Andrew H Sinclair

List of Publications by Year
in descending order

199 papers	15,777 citations	25034 57 h-index	18130 120 g-index
212 all docs	212 docs citations	212 times ranked	9928 citing authors

#	Article	IF	Citations
1	A gene from the human sex-determining region encodes a protein with homology to a conserved DNA-binding motif. Nature, 1990, 346, 240-244.	27.8	3,014
2	Genetic evidence equating SRY and the testis-determining factor. Nature, 1990, 348, 448-450.	27.8	907
3	The avian Z-linked gene DMRT1 is required for male sex determination in the chicken. Nature, 2009, 461, 267-271.	27.8	728
4	A male-specific role for SOX9 in vertebrate sex determination. Development (Cambridge), 1996, 122, 2813-2822.	2.5	623
5	Conservation of a sex-determining gene. Nature, 1999, 402, 601-602.	27.8	359
6	Genetic evidence that ZFY is not the testis-determining factor. Nature, 1989, 342, 937-939.	27.8	305
7	Sex determination: insights from the chicken. BioEssays, 2004, 26, 120-132.	2.5	303
8	Dynamic Regulation of Mitotic Arrest in Fetal Male Germ Cells. Stem Cells, 2008, 26, 339-347.	3.2	247
9	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. Genome Biology, 2016, 17, 243.	8.8	241
10	Identification of SOX3 as an XX male sex reversal gene in mice and humans. Journal of Clinical Investigation, 2011, 121, 328-341.	8.2	234
11	Evolution of sex determination and the Y chromosome: SRY-related sequences in marsupials. Nature, 1992, 359, 531-533.	27.8	224
12	The business impact of an integrated continuous biomanufacturing platform for recombinant protein production. Journal of Biotechnology, 2015, 213, 3-12.	3.8	207
13	Sequences homologous to ZFY, a candidate human sex-determining gene, are autosomal in marsupials. Nature, 1988, 336, 780-783.	27.8	182
14	Premature Ovarian Insufficiency: New Perspectives on Genetic Cause and Phenotypic Spectrum. Endocrine Reviews, 2016, 37, 609-635.	20.1	170
15	Sites ofEstrogen ReceptorandAromataseExpression in the Chicken Embryo. General and Comparative Endocrinology, 1997, 108, 182-190.	1.8	169
16	Mutations inSRY andSOX9: Testis-determining genes. Human Mutation, 1997, 9, 388-395.	2.5	165
17	Endothelial cell migration directs testis cord formation. Developmental Biology, 2009, 326, 112-120.	2.0	164
18	The human SRY transcript. Human Molecular Genetics, 1993, 2, 2007-2012.	2.9	162

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19	Genetic regulation of mammalian gonad development. Nature Reviews Endocrinology, 2014, 10, 673-683.	9.6	162
20	Vertebrate sex determination: many means to an end. Reproduction, 2002, 124, 447-457.	2.6	161
21	DMRT1 Is Upregulated in the Gonads During Female-to-Male Sex Reversal in ZW Chicken Embryos1. Biology of Reproduction, 2003, 68, 560-570.	2.7	161
22	Mutations in MAP3K1 Cause 46,XY Disorders of Sex Development and Implicate a Common Signal Transduction Pathway in Human Testis Determination. American Journal of Human Genetics, 2010, 87, 898-904.	6.2	155
23	Gonadal sex differentiation in chicken embryos: Expression of estrogen receptor and aromatase genes. Journal of Steroid Biochemistry and Molecular Biology, 1997, 60, 295-302.	2.5	154
24	The Genetic and Environmental Factors Underlying Hypospadias. Sexual Development, 2015, 9, 239-259.	2.0	142
25	A Long-term Outcome Study of Intersex Conditions. Journal of Pediatric Endocrinology and Metabolism, 2005, 18, 555-67.	0.9	139
26	Mammalian sex determinationâ€"insights from humans and mice. Chromosome Research, 2012, 20, 215-238.	2.2	139
27	Second report on chicken genes and chromosomes 2005. Cytogenetic and Genome Research, 2005, 109, 415-479.	1.1	136
28	Gene expression during gonadogenesis in the chicken embryo. Gene, 1999, 234, 395-402.	2.2	134
29	Temperature-dependent sex determination: Upregulation ofSOX9 expression after commitment to male development. Developmental Dynamics, 1999, 214, 171-177.	1.8	132
30	Temperature-dependent sex determination in the american alligator:AMH precedesSOX9 expression. Developmental Dynamics, 1999, 216, 411-419.	1.8	128
31	Onset of meiosis in the chicken embryo; evidence of a role for retinoic acid. BMC Developmental Biology, 2008, 8, 85.	2.1	125
32	Rapid DNA extraction and PCR-sexing of mouse embryos. Molecular Reproduction and Development, 2001, 60, 225-226.	2.0	122
33	Aromatase inhibition reduces expression of FOXL2 in the embryonic chicken ovary. Developmental Dynamics, 2005, 233, 1052-1055.	1.8	120
34	Cloning and expression of R-Spondin1in different vertebrates suggests a conserved role in ovarian development. BMC Developmental Biology, 2008, 8, 72.	2.1	120
35	Human sex reversal is caused by duplication or deletion of core enhancers upstream of SOX9. Nature Communications, 2018, 9, 5319.	12.8	116
36	Copy Number Variation in Patients with Disorders of Sex Development Due to 46,XY Gonadal Dysgenesis. PLoS ONE, 2011, 6, e17793.	2.5	116

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37	ASW : a gene with conserved avian W-linkage and female specific expression in chick embryonic gonad. Development Genes and Evolution, 2000, 210, 243-249.	0.9	112
38	Dppa2 and Dppa4 Are Closely Linked SAP Motif Genes Restricted to Pluripotent Cells and the Germ Line. Stem Cells, 2007, 25, 19-28.	3.2	109
39	Temperature-dependent sex determination in the American alligator: expression of SF1, WT1 and DAX1 during gonadogenesis. Gene, 2000, 241, 223-232.	2.2	100
40	RNA sequencing reveals sexually dimorphic gene expression before gonadal differentiation in chicken and allows comprehensive annotation of the W-chromosome. Genome Biology, 2013, 14, R26.	9.6	98
41	Sexually Dimorphic MicroRNA Expression During Chicken Embryonic Gonadal Development1. Biology of Reproduction, 2009, 81, 165-176.	2.7	92
42	Comparison of human ZFY and ZFX transcripts.. Proceedings of the National Academy of Sciences of the United States of America, 1990, 87, 1681-1685.	7.1	87
43	Conserved regulatory modules in the Sox9 testis-specific enhancer predict roles for SOX, TCF/LEF, Forkhead, DMRT, and GATA proteins in vertebrate sex determination. International Journal of Biochemistry and Cell Biology, 2010, 42, 472-477.	2.8	84
44	The Genetics of Disorders of Sex Development in Humans. Sexual Development, 2014, 8, 262-272.	2.0	83
45	A novel germ line mutation in SOX9 causes familial campomelic dysplasia and sex reversal. Human Molecular Genetics, 1996, 5, 1625-1630.	2.9	82
46	Threeâ€dimensional visualization of testis cord morphogenesis, a novel tubulogenic mechanism in development. Developmental Dynamics, 2009, 238, 1033-1041.	1.8	82
47	Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. American Journal of Human Genetics, 2019, 105, 7-14.	6.2	75
48	Signaling through the TGF Beta-Activin Receptors ALK4/5/7 Regulates Testis Formation and Male Germ Cell Development. PLoS ONE, 2013, 8, e54606.	2.5	75
49	Overexpression of Aromatase Alone is Sufficient for Ovarian Development in Genetically Male Chicken Embryos. PLoS ONE, 2013, 8, e68362.	2.5	73
50	Preparing for genomic medicine: a real world demonstration of health system change. Npj Genomic Medicine, 2017, 2, 16.	3.8	73
51	SRY protein enhances transcription of Fos-related antigen 1 promoter constructs.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 4372-4376.	7.1	68
52	Male fetal germ cell differentiation involves complex repression of the regulatory network controlling pluripotency. FASEB Journal, 2010, 24, 3026-3035.	0.5	68
53	The cell biology and molecular genetics of MÃ¼llerian duct development. Wiley Interdisciplinary Reviews: Developmental Biology, 2018, 7, e310.	5.9	67
54	Avian sex determination: what, when and where?. Cytogenetic and Genome Research, 2007, 117, 165-173.	1.1	65

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55	Expression of Chicken Steroidogenic Factor-1 during Gonadal Sex Differentiation. General and Comparative Endocrinology, 1999, 113, 187-196.	1.8	63
56	The long non-coding RNA, MHM, plays a role in chicken embryonic development, including gonadogenesis. Developmental Biology, 2012, 366, 317-326.	2.0	63
57	Wnt Signaling in Ovarian Development Inhibits Sf1 Activation of Sox9 via the Tesco Enhancer. Endocrinology, 2012, 153, 901-912.	2.8	62
58	Manipulation of Estrogen Synthesis Alters MIR202* Expression in Embryonic Chicken Gonads1. Biology of Reproduction, 2011, 85, 22-30.	2.7	61
59	Defective survival of proliferating Sertoli cells and androgen receptor function in a mouse model of the ATR-X syndrome. Human Molecular Genetics, 2011, 20, 2213-2224.	2.9	59
60	An environmental life cycle assessment comparison of single-use and conventional process technology for the production of monoclonal antibodies. Journal of Cleaner Production, 2013, 41, 150-162.	9.3	58
61	Cloning and expression of a DAX1 homologue in the chicken embryo. Journal of Molecular Endocrinology, 2000, 24, 23-32.	2.5	57
62	Male-specific cell migration into the developing gonad is a conserved process involving PDGF signalling. Developmental Biology, 2005, 284, 337-350.	2.0	57
63	Regulation of the female mouse germ cell cycle during entry into meiosis. Cell Cycle, 2010, 9, 408-418.	2.6	57
64	Sex determination in the chicken embryo. The Journal of Experimental Zoology, 2001, 290, 691-699.	1.4	56
65	Identification of candidate gonadal sex differentiation genes in the chicken embryo using RNA-seq. BMC Genomics, 2015, 16, 704.	2.8	54
66	Isolation of rare transcripts by representational difference analysis. Nucleic Acids Research, 1997, 25, 2681-2682.	14.5	53
67	Genetic evidence against a role for W-linked histidine triad nucleotide binding protein (HINTW) in avian sex determination. International Journal of Developmental Biology, 2009, 53, 59-67.	0.6	53
68	Fertile females of the mole Talpa occidentalis are phenotypic intersexes with ovotestes. Development (Cambridge), 1993, 118, 1303-1311.	2.5	53
69	Purification and Transcriptomic Analysis of Mouse Fetal Leydig Cells Reveals Candidate Genes for Specification of Gonadal Steroidogenic Cells1. Biology of Reproduction, 2015, 92, 145.	2.7	51
70	Application of a Decision-Support Tool to Assess Pooling Strategies in Perfusion Culture Processes under Uncertainty. Biotechnology Progress, 2008, 21, 1231-1242.	2.6	50
71	Genes on the short arm of the human X chromosome are not shared with the marsupial X. Genomics, 1991, 11, 339-345.	2.9	48
72	Evaluation of candidate markers for the peritubular myoid cell lineage in the developing mouse testis. Reproduction, 2005, 130, 509-516.	2.6	48

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73	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.	2.8	48
74	The Molecular Genetics of Ovarian Differentiation in the Avian Model. Sexual Development, 2013, 7, 80-94.	2.0	48
75	Gonadal defects in Cited2 -mutant mice indicate a role for SF1 in both testis and ovary differentiation. International Journal of Developmental Biology, 2010, 54, 683-689.	0.6	46
76	Retinoic Acid Antagonizes Testis Development in Mice. Cell Reports, 2018, 24, 1330-1341.	6.4	46
77	Subtractive hybridisation screen identifies sexually dimorphic gene expression in the embryonic mouse gonad. Genesis, 2003, 37, 84-90.	1.6	44
78	The Cerebellin 4 Precursor Gene Is a Direct Target of SRY and SOX9 in Mice1. Biology of Reproduction, 2009, 80, 1178-1188.	2.7	44
79	Normalizing Gene Expression Levels in Mouse Fetal Germ Cells1. Biology of Reproduction, 2009, 81, 362-370.	2.7	44
80	Transgenic Chickens Overexpressing Aromatase Have High Estrogen Levels but Maintain a Predominantly Male Phenotype. Endocrinology, 2016, 157, 83-90.	2.8	44
81	Maleâ€specific expression of Aldh1a1 in mouse and chicken fetal testes: Implications for retinoid balance in gonad development. Developmental Dynamics, 2009, 238, 2073-2080.	1.8	43
82	The potential role of microRNAs in regulating gonadal sex differentiation in the chicken embryo. Chromosome Research, 2012, 20, 201-213.	2.2	43
83	WNT/Î²-catenin and p27/FOXL2 differentially regulate supporting cell proliferation in the developing ovary. Developmental Biology, 2016, 412, 250-260.	2.0	43
84	SOX8 expression during chick embryogenesis. Mechanisms of Development, 2000, 94, 257-260.	1.7	42
85	New insights into the genetic basis of premature ovarian insufficiency: Novel causative variants and candidate genes revealed by genomic sequencing. Maturitas, 2020, 141, 9-19.	2.4	41
86	FET-1: a novel W-linked, female specific gene up-regulated in the embryonic chicken ovary. Mechanisms of Development, 2002, 119, S87-S90.	1.7	39
87	Occasional Article. Journal of Paediatrics and Child Health, 2003, 39, 406-413.	0.8	38
88	Functional characterization of novelÂNR5A1 Âvariants reveals multiple complex roles in disorders of sex development. Human Mutation, 2018, 39, 124-139.	2.5	38
89	Copy number variation associated with meiotic arrest in idiopathic male infertility. Fertility and Sterility, 2015, 103, 214-219.	1.0	37
90	The Role of Copy Number Variants in Disorders of Sex Development. Sexual Development, 2018, 12, 19-29.	2.0	37

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91	Autosomal assignment of OTC in marsupials and monotremes: implications for the evolution of sex chromosomes. Genetical Research, 1987, 50, 131-136.	0.9	36
92	Heterogeneity of Human Neutrophil CD177 Expression Results from CD177P1 Pseudogene Conversion. PLoS Genetics, 2016, 12, e1006067.	3.5	36
93	Type II and Type IX Collagen Transcript Isoforms Are Expressed During Mouse Testis Development. Biology of Reproduction, 2003, 68, 1742-1747.	2.7	35
94	Restricted expression of DMRT3 in chicken and mouse embryos. Mechanisms of Development, 2002, 119, S73-S76.	1.7	34
95	Sex, genes, and heat: Triggers of diversity. The Journal of Experimental Zoology, 2001, 290, 624-631.	1.4	31
96	Robust and ubiquitous GFP expression in a single generation of chicken embryos using the avian retroviral vector, RCASBP. Differentiation, 2009, 77, 473-482.	1.9	31
97	Identification of variants in pleiotropic genes causing â€œisolatedâ€•premature ovarian insufficiency: implications for medical practice. European Journal of Human Genetics, 2018, 26, 1319-1328.	2.8	31
98	The Rhox Homeobox Gene Family Shows Sexually Dimorphic and Dynamic Expression During Mouse Embryonic Gonad Development1. Biology of Reproduction, 2008, 79, 468-474.	2.7	30
99	Pre-Sertoli Specific Gene Expression Profiling Reveals Differential Expression of Ppt1 and Brd3 Genes Within the Mouse Genital Ridge at the Time of Sex Determination1. Biology of Reproduction, 2004, 71, 820-827.	2.7	29
100	Anti-MÃ¼llerian Hormone Is Required for Chicken Embryonic Urogenital System Growth but Not Sexual Differentiation1. Biology of Reproduction, 2015, 93, 138.	2.7	29
101	TP63â€truncating variants cause isolated premature ovarian insufficiency. Human Mutation, 2019, 40, 886-892.	2.5	29
102	Identification of Candidate Genes for Mayer-Rokitansky-KÃ¼ster-Hauser Syndrome Using Genomic Approaches. Sexual Development, 2019, 13, 26-34.	2.0	29
103	Identification, Expression, and Regulation of Anti-MÃ¼llerian Hormone Type-II Receptor in the Embryonic Chicken Gonad1. Biology of Reproduction, 2014, 90, 106.	2.7	28
104	Overexpression of Anti-MÃ¼llerian Hormone Disrupts Gonadal Sex Differentiation, Blocks Sex Hormone Synthesis, and Supports Cell Autonomous Sex Development in the Chicken. Endocrinology, 2016, 157, 1258-1275.	2.8	28
105	DMRT1 is required for MÃ¼llerian duct formation in the chicken embryo. Developmental Biology, 2015, 400, 224-236.	2.0	27
106	Whole exome sequencing combined with linkage analysis identifies a novel 3â€‰bp deletion in NR5A1. European Journal of Human Genetics, 2015, 23, 486-493.	2.8	27
107	Mitotic Arrest in Teratoma Susceptible Fetal Male Germ Cells. PLoS ONE, 2011, 6, e20736.	2.5	27
108	GATA4 Variants in Individuals With a 46,XY Disorder of Sex Development (DSD) May or May Not Be Associated With Cardiac Defects Depending on Second Hits in Other DSD Genes. Frontiers in Endocrinology, 2018, 9, 142.	3.5	26

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109	Analysis of NR5A1 in 142 patients with premature ovarian insufficiency, diminished ovarian reserve, or unexplained infertility. Maturitas, 2020, 131, 78-86.	2.4	26
110	STAG3 homozygous missense variant causes primary ovarian insufficiency and male non-obstructive azoospermia. Molecular Human Reproduction, 2020, 26, 665-677.	2.8	26
111	Characterisation of Urogenital Ridge Gene Expression in the Human Embryonal Carcinoma Cell Line NT2/D1. Sexual Development, 2007, 1, 114-126.	2.0	25
112	Hormonal evaluation in relation to phenotype and genotype in 286 patients with a disorder of sex development from Indonesia. Clinical Endocrinology, 2016, 85, 247-257.	2.4	24
113	NR5A1 gene variants repress the ovarian-specific WNT signaling pathway in 46,XX disorders of sex development patients. Human Mutation, 2019, 40, 207-216.	2.5	24
114	Review disorders of sex development: The evolving role of genomics in diagnosis and gene discovery. Birth Defects Research Part C: Embryo Today Reviews, 2016, 108, 337-350.	3.6	24
115	A duplication in a patient with 46,<scp>XX</scp> ovoâ€testicular disorder of sex development refines the <scp>SOX9</scp> testisâ€specific regulatory region to 24 kb. Clinical Genetics, 2017, 92, 347-349.	2.0	23
116	Variants in congenital hypogonadotrophic hypogonadism genes identified in an Indonesian cohort of 46,XY under-virilised boys. Human Genomics, 2017, 11, 1.	2.9	23
117	A Framework for the Prediction of Scale-Up When Using Compressible Chromatographic Packings. Biotechnology Progress, 2007, 23, 413-422.	2.6	22
118	An InÂVitro Differentiation Protocol for Human Embryonic Bipotential Gonad and Testis Cell Development. Stem Cell Reports, 2020, 15, 1377-1391.	4.8	22
119	Mutation analysis of the SOX9 gene in a patient with campomelic dysplasia. Human Mutation, 1998, 11, S112-S113.	2.5	21
120	Familial bilateral cryptorchidism is caused by recessive variants in RXFP2. Journal of Medical Genetics, 2019, 56, 727-733.	3.2	21
121	Genomic sequencing highlights the diverse molecular causes of Perrault syndrome: a peroxisomal disorder (PEX6), metabolic disorders (CLPP, GGPS1), and mtDNA maintenance/translation disorders (LARS2, TFAM). Human Genetics, 2020, 139, 1325-1343.	3.8	21
122	Isolation and expression of a novel member of the CITED family. Mechanisms of Development, 2000, 95, 305-308.	1.7	20
123	Expression and evolutionary conservation of the tescalcin gene during development. Gene Expression Patterns, 2009, 9, 273-281.	0.8	20
124	The proto-oncogene Ret is required for male foetal germ cell survival. Developmental Biology, 2012, 365, 101-109.	2.0	20
125	Expression profile of the RNA-binding protein genehermes during chicken embryonic development. Developmental Dynamics, 2005, 233, 1045-1051.	1.8	19
126	Ex vivo magnetofection: A novel strategy for the study of gene function in mouse organogenesis. Developmental Dynamics, 2009, 238, 956-964.	1.8	19

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127	FGF9, activin and TGFÎ² promote testicular characteristics in an XX gonad organ culture model. Reproduction, 2016, 152, 529-543.	2.6	19
128	Sox15 is up regulated in the embryonic mouse testis. Gene Expression Patterns, 2003, 3, 413-417.	0.8	18
129	Temporal and spatial expression profile of the novelarmadillo-related gene,Alex2, during testicular differentiation in the mouse embryo. Developmental Dynamics, 2005, 233, 188-193.	1.8	18
130	Of sex and determination: marking 25â€...years of Randy, the sex-reversed mouse. Development (Cambridge), 2016, 143, 1633-1637.	2.5	18
131	Mutations in SRY and SOX9: Testisâ€determining genes. Human Mutation, 1997, 9, 388-395.	2.5	18
132	Meiotic genes in premature ovarian insufficiency: variants in HROB and REC8 as likely genetic causes. European Journal of Human Genetics, 2022, 30, 219-228.	2.8	18
133	SOX14 is a candidate gene for limb defects associated with BPES and MÃ¶bius syndrome. Human Genetics, 2000, 106, 269-276.	3.8	17
134	Sex-specific expression of a novel gene Tmem184a during mouse testis differentiation. Reproduction, 2007, 133, 983-989.	2.6	16
135	Females Battle to Suppress Their Inner Male. Cell, 2009, 139, 1051-1053.	28.9	16
136	Molecular mechanisms associated with 46,XX disorders of sex development. Clinical Science, 2016, 130, 421-432.	4.3	16
137	Analysis of variants in GATA4 and FOG2/ZFPM2 demonstrates benign contribution to 46,XY disorders of sex development. Molecular Genetics & Genomic Medicine, 2020, 8, e1095.	1.2	16
138	Gene mapping in marsupials: Detection of an ancient autosomal gene cluster. Genomics, 1991, 9, 581-586.	2.9	15
139	Human sex determination. , 1998, 281, 501-505.		15
140	SOX14 is a candidate gene for limb defects associated with BPES and Mï¿½bius syndrome. Human Genetics, 2000, 106, 269-276.	3.8	15
141	Novel scavenger receptor gene is differentially expressed in the embryonic and adult mouse testis. Developmental Dynamics, 2005, 234, 1026-1033.	1.8	15
142	Functional analysis of the SRYâ€"KRAB interaction in mouse sex determination. Biology of the Cell, 2009, 101, 55-67.	2.0	15
143	CITED2 mutations potentially cause idiopathic premature ovarian failure. Translational Research, 2012, 160, 384-388.	5.0	15
144	SRY mutation analysis by next generation (deep) sequencing in a cohort of chromosomal Disorders of Sex Development (DSD) patients with a mosaic karyotype. BMC Medical Genetics, 2012, 13, 108.	2.1	15

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145	A recessive variant in TFAM causes mtDNA depletion associated with primary ovarian insufficiency, seizures, intellectual disability and hearing loss. Human Genetics, 2021, 140, 1733-1751.	3.8	15
146	Annexin XI co-localises with calcyclin in proliferating cells of the embryonic mouse testis. Developmental Dynamics, 2005, 234, 432-437.	1.8	14
147	A novel, homozygous mutation in desert hedgehog (DHH) in a 46, XY patient with dysgenetic testes presenting with primary amenorrhoea: a case report. International Journal of Pediatric Endocrinology (Springer), 2018, 2018, 2.	1.6	14
148	A framework for assessing the solutions in chromatographic process design and operation for large-scale manufacture. Journal of Chemical Technology and Biotechnology, 2006, 81, 1009-1020.	3.2	13
149	Inhibition of SRY-Calmodulin Complex Formation Induces Ectopic Expression of Ovarian Cell Markers in Developing XY Gonads. Endocrinology, 2011, 152, 2883-2893.	2.8	13
150	Analysis of Gene Function in Cultured Embryonic Mouse Gonads Using Nucleofection. Sexual Development, 2011, 5, 7-15.	2.0	12
151	Using ROADMAP Data to Identify Enhancers Associated with Disorders of Sex Development. Sexual Development, 2016, 10, 59-65.	2.0	12
152	XX Disorder of Sex Development is associated with an insertion on chromosome 9 and downregulation of RSPO1 in dogs (Canis lupus familiaris). PLoS ONE, 2017, 12, e0186331.	2.5	12
153	Mutant NR5A1/SF-1 in patients with disorders of sex development shows defective activation of theSOX9TESCO enhancer. Human Mutation, 2018, 39, 1861-1874.	2.5	12
154	The gene encoding the ketogenic enzyme HMGCS2 displays a unique expression during gonad development in mice. PLoS ONE, 2020, 15, e0227411.	2.5	12
155	Functional analysis of novel desert hedgehog gene variants improves the clinical interpretation of genomic data and provides a more accurate diagnosis for patients with 46,XY differences of sex development. Journal of Medical Genetics, 2019, 56, 434-443.	3.2	11
156	A comparative analysis of vertebrate sex determination. Novartis Foundation Symposium, 2002, 244, 102-11; discussion 111-4, 203-6, 253-7.	1.1	11
157	Sox9-dependent expression of Gstm6 in Sertoli cells during testis development in mice. Reproduction, 2009, 137, 481-486.	2.6	10
158	Expression of Wsb2 in the developing and adult mouse testis. Reproduction, 2007, 133, 753-761.	2.6	9
159	Protein tyrosine kinase 2 beta (PTK2B), but not focal adhesion kinase (FAK), is expressed in a sexually dimorphic pattern in developing mouse gonads. Developmental Dynamics, 2010, 239, 2735-2741.	1.8	9
160	SOX Genes and Their Role in Disorders of Sex Development. Sexual Development, 2022, 16, 80-91.	2.0	9
161	Analysis of In-situ Hybridization Data for Unique Genes Using GLIM. Biometrics, 1989, 45, 601.	1.4	8
162	Conserved expression of a novel gene during gonadal development. Developmental Dynamics, 2005, 233, 1083-1090.	1.8	8

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163	Testis Development, Fertility, and Survival in Ethanolamine Kinase 2-Deficient Mice. Endocrinology, 2008, 149, 6176-6186.	2.8	8
164	Rapid high-throughput analysis of DNaseI hypersensitive sites using a modified Multiplex Ligation-dependent Probe Amplification approach. BMC Genomics, 2009, 10, 412.	2.8	8
165	Development of Retroviral Vectors for Tissue-Restricted Expression in Chicken Embryonic Gonads. PLoS ONE, 2014, 9, e101811.	2.5	8
166	Dominant TP63 missense variants lead to constitutive activation and premature ovarian insufficiency. Human Mutation, 2022, 43, 1443-1453.	2.5	8
167	Phosphoglycerate kinase pseudogenes in the tammar wallaby and other macropodid marsupials. Mammalian Genome, 1994, 5, 531-537.	2.2	7
168	Kallmann syndrome gene (KAL-X) is not mutated in schizophrenia. , 1999, 88, 34-37.		7
169	Eki2 is upregulated specifically in the testis during mouse sex determination. Gene Expression Patterns, 2004, 4, 135-140.	0.8	7
170	Human Embryonic Stem Cell Research: An Australian Perspective. Cell, 2007, 128, 221-223.	28.9	7
171	Rapid and reliable determination of transgene zygosity in mice by multiplex ligation-dependent probe amplification. Transgenic Research, 2009, 18, 987-991.	2.4	7
172	Gonadal Dysgenesis: Associations between Clinical Features and Sex of Rearing.. Endocrine Journal, 1997, 44, 95-104.	1.6	6
173	The Cloning of SRY. , 1994, , 23-41.		5
174	Painful ovulation in a 46,XX SRY â^'ve adult male with SOX9 duplication. Endocrinology, Diabetes and Metabolism Case Reports, 2017, 2017, .	0.5	5
175	Novel mutation in the SRY gene results in 46,XY gonadal dysgenesis. Human Mutation, 1998, 11, S110-S111.	2.5	4
176	Redd1 Is a Novel Marker of Testis Development but Is Not Required for Normal Male Reproduction. Sexual Development, 2012, 6, 223-230.	2.0	4
177	Genetic Analysis Reveals Complete Androgen Insensitivity Syndrome in Female Children Surgically Treated for Inguinal Hernia. Journal of Investigative Surgery, 2021, 34, 227-233.	1.3	4
178	Analysis of the androgen receptor (AR) gene in a cohort of Indonesian undermasculinized 46, XY DSD patients. Egyptian Journal of Medical Human Genetics, 2021, 22, .	1.0	4
179	Whole exome sequencing reveals copy number variants in individuals with disorders of sex development. Molecular and Cellular Endocrinology, 2022, 546, 111570.	3.2	4
180	Establishing a Molecular Genetic Diagnosis in Children with Differences of Sex Development: A Clinical Approach. Hormone Research in Paediatrics, 2023, 96, 128-143.	1.8	4

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181	The Molecular Basis of Gonadal Development and Disorders of Sex Development. , 2012, , 1-9.		3
182	Genetics and Genomics of Primary Ovarian Insufficiency. , 2019, , 427-445.		3
183	The Cell Biology and Molecular Genetics of Testis Determination. Results and Problems in Cell Differentiation, 2000, 28, 23-52.	0.7	3
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