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	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 476-500.	9.6	224
2	Epidemiology and Initial Management of Ambiguous Genitalia at Birth in Germany. Hormone Research in Paediatrics, 2006, 66, 195-203.	1.8	171
3	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
4	Analysis ofRET protooncogene point mutations distinguishes heritable from nonheritable medullary thyroid carcinomas. Cancer, 1995, 76, 479-489.	4.1	145
5	Deletion and Point Mutations of PTHLH Cause Brachydactyly Type E. American Journal of Human Genetics, 2010, 86, 434-439.	6.2	127
6	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
7	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
8	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
9	Management of disorders of sex development. Nature Reviews Endocrinology, 2014, 10, 520-529.	9.6	114
10	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
11	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
12	Inherited and de novo androgen receptor gene mutations: Investigation of single-case families. Journal of Pediatrics, 1998, 132, 939-943.	1.8	98
13	Changes Over Time in Sex Assignment for Disorders of Sex Development. Pediatrics, 2014, 134, e710-e715.	2.1	98
14	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
15	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
16	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
17	Etiologic classification of severe hypospadias: Implications for prognosis and management. Journal of Pediatrics, 1997, 131, 386-392.	1.8	84
18	Detection of point mutations in the androgen receptor gene using non-isotopic single strand conformation polymorphism analysis. Human Molecular Genetics, 1994, 3, 1163-1166.	2.9	81

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19	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
20	Gender role behavior in children with XY karyotype and disorders of sex development. Hormones and Behavior, 2007, 51, 443-453.	2.1	79
21	The differential role of androgens in early human sex development. BMC Medicine, 2013, 11, 152.	5.5	67
22	Partial deletion of DMRT1 causes 46,XY ovotesticular disorder of sexual development. European Journal of Endocrinology, 2012, 167, 119-124.	3.7	66
23	Androgens and Fetal Growth. Hormone Research in Paediatrics, 1998, 50, 243-244.	1.8	65
24	Androgens and Puberty. Best Practice and Research in Clinical Endocrinology and Metabolism, 2002, 16, 31-41.	4.7	64
25	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
26	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
27	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
28	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
29	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
30	Gonadectomy in Complete Androgen Insensitivity Syndrome: Why and When?. Sexual Development, 2017, 11, 171-174.	2.0	56
31	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
32	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
33	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
34	Association of Parvovirus B19 Infection and Hashimoto's Thyroiditis in Children. Viral Immunology, 2008, 21, 379-384.	1.3	51
35	The External Genitalia Score (EGS): A European Multicenter Validation Study. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e222-e230.	3.6	51
36	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

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37	Deciding on Gender in Children with Intersex Conditions. Treatments in Endocrinology: Guiding Your Management of Endocrine Disorders, 2005, 4, 1-8.	1.8	47
38	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
39	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
40	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
41	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
42	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
43	Cell-line and tissue-specific signatures of androgen receptor-coregulator transcription. Journal of Molecular Medicine, 2006, 84, 919-931.	3.9	44
44	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
45	Clinical and Molecular Spectrum of Somatic Mosaicism in Androgen Insensitivity Syndrome. Pediatric Research, 1999, 46, 684-684.	2.3	43
46	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
47	True hermaphroditism with 46,XY karyotype and a point mutation in the SRY gene. Journal of Pediatrics, 1995, 126, 1022.	1.8	41
48	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
49	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
50	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
51	Hormonal Management of Complete Androgen Insensitivity Syndrome from Adolescence Onward. Hormone Research in Paediatrics, 2011, 76, 428-433.	1.8	37
52	New NR5A1 mutations and phenotypic variations of gonadal dysgenesis. PLoS ONE, 2017, 12, e0176720.	2.5	37
53	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
54	Minor Hypospadias: The â€œTip of the Icebergâ€of the Partial Androgen Insensitivity Syndrome. PLoS ONE, 2013, 8, e61824.	2.5	36

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55	Health-related quality of life in children with disorders of sex development (DSD). European Journal of Pediatrics, 2014, 173, 893-903.	2.7	36
56	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
57	Clinical and Molecular Aspects of Androgen Insensitivity. Endocrine Development, 2013, 24, 33-40.	1.3	35
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	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
60	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
61	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
62	We used to call them hermaphrodites. Genetics in Medicine, 2007, 9, 65-66.	2.4	33
63	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
64	Clinical, Endocrine, and Molecular Genetic Findings in Patients with 17Î²-Hydroxysteroid Dehydrogenase Deficiency. Hormone Research in Paediatrics, 2000, 53, 26-31.	1.8	31
65	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
66	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
67	Addressing gaps in care of people with conditions affecting sex development and maturation. Nature Reviews Endocrinology, 2019, 15, 615-622.	9.6	30
68	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
69	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
70	Physiology and pathophysiology of androgen action. Bailliere's Clinical Endocrinology and Metabolism, 1998, 12, 115-132.	1.0	27
71	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
72	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

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73	Androgen Receptor Function Links Human Sexual Dimorphism to DNA Methylation. PLoS ONE, 2013, 8, e73288.	2.5	26
74	The Molecular Basis of Androgen Insensitivity. Hormone Research in Paediatrics, 2000, 54, 327-333.	1.8	24
75	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
76	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
77	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
78	17Î²-Hydroxysteroid dehydrogenase-3 deficiency: A rare endocrine cause of male-to-female sex reversal. Gynecological Endocrinology, 2006, 22, 488-494.	1.7	22
79	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
80	Molecular diagnosis of multiple endocrine neoplasia (MEN) in paraffin-embedded specimens. Endocrine Pathology, 1995, 6, 267-278.	9.0	21
81	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
82	Diagnosis of 17Î²-hydroxysteroid dehydrogenase deficiency. Expert Review of Endocrinology and Metabolism, 2009, 4, 53-65.	2.4	21
83	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
84	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
85	Immunohistochemistry and in situ hybridization of the androgen receptor in the developing human prostate. Anatomy and Embryology, 1998, 197, 199-208.	1.5	20
86	Long-term follow-up of bone mineral density in childhood hypophosphatasia. Joint Bone Spine, 2007, 74, 263-269.	1.6	19
87	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
88	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
89	Pubertal Development inâ€¨17Beta-Hydroxysteroid Dehydrogenase Type 3 Deficiency. Hormone Research in Paediatrics, 2017, 87, 354-358.	1.8	17
90	Progression of Mineral Ion Abnormalities in Patients With Jansen Metaphyseal Chondrodysplasia. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2660-2669.	3.6	17

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91	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
92	Birth Weight in Different Etiologies of Disorders of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1044-1050.	3.6	16
93	Clinical Findings and Follow-Up of 46,XY and 45,X/46,XY Testicular Dysgenesis. Sexual Development, 2019, 13, 171-177.	2.0	16
94	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
95	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
96	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
97	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
98	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
99	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
100	Requirements for a multicentric multidisciplinary registry on patients with disorders of sex development. Journal of Pediatric Urology, 2012, 8, 624-628.	1.1	13
101	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
102	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
103	CPMSâ€“improving patient care in Europe via virtual case discussions. Endocrine, 2021, 71, 549-554.	2.3	13
104	Functional characterization of fiveNR5A1gene mutations found in patients with 46,XY disorders of sex development. Human Mutation, 2018, 39, 114-123.	2.5	12
105	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
106	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
107	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
108	Diabetes Type 1 Negatively Influences Leydig Cell Function in Rats, Which is Partially Reversible By Insulin Treatment. Endocrinology, 2021, 162, .	2.8	10

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109	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
110	Functional Impact of Novel Androgen Receptor Mutations on the Clinical Manifestation of Androgen Insensitivity Syndrome. Sexual Development, 2017, 11, 238-247.	2.0	9
111	Assessing the health-related management of people with differences of sex development. Endocrine, 2021, 71, 675-680.	2.3	9
112	Gonadectomy in conditions affecting sex development: a registry-based cohort study. European Journal of Endocrinology, 2021, 184, 791-801.	3.7	9
113	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
114	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
115	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
116	Long-term management of patients with disorders of sex development (DSD). Annales D'Endocrinologie, 2014, 75, 64-66.	1.4	7
117	Introduction to Endo-ERNâ€“scope and mission. Endocrine, 2021, 71, 537-538.	2.3	7
118	46,XY Karyotype in a Female Phenotype Fetus: A Challenging Diagnosis. Journal of Pediatric and Adolescent Gynecology, 2012, 25, e77-e79.	0.7	6
119	Endoscopy and Laparoscopy in Disorders of Sex Development. Sexual Development, 2018, 12, 100-105.	2.0	6
120	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
121	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
122	Mutational Analysis of Hungarian Patients with Androgen Insensitivity Syndrome. Journal of Pediatric Endocrinology and Metabolism, 2003, 16, 367-73.	0.9	5
123	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
124	Clinical, Biochemical, and Molecular Characterization of Indian Children with Clinically Suspected Androgen Insensitivity Syndrome. Sexual Development, 2022, 16, 34-45.	2.0	5
125	Clinical spectrum and management of imprinting disorders. Medizinische Genetik, 2020, 32, 321-334.	0.2	5
126	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

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127	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
128	Patients with rare endocrine conditions have corresponding views on unmet needs in clinical research. Endocrine, 2021, 71, 561-568.	2.3	4
129	Pubertal development in 46,XY patients with NR5A1 mutations. Endocrine, 2022, 75, 601-613.	2.3	4
130	Metabolic effects of estradiol versus testosterone in complete androgen insensitivity syndrome. Endocrine, 2022, 76, 722-732.	2.3	4
131	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
132	Molecular characterization of the androgen receptor gene in boys with hypospadias. European Journal of Pediatrics, 1994, 153, 317-321.	2.7	4
133	Can Non-Coding NR5A1 Gene Variants Explain Phenotypes of Disorders of Sex Development?. Sexual Development, 2022, 16, 252-260.	2.0	4
134	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
135	Diagnostic pathways in disorders of sex development. Clinical Biochemistry, 2011, 44, 509.	1.9	3
136	Educational and knowledge gaps within the European reference network on rare endocrine conditions. Endocrine Connections, 2021, 10, 37-44.	1.9	3
137	Access to patient oriented informationâ€"a baseline Endo-ERN survey among patients with rare endocrine disorders. Endocrine, 2021, 71, 542-548.	2.3	3
138	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
139	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
140	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
141	Pseudohypoparathyroidism Type IA (PHP-Ia): Maternally Inherited GNAS Gene Mutation. Journal of Pediatric Endocrinology and Metabolism, 2009, 22, 107-8.	0.9	1
142	Normal and Variant Sex Development. , 2017, , 1-16.		1
143	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
144	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

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145	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
146	Intersex, DSD, and the Childâ€™s Well-Being: Changing Perceptions. Hormone Research in Paediatrics, 2022, 95, 21-24.	1.8	1
147	Prof. Dr. Klaus Kruse. Hormone Research in Paediatrics, 2002, 57, 64-64.	1.8	0
148	Ã‰volution auÂlong cours deÂlaÂdensitÃ© minÃ©rale osseuse dansÂl'hypophosphatasie juvÃ©nile. Revue Du Rhumatisme (Edition Francaise), 2007, 74, 479-485.	0.0	0
149	Alternative Gender Categories in Different Cultures: Lessons to be Learnt. Hormone Research in Paediatrics, 2008, 69, 251-252.	1.8	0
150	First Steps Towards the Engineering of Genital Skin. Journal of Pediatric Urology, 2010, 6, S22-S23.	1.1	0
151	Reasonable diagnostic pathways in disorders of sex development. Laboratoriums Medizin, 2012, 36, 1-6.	0.6	0
152	Uniparental Disomy in Somatic Mosaicism 45,X/46,XY/46,XX Associated with Ambiguous Genitalia. Sexual Development, 2015, 9, 136-143.	2.0	0
153	Concluding Remarks - From Bench to Bed. Sexual Development, 2018, 12, 155-157.	2.0	0
154	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
155	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
156	Corrigendum to: Clinical spectrum and management of imprinting disorders. Medizinische Genetik, 2021, 33, 61-63.	0.2	0
157	Combien y a-t-il de sexesÂ?. , 2022, NÂ° 140, 86-87.		0