

# Martine Cools

## List of Publications by Year in descending order

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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	Global Disorders of Sex Development Update since 2006: Perceptions, Approach and Care. <i>Hormone Research in Paediatrics</i> , 2016, 85, 158-180.	0.8	852
2	Germ Cell Tumors in the Intersex Gonad: Old Paths, New Directions, Moving Frontiers. <i>Endocrine Reviews</i> , 2006, 27, 468-484.	8.9	424
3	Caring for individuals with a difference of sex development (DSD): a Consensus Statement. <i>Nature Reviews Endocrinology</i> , 2018, 14, 415-429.	4.3	264
4	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. <i>Genome Biology</i> , 2016, 17, 243.	3.8	241
5	FOXL2 and BPES: Mutational Hotspots, Phenotypic Variability, and Revision of the Genotype-Phenotype Correlation. <i>American Journal of Human Genetics</i> , 2003, 72, 478-487.	2.6	219
6	Gonadoblastoma Arising in Undifferentiated Gonadal Tissue within Dysgenetic Gonads. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006, 91, 2404-2413.	1.8	190
7	Morphological and Immunohistochemical Differences between Gonadal Maturation Delay and Early Germ Cell Neoplasia in Patients with Undervirilization Syndromes. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005, 90, 5295-5303.	1.8	184
8	Tumor risk in disorders of sex development (DSD). <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2007, 21, 480-495.	2.2	174
9	Identification of germ cells at risk for neoplastic transformation in gonadoblastoma. <i>Human Pathology</i> , 2005, 36, 512-521.	1.1	155
10	Tumor Risk in Disorders of Sex Development. <i>Sexual Development</i> , 2010, 4, 259-269.	1.1	155
11	An update on surgical and non-surgical treatments for vaginal hypoplasia. <i>Human Reproduction Update</i> , 2014, 20, 775-801.	5.2	145
12	Gonadal Pathology and Tumor Risk in Relation to Clinical Characteristics in Patients with 45,X/46,XY Mosaicism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011, 96, E1171-E1180.	1.8	131
13	Stem cell factor as a novel diagnostic marker for early malignant germ cells. <i>Journal of Pathology</i> , 2008, 216, 43-54.	2.1	126
14	Differentiation and development of human female germ cells during prenatal gonadogenesis: an immunohistochemical study. <i>Human Reproduction</i> , 2005, 20, 1466-1476.	0.4	124
15	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 "DSDnet". <i>European Journal of Endocrinology</i> , 2018, 179, R197-R206.	1.9	105
16	Changes Over Time in Sex Assignment for Disorders of Sex Development. <i>Pediatrics</i> , 2014, 134, e710-e715.	1.0	98
17	Gonadal Maldevelopment as Risk Factor for Germ Cell Cancer: Towards a Clinical Decision Model. <i>European Urology</i> , 2015, 67, 692-701.	0.9	92
18	Gonadal tumours and DSD. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2010, 24, 291-310.	2.2	90

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19	NR5A1 is a novel disease gene for 46,XX testicular and ovotesticular disorders of sex development. <i>Genetics in Medicine</i> , 2017, 19, 367-376.	1.1	87
20	Novel Associations in Disorders of Sex Development: Findings From the I-DSD Registry. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, E348-E355.	1.8	85
21	The Long-Term Outcome of Boys With Partial Androgen Insensitivity Syndrome and a Mutation in the Androgen Receptor Gene. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 3959-3967.	1.8	81
22	Male Gender Identity in Complete Androgen Insensitivity Syndrome. <i>Archives of Sexual Behavior</i> , 2011, 40, 635-638.	1.2	80
23	Gonadal Development and Tumor Formation at the Crossroads of Male and Female Sex Determination. <i>Sexual Development</i> , 2011, 5, 167-180.	1.1	77
24	New insights into type II germ cell tumor pathogenesis based on studies of patients with various forms of disorders of sex development (DSD). <i>Molecular and Cellular Endocrinology</i> , 2008, 291, 1-10.	1.6	71
25	Managing the Risk of Germ Cell Tumourigenesis in Disorders of Sex Development Patients. <i>Endocrine Development</i> , 2014, 27, 185-196.	1.3	71
26	Disorders of sex development: update on the genetic background, terminology and risk for the development of germ cell tumors. <i>World Journal of Pediatrics</i> , 2009, 5, 93-102.	0.8	66
27	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. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 2000, 55, 69-73.	2.7	65
28	Identification of an <i>AR</i> Mutation-Negative Class of Androgen Insensitivity by Determining Endogenous AR Activity. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 4468-4477.	1.8	64
29	Cognitive, Emotional, and Psychosocial Functioning of Girls Treated with Pharmacological Puberty Blockage for Idiopathic Central Precocious Puberty. <i>Frontiers in Psychology</i> , 2016, 7, 1053.	1.1	58
30	Update on the Pathophysiology and Risk Factors for the Development of Malignant Testicular Germ Cell Tumors in Complete Androgen Insensitivity Syndrome. <i>Sexual Development</i> , 2017, 11, 175-181.	1.1	55
31	Calcium and bone homeostasis in heterozygous carriers of CYP24A1 mutations: A cross-sectional study. <i>Bone</i> , 2015, 81, 89-96.	1.4	54
32	Long-term Psychosexual and Anatomical Outcome after Vaginal Dilatation or Vaginoplasty: A Comparative Study. <i>Journal of Sexual Medicine</i> , 2012, 9, 1842-1851.	0.3	52
33	Complete androgen insensitivity syndrome: factors influencing gonadal histology including germ cell pathology. <i>Modern Pathology</i> , 2014, 27, 721-730.	2.9	52
34	Maturation delay of germ cells in fetuses with trisomy 21 results in increased risk for the development of testicular germ cell tumors. <i>Human Pathology</i> , 2006, 37, 101-111.	1.1	51
35	Gonadal dysgenesis in disorders of sex development: Diagnosis and surgical management. <i>Journal of Pediatric Urology</i> , 2016, 12, 411-416.	0.6	51
36	The External Genitalia Score (EGS): A European Multicenter Validation Study. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e222-e230.	1.8	51

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37	Malignant testicular germ cell tumors in postpubertal individuals with androgen insensitivity: prevalence, pathology and relevance of single nucleotide polymorphism-based susceptibility profiling. <i>Human Reproduction</i> , 2017, 32, 2561-2573.	0.4	50
38	A multi-exon deletion within WWOX is associated with a 46,XY disorder of sex development. <i>European Journal of Human Genetics</i> , 2012, 20, 348-351.	1.4	48
39	Consecutive lynestrenol and cross-sex hormone treatment in biological female adolescents with gender dysphoria: a retrospective analysis. <i>Biology of Sex Differences</i> , 2016, 7, 14.	1.8	47
40	Pubertal androgenization and gonadal histology in two 46,XY adolescents with NR5A1 mutations and predominantly female phenotype at birth. <i>European Journal of Endocrinology</i> , 2012, 166, 341-349.	1.9	45
41	Extensive clinical, hormonal and genetic screening in a large consecutive series of 46,XY neonates and infants with atypical sexual development. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 209.	1.2	44
42	The biology of germ cell tumors in disorders of sex development. <i>Clinical Genetics</i> , 2017, 91, 292-301.	1.0	42
43	A Recurrent Germline Mutation in the 5'UTR of the Androgen Receptor Causes Complete Androgen Insensitivity by Activating Aberrant uORF Translation. <i>PLoS ONE</i> , 2016, 11, e0154158.	1.1	41
44	Update on the genetics of differences of sex development (DSD). <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2019, 33, 101271.	2.2	40
45	Non-coding variation in disorders of sex development. <i>Clinical Genetics</i> , 2017, 91, 163-172.	1.0	39
46	Sexual Quality of Life after Hormonal and Surgical Treatment, Including Phalloplasty, in Men with Micropenis: A Review. <i>Journal of Sexual Medicine</i> , 2013, 10, 2890-2903.	0.3	38
47	Consecutive Cyproterone Acetate and Estradiol Treatment in Late-Pubertal Transgender Female Adolescents. <i>Journal of Sexual Medicine</i> , 2017, 14, 747-757.	0.3	37
48	Adverse neonatal outcome after maternal biliopancreatic diversion operation: report of nine cases. <i>European Journal of Pediatrics</i> , 2006, 165, 199-202.	1.3	36
49	Response to the Council of Europe Human Rights Commissioner's Issue Paper on Human Rights and Intersex People. <i>European Urology</i> , 2016, 70, 407-409.	0.9	35
50	Histological Assessment of Gonads in DSD: Relevance for Clinical Management. <i>Sexual Development</i> , 2018, 12, 106-122.	1.1	35
51	Sexual quality of life after total phalloplasty in men with penile deficiency: an exploratory study. <i>World Journal of Urology</i> , 2015, 33, 137-143.	1.2	34
52	Management of Gonads in Adults with Androgen Insensitivity: An International Survey. <i>Hormone Research in Paediatrics</i> , 2018, 90, 236-246.	0.8	34
53	A nonmosaic 45,X karyotype in a mother with Turner's syndrome and in her daughter. <i>Fertility and Sterility</i> , 2004, 82, 923-925.	0.5	33
54	Impact of the Y-containing cell line on histological differentiation patterns in dysgenetic gonads. <i>Clinical Endocrinology</i> , 2007, 67, 184-192.	1.2	32

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55	Proandrogenic and Antiandrogenic Progestins in Transgender Youth: Differential Effects on Body Composition and Bone Metabolism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 2147-2156.	1.8	32
56	Addressing gaps in care of people with conditions affecting sex development and maturation. <i>Nature Reviews Endocrinology</i> , 2019, 15, 615-622.	4.3	30
57	Partial Deletion of the <i>NR5A1 (SF1)</i> Gene Detected by Synthetic Probe MLPA in a Patient with XY Gonadal Disorder of Sex Development. <i>Sexual Development</i> , 2011, 5, 181-187.	1.1	29
58	Cardiovascular Pathology in Males and Females with 45,X/46,XY Mosaicism. <i>PLoS ONE</i> , 2013, 8, e54977.	1.1	28
59	Biallelic and monoallelic <i>ESR2</i> variants associated with 46,XY disorders of sex development. <i>Genetics in Medicine</i> , 2018, 20, 717-727.	1.1	28
60	Fertility and sexuality issues in congenital lifelong urology patients: male aspects. <i>World Journal of Urology</i> , 2021, 39, 1013-1019.	1.2	28
61	Psychosexual Outcome, Sexual Function, and Long-Term Satisfaction of Adolescent and Young Adult Men After Childhood Hypospadias Repair. <i>Journal of Sexual Medicine</i> , 2020, 17, 1665-1675.	0.3	28
62	Do Surgical Interventions Influence Psychosexual and Cosmetic Outcomes in Women with Disorders of Sex Development?. <i>Isrn Endocrinology</i> , 2012, 2012, 1-8.	2.0	27
63	Clinical but Not Histological Outcomes in Males With 45,X/46,XY Mosaicism Vary Depending on Reason for Diagnosis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 4366-4381.	1.8	27
64	Developing and evaluating rare disease educational materials co-created by expert clinicians and patients: the paradigm of congenital hypogonadotropic hypogonadism. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 57.	1.2	26
65	Androgen Receptor Function Links Human Sexual Dimorphism to DNA Methylation. <i>PLoS ONE</i> , 2013, 8, e73288.	1.1	26
66	The current landscape of European registries for rare endocrine conditions. <i>European Journal of Endocrinology</i> , 2019, 180, 89-98.	1.9	25
67	Vaginal dilation treatment in women with vaginal hypoplasia: a prospective one-year follow-up study. <i>American Journal of Obstetrics and Gynecology</i> , 2014, 211, 228.e1-228.e12.	0.7	24
68	Epigenetic Repression of Androgen Receptor Transcription in Mutation-Negative Androgen Insensitivity Syndrome (AIS Type II). <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 4617-4627.	1.8	22
69	Evaluation of the hypothalamic-pituitary-adrenal axis and its relationship with central respiratory dysfunction in children with Prader-Willi syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 106.	1.2	21
70	International practice of corticosteroid replacement therapy in congenital adrenal hyperplasia: data from the I-CAH registry. <i>European Journal of Endocrinology</i> , 2021, 184, 553-563.	1.9	21
71	Standardised data collection for clinical follow-up and assessment of outcomes in differences of sex development (DSD): recommendations from the COST action DSDnet. <i>European Journal of Endocrinology</i> , 2019, 181, 545-564.	1.9	21
72	Families with pediatric type 1 diabetes: A comparison with the general population on child well-being, parental distress, and parenting behavior. <i>Pediatric Diabetes</i> , 2020, 21, 395-408.	1.2	20

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73	Real-World Estimates of Adrenal Insufficiency-Related Adverse Events in Children With Congenital Adrenal Hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e192-e203.	1.8	20
74	ENDO-ERN expert opinion on the differential diagnosis of pubertal delay. <i>Endocrine</i> , 2021, 71, 681-688.	1.1	19
75	The contribution of the androgen receptor (AR) in human spatial learning and memory: A study in women with complete androgen insensitivity syndrome (CAIS). <i>Hormones and Behavior</i> , 2016, 78, 121-126.	1.0	18
76	Birth Weight in Different Etiologies of Disorders of Sex Development. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 1044-1050.	1.8	16
77	Clinical Findings and Follow-Up of 46,XY and 45,X/46,XY Testicular Dysgenesis. <i>Sexual Development</i> , 2019, 13, 171-177.	1.1	16
78	Germ cell cancer risk in DSD patients. <i>Annales D'Endocrinologie</i> , 2014, 75, 67-71.	0.6	15
79	<sc>SF1</sc> and spleen development: new heterozygous mutation, literature review and consequences for <i><sc>NR5A1</sc></i> mutated patient's management. <i>Clinical Genetics</i> , 2017, 92, 99-103.	1.0	15
80	Adolescent and Young Adult Urogenital Outcome following Childhood Hypospadias Repair: Perfection Revisited. <i>Journal of Urology</i> , 2021, 206, 734-744.	0.2	14
81	Requirements for a multicentric multidisciplinary registry on patients with disorders of sex development. <i>Journal of Pediatric Urology</i> , 2012, 8, 624-628.	0.6	13
82	Self-Assessment of Genital Anatomy and Sexual Function within a Belgian, Dutch-Speaking Female Population: A Validation Study. <i>Journal of Sexual Medicine</i> , 2013, 10, 3006-3018.	0.3	13
83	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. <i>Hormone Research in Paediatrics</i> , 2017, 88, 127-139.	0.8	13
84	Involving Individuals with Disorders of Sex Development and Their Parents in Exploring New Models of Shared Learning: Proceedings from a DSDnet COST Action Workshop. <i>Sexual Development</i> , 2018, 12, 225-231.	1.1	13
85	Mindfulness, Worries, and Parenting in Parents of Children With Type 1 Diabetes. <i>Journal of Pediatric Psychology</i> , 2019, 44, 499-508.	1.1	13
86	The EuRRECa Project as a Model for Data Access and Governance Policies for Rare Disease Registries That Collect Clinical Outcomes. <i>International Journal of Environmental Research and Public Health</i> , 2020, 17, 8743.	1.2	13
87	Gonadal malignancy in 13 consecutive collected patients with disorders of sex development (DSD) from Semarang (Indonesia). <i>Journal of Clinical Pathology</i> , 2013, 66, 198-204.	1.0	12
88	Desmopressin Lyophilisate for the Treatment of Central Diabetes Insipidus: First Experience in Very Young Infants. <i>International Journal of Endocrinology and Metabolism</i> , 2014, 12, e16120.	0.3	9
89	Conditional islet hypovascularisation does not preclude beta cell expansion during pregnancy in mice. <i>Diabetologia</i> , 2017, 60, 1051-1056.	2.9	9
90	Androgen receptor expression in preputial dartos tissue correlates with physiological androgen exposure in congenital malformations of the penis and in controls. <i>Journal of Pediatric Urology</i> , 2020, 16, 43.e1-43.e8.	0.6	9

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91	Assessing the health-related management of people with differences of sex development. <i>Endocrine</i> , 2021, 71, 675-680.	1.1	9
92	Variable Loss of Functional Activities of Androgen Receptor Mutants in Patients with Androgen Insensitivity Syndrome. <i>Sexual Development</i> , 2013, 7, 223-34.	1.1	8
93	Clinical presentation and outcome of children with central diabetes insipidus associated with a self-limited or transient pituitary stalk thickening, diagnosed as infundibuloneurohypophysitis. <i>Clinical Endocrinology</i> , 2017, 87, 171-176.	1.2	8
94	Under-reported aspects of diagnosis and treatment addressed in the Dutch-Flemish guideline for comprehensive diagnostics in disorders/differences of sex development. <i>Journal of Medical Genetics</i> , 2020, 57, 581-589.	1.5	8
95	Novel model to study the physiological effects of temporary or prolonged sex steroid deficiency in male mice. <i>American Journal of Physiology - Endocrinology and Metabolism</i> , 2021, 320, E415-E424.	1.8	7
96	Ovotesticular Difference of Sex Development: Genetic Background, Histological Features, and Clinical Management. <i>Hormone Research in Paediatrics</i> , 2023, 96, 180-189.	0.8	7
97	Approach to the Virilizing Girl at Puberty. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, 1530-1539.	1.8	7
98	Prenatal dexamethasone treatment for classic 21-hydroxylase deficiency in Europe. <i>European Journal of Endocrinology</i> , 2022, 186, K17-K24.	1.9	7
99	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. <i>Medical Science Educator</i> , 2016, 26, 679-689.	0.7	6
100	Multidisciplinary Approach to the Child with Sex Chromosomal Mosaicism Including a Y-Containing Cell Line. <i>International Journal of Environmental Research and Public Health</i> , 2021, 18, 917.	1.2	6
101	Growth, puberty and testicular function in boys born small for gestational age with a nonspecific disorder of sex development. <i>Clinical Endocrinology</i> , 2022, 96, 165-174.	1.2	6
102	Rare and special robotic surgery indications in the pediatric population: ectopic organs and differences of sexual development. <i>World Journal of Urology</i> , 2020, 38, 1865-1868.	1.2	5
103	Tumor risk and clinical follow-up in patients with disorders of sex development. <i>Pediatric Endocrinology Reviews</i> , 2011, 9 Suppl 1, 519-24.	1.2	5
104	Genetische basis, terminologie en het risico voor de ontwikkeling van kiemceltumoren bij stoornissen in de geslachtsontwikkeling. <i>Tijdschrift Voor Kindergeneeskunde</i> , 2008, 76, 92-104.	0.0	4
105	Parental stress, anxiety and trait mindfulness: associations with parent-child mealtime interactions in children with type 1 diabetes. <i>Journal of Behavioral Medicine</i> , 2020, 43, 448-459.	1.1	4
106	Testosterone Therapy and Its Monitoring in Adolescent Boys with Hypogonadism: Results of an International Survey from the I-DSD Registry. <i>Sexual Development</i> , 2021, 15, 236-243.	1.1	4
107	Patients with rare endocrine conditions have corresponding views on unmet needs in clinical research. <i>Endocrine</i> , 2021, 71, 561-568.	1.1	4
108	Evaluation of DSD training schools organized by cost action BM1303 "DSDnet". <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 227.	1.2	3

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109	Advances in Molecular Markers of Germ Cell Cancer in Patients with Disorders of Sex Development. <i>Endocrine Development</i> , 2014, 27, 172-184.	1.3	2
110	536 VAGINAL AGENESIS AND PSYCHOSEXUAL FUNCTIONING: LONG TERM OUTCOME OF A VAGINAL SUBSTITUTION TREATMENT. <i>Journal of Urology</i> , 2010, 183, .	0.2	1
111	Costs of pleasure and the benefits of pain: self-perceived genital sensation, anatomy and sexual dysfunction. <i>Sexual Health</i> , 2016, 13, 63.	0.4	1
112	Genetic Defects of Female Sexual Differentiation. , 2017, , 105-134.		0
113	45,X/46,XY Gonadal Dysgenesis, 46,XX/46,XY Chimerism (and Variants), and 46,XX Testicular and Ovotesticular DSD. , 2019, , 568-574.		0
114	Response to Letter to the Editor: "Clinical but Not Histological Outcomes in Males With 45,X/46,XY Mosaicism Vary Depending on Reason for Diagnosis" <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 5812-5813.	1.8	0
115	SUN-070 European Registries for Rare Endocrine Conditions (EuRRECa): Results from the Platform for E-reporting of Rare Endocrine Conditions (e-REC). <i>Journal of the Endocrine Society</i> , 2020, 4, .	0.1	0
116	Reply by Authors. <i>Journal of Urology</i> , 2021, 206, 744-744.	0.2	0