Olaf Hiort

List of Publications by Year
in descending order

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157	5,383	66336	106340
papers	citations	42 h-index	65 g-index

168	168	168	4055
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Pubertal development in 46,XY patients with NR5A1 mutations. Endocrine, 2022, 75, 601-613.	2.3	4
2	Clinical, Biochemical, and Molecular Characterization of Indian Children with Clinically Suspected Androgen Insensitivity Syndrome. Sexual Development, 2022, 16, 34-45.	2.0	5
3	Combien y a-t-il de sexesÂ?. , 2022, NÂ° 140, 86-87.		0
4	ENDO-ERN ON RARE ENDOCRINE CONDITIONS: Endo-ERN in its fifth year: a pinch of care, science, curiosity and new horizons. Endocrine Connections, 2022, 11, .	1.9	1
5	Metabolic effects of estradiol versus testosterone in complete androgen insensitivity syndrome. Endocrine, 2022, 76, 722-732.	2.3	4
6	Intersex, DSD, and the Childâ€™s Well-Being: Changing Perceptions. Hormone Research in Paediatrics, 2022, 95, 21-24.	1.8	1
7	Can Non-Coding NR5A1 Gene Variants Explain Phenotypes of Disorders of Sex Development?. Sexual Development, 2022, 16, 252-260.	2.0	4
8	Assessing the benefits and challenges of video consultations for the treatment of children with type 1 diabetes â€“ A qualitative study among diabetes professionals. Experimental and Clinical Endocrinology and Diabetes, 2021, 129, 831-836.	1.2	15
9	Assessing the health-related management of people with differences of sex development. Endocrine, 2021, 71, 675-680.	2.3	9
10	Diabetes Type 1 Negatively Influences Leydig Cell Function in Rats, Which is Partially Reversible By Insulin Treatment. Endocrinology, 2021, 162, .	2.8	10
11	Educational and knowledge gaps within the European reference network on rare endocrine conditions. Endocrine Connections, 2021, 10, 37-44.	1.9	3
12	Introduction to Endo-ERNâ€“scope and mission. Endocrine, 2021, 71, 537-538.	2.3	7
13	Patients with rare endocrine conditions have corresponding views on unmet needs in clinical research. Endocrine, 2021, 71, 561-568.	2.3	4
14	Access to patient oriented informationâ€”a baseline Endo-ERN survey among patients with rare endocrine disorders. Endocrine, 2021, 71, 542-548.	2.3	3
15	CPMSâ€“improving patient care in Europe via virtual case discussions. Endocrine, 2021, 71, 549-554.	2.3	13
16	In vitro functional characterization of androgen receptor gene mutations at arginine p.856 of the ligand-binding-domain associated with androgen insensitivity syndrome. Journal of Steroid Biochemistry and Molecular Biology, 2021, 208, 105834.	2.5	1
17	Gonadectomy in conditions affecting sex development: a registry-based cohort study. European Journal of Endocrinology, 2021, 184, 791-801.	3.7	9
18	Corrigendum to: Clinical spectrum and management of imprinting disorders. Medizinische Genetik, 2021, 33, 61-63.	0.2	0

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19	Diagnosis and Therapy of Female Genital Malformations (Part 1). Guideline of the DGGG, OEGGG and SGGG (S2k Level, AWMF Registry Number 015/052, May 2019). Geburtshilfe Und Frauenheilkunde, 2021, 81, 1307-1328.	1.8	4
20	Diagnosis and Therapy of Female Genital Malformations (Part 2). Guideline of the DGGG, OEGGG and SGGG (S2k Level, AWMF Registry Number 015/052, May 2019). Geburtshilfe Und Frauenheilkunde, 2021, 81, 1329-1347.	1.8	2
21	Monthly Video-Consultation for Children With Type 1 Diabetes Using a Continuous Glucose Monitoring System: Design of ViDiKi, a Multimethod Intervention Study to Evaluate the Benefit of Telemedicine. Journal of Diabetes Science and Technology, 2020, 14, 105-111.	2.2	24
22	The External Genitalia Score (EGS): A European Multicenter Validation Study. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e222-e230.	3.6	51
23	Outcomes of monthly video consultations as an addâ€on to regular care for children with type 1 diabetes: A 6â€month quasiâ€randomized clinical trial followed by an extension phase. Pediatric Diabetes, 2020, 21, 1502-1515.	2.9	28
24	The EuRRECa Project as a Model for Data Access and Governance Policies for Rare Disease Registries That Collect Clinical Outcomes. International Journal of Environmental Research and Public Health, 2020, 17, 8743.	2.6	13
25	Recommendations for Diagnosis and Treatment of Pseudohypoparathyroidism and Related Disorders: An Updated Practical Tool for Physicians and Patients. Hormone Research in Paediatrics, 2020, 93, 182-196.	1.8	42
26	Genetic testing in inherited endocrine disorders: joint position paper of the European reference network on rare endocrine conditions (Endo-ERN). Orphanet Journal of Rare Diseases, 2020, 15, 144.	2.7	15
27	An overview of clinical activities in Endo-ERN: the need for alignment of future network criteria. European Journal of Endocrinology, 2020, 183, 141-148.	3.7	9
28	Validation of a next-generation sequencing (NGS) panel to improve the diagnosis of X-linked hypophosphataemia (XLH) and other genetic disorders of renal phosphate wasting. European Journal of Endocrinology, 2020, 183, 497-504.	3.7	11
29	Clinical spectrum and management of imprinting disorders. Medizinische Genetik, 2020, 32, 321-334.	0.2	5
30	Addressing gaps in care of people with conditions affecting sex development and maturation. Nature Reviews Endocrinology, 2019, 15, 615-622.	9.6	30
31	Reduced Androgen Receptor Expression in Genital Skin Fibroblasts From Patients With 45,X/46,XY Mosaicism. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 4630-4638.	3.6	5
32	Resistance to GHRH but Not to PTH in a 15-Year-Old Boy With Pseudohypoparathyroidism 1A. Journal of the Endocrine Society, 2019, 3, 1383-1389.	0.2	4
33	Clinical but Not Histological Outcomes in Males With 45,X/46,XY Mosaicism Vary Depending on Reason for Diagnosis. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 4366-4381.	3.6	27
34	Spectrum of Pathogenic Variants in <i>SRD5A2</i> in Indian Children with 46,XY Disorders of Sex Development and Clinically Suspected Steroid 5Î±-Reductase 2 Deficiency. Sexual Development, 2019, 13, 228-239.	2.0	6
35	Response to Letter to the Editor: â€œClinical but Not Histological Outcomes in Males With 45,X/46,XY Mosaicism Vary Depending on Reason for Diagnosisâ€• Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5812-5813.	3.6	0
36	Clinical Findings and Follow-Up of 46,XY and 45,X/46,XY Testicular Dysgenesis. Sexual Development, 2019, 13, 171-177.	2.0	16

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37	Standardised data collection for clinical follow-up and assessment of outcomes in differences of sex development (DSD): recommendations from the COST action DSDnet. European Journal of Endocrinology, 2019, 181, 545-564.	3.7	21
38	Concluding Remarks - From Bench to Bed. Sexual Development, 2018, 12, 155-157.	2.0	0
39	Endoscopy and Laparoscopy in Disorders of Sex Development. Sexual Development, 2018, 12, 100-105.	2.0	6
40	Effects of growth hormone treatment on adult height in severely short children with X-linked hypophosphatemic rickets. Pediatric Nephrology, 2018, 33, 447-456.	1.7	35
41	Functional characterization of fiveNR5A1gene mutations found in patients with 46,XY disorders of sex development. Human Mutation, 2018, 39, 114-123.	2.5	12
42	Epigenetic Repression of Androgen Receptor Transcription in Mutation-Negative Androgen Insensitivity Syndrome (AIS Type II). Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4617-4627.	3.6	22
43	In vitrofunctional characterization of the novelDHHmutations p.(Asn337Lysfs*24) and p.(Glu212Lys) associated with gonadal dysgenesis. Human Mutation, 2018, 39, 2097-2109.	2.5	12
44	Involving Individuals with Disorders of Sex Development and Their Parents in Exploring New Models of Shared Learning: Proceedings from a DSDnet COST Action Workshop. Sexual Development, 2018, 12, 225-231.	2.0	13
45	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 476-500.	9.6	224
46	Oestrogen versus androgen in hormone-replacement therapy for complete androgen insensitivity syndrome: a multicentre, randomised, double-dummy, double-blind crossover trial. Lancet Diabetes and Endocrinology,the, 2018, 6, 771-780.	11.4	35
47	Progression of Mineral Ion Abnormalities in Patients With Jansen Metaphyseal Chondrodysplasia. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2660-2669.	3.6	17
48	A Case of Homozygous Disruption of P450 Side-Chain Cleavage (CYP11A1): Cerebral MRI and CSF Neurotransmitter Findings. Journal of Neurology Research, 2018, 8, 13-15.	0.5	1
49	Pubertal Development inâ€¨17Beta-Hydroxysteroid Dehydrogenase Type 3 Deficiency. Hormone Research in Paediatrics, 2017, 87, 354-358.	1.8	17
50	Birth Weight in Different Etiologies of Disorders of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1044-1050.	3.6	16
51	Functional Impact of Novel Androgen Receptor Mutations on the Clinical Manifestation of Androgen Insensitivity Syndrome. Sexual Development, 2017, 11, 238-247.	2.0	9
52	Gonadectomy in Complete Androgen Insensitivity Syndrome: Why and When?. Sexual Development, 2017, 11, 171-174.	2.0	56
53	Understanding the needs of professionals who provide psychosocial care for children and adults with disorders of sex development. BMJ Paediatrics Open, 2017, 1, e000132.	1.4	19
54	New NR5A1 mutations and phenotypic variations of gonadal dysgenesis. PLoS ONE, 2017, 12, e0176720.	2.5	37

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55	Normal and Variant Sex Development. , 2017, , 1-16.		1
56	Follow-up Findings in a Turkish Girl with Pseudohypoparathyroidism Type Ia Caused by a Novel Heterozygous Mutation in the GNAS Gene. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 74-79.	0.9	0
57	A Recurrent Germline Mutation in the 5â€™UTR of the Androgen Receptor Causes Complete Androgen Insensitivity by Activating Aberrant uORF Translation. PLoS ONE, 2016, 11, e0154158.	2.5	41
58	Response to the Council of Europe Human Rights Commissioner's Issue Paper on Human Rights and Intersex People. European Urology, 2016, 70, 407-409.	1.9	35
59	From pseudohypoparathyroidism to inactivating PTH/PTHrP signalling disorder (iPPSD), a novel classification proposed by the EuroPHP network. European Journal of Endocrinology, 2016, 175, P1-P17.	3.7	117
60	Current models of care for disorders of sex development â€“ results from an International survey of specialist centres. Orphanet Journal of Rare Diseases, 2016, 11, 155.	2.7	63
61	A positive genotypeâ€“phenotype correlation in a large cohort of patients with Pseudohypoparathyroidism Type Ia and Pseudoâ€pseudohypoparathyroidism and 33 newly identified mutations in the <scp>GNAS</scp> gene. Molecular Genetics & Genomic Medicine, 2015, 3, 111-120.	1.2	46
62	Copy number variation of two separate regulatory regions upstream ofSOX9causes isolated 46,XY or 46,XX disorder of sex development. Journal of Medical Genetics, 2015, 52, 240-247.	3.2	88
63	Uniparental Disomy in Somatic Mosaicism 45,X/46,XY/46,XX Associated with Ambiguous Genitalia. Sexual Development, 2015, 9, 136-143.	2.0	0
64	Characteristic Features of Reproductive Hormone Profiles in Late Adolescent and Adult Females with Complete Androgen Insensitivity Syndrome. Sexual Development, 2015, 9, 69-74.	2.0	49
65	46,XY Gonadal Dysgenesis due to a Homozygous Mutation in Desert Hedgehog (DHH) Identified by Exome Sequencing. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1022-E1029.	3.6	59
66	Novel Insights into 46,XY Disorders of Sex Development due to <i>NR5A1</i> Gene Mutation. Sexual Development, 2015, 9, 260-268.	2.0	17
67	Severe Undervirilisation in a 46,XY Case Due to a Novel Mutation in HSD17B3 Gene. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2015, 7, 249-252.	0.9	6
68	46,XY Disorder of Sex Development in a Sudanese Patient Caused by a Novel Mutation in theHSD17B3Gene. Sexual Development, 2014, 8, 151-155.	2.0	8
69	Preserved Fertility in a Patient with Gynecomastia Associated with the p.Pro695Ser Mutation in the Androgen Receptor. Sexual Development, 2014, 8, 350-355.	2.0	8
70	Health-related quality of life in children with disorders of sex development (DSD). European Journal of Pediatrics, 2014, 173, 893-903.	2.7	36
71	Novel Associations in Disorders of Sex Development: Findings From the I-DSD Registry. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E348-E355.	3.6	85
72	Utilization of Health Care Services and Satisfaction with Care in Adults Affected by Disorders of Sex Development (DSD). Journal of General Internal Medicine, 2014, 29, 752-759.	2.6	46

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73	Management of disorders of sex development. Nature Reviews Endocrinology, 2014, 10, 520-529.	9.6	114
74	Changes Over Time in Sex Assignment for Disorders of Sex Development. Pediatrics, 2014, 134, e710-e715.	2.1	98
75	Different Pattern of Epigenetic Changes of theGNASGene Locus in Patients With Pseudohypoparathyroidism Type Ic Confirm the Heterogeneity of Underlying Pathomechanisms in This Subgroup of Pseudohypoparathyroidism and the Demand for a New Classification ofGNAS-Related Disorders. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1564-E1570.	3.6	28
76	Long-term management of patients with disorders of sex development (DSD). Annales D'Endocrinologie, 2014, 75, 64-66.	1.4	7
77	Psychosexual Development in Adolescents and Adults with Disorders of Sex Developmentâ€"Results from the German Clinical Evaluation Study. Journal of Sexual Medicine, 2013, 10, 2703-2714.	0.6	64
78	The differential role of androgens in early human sex development. BMC Medicine, 2013, 11, 152.	5.5	67
79	Clinical and Molecular Aspects of Androgen Insensitivity. Endocrine Development, 2013, 24, 33-40.	1.3	35
80	Minor Hypospadias: The â€œTip of the Icebergâ€•of the Partial Androgen Insensitivity Syndrome. PLoS ONE, 2013, 8, e61824.	2.5	36
81	Androgen Receptor Function Links Human Sexual Dimorphism to DNA Methylation. PLoS ONE, 2013, 8, e73288.	2.5	26
82	Partial deletion of DMRT1 causes 46,XY ovotesticular disorder of sexual development. European Journal of Endocrinology, 2012, 167, 119-124.	3.7	66
83	17Î²-Hydroxysteroid dehydrogenase type 3 deficiency as a result of a homozygous 7 base pair deletion in 17Î²HSD3 gene. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 561-3.	0.9	13
84	Reasonable diagnostic pathways in disorders of sex development. Laboratoriums Medizin, 2012, 36, 1-6.	0.6	0
85	PRKAR1AandPDE4DMutations Cause Acrodysostosis but Two Distinct Syndromes with or without GPCR-Signaling Hormone Resistance. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E2328-E2338.	3.6	100
86	Requirements for a multicentric multidisciplinary registry on patients with disorders of sex development. Journal of Pediatric Urology, 2012, 8, 624-628.	1.1	13
87	Satisfaction with Genital Surgery and Sexual Life of Adults with XY Disorders of Sex Development: Results from the German Clinical Evaluation Study. Journal of Clinical Endocrinology and Metabolism, 2012, 97, 577-588.	3.6	126
88	46,XY Karyotype in a Female Phenotype Fetus: A Challenging Diagnosis. Journal of Pediatric and Adolescent Gynecology, 2012, 25, e77-e79.	0.7	6
89	RWDD1 interacts with the ligand binding domain of the androgen receptor and acts as a coactivator of androgen-dependent transactivation. Molecular and Cellular Endocrinology, 2012, 358, 53-62.	3.2	9
90	Patients with disorders of sex development (DSD) at risk of gonadal tumour development: management based on laparoscopic biopsy and molecular diagnosis. BJU International, 2012, 110, E958-E965.	2.5	80

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91	Functional characterization of GNAS mutations found in patients with pseudohypoparathyroidism type Ic defines a new subgroup of pseudohypoparathyroidism affecting selectively GsÎ±-receptor interaction. Human Mutation, 2011, 32, 653-660.	2.5	62
92	GsÎ± activity is reduced in erythrocyte membranes of patients with psedohypoparathyroidism due to epigenetic alterations at the GNAS locus. Journal of Bone and Mineral Research, 2011, 26, 1864-1870.	2.8	52
93	Hereditary vitamin Dâ€“resistant rickets (HVDRR) owing to a heterozygous mutation in the vitamin D receptor. Journal of Bone and Mineral Research, 2011, 26, 2710-2718.	2.8	36
94	Diagnostic pathways in disorders of sex development. Clinical Biochemistry, 2011, 44, 509.	1.9	3
95	Hormonal Management of Complete Androgen Insensitivity Syndrome from Adolescence Onward. Hormone Research in Paediatrics, 2011, 76, 428-433.	1.8	37
96	Deletion and Point Mutations of PTHLH Cause Brachydactyly Type E. American Journal of Human Genetics, 2010, 86, 434-439.	6.2	127
97	A de novo unbalanced translocation leading to partial monosomy 9p23-pter and partial trisomy 15q25.3-qter associated with 46,XY complete gonadal dysgenesis, tall stature and mental retardation. Clinical Dysmorphology, 2010, 19, 190-194.	0.3	2
98	Long-term follow-up of a pseudohypoparathyroidism type 1A patient with missense mutation (Pro115Ser) in exon 5. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2010, 2, 85-88.	0.9	3
99	First Steps Towards the Engineering of Genital Skin. Journal of Pediatric Urology, 2010, 6, S22-S23.	1.1	0
100	46,XY disorders of sex development â€“ the undermasculinised male with disorders of androgen action. Best Practice and Research in Clinical Endocrinology and Metabolism, 2010, 24, 263-277.	4.7	30
101	Difficulties in Diagnosis and Treatment of 5Î±-Reductase Type 2 Deficiency in a Newborn with 46,XY DSD. Hormone Research in Paediatrics, 2010, 74, 67-71.	1.8	53
102	Diagnosis of 17Î²-hydroxysteroid dehydrogenase deficiency. Expert Review of Endocrinology and Metabolism, 2009, 4, 53-65.	2.4	21
103	Pseudohypoparathyroidism Type IA (PHP-Ia): Maternally Inherited GNAS Gene Mutation. Journal of Pediatric Endocrinology and Metabolism, 2009, 22, 107-8.	0.9	1
104	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.	3.7	115
105	Disorders of sex development expose transcriptional autonomy of genetic sex and androgen-programmed hormonal sex in human blood leukocytes. BMC Genomics, 2009, 10, 292.	2.8	21
106	Transition from insulin to sulfonylurea in a child with diabetes due to a mutation in KCNJ11 encoding Kir6.2â€”initial and long-term response to sulfonylurea therapy. European Journal of Pediatrics, 2009, 168, 359-361.	2.7	14
107	Apolipoprotein D (APOD) is a putative biomarker of androgen receptor function in androgen insensitivity syndrome. Journal of Molecular Medicine, 2009, 87, 623-632.	3.9	35
108	Alternative Gender Categories in Different Cultures: Lessons to be Learnt. Hormone Research in Paediatrics, 2008, 69, 251-252.	1.8	0

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109	PCR-Based Analysis of Differentially Methylated Regions of GNAS Enables Convenient Diagnostic Testing of Pseudohypoparathyroidism Type Ib. Clinical Chemistry, 2008, 54, 1537-1545.	3.2	23
110	Functional and Structural Consequences of a Novel Point Mutation in theCYP21A2Gene Causing Congenital Adrenal Hyperplasia: Potential Relevance of Helix C for P450 Oxidoreductase-21-Hydroxylase Interaction. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 2891-2895.	3.6	14
111	Association of Parvovirus B19 Infection and Hashimoto's Thyroiditis in Children. Viral Immunology, 2008, 21, 379-384.	1.3	51
112	Classification of Distinct Baseline Insulin Infusion Patterns in Children and Adolescents With Type 1 Diabetes on Continuous Subcutaneous Insulin Infusion Therapy. Diabetes Care, 2007, 30, 568-573.	8.6	40
113	Epigenetic Defects ofGNASin Patients with Pseudohypoparathyroidism and Mild Features of Albrightâ€™s Hereditary Osteodystrophy. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 2370-2373.	3.6	157
114	Gender role behavior in children with XY karyotype and disorders of sex development. Hormones and Behavior, 2007, 51, 443-453.	2.1	79
115	We used to call them hermaphrodites. Genetics in Medicine, 2007, 9, 65-66.	2.4	33
116	A Disruptive Mutation in Exon 3 of the GNAS Gene with Albright Hereditary Osteodystrophy, Normocalcemic Pseudohypoparathyroidism, and Selective Long Transcript Variant Gsî±-L Deficiency. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 1764-1768.	3.6	34
117	Ã‰volution auÂlong cours deÂlaÂdensitÃ© minÃ©rale osseuse dansÂl'hypophosphatasie juvÃ©nile. Revue Du Rhumatisme (Edition Francaise), 2007, 74, 479-485.	0.0	0
118	Intrinsic androgen-dependent gene expression patterns revealed by comparison of genital fibroblasts from normal males and individuals with complete and partial androgen insensitivity syndrome. BMC Genomics, 2007, 8, 376.	2.8	38
119	Long-term follow-up of bone mineral density in childhood hypophosphatasia. Joint Bone Spine, 2007, 74, 263-269.	1.6	19
120	17Î²-Hydroxysteroid dehydrogenase-3 deficiency: A rare endocrine cause of male-to-female sex reversal. Gynecological Endocrinology, 2006, 22, 488-494.	1.7	22
121	Androgen receptor gene mutations in androgen insensitivity syndrome cause distinct patterns of reduced activation of androgen-responsive promoter constructs. Journal of Steroid Biochemistry and Molecular Biology, 2006, 101, 1-10.	2.5	16
122	Cell-line and tissue-specific signatures of androgen receptor-coregulator transcription. Journal of Molecular Medicine, 2006, 84, 919-931.	3.9	44
123	The A645D Mutation in the Hinge Region of the Human Androgen Receptor (AR) Gene Modulates AR Activity, Depending on the Context of the Polymorphic Glutamine and Glycine Repeats. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 3515-3520.	3.6	56
124	Epidemiology and Initial Management of Ambiguous Genitalia at Birth in Germany. Hormone Research in Paediatrics, 2006, 66, 195-203.	1.8	171
125	The Approach to a Neonate with a Possible Prenatal Diagnosis of Androgen Insensitivity Syndrome. Journal of Pediatric Endocrinology and Metabolism, 2006, 19, 1437-43.	0.9	1
126	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.	3.6	112

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127	Isoenzyme type 1 of 5alpha-reductase is abundantly transcribed in normal human genital skin fibroblasts and may play an important role in masculinization of 5alpha-reductase type 2 deficient males. European Journal of Endocrinology, 2005, 152, 875-880.	3.7	43
128	Early manifestation of calcinosis cutis in pseudohypoparathyroidism type Ia associated with a novel mutation in the GNAS gene. European Journal of Endocrinology, 2005, 152, 515-519.	3.7	44
129	A novel missense mutation of 5-alpha reductase type 2 gene (SRD5A2) leads to severe male pseudohermaphroditism in a Turkish family. Urology, 2005, 66, 407-410.	1.0	26
130	Deciding on Gender in Children with Intersex Conditions. Treatments in Endocrinology: Guiding Your Management of Endocrine Disorders, 2005, 4, 1-8.	1.8	47
131	Steroid 5Î±-Reductase 1 Polymorphisms and Testosterone/Dihydrotestosterone Ratio in Male Patients with Hypospadias. Hormone Research in Paediatrics, 2004, 61, 180-183.	1.8	21
132	Differential gene-expression patterns in genital fibroblasts of normal males and 46,XY females with androgen insensitivity syndrome: evidence for early programming involving the androgen receptor. Genome Biology, 2003, 4, R37.	9.6	45
133	A new heterozygous mutation (L338N) in the human Gsalpha (GNAS1) gene as a cause for congenital hypothyroidism in Albright's hereditary osteodystrophy. European Journal of Endocrinology, 2003, 148, 463-468.	3.7	30
134	Effect of GH replacement therapy in two male siblings with combined X-linked hypophosphatemia and partial GH deficiency. European Journal of Endocrinology, 2003, 149, 317-321.	3.7	14
135	A Novel Point Mutation in the Hormone Binding Domain of the Androgen Receptor Associated with Partial and Minimal Androgen Insensitivity Syndrome. Journal of Pediatric Endocrinology and Metabolism, 2003, 16, 149-54.	0.9	11
136	Mutational Analysis of Hungarian Patients with Androgen Insensitivity Syndrome. Journal of Pediatric Endocrinology and Metabolism, 2003, 16, 367-73.	0.9	5
137	Prof. Dr. Klaus Kruse. Hormone Research in Paediatrics, 2002, 57, 64-64.	1.8	0
138	Androgens and Puberty. Best Practice and Research in Clinical Endocrinology and Metabolism, 2002, 16, 31-41.	4.7	64
139	A novel homozygous disruptive mutation in the SRD5A2-gene in a partially virilized patient with 5alpha-reductase deficiency. Journal of Developmental and Physical Disabilities, 2002, 25, 55-58.	3.6	24
140	Basal Inhibin B and the Testosterone Response to Human Chorionic Gonadotropin Correlate in Prepubertal Boys1. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 134-138.	3.6	87
141	Clinical, Endocrine, and Molecular Genetic Findings in Patients with 17Î²-Hydroxysteroid Dehydrogenase Deficiency. Hormone Research in Paediatrics, 2000, 53, 26-31.	1.8	31
142	The Molecular Basis of Androgen Insensitivity. Hormone Research in Paediatrics, 2000, 54, 327-333.	1.8	24
143	Transcription of androgen receptor and 5Î±-reductase II in genital fibroblasts from patients with androgen insensitivity syndrome. Journal of Steroid Biochemistry and Molecular Biology, 2000, 75, 213-218.	2.5	4
144	Gonadal Histology with Testicular Carcinomain Situin a 15-Year-Old 46,XY Female Patient with a Premature Termination in the Steroidogenic Acute Regulatory Protein Causing Congenital Lipoid Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 1628-1632.	3.6	46

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145	Complete androgen insensitivity caused by a splice donor site mutation in intron 2 of the human androgen receptor gene resulting in an exon 2-lacking transcript with premature stop-codon and reduced expression. Journal of Steroid Biochemistry and Molecular Biology, 1999, 68, 1-9.	2.5	31
146	Clinical and Molecular Spectrum of Somatic Mosaicism in Androgen Insensitivity Syndrome. Pediatric Research, 1999, 46, 684-684.	2.3	43
147	Immunohistochemistry and in situ hybridization of the androgen receptor in the developing human prostate. Anatomy and Embryology, 1998, 197, 199-208.	1.5	20
148	Physiology and pathophysiology of androgen action. Bailliere's Clinical Endocrinology and Metabolism, 1998, 12, 115-132.	1.0	27
149	Inherited and de novo androgen receptor gene mutations: Investigation of single-case families. Journal of Pediatrics, 1998, 132, 939-943.	1.8	98
150	Androgens and Fetal Growth. Hormone Research in Paediatrics, 1998, 50, 243-244.	1.8	65
151	Etiologic classification of severe hypospadias: Implications for prognosis and management. Journal of Pediatrics, 1997, 131, 386-392.	1.8	84
152	Molecular genetic analysis and human chorionic gonadotropin stimulation tests in the diagnosis of prepubertal patients with partial 5Î±-reductase deficiency. European Journal of Pediatrics, 1996, 155, 445-451.	2.7	53
153	Analysis ofRET protooncogene point mutations distinguishes heritable from nonheritable medullary thyroid carcinomas. Cancer, 1995, 76, 479-489.	4.1	145
154	Molecular diagnosis of multiple endocrine neoplasia (MEN) in paraffin-embedded specimens. Endocrine Pathology, 1995, 6, 267-278.	9.0	21
155	True hermaphroditism with 46,XY karyotype and a point mutation in the SRY gene. Journal of Pediatrics, 1995, 126, 1022.	1.8	41
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