Martine Cools

List of Publications by Year
in descending order

116 papers	6,478 citations	76196 40 h-index	69108 77 g-index
119 all docs	119 docs citations	119 times ranked	4189 citing authors

#	Article	IF	Citations
1	Ovotesticular Difference of Sex Development: Genetic Background, Histological Features, and Clinical Management. Hormone Research in Paediatrics, 2023, 96, 180-189.	0.8	7
2	Growth, puberty and testicular function in boys born small for gestational age with a nonspecific disorder of sex development. Clinical Endocrinology, 2022, 96, 165-174.	1.2	6
3	Prenatal dexamethasone treatment for classic 21-hydroxylase deficiency in Europe. European Journal of Endocrinology, 2022, 186, K17-K24.	1.9	7
4	Fertility and sexuality issues in congenital lifelong urology patients: male aspects. World Journal of Urology, 2021, 39, 1013-1019.	1.2	28
5	Real-World Estimates of Adrenal Insufficiencyâ€“Related Adverse Events in Children With Congenital Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e192-e203.	1.8	20
6	Assessing the health-related management of people with differences of sex development. Endocrine, 2021, 71, 675-680.	1.1	9
7	ENDO-ERN expert opinion on the differential diagnosis of pubertal delay. Endocrine, 2021, 71, 681-688.	1.1	19
8	Multidisciplinary Approach to the Child with Sex Chromosomal Mosaicism Including a Y-Containing Cell Line. International Journal of Environmental Research and Public Health, 2021, 18, 917.	1.2	6
9	Testosterone Therapy and Its Monitoring in Adolescent Boys with Hypogonadism: Results of an International Survey from the I-DSD Registry. Sexual Development, 2021, 15, 236-243.	1.1	4
10	Patients with rare endocrine conditions have corresponding views on unmet needs in clinical research. Endocrine, 2021, 71, 561-568.	1.1	4
11	Novel model to study the physiological effects of temporary or prolonged sex steroid deficiency in male mice. American Journal of Physiology - Endocrinology and Metabolism, 2021, 320, E415-E424.	1.8	7
12	International practice of corticosteroid replacement therapy in congenital adrenal hyperplasia: data from the I-CAH registry. European Journal of Endocrinology, 2021, 184, 553-563.	1.9	21
13	Reply by Authors. Journal of Urology, 2021, 206, 744-744.	0.2	0
14	Adolescent and Young Adult Urogenital Outcome following Childhood Hypospadias Repair: Perfection Revisited. Journal of Urology, 2021, 206, 734-744.	0.2	14
15	Approach to the Virilizing Girl at Puberty. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 1530-1539.	1.8	7
16	Rare and special robotic surgery indications in the pediatric population: ectopic organs and differences of sexual development. World Journal of Urology, 2020, 38, 1865-1868.	1.2	5
17	Androgen receptor expression in preputial dartos tissue correlates with physiological androgen exposure in congenital malformations of the penis and in controls. Journal of Pediatric Urology, 2020, 16, 43.e1-43.e8.	0.6	9
18	The External Genitalia Score (EGS): A European Multicenter Validation Study. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e222-e230.	1.8	51

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19	Families with pediatric type 1 diabetes: A comparison with the general population on child wellâ€being, parental distress, and parenting behavior. Pediatric Diabetes, 2020, 21, 395-408.	1.2	20
20	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.	1.2	13
21	SUN-070 European Registries for Rare Endocrine Conditions (EuRRECa): Results from the Platform for E-reporting of Rare Endocrine Conditions (e-REC). Journal of the Endocrine Society, 2020, 4, .	0.1	0
22	Parental stress, anxiety and trait mindfulness: associations with parentâ€“child mealtime interactions in children with type 1 diabetes. Journal of Behavioral Medicine, 2020, 43, 448-459.	1.1	4
23	Under-reported aspects of diagnosis and treatment addressed in the Dutch-Flemish guideline for comprehensive diagnostics in disorders/differences of sex development. Journal of Medical Genetics, 2020, 57, 581-589.	1.5	8
24	Psychosexual Outcome, Sexual Function, and Long-Term Satisfaction of Adolescent and Young Adult Men After Childhood Hypospadias Repair. Journal of Sexual Medicine, 2020, 17, 1665-1675.	0.3	28
25	Addressing gaps in care of people with conditions affecting sex development and maturation. Nature Reviews Endocrinology, 2019, 15, 615-622.	4.3	30
26	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.	1.8	27
27	45,X/46,XY Gonadal Dysgenesis, 46,XX/46,XY Chimerism (and Variants), and 46,XX Testicular and Ovotesticular DSD. , 2019, , 568-574.		0
28	Update on the genetics of differences of sex development (DSD). Best Practice and Research in Clinical Endocrinology and Metabolism, 2019, 33, 101271.	2.2	40
29	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.	1.8	0
30	Clinical Findings and Follow-Up of 46,XY and 45,X/46,XY Testicular Dysgenesis. Sexual Development, 2019, 13, 171-177.	1.1	16
31	Mindfulness, Worries, and Parenting in Parents of Children With Type 1 Diabetes. Journal of Pediatric Psychology, 2019, 44, 499-508.	1.1	13
32	The current landscape of European registries for rare endocrine conditions. European Journal of Endocrinology, 2019, 180, 89-98.	1.9	25
33	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.	1.9	21
34	Biallelic and monoallelic ESR2 variants associated with 46,XY disorders of sex development. Genetics in Medicine, 2018, 20, 717-727.	1.1	28
35	Histological Assessment of Gonads in DSD: Relevance for Clinical Management. Sexual Development, 2018, 12, 106-122.	1.1	35
36	Evaluation of DSD training schools organized by cost action BM1303 â€œDSDnetâ€• Orphanet Journal of Rare Diseases, 2018, 13, 227.	1.2	3

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37	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.	1.8	22
38	Management of Gonads in Adults with Androgen Insensitivity: An International Survey. Hormone Research in Paediatrics, 2018, 90, 236-246.	0.8	34
39	Caring for individuals with a difference of sex development (DSD): a Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 415-429.	4.3	264
40	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.	1.1	13
41	GENETICS IN ENDOCRINOLOGY: Approaches to molecular genetic diagnosis in the management of differences/disorders of sex development (DSD): position paper of EU COST Action BM 1303 â€˜DSDnetâ€™. European Journal of Endocrinology, 2018, 179, R197-R206.	1.9	105
42	Proandrogenic and Antiandrogenic Progestins in Transgender Youth: Differential Effects on Body Composition and Bone Metabolism. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2147-2156.	1.8	32
43	Birth Weight in Different Etiologies of Disorders of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1044-1050.	1.8	16
44	Consecutive Cyproterone Acetate and Estradiol Treatment in Late-Pubertal Transgender Female Adolescents. Journal of Sexual Medicine, 2017, 14, 747-757.	0.3	37
45	Clinical presentation and outcome of children with central diabetes insipidus associated with a selfâ€limited or transient pituitary stalk thickening, diagnosed as infundibuloneurohypophysitis. Clinical Endocrinology, 2017, 87, 171-176.	1.2	8
46	Conditional islet hypovascularisation does not preclude beta cell expansion during pregnancy in mice. Diabetologia, 2017, 60, 1051-1056.	2.9	9
47	<scp>SF1</scp> and spleen development: new heterozygous mutation, literature review and consequences for <scp>NR5A1</scp>â€mutated patient's management. Clinical Genetics, 2017, 92, 99-103.	1.0	15
48	Global Application of the Assessment of Communication Skills of Paediatric Endocrinology Fellows in the Management of Differences in Sex Development Using the ESPE E-Learning.Org Portal. Hormone Research in Paediatrics, 2017, 88, 127-139.	0.8	13
49	Update on the Pathophysiology and Risk Factors for the Development of Malignant Testicular Germ Cell Tumors in Complete Androgen Insensitivity Syndrome. Sexual Development, 2017, 11, 175-181.	1.1	55
50	Developing and evaluating rare disease educational materials co-created by expert clinicians and patients: the paradigm of congenital hypogonadotropic hypogonadism. Orphanet Journal of Rare Diseases, 2017, 12, 57.	1.2	26
51	Nonâ€coding variation in disorders of sex development. Clinical Genetics, 2017, 91, 163-172.	1.0	39
52	NR5A1 is a novel disease gene for 46,XX testicular and ovotesticular disorders of sex development. Genetics in Medicine, 2017, 19, 367-376.	1.1	87
53	The biology of germ cell tumors in disorders of sex development. Clinical Genetics, 2017, 91, 292-301.	1.0	42
54	Malignant testicular germ cell tumors in postpubertal individuals with androgen insensitivity: prevalence, pathology and relevance of single nucleotide polymorphism-based susceptibility profiling. Human Reproduction, 2017, 32, 2561-2573.	0.4	50

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55	Genetic Defects of Female Sexual Differentiation. , 2017, , 105-134.		0
56	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.	1.1	41
57	Cognitive, Emotional, and Psychosocial Functioning of Girls Treated with Pharmacological Puberty Blockage for Idiopathic Central Precocious Puberty. Frontiers in Psychology, 2016, 7, 1053.	1.1	58
58	Global Application of Assessment of Competencies of Paediatric Endocrinology Fellows in the Management of Differences of Sex Development (DSD) Using the ESPE e-learning.org Portal. Medical Science Educator, 2016, 26, 679-689.	0.7	6
59	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. Genome Biology, 2016, 17, 243.	3.8	241
60	Identification of an AR Mutation-Negative Class of Androgen Insensitivity by Determining Endogenous AR Activity. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 4468-4477.	1.8	64
61	Response to the Council of Europe Human Rights Commissioner's Issue Paper on Human Rights and Intersex People. European Urology, 2016, 70, 407-409.	0.9	35
62	The Long-Term Outcome of Boys With Partial Androgen Insensitivity Syndrome and a Mutation in the Androgen Receptor Gene. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 3959-3967.	1.8	81
63	Gonadal dysgenesis in disorders of sex development: Diagnosis and surgical management. Journal of Pediatric Urology, 2016, 12, 411-416.	0.6	51
64	Consecutive lynestrenol and cross-sex hormone treatment in biological female adolescents with gender dysphoria: a retrospective analysis. Biology of Sex Differences, 2016, 7, 14.	1.8	47
65	Global Disorders of Sex Development Update since 2006: Perceptions, Approach and Care. Hormone Research in Paediatrics, 2016, 85, 158-180.	0.8	852
66	Costs of pleasure and the benefits of pain: self-perceived genital sensation, anatomy and sexual dysfunction. Sexual Health, 2016, 13, 63.	0.4	1
67	The contribution of the androgen receptor (AR) in human spatial learning and memory: A study in women with complete androgen insensitivity syndrome (CAIS). Hormones and Behavior, 2016, 78, 121-126.	1.0	18
68	Evaluation of the hypothalamic-pituitary-adrenal axis and its relationship with central respiratory dysfunction in children with Prader-Willi syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 106.	1.2	21
69	Calcium and bone homeostasis in heterozygous carriers of CYP24A1 mutations: A cross-sectional study. Bone, 2015, 81, 89-96.	1.4	54
70	Gonadal Maldevelopment as Risk Factor for Germ Cell Cancer: Towards a Clinical Decision Model. European Urology, 2015, 67, 692-701.	0.9	92
71	Sexual quality of life after total phalloplasty in men with penile deficiency: an exploratory study. World Journal of Urology, 2015, 33, 137-143.	1.2	34
72	Desmopressin Lyophilisate for the Treatment of Central Diabetes Insipidus: First Experience in Very Young Infants. International Journal of Endocrinology and Metabolism, 2014, 12, e16120.	0.3	9

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73	Advances in Molecular Markers of Germ Cell Cancer in Patients with Disorders of Sex Development. Endocrine Development, 2014, 27, 172-184.	1.3	2
74	Managing the Risk of Germ Cell Tumourigenesis in Disorders of Sex Development Patients. Endocrine Development, 2014, 27, 185-196.	1.3	71
75	Extensive clinical, hormonal and genetic screening in a large consecutive series of 46,XY neonates and infants with atypical sexual development. Orphanet Journal of Rare Diseases, 2014, 9, 209.	1.2	44
76	Germ cell cancer risk in DSD patients. Annales D'Endocrinologie, 2014, 75, 67-71.	0.6	15
77	Complete androgen insensitivity syndrome: factors influencing gonadal histology including germ cell pathology. Modern Pathology, 2014, 27, 721-730.	2.9	52
78	Novel Associations in Disorders of Sex Development: Findings From the I-DSD Registry. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E348-E355.	1.8	85
79	An update on surgical and non-surgical treatments for vaginal hypoplasia. Human Reproduction Update, 2014, 20, 775-801.	5.2	145
80	Changes Over Time in Sex Assignment for Disorders of Sex Development. Pediatrics, 2014, 134, e710-e715.	1.0	98
81	Vaginal dilation treatment in women with vaginal hypoplasia: a prospective one-year follow-up study. American Journal of Obstetrics and Gynecology, 2014, 211, 228.e1-228.e12.	0.7	24
82	Variable Loss of Functional Activities of Androgen Receptor Mutants in Patients with Androgen Insensitivity Syndrome. Sexual Development, 2013, 7, 223-34.	1.1	8
83	Selfâ€Assessment of Genital Anatomy and Sexual Function within a Belgian, Dutchâ€Speaking Female Population: A Validation Study. Journal of Sexual Medicine, 2013, 10, 3006-3018.	0.3	13
84	Cardiovascular Pathology in Males and Females with 45,X/46,XY Mosaicism. PLoS ONE, 2013, 8, e54977.	1.1	28
85	Gonadal malignancy in 13 consecutive collected patients with disorders of sex development (DSD) from Semarang (Indonesia). Journal of Clinical Pathology, 2013, 66, 198-204.	1.0	12
86	Sexual Quality of Life after Hormonal and Surgical Treatment, Including Phalloplasty, in Men with Micropenis: A Review. Journal of Sexual Medicine, 2013, 10, 2890-2903.	0.3	38
87	Androgen Receptor Function Links Human Sexual Dimorphism to DNA Methylation. PLoS ONE, 2013, 8, e73288.	1.1	26
88	Do Surgical Interventions Influence Psychosexual and Cosmetic Outcomes in Women with Disorders of Sex Development?. Isrn Endocrinology, 2012, 2012, 1-8.	2.0	27
89	Pubertal androgenization and gonadal histology in two 46,XY adolescents with NR5A1 mutations and predominantly female phenotype at birth. European Journal of Endocrinology, 2012, 166, 341-349.	1.9	45
90	A multi-exon deletion within WWOX is associated with a 46,XY disorder of sex development. European Journal of Human Genetics, 2012, 20, 348-351.	1.4	48

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91	Requirements for a multicentric multidisciplinary registry on patients with disorders of sex development. Journal of Pediatric Urology, 2012, 8, 624-628.	0.6	13
92	Longâ€Term Psychosexual and Anatomical Outcome after Vaginal Dilation or Vaginoplasty: A Comparative Study. Journal of Sexual Medicine, 2012, 9, 1842-1851.	0.3	52
93	Gonadal Pathology and Tumor Risk in Relation to Clinical Characteristics in Patients with 45,X/46,XY Mosaicism. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1171-E1180.	1.8	131
94	Gonadal Development and Tumor Formation at the Crossroads of Male and Female Sex Determination. Sexual Development, 2011, 5, 167-180.	1.1	77
95	Male Gender Identity in Complete Androgen Insensitivity Syndrome. Archives of Sexual Behavior, 2011, 40, 635-638.	1.2	80
96	Partial Deletion of the <i>NR5A1 (SF1)</i> Gene Detected by Synthetic Probe MLPA in a Patient with XY Gonadal Disorder of Sex Development. Sexual Development, 2011, 5, 181-187.	1.1	29
97	Tumor risk and clinical follow-up in patients with disorders of sex development. Pediatric Endocrinology Reviews, 2011, 9 Suppl 1, 519-24.	1.2	5
98	Gonadal tumours and DSD. Best Practice and Research in Clinical Endocrinology and Metabolism, 2010, 24, 291-310.	2.2	90
99	536 VAGINAL AGENESIS AND PSYCHOSEXUAL FUNCTIONING: LONG TERM OUTCOME OF A VAGINAL SUBSTITUTION TREATMENT. Journal of Urology, 2010, 183, .	0.2	1
100	Tumor Risk in Disorders of Sex Development. Sexual Development, 2010, 4, 259-269.	1.1	155
101	Disorders of sex development: update on the genetic background, terminology and risk for the development of germ cell tumors. World Journal of Pediatrics, 2009, 5, 93-102.	0.8	66
102	Genetische basis, terminologie en het risico voor de ontwikkeling van kiemceltumoren bij stoornissen in de geslachtsontwikkeling. Tijdschrift Voor Kindergeneeskunde, 2008, 76, 92-104.	0.0	4
103	Stem cell factor as a novel diagnostic marker for early malignant germ cells. Journal of Pathology, 2008, 216, 43-54.	2.1	126
104	New insights into type II germ cell tumor pathogenesis based on studies of patients with various forms of disorders of sex development (DSD). Molecular and Cellular Endocrinology, 2008, 291, 1-10.	1.6	71
105	Tumor risk in disorders of sex development (DSD). Best Practice and Research in Clinical Endocrinology and Metabolism, 2007, 21, 480-495.	2.2	174
106	Impact of the Y-containing cell line on histological differentiation patterns in dysgenetic gonads. Clinical Endocrinology, 2007, 67, 184-192.	1.2	32
107	Germ Cell Tumors in the Intersex Gonad: Old Paths, New Directions, Moving Frontiers. Endocrine Reviews, 2006, 27, 468-484.	8.9	424
108	Maturation delay of germ cells in fetuses with trisomy 21 results in increased risk for the development of testicular germ cell tumors. Human Pathology, 2006, 37, 101-111.	1.1	51

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109	Adverse neonatal outcome after maternal biliopancreatic diversion operation: report of nine cases. European Journal of Pediatrics, 2006, 165, 199-202.	1.3	36
110	Gonadoblastoma Arising in Undifferentiated Gonadal Tissue within Dysgenetic Gonads. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 2404-2413.	1.8	190
111	Differentiation and development of human female germ cells during prenatal gonadogenesis: an immunohistochemical study. Human Reproduction, 2005, 20, 1466-1476.	0.4	124
112	Identification of germ cells at risk for neoplastic transformation in gonadoblastoma. Human Pathology, 2005, 36, 512-521.	1.1	155
113	Morphological and Immunohistochemical Differences between Gonadal Maturation Delay and Early Germ Cell Neoplasia in Patients with Undervirilization Syndromes. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 5295-5303.	1.8	184
114	A nonmosaic 45,X karyotype in a mother with Turner's syndrome and in her daughter. Fertility and Sterility, 2004, 82, 923-925.	0.5	33
115	FOXL2 and BPES: Mutational Hotspots, Phenotypic Variability, and Revision of the Genotype-Phenotype Correlation. American Journal of Human Genetics, 2003, 72, 478-487.	2.6	219
116	Long-term effects of specific immunotherapy, administered during childhood, in asthmatic patients allergic to either house-dust mite or to both house-dust mite and grass pollen. Allergy: European Journal of Allergy and Clinical Immunology, 2000, 55, 69-73.	2.7	65