

Andrew H Sinclair

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196
papers

12,790
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51
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110
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211
ext. papers

14,210
ext. citations

6.6
avg, IF

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L-index

#	Paper	IF	Citations
196	A gene from the human sex-determining region encodes a protein with homology to a conserved DNA-binding motif. <i>Nature</i> , 1990 , 346, 240-4	50.4	2617
195	Genetic evidence equating SRY and the testis-determining factor. <i>Nature</i> , 1990 , 348, 448-50	50.4	777
194	The avian Z-linked gene DMRT1 is required for male sex determination in the chicken. <i>Nature</i> , 2009 , 461, 267-71	50.4	575
193	A male-specific role for SOX9 in vertebrate sex determination. <i>Development (Cambridge)</i> , 1996 , 122, 2813-2822	50.4	504
192	Conservation of a sex-determining gene. <i>Nature</i> , 1999 , 402, 601-2	50.4	318
191	Genetic evidence that ZFY is not the testis-determining factor. <i>Nature</i> , 1989 , 342, 937-9	50.4	272
190	Sex determination: insights from the chicken. <i>BioEssays</i> , 2004 , 26, 120-32	4.1	248
189	Dynamic regulation of mitotic arrest in fetal male germ cells. <i>Stem Cells</i> , 2008 , 26, 339-47	5.8	213
188	Evolution of sex determination and the Y chromosome: SRY-related sequences in marsupials. <i>Nature</i> , 1992 , 359, 531-3	50.4	210
187	Identification of SOX3 as an XX male sex reversal gene in mice and humans. <i>Journal of Clinical Investigation</i> , 2011 , 121, 328-41	15.9	196
186	The business impact of an integrated continuous biomanufacturing platform for recombinant protein production. <i>Journal of Biotechnology</i> , 2015 , 213, 3-12	3.7	169
185	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. <i>Genome Biology</i> , 2016 , 17, 243	18.3	166
184	Sequences homologous to ZFY, a candidate human sex-determining gene, are autosomal in marsupials. <i>Nature</i> , 1988 , 336, 780-3	50.4	163
183	Sites of estrogen receptor and aromatase expression in the chicken embryo. <i>General and Comparative Endocrinology</i> , 1997 , 108, 182-90	3	147
182	The human SRY transcript. <i>Human Molecular Genetics</i> , 1993 , 2, 2007-12	5.6	146
181	Vertebrate sex determination: many means to an end. <i>Reproduction</i> , 2002 , 124, 447-57	3.8	139
180	Endothelial cell migration directs testis cord formation. <i>Developmental Biology</i> , 2009 , 326, 112-20	3.1	136

179	Gonadal sex differentiation in chicken embryos: expression of estrogen receptor and aromatase genes. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 1997 , 60, 295-302	5.1	136
178	DMRT1 is upregulated in the gonads during female-to-male sex reversal in ZW chicken embryos. <i>Biology of Reproduction</i> , 2003 , 68, 560-70	3.9	136
177	Mutations in MAP3K1 cause 46,XY disorders of sex development and implicate a common signal transduction pathway in human testis determination. <i>American Journal of Human Genetics</i> , 2010 , 87, 898-904	11	133
176	Temperature-dependent sex determination: upregulation of SOX9 expression after commitment to male development. <i>Developmental Dynamics</i> , 1999 , 214, 171-7	2.9	123
175	Genetic regulation of mammalian gonad development. <i>Nature Reviews Endocrinology</i> , 2014 , 10, 673-83	15.2	122
174	Mutations in SRY and SOX9: testis-determining genes. <i>Human Mutation</i> , 1997 , 9, 388-95	4.7	121
173	Premature Ovarian Insufficiency: New Perspectives on Genetic Cause and Phenotypic Spectrum. <i>Endocrine Reviews</i> , 2016 , 37, 609-635	27.2	119
172	Rapid DNA extraction and PCR-sexing of mouse embryos. <i>Molecular Reproduction and Development</i> , 2001 , 60, 225-6	2.6	116
171	Temperature-dependent sex determination in the American alligator: AMH precedes SOX9 expression. <i>Developmental Dynamics</i> , 1999 , 216, 411-9	2.9	116
170	Gene expression during gonadogenesis in the chicken embryo. <i>Gene</i> , 1999 , 234, 395-402	3.8	116
169	Mammalian sex determination insights from humans and mice. <i>Chromosome Research</i> , 2012 , 20, 215-38	4.4	115
168	A long-term outcome study of intersex conditions. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2005 , 18, 555-67	1.6	113
167	Aromatase inhibition reduces expression of FOXL2 in the embryonic chicken ovary. <i>Developmental Dynamics</i> , 2005 , 233, 1052-5	2.9	105
166	Cloning and expression of R-Spondin1 in different vertebrates suggests a conserved role in ovarian development. <i>BMC Developmental Biology</i> , 2008 , 8, 72	3.1	99
165	ASW: a gene with conserved avian W-linkage and female specific expression in chick embryonic gonad. <i>Development Genes and Evolution</i> , 2000 , 210, 243-9	1.8	95
164	Onset of meiosis in the chicken embryo; evidence of a role for retinoic acid. <i>BMC Developmental Biology</i> , 2008 , 8, 85	3.1	92
163	Copy number variation in patients with disorders of sex development due to 46,XY gonadal dysgenesis. <i>PLoS ONE</i> , 2011 , 6, e17793	3.7	88
162	Sexually dimorphic microRNA expression during chicken embryonic gonadal development. <i>Biology of Reproduction</i> , 2009 , 81, 165-76	3.9	87

161	Temperature-dependent sex determination in the American alligator: expression of SF1, WT1 and DAX1 during gonadogenesis. <i>Gene</i> , 2000 , 241, 223-32	3.8	87
160	Dppa2 and Dppa4 are closely linked SAP motif genes restricted to pluripotent cells and the germ line. <i>Stem Cells</i> , 2007 , 25, 19-28	5.8	86
159	The Genetic and Environmental Factors Underlying Hypospadias. <i>Sexual Development</i> , 2015 , 9, 239-259	1.6	82
158	Comparison of human ZFY and ZFX transcripts. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1990 , 87, 1681-5	11.5	74
157	RNA sequencing reveals sexually dimorphic gene expression before gonadal differentiation in chicken and allows comprehensive annotation of the W-chromosome. <i>Genome Biology</i> , 2013 , 14, R26	18.3	72
156	A novel germ line mutation in SOX9 causes familial campomelic dysplasia and sex reversal. <i>Human Molecular Genetics</i> , 1996 , 5, 1625-30	5.6	69
155	Three-dimensional visualization of testis cord morphogenesis, a novel tubulogenic mechanism in development. <i>Developmental Dynamics</i> , 2009 , 238, 1033-41	2.9	67
154	Human sex reversal is caused by duplication or deletion of core enhancers upstream of SOX9. <i>Nature Communications</i> , 2018 , 9, 5319	17.4	65
153	Signaling through the TGF beta-activin receptors ALK4/5/7 regulates testis formation and male germ cell development. <i>PLoS ONE</i> , 2013 , 8, e54606	3.7	64
152	SRY protein enhances transcription of Fos-related antigen 1 promoter constructs. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1994 , 91, 4372-6	11.5	63
151	Male fetal germ cell differentiation involves complex repression of the regulatory network controlling pluripotency. <i>FASEB Journal</i> , 2010 , 24, 3026-35	0.9	62
150	Conserved regulatory modules in the Sox9 testis-specific enhancer predict roles for SOX, TCF/LEF, Forkhead, DMRT, and GATA proteins in vertebrate sex determination. <i>International Journal of Biochemistry and Cell Biology</i> , 2010 , 42, 472-7	5.6	61
149	Wnt signaling in ovarian development inhibits Sf1 activation of Sox9 via the Tesco enhancer. <i>Endocrinology</i> , 2012 , 153, 901-12	4.8	58
148	The long non-coding RNA, MHM, plays a role in chicken embryonic development, including gonadogenesis. <i>Developmental Biology</i> , 2012 , 366, 317-26	3.1	52
147	Expression of chicken steroidogenic factor-1 during gonadal sex differentiation. <i>General and Comparative Endocrinology</i> , 1999 , 113, 187-96	3	52
146	Preparing for genomic medicine: a real world demonstration of health system change. <i>Npj Genomic Medicine</i> , 2017 , 2, 16	6.2	51
145	Manipulation of estrogen synthesis alters MIR202* expression in embryonic chicken gonads. <i>Biology of Reproduction</i> , 2011 , 85, 22-30	3.9	51
144	Defective survival of proliferating Sertoli cells and androgen receptor function in a mouse model of the ATR-X syndrome. <i>Human Molecular Genetics</i> , 2011 , 20, 2213-24	5.6	51

143	Male-specific cell migration into the developing gonad is a conserved process involving PDGF signalling. <i>Developmental Biology</i> , 2005 , 284, 337-50	3.1	51
142	Overexpression of aromatase alone is sufficient for ovarian development in genetically male chicken embryos. <i>PLoS ONE</i> , 2013 , 8, e68362	3.7	50
141	Avian sex determination: what, when and where?. <i>Cytogenetic and Genome Research</i> , 2007 , 117, 165-73	1.9	50
140	Sex determination in the chicken embryo. <i>The Journal of Experimental Zoology</i> , 2001 , 290, 691-9		50
139	Cloning and expression of a DAX1 homologue in the chicken embryo. <i>Journal of Molecular Endocrinology</i> , 2000 , 24, 23-32	4.5	50
138	An environmental life cycle assessment comparison of single-use and conventional process technology for the production of monoclonal antibodies. <i>Journal of Cleaner Production</i> , 2013 , 41, 150-162	10.3	46
137	Isolation of rare transcripts by representational difference analysis. <i>Nucleic Acids Research</i> , 1997 , 25, 2681-2	20.1	46
136	Genes on the short arm of the human X chromosome are not shared with the marsupial X. <i>Genomics</i> , 1991 , 11, 339-45	4.3	46
135	Regulation of the female mouse germ cell cycle during entry into meiosis. <i>Cell Cycle</i> , 2010 , 9, 408-18	4.7	45
134	Application of a decision-support tool to assess pooling strategies in perfusion culture processes under uncertainty. <i>Biotechnology Progress</i> , 2005 , 21, 1231-42	2.8	44
133	Fertile females of the mole <i>Talpa occidentalis</i> are phenotypic intersexes with ovotestes. <i>Development (Cambridge)</i> , 1993 , 118, 1303-1311	6.6	44
132	Subtractive hybridisation screen identifies sexually dimorphic gene expression in the embryonic mouse gonad. <i>Genesis</i> , 2003 , 37, 84-90	1.9	43
131	Male-specific expression of <i>Aldh1a1</i> in mouse and chicken fetal testes: implications for retinoid balance in gonad development. <i>Developmental Dynamics</i> , 2009 , 238, 2073-80	2.9	42
130	The genetics of disorders of sex development in humans. <i>Sexual Development</i> , 2014 , 8, 262-72	1.6	40
129	Normalizing gene expression levels in mouse fetal germ cells. <i>Biology of Reproduction</i> , 2009 , 81, 362-70	3.9	40
128	A multi-exon deletion within <i>WFOX1</i> is associated with a 46,XY disorder of sex development. <i>European Journal of Human Genetics</i> , 2012 , 20, 348-51	5.3	40
127	Evaluation of candidate markers for the peritubular myoid cell lineage in the developing mouse testis. <i>Reproduction</i> , 2005 , 130, 509-16	3.8	40
126	Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. <i>American Journal of Human Genetics</i> , 2019 , 105, 7-14	11	39

125	SOX8 expression during chick embryogenesis. <i>Mechanisms of Development</i> , 2000 , 94, 257-60	1.7	39
124	Gonadal defects in Cited2-mutant mice indicate a role for SF1 in both testis and ovary differentiation. <i>International Journal of Developmental Biology</i> , 2010 , 54, 683-9	1.9	38
123	The potential role of microRNAs in regulating gonadal sex differentiation in the chicken embryo. <i>Chromosome Research</i> , 2012 , 20, 201-13	4.4	37
122	Genetic evidence against a role for W-linked histidine triad nucleotide binding protein (HINTW) in avian sex determination. <i>International Journal of Developmental Biology</i> , 2009 , 53, 59-67	1.9	37
121	The cerebellin 4 precursor gene is a direct target of SRY and SOX9 in mice. <i>Biology of Reproduction</i> , 2009 , 80, 1178-88	3.9	37
120	The molecular genetics of ovarian differentiation in the avian model. <i>Sexual Development</i> , 2013 , 7, 80-94	1.6	34
119	Autosomal assignment of OTC in marsupials and monotremes: implications for the evolution of sex chromosomes. <i>Genetical Research</i> , 1987 , 50, 131-6	1.1	34
118	WNT/ β -catenin and p27/FOXL2 differentially regulate supporting cell proliferation in the developing ovary. <i>Developmental Biology</i> , 2016 , 412, 250-60	3.1	33
117	Transgenic Chickens Overexpressing Aromatase Have High Estrogen Levels but Maintain a Predominantly Male Phenotype. <i>Endocrinology</i> , 2016 , 157, 83-90	4.8	33
116	FET-1: a novel W-linked, female specific gene up-regulated in the embryonic chicken ovary. <i>Mechanisms of Development</i> , 2002 , 119 Suppl 1, S87-90	1.7	33
115	The cell biology and molecular genetics of Müllerian duct development. <i>Wiley Interdisciplinary Reviews: Developmental Biology</i> , 2018 , 7, e310	5.9	32
114	Retinoic Acid Antagonizes Testis Development in Mice. <i>Cell Reports</i> , 2018 , 24, 1330-1341	10.6	30
113	Identification of candidate gonadal sex differentiation genes in the chicken embryo using RNA-seq. <i>BMC Genomics</i> , 2015 , 16, 704	4.5	30
112	Copy number variation associated with meiotic arrest in idiopathic male infertility. <i>Fertility and Sterility</i> , 2015 , 103, 214-9	4.8	29
111	Type II and type IX collagen transcript isoforms are expressed during mouse testis development. <i>Biology of Reproduction</i> , 2003 , 68, 1742-7	3.9	29
110	Sex, genes, and heat: triggers of diversity. <i>The Journal of Experimental Zoology</i> , 2001 , 290, 624-31		29
109	Purification and Transcriptomic Analysis of Mouse Fetal Leydig Cells Reveals Candidate Genes for Specification of Gonadal Steroidogenic Cells. <i>Biology of Reproduction</i> , 2015 , 92, 145	3.9	28
108	Heterogeneity of Human Neutrophil CD177 Expression Results from CD177P1 Pseudogene Conversion. <i>PLoS Genetics</i> , 2016 , 12, e1006067	6	28

107	Functional characterization of novel NR5A1 variants reveals multiple complex roles in disorders of sex development. <i>Human Mutation</i> , 2018 , 39, 124-139	4.7	27
106	Pre-sertoli specific gene expression profiling reveals differential expression of Ppt1 and Brd3 genes within the mouse genital ridge at the time of sex determination. <i>Biology of Reproduction</i> , 2004 , 71, 820-7	3.9	27
105	The rhox homeobox gene family shows sexually dimorphic and dynamic expression during mouse embryonic gonad development. <i>Biology of Reproduction</i> , 2008 , 79, 468-74	3.9	24
104	The Role of Copy Number Variants in Disorders of Sex Development. <i>Sexual Development</i> , 2018 , 12, 19-29	3.6	24
103	Whole exome sequencing combined with linkage analysis identifies a novel 3 bp deletion in NR5A1. <i>European Journal of Human Genetics</i> , 2015 , 23, 486-93	5.3	23
102	Robust and ubiquitous GFP expression in a single generation of chicken embryos using the avian retroviral vector, RCASBP. <i>Differentiation</i> , 2009 , 77, 473-82	3.5	23
101	DMRT1 is required for Müllerian duct formation in the chicken embryo. <i>Developmental Biology</i> , 2015 , 400, 224-36	3.1	22
100	Overexpression of Anti-Müllerian Hormone Disrupts Gonadal Sex Differentiation, Blocks Sex Hormone Synthesis, and Supports Cell Autonomous Sex Development in the Chicken. <i>Endocrinology</i> , 2016 , 157, 1258-75	4.8	22
99	Characterisation of urogenital ridge gene expression in the human embryonal carcinoma cell line NT2/D1. <i>Sexual Development</i> , 2007 , 1, 114-26	1.6	22
98	A duplication in a patient with 46,XX ovo-testicular disorder of sex development refines the SOX9 testis-specific regulatory region to 24 kb. <i>Clinical Genetics</i> , 2017 , 92, 347-349	4	21
97	Mitotic arrest in teratoma susceptible fetal male germ cells. <i>PLoS ONE</i> , 2011 , 6, e20736	3.7	21
96	Anti-Müllerian Hormone Is Required for Chicken Embryonic Urogenital System Growth but Not Sexual Differentiation. <i>Biology of Reproduction</i> , 2015 , 93, 138	3.9	20
95	Identification of variants in pleiotropic genes causing "isolated" premature ovarian insufficiency: implications for medical practice. <i>European Journal of Human Genetics</i> , 2018 , 26, 1319-1328	5.3	20
94	Restricted expression of DMRT3 in chicken and mouse embryos. <i>Gene Expression Patterns</i> , 2002 , 2, 69-72	1.5	20
93	Identification, expression, and regulation of anti-Müllerian hormone type-II receptor in the embryonic chicken gonad. <i>Biology of Reproduction</i> , 2014 , 90, 106	3.9	19
92	Expression and evolutionary conservation of the tescalcin gene during development. <i>Gene Expression Patterns</i> , 2009 , 9, 273-81	1.5	19
91	A framework for the prediction of scale-up when using compressible chromatographic packings. <i>Biotechnology Progress</i> , 2007 , 23, 413-22	2.8	19
90	Restricted expression of DMRT3 in chicken and mouse embryos. <i>Mechanisms of Development</i> , 2002 , 119 Suppl 1, S73-6	1.7	19

89	Isolation and expression of a novel member of the CITED family. <i>Mechanisms of Development</i> , 2000 , 95, 305-8	1.7	19
88	Sox15 is up regulated in the embryonic mouse testis. <i>Gene Expression Patterns</i> , 2003 , 3, 413-7	1.5	18
87	Rules for clinical diagnosis in babies with ambiguous genitalia. <i>Journal of Paediatrics and Child Health</i> , 2003 , 39, 406-13	1.3	17
86	The proto-oncogene Ret is required for male foetal germ cell survival. <i>Developmental Biology</i> , 2012 , 365, 101-9	3.1	16
85	Ex vivo magnetofection: a novel strategy for the study of gene function in mouse organogenesis. <i>Developmental Dynamics</i> , 2009 , 238, 956-64	2.9	16
84	Sex-specific expression of a novel gene Tmem184a during mouse testis differentiation. <i>Reproduction</i> , 2007 , 133, 983-9	3.8	16
83	Temporal and spatial expression profile of the novel armadillo-related gene, Alex2, during testicular differentiation in the mouse embryo. <i>Developmental Dynamics</i> , 2005 , 233, 188-93	2.9	16
82	Of sex and determination: marking 25 years of Randy, the sex-reversed mouse. <i>Development (Cambridge)</i> , 2016 , 143, 1633-7	6.6	16
81	TP63-truncating variants cause isolated premature ovarian insufficiency. <i>Human Mutation</i> , 2019 , 40, 886-892	4.7	15
80	Expression profile of the RNA-binding protein gene hermes during chicken embryonic development. <i>Developmental Dynamics</i> , 2005 , 233, 1045-51	2.9	15
79	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. <i>Frontiers in Endocrinology</i> , 2018 , 9, 142	5.7	14
78	Functional analysis of the SRY-KRAB interaction in mouse sex determination. <i>Biology of the Cell</i> , 2009 , 101, 55-67	3.5	14
77	Analysis of NR5A1 in 142 patients with premature ovarian insufficiency, diminished ovarian reserve, or unexplained infertility. <i>Maturitas</i> , 2020 , 131, 78-86	5	14
76	Hormonal evaluation in relation to phenotype and genotype in 286 patients with a disorder of sex development from Indonesia. <i>Clinical Endocrinology</i> , 2016 , 85, 247-57	3.4	14
75	FGF9, activin and TGF β promote testicular characteristics in an XX gonad organ culture model. <i>Reproduction</i> , 2016 , 152, 529-43	3.8	14
74	New insights into the genetic basis of premature ovarian insufficiency: Novel causative variants and candidate genes revealed by genomic sequencing. <i>Maturitas</i> , 2020 , 141, 9-19	5	13
73	Females battle to suppress their inner male. <i>Cell</i> , 2009 , 139, 1051-3	56.2	13
72	Inhibition of SRY-calmodulin complex formation induces ectopic expression of ovarian cell markers in developing XY gonads. <i>Endocrinology</i> , 2011 , 152, 2883-93	4.8	13

71	Mutation analysis of the SOX9 gene in a patient with campomelic dysplasia. <i>Human Mutation</i> , 1998 , Suppl 1, S112-3	4.7	13
70	Novel scavenger receptor gene is differentially expressed in the embryonic and adult mouse testis. <i>Developmental Dynamics</i> , 2005 , 234, 1026-33	2.9	13
69	Gene mapping in marsupials: detection of an ancient autosomal gene cluster. <i>Genomics</i> , 1991 , 9, 581-6	4.3	13
68	NR5A1 gene variants repress the ovarian-specific WNT signaling pathway in 46,XX disorders of sex development patients. <i>Human Mutation</i> , 2019 , 40, 207-216	4.7	13
67	Human sex determination. <i>The Journal of Experimental Zoology</i> , 1998 , 281, 501-5		12
66	A framework for assessing the solutions in chromatographic process design and operation for large-scale manufacture. <i>Journal of Chemical Technology and Biotechnology</i> , 2006 , 81, 1009-1020	3.5	12
65	Annexin XI co-localises with calyculin in proliferating cells of the embryonic mouse testis. <i>Developmental Dynamics</i> , 2005 , 234, 432-7	2.9	12
64	SOX14 is a candidate gene for limb defects associated with BPES and MBius syndrome. <i>Human Genetics</i> , 2000 , 106, 269-276	6.3	12
63	Identification of Candidate Genes for Mayer-Rokitansky-Küster-Hauser Syndrome Using Genomic Approaches. <i>Sexual Development</i> , 2019 , 13, 26-34	1.6	12
62	STAG3 homozygous missense variant causes primary ovarian insufficiency and male non-obstructive azoospermia. <i>Molecular Human Reproduction</i> , 2020 , 26, 665-677	4.4	11
61	XX Disorder of Sex Development is associated with an insertion on chromosome 9 and downregulation of RSPO1 in dogs (<i>Canis lupus familiaris</i>). <i>PLoS ONE</i> , 2017 , 12, e0186331	3.7	11
60	Variants in congenital hypogonadotrophic hypogonadism genes identified in an Indonesian cohort of 46,XY under-virilised boys. <i>Human Genomics</i> , 2017 , 11, 1	6.8	11
59	Analysis of gene function in cultured embryonic mouse gonads using nucleofection. <i>Sexual Development</i> , 2011 , 5, 7-15	1.6	11
58	FET-1: a novel W-linked, female specific gene up-regulated in the embryonic chicken ovary. <i>Gene Expression Patterns</i> , 2002 , 2, 83-6	1.5	11
57	Using ROADMAP Data to Identify Enhancers Associated with Disorders of Sex Development. <i>Sexual Development</i> , 2016 , 10, 59-65	1.6	11
56	A comparative analysis of vertebrate sex determination. <i>Novartis Foundation Symposium</i> , 2002 , 244, 102-11; discussion 111-4, 203-6, 253-7		11
55	CITED2 mutations potentially cause idiopathic premature ovarian failure. <i>Translational Research</i> , 2012 , 160, 384-8	11	10
54	SRY mutation analysis by next generation (deep) sequencing in a cohort of chromosomal Disorders of Sex Development (DSD) patients with a mosaic karyotype. <i>BMC Medical Genetics</i> , 2012 , 13, 108	2.1	10

53	A Comparative Analysis of Vertebrate Sex Determination. <i>Novartis Foundation Symposium</i> , 2008 , 102-114		10
52	SOX14 is a candidate gene for limb defects associated with BPES and Mbius syndrome. <i>Human Genetics</i> , 2000 , 106, 269-76	6.3	10
51	Review disorders of sex development: The evolving role of genomics in diagnosis and gene discovery. <i>Birth Defects Research Part C: Embryo Today Reviews</i> , 2016 , 108, 337-350		10
50	Familial bilateral cryptorchidism is caused by recessive variants in. <i>Journal of Medical Genetics</i> , 2019 , 56, 727-733	5.8	9
49	Expression of Wsb2 in the developing and adult mouse testis. <i>Reproduction</i> , 2007 , 133, 753-61	3.8	9
48	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). <i>Human Genetics</i> , 2020 , 139, 1325-1343	6.3	8
47	A novel, homozygous mutation in () in a 46, XY patient with dysgenetic testes presenting with primary amenorrhoea: a case report. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2018 , 2018, 2	1.5	8
46	Molecular mechanisms associated with 46,XX disorders of sex development. <i>Clinical Science</i> , 2016 , 130, 421-32	6.5	8
45	Testis development, fertility, and survival in Ethanolamine kinase 2-deficient mice. <i>Endocrinology</i> , 2008 , 149, 6176-86	4.8	8
44	An In Vitro Differentiation Protocol for Human Embryonic Bipotential Gonad and Testis Cell Development. <i>Stem Cell Reports</i> , 2020 , 15, 1377-1391	8	8
43	Mutant NR5A1/SF-1 in patients with disorders of sex development shows defective activation of the SOX9 TESCO enhancer. <i>Human Mutation</i> , 2018 , 39, 1861-1874	4.7	7
42	Rapid high-throughput analysis of DNaseI hypersensitive sites using a modified Multiplex Ligation-dependent Probe Amplification approach. <i>BMC Genomics</i> , 2009 , 10, 412	4.5	7
41	Human embryonic stem cell research: an Australian perspective. <i>Cell</i> , 2007 , 128, 221-3	56.2	7
40	Mutations in SRY and SOX9: Testis-determining genes 1997 , 9, 388		7
39	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. <i>Journal of Medical Genetics</i> , 2019 , 56, 434-443	5.8	6
38	The gene encoding the ketogenic enzyme HMGCS2 displays a unique expression during gonad development in mice. <i>PLoS ONE</i> , 2020 , 15, e0227411	3.7	6
37	Sox9-dependent expression of Gstm6 in Sertoli cells during testis development in mice. <i>Reproduction</i> , 2009 , 137, 481-6	3.8	6
36	Protein tyrosine kinase 2 beta (PTK2B), but not focal adhesion kinase (FAK), is expressed in a sexually dimorphic pattern in developing mouse gonads. <i>Developmental Dynamics</i> , 2010 , 239, 2735-41	2.9	6

35	Gonadal dysgenesis: associations between clinical features and sex of rearing. <i>Endocrine Journal</i> , 1997 , 44, 95-104	2.9	6
34	Eki2 is upregulated specifically in the testis during mouse sex determination. <i>Gene Expression Patterns</i> , 2004 , 4, 135-40	1.5	6
33	Conserved expression of a novel gene during gonadal development. <i>Developmental Dynamics</i> , 2005 , 233, 1083-90	2.9	6
32	Analysis of In-situ Hybridization Data for Unique Genes Using GLIM. <i>Biometrics</i> , 1989 , 45, 601	1.8	6
31	Phosphoglycerate kinase pseudogenes in the tammar wallaby and other macropodid marsupials. <i>Mammalian Genome</i> , 1994 , 5, 531-7	3.2	5
30	Development of retroviral vectors for tissue-restricted expression in chicken embryonic gonads. <i>PLoS ONE</i> , 2014 , 9, e101811	3.7	4
29	Rapid and reliable determination of transgene zygosity in mice by multiplex ligation-dependent probe amplification. <i>Transgenic Research</i> , 2009 , 18, 987-91	3.3	4
28	Kallmann syndrome gene (KAL-X) is not mutated in schizophrenia. <i>American Journal of Medical Genetics Part A</i> , 1999 , 88, 34-7		4
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