of Human Genetics, 2010, 87, 505-512. | 2.6 | 210 |
| 61 | New Technologies for the Identification of Novel Genetic Markers of Disorders of Sex Development (DSD). Sexual Development, 2010, 4, 213-224. | 1.1 | 53 |
| 62 | Clinical, Genetic, and Functional Characterization of Four Patients Carrying Partial Loss-of-Function Mutations in the Steroidogenic Acute Regulatory Protein (StAR). Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3352-3359. | 1.8 | 73 |
| 63 | Holistic management of DSD. Best Practice and Research in Clinical Endocrinology and Metabolism, 2010, 24, 335-354. | 2.2 | 101 |
| 64 | Puberty, stress, and sudden death. Lancet, The, 2010, 376, 1512. | 6.3 | 19 |
| 65 | Adrenals. Yearbook of Paediatric Endocrinology, 2009, , 93-110. | 0.0 | 0 |
| 66 | 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. European Journal of Endocrinology, 2009, 161, 237-242. | 1.9 | 115 |
| 67 | 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 |
| 68 | Nonclassic Lipoid Congenital Adrenal Hyperplasia Masquerading as Familial Glucocorticoid Deficiency. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 3865-3871. | 1.8 | 138 |
| 69 | Mutations inNR5A1Associated with Ovarian Insufficiency. New England Journal of Medicine, 2009, 360, 1200-1210. | 13.9 | 339 |
| 70 | Mutations in NR5A1 Associated With Ovarian Insufficiency. Obstetrical and Gynecological Survey, 2009, 64, 665-666. | 0.2 | 3 |
| 71 | Five novel mutations in steroidogenic factor 1 (SF1,NR5A1) in 46,XY patients with severe underandrogenization but without adrenal insufficiency. Human Mutation, 2008, 29, 59-64. | 1.1 | 141 |
| 72 | SOX2 Plays a Critical Role in the Pituitary, Forebrain, and Eye during Human Embryonic Development. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 1865-1873. | 1.8 | 154 |

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|---|---|---|---|
| 73 | Disorders of Adrenal Development. , 2008, 13, 19-32. | | 41 |
| 74 | Steroidogenic Factor-1 <i>(SF-1, Ad4BP, NR5A1)</i> and Disorders of Testis Development. Sexual Development, 2008, 2, 200-209. | 1.1 | 163 |
| 75 | Severe Combined Adrenal and Gonadal Deficiency Caused by Novel Mutations in the Cholesterol Side Chain Cleavage Enzyme, P450scc. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 696-702. | 1.8 | 125 |
| 76 | 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 |
| 77 | The Adrenal Cortex and Its Disorders. , 2008, , 444-511. | | 19 |
| 78 | Mutational analysis of steroidogenic factor 1 (NR5a1) in 24 boys with bilateral anorchia: a French collaborative study. Human Reproduction, 2007, 22, 3255-3261. | 0.4 | 131 |
| 79 | Variable Phenotypes Associated with Aromatase (CYP19) Insufficiency in Humans. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 982-990. | 1.8 | 108 |
| 80 | Heterozygous Missense Mutations in Steroidogenic Factor 1 (SF1/Ad4BP, NR5A1) Are Associated with 46,XY Disorders of Sex Development with Normal Adrenal Function. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 991-999. | 1.8 | 189 |
| 81 | 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 |
| 82 | Genetic Disorders Involving Adrenal Development. , 2007, 11, 36-46. | | 5 |
| 83 | We used to call them hermaphrodites. Genetics in Medicine, 2007, 9, 65-66. | 1.1 | 33 |
| 84 | Severe loss-of-function mutations in the adrenocorticotropin receptor (ACTHR, MC2R) can be found in patients diagnosed with salt-losing adrenal hypoplasia. Clinical Endocrinology, 2007, 66, 205-210. | 1.2 | 79 |
| 85 | Consensus statement on management of intersex disorders. Journal of Pediatric Urology, 2006, 2, 148-162. | 0.6 | 516 |
| 86 | Analysis of DAX1 (NR0B1) and Steroidogenic Factor-1 (NR5A1) in Children and Adults with Primary Adrenal Failure: Ten Yearsâ€™ Experience. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 3048-3054. | 1.8 | 183 |
| 87 | Consensus Statement on Management of Intersex Disorders. Pediatrics, 2006, 118, e488-e500. | 1.0 | 1,378 |
| 88 | IMAGe association and congenital adrenal hypoplasia: No disease-causing mutations found in the ACD gene. Molecular Genetics and Metabolism, 2006, 88, 66-70. | 0.5 | 12 |
| 89 | Nonclassic Congenital Lipoid Adrenal Hyperplasia: A New Disorder of the Steroidogenic Acute Regulatory Protein with Very Late Presentation and Normal Male Genitalia. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 4781-4785. | 1.8 | 164 |
| 90 | Summary of Consensus Statement on Intersex Disorders and Their Management. Pediatrics, 2006, 118, 753-757. | 1.0 | 200 |

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|---|---|---|---|
| 91 | Mutations within Sox2/SOX2 are associated with abnormalities in the hypothalamo-pituitary-gonadal axis in mice and humans. Journal of Clinical Investigation, 2006, 116, 2442-55. | 3.9 | 285 |
| 92 | A Novel Point Mutation in P450c17 (CYP17) Causing Combined 17Î±-Hydroxylase/17,20-Lyase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 2428-2431. | 1.8 | 38 |
| 93 | A Homozygous R262Q Mutation in the Gonadotropin-Releasing Hormone Receptor Presenting as Constitutional Delay of Growth and Puberty with Subsequent Borderline Oligospermia. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 5117-5121. | 1.8 | 62 |
| 94 | Steroidogenic factor-1 (SF-1) and its relevance to pediatric endocrinology. Pediatric Endocrinology Reviews, 2006, 3, 359-64. | 1.2 | 9 |
| 95 | Reproductive Endocrinology. , 2005, , 89-106. | | 0 |
| 96 | Two Novel Missense Mutations in G Protein-Coupled Receptor 54 in a Patient with Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 1849-1855. | 1.8 | 264 |
| 97 | Homozygous Disruption of P450 Side-Chain Cleavage (CYP11A1) Is Associated with Prematurity, Complete 46,XY Sex Reversal, and Severe Adrenal Failure. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 538-541. | 1.8 | 112 |
| 98 | A Novel Mutation L260P of the Steroidogenic Acute Regulatory Protein Gene in Three Unrelated Patients of Swiss Ancestry with Congenital Lipoid Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 5304-5308. | 1.8 | 47 |
| 99 | 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 |
| 100 | Consensus statement on management of intersex disorders. Archives of Disease in Childhood, 2005, 91, 554-563. | 1.0 | 900 |
| 101 | The Adrenal. Hormone Research in Paediatrics, 2004, 62, 22-29. | 0.8 | 11 |
| 102 | Inherited adrenal hypoplasia: not just for kids!. Clinical Endocrinology, 2004, 60, 529-537. | 1.2 | 26 |
| 103 | SF1 in the Development of the Adrenal Gland and Gonads. Hormone Research in Paediatrics, 2003, 59, 94-98. | 0.8 | 55 |
| 104 | Regarding the Consensus Statement on 21-Hydroxylase Deficiency from the Lawson Wilkins Pediatric Endocrine Society and The European Society for Paediatric Endocrinology. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 3455-3455. | 1.8 | 9 |
| 105 | A novel loss of function mutation in exon 10 of the FSH receptor gene causing hypergonadotrophic hypogonadism: clinical and molecular characteristics. Human Reproduction, 2003, 18, 251-256. | 0.4 | 100 |
| 106 | An Alternate Translation Initiation Site Circumvents an Amino-Terminal DAX1 Nonsense Mutation Leading to a Mild Form of X-Linked Adrenal Hypoplasia Congenita. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 417-423. | 1.8 | 103 |
| 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 | 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 |

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|---|---|---|---|
| 109 | Hypogonadotropic Hypogonadism as a Presenting Feature of Late-Onset X-Linked Adrenal Hypoplasia Congenita. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 44-48. | 1.8 | 94 |
| 110 | Gonadal Determination and Adrenal Development Are Regulated by the Orphan Nuclear Receptor Steroidogenic Factor-1, in a Dose-Dependent Manner. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 1829-1833. | 1.8 | 251 |
| 111 | Genetic Causes of Human Reproductive Disease. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 2447-2454. | 1.8 | 70 |
| 112 | The role of SF1 in adrenal and reproductive function: insight from naturally occurring mutations in humans. Molecular Genetics and Metabolism, 2002, 76, 85-91. | 0.5 | 73 |
| 113 | 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 |
| 114 | Inherited disorders of the gonadotropin hormones. Molecular and Cellular Endocrinology, 2001, 179, 89-96. | 1.6 | 77 |
| 115 | Phenotypic spectrum of mutations in DAX-1 and SF-1. Molecular and Cellular Endocrinology, 2001, 185, 17-25. | 1.6 | 146 |
| 116 | Dehydroepiandrosterone replacement for patients with adrenal insufficiency. Lancet, The, 2001, 357, 1381-1382. | 6.3 | 34 |
| 117 | Variable Presentation of X-linked Adrenal Hypoplasia Congenita. Journal of Pediatric Endocrinology and Metabolism, 2001, 14, 1093-6. | 0.4 | 40 |
| 118 | Missense Mutations Cluster within the Carboxyl-Terminal Region of DAX-1 and Impair Transcriptional Repression1. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 3171-3175. | 1.8 | 69 |
| 119 | X-Linked Adrenal Hypoplasia Congenita and DAX-1. , 2000, 10, 289-299. | | 13 |
| 120 | A Naturally Occurring Steroidogenic Factor-1 Mutation Exhibits Differential Binding and Activation of Target Genes. Journal of Biological Chemistry, 2000, 275, 31708-31714. | 1.6 | 75 |
| 121 | Presymptomatic diagnosis of X-linked adrenal hypoplasia congenita by analysis of DAX1. Journal of Pediatrics, 2000, 137, 878-881. | 0.9 | 57 |
| 122 | A novel mutation in DAX1 causes delayed-onset adrenal insufficiency and incomplete hypogonadotropic hypogonadism. Journal of Clinical Investigation, 2000, 105, 321-328. | 3.9 | 171 |
| 123 | Clinical and Functional Effects of Mutations in theDAX-1Gene in Patients with Adrenal Hypoplasia Congenita<sup>1</sup>. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 504-511. | 1.8 | 143 |
| 124 | X-Linked Adrenal Hypoplasia Congenita: A Mutation inDAX1Expands the Phenotypic Spectrum in Males and Females1. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 4501-4509. | 1.8 | 157 |
| 125 | Fertility and Infertility: Genetic Contributions from the Hypothalamic-Pituitary- Gonadal Axis. Molecular Endocrinology, 1999, 13, 812-818. | 3.7 | 51 |
| 126 | Mutational Analysis of DAX1 in Patients with Hypogonadotropic Hypogonadism or Pubertal Delay1. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 4497-4500. | 1.8 | 77 |

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| 127 | A mutation in the gene encoding steroidogenic factor-1 causes XY sex reversal and adrenal failure in humans. Nature Genetics, 1999, 22, 125-126. | 9.4 | 642 |
| 128 | X-Linked Adrenal Hypoplasia Congenita: A Mutation in DAX1Expands the Phenotypic Spectrum in Males and Females. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 4501-4509. | 1.8 | 124 |
| 129 | Clinical and Functional Effects of Mutations in the DAX-1 Gene in Patients with Adrenal Hypoplasia Congenita. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 504-511. | 1.8 | 110 |
| 130 | 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 |
| 131 | The Role of DAX-1 in Reproduction. Trends in Endocrinology and Metabolism, 1998, 9, 169-175. | 3.1 | 36 |
| 132 | Birth Weight Influences the Initial Response to Growth Hormone Treatment in Growth Hormone-insufficient Children. Pediatrics, 1998, 102, 342-345. | 1.0 | 16 |
| 133 | Clearing up at 2.5 years. British Journal of Developmental Psychology, 1991, 9, 365-376. | 0.9 | 21 |
| 134 | Disorders of sex development. , 0, , 15-34. | | 1 |
| 135 | A genomic atlas of human adrenal and gonad development. Wellcome Open Research, 0, 2, 25. | 0.9 | 33 |