Pierre F Ray

List of Publications by Year
in descending order

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Version: 2024-02-01

109 papers	6,501 citations	81900 39 h-index	76900 74 g-index
122 all docs	122 docs citations	122 times ranked	4775 citing authors

#	Article	IF	Citations
1	Polyalanine expansion and frameshift mutations of the paired-like homeobox gene PHOX2B in congenital central hypoventilation syndrome. Nature Genetics, 2003, 33, 459-461.	21.4	771
2	Mutations in DNAH1, which Encodes an Inner Arm Heavy Chain Dynein, Lead to Male Infertility from Multiple Morphological Abnormalities of the Sperm Flagella. American Journal of Human Genetics, 2014, 94, 95-104.	6.2	328
3	Teratozoospermia: spotlight on the main genetic actors in the human. Human Reproduction Update, 2015, 21, 455-485.	10.8	255
4	Homozygous mutation of AURKC yields large-headed polyploid spermatozoa and causes male infertility. Nature Genetics, 2007, 39, 661-665.	21.4	248
5	Absence of triadin, a protein of the calcium release complex, is responsible for cardiac arrhythmia with sudden death in human. Human Molecular Genetics, 2012, 21, 2759-2767.	2.9	227
6	Mutations in CFAP43 and CFAP44 cause male infertility and flagellum defects in Trypanosoma and human. Nature Communications, 2018, 9, 686.	12.8	173
7	DPY19L2 Deletion as a Major Cause of Globozoospermia. American Journal of Human Genetics, 2011, 88, 344-350.	6.2	172
8	A Recurrent Deletion of DPY19L2 Causes Infertility in Man by Blocking Sperm Head Elongation and Acrosome Formation. American Journal of Human Genetics, 2011, 88, 351-361.	6.2	165
9	Absence of Dpy19l2, a new inner nuclear membrane protein, causes globozoospermia in mice by preventing the anchoring of the acrosome to the nucleus. Development (Cambridge), 2012, 139, 2955-2965.	2.5	144
10	The genetic architecture of morphological abnormalities of the sperm tail. Human Genetics, 2021, 140, 21-42.	3.8	130
11	Genetic abnormalities leading to qualitative defects of sperm morphology or function. Clinical Genetics, 2017, 91, 217-232.	2.0	127
12	Absence of CFAP69 Causes Male Infertility due to Multiple Morphological Abnormalities of the Flagella in Human and Mouse. American Journal of Human Genetics, 2018, 102, 636-648.	6.2	121
13	Bi-allelic Mutations in ARMC2 Lead to Severe Astheno-Teratozoospermia Due to Sperm Flagellum Malformations in Humans and Mice. American Journal of Human Genetics, 2019, 104, 331-340.	6.2	113
14	Homozygous mutation of PLCZ1 leads to defective human oocyte activation and infertility that is not rescued by the WW-binding protein PAWP. Human Molecular Genetics, 2016, 25, 878-891.	2.9	112
15	Increasing the denaturation temperature during the first cycles of amplification reduced allele dropout from single cells for preimplantation genetic diagnosis. Molecular Human Reproduction, 1996, 2, 213-218.	2.8	111
16	Bi-allelic DNAH8 Variants Lead to Multiple Morphological Abnormalities of the Sperm Flagella and Primary Male Infertility. American Journal of Human Genetics, 2020, 107, 330-341.	6.2	111
17	Bi-allelic Mutations in TTC21A Induce Asthenoteratospermia in Humans and Mice. American Journal of Human Genetics, 2019, 104, 738-748.	6.2	103
18	The Aurora Kinase C c.144delC mutation causes meiosis I arrest in men and is frequent in the North African population. Human Molecular Genetics, 2009, 18, 1301-1309.	2.9	97

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19	Whole-exome sequencing of familial cases of multiple morphological abnormalities of the sperm flagella (MMAF) reveals newDNAH1mutations. Human Reproduction, 2016, 31, 2872-2880.	0.9	96
20	<scp>SPINK</scp>2 deficiency causes infertility by inducing sperm defects in heterozygotes and azoospermia inÂhomozygotes. EMBO Molecular Medicine, 2017, 9, 1132-1149.	6.9	95
21	Homozygous missense mutation L673P in adenylate kinase 7 (AK7) leads to primary male infertility and multiple morphological anomalies of the flagella but not to primary ciliary dyskinesia. Human Molecular Genetics, 2018, 27, 1196-1211.	2.9	95
22	Whole-exome sequencing identifies mutations in FSIP2 as a recurrent cause of multiple morphological abnormalities of the sperm flagella. Human Reproduction, 2018, 33, 1973-1984.	0.9	93
23	Missense Mutations in SLC26A8, Encoding a Sperm-Specific Activator of CFTR, Are Associated with Human Asthenozoospermia. American Journal of Human Genetics, 2013, 92, 760-766.	6.2	92
24	Patients with multiple morphological abnormalities of the sperm flagella due toDNAH1mutations have a good prognosis following intracytoplasmic sperm injection. Human Reproduction, 2016, 31, 1164-1172.	0.9	85
25	Subcellular localization of phospholipase CÎ¶ in human sperm and its absence in DPY19L2-deficient sperm are consistent with its role in oocyte activation. Molecular Human Reproduction, 2015, 21, 157-168.	2.8	83
26	A Homozygous Ancestral SVA-Insertion-Mediated Deletion in WDR66 Induces Multiple Morphological Abnormalities of the Sperm Flagellum and Male Infertility. American Journal of Human Genetics, 2018, 103, 400-412.	6.2	81
27	XIST expression from the maternal X chromosome in human male preimplantation embryos at the blastocyst stage. Human Molecular Genetics, 1997, 6, 1323-1327.	2.9	77
28	Deleterious variants in X-linked CFAP47 induce asthenoteratozoospermia and primary male infertility. American Journal of Human Genetics, 2021, 108, 309-323.	6.2	74
29	Paternal transcripts for glucose-6-phosphate dehydrogenase and adenosine deaminase are first detectable in the human preimplantation embryo at the Three- to Four-Cell stage. Molecular Reproduction and Development, 1997, 48, 442-448.	2.0	67
30	A new AURKC mutation causing macrozoospermia: implications for human spermatogenesis and clinical diagnosis. Molecular Human Reproduction, 2011, 17, 762-768.	2.8	65
31	MLPA and sequence analysis of DPY19L2 reveals point mutations causing globozoospermia. Human Reproduction, 2012, 27, 2549-2558.	0.9	62
32	Dpy19l2-deficient globozoospermic sperm display altered genome packaging and DNA damage that compromises the initiation of embryo development. Molecular Human Reproduction, 2015, 21, 169-185.	2.8	61
33	Homozygous mutations inSPEF2induce multiple morphological abnormalities of the sperm flagella and male infertility. Journal of Medical Genetics, 2020, 57, 31-37.	3.2	57
34	Biallelic mutations inCFAP65cause male infertility with multiple morphological abnormalities of the sperm flagella in humans and mice. Journal of Medical Genetics, 2020, 57, 89-95.	3.2	55
35	CLINICAL EXPERIENCE WITH PREIMPLANTATION GENETIC DIAGNOSIS OF CYSTIC FIBROSIS (Î”F508). , 1996, 16, 137-142.		53
36	Assessment of the reliability of single blastomere analysis for preimplantation diagnosis of the Î”F508 deletion causing cystic fibrosis in clinical practice. , 1998, 18, 1402-1412.		53

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37	<scp>PATL</scp> 2 is a key actor of oocyte maturation whose invalidation causes infertility in women and mice. EMBO Molecular Medicine, 2018, 10, .	6.9	53
38	Identification of a new recurrent Aurora kinase C mutation in both European and African men with macrozoospermia. Human Reproduction, 2012, 27, 3337-3346.	0.9	52
39	Single cell co-amplification of polymorphic markers for the indirect preimplantation genetic diagnosis of hemophiliaï¿½A, X-linked adrenoleukodystrophy, X-linked hydrocephalus and incontinentia pigmenti loci on Xq28. Human Genetics, 2004, 114, 298-305.	3.8	50
40	Diversity of RNA-Binding Proteins Modulating Post-Transcriptional Regulation of Protein Expression in the Maturing Mammalian Oocyte. Cells, 2020, 9, 662.	4.1	50
41	First specific preimplantation genetic diagnosis for ornithine transcarbamylase deficiency. Prenatal Diagnosis, 2000, 20, 1048-1054.	2.3	49
42	Reduced allele dropout in single-cell analysis for preimplantation genetic diagnosis of cystic fibrosis. Journal of Assisted Reproduction and Genetics, 1996, 13, 104-106.	2.5	48
43	PBX1haploinsufficiency leads to syndromic congenital anomalies of the kidney and urinary tract (CAKUT) in humans. Journal of Medical Genetics, 2017, 54, 502-510.	3.2	46
44	Mutations in TTC29, Encoding an Evolutionarily Conserved Axonemal Protein, Result in Asthenozoospermia and Male Infertility. American Journal of Human Genetics, 2019, 105, 1148-1167.	6.2	44
45	Paternal epigenetics: Mammalian sperm provide much more than DNA at fertilization. Molecular and Cellular Endocrinology, 2020, 518, 110964.	3.2	44
46	Single gene defects leading to sperm quantitative anomalies. Clinical Genetics, 2017, 91, 208-216.	2.0	43
47	CFAP70 mutations lead to male infertility due to severe astheno-teratozoospermia. A case report. Human Reproduction, 2019, 34, 2071-2079.	0.9	43
48	Genetic causes of male infertility: snapshot on morphological abnormalities of the sperm flagellum. Basic and Clinical Andrology, 2019, 29, 2.	1.9	43
49	Biallelic variants in MAATS1 encoding CFAP91, a calmodulin-associated and spoke-associated complex protein, cause severe astheno-teratozoospermia and male infertility. Journal of Medical Genetics, 2020, 57, 708-716.	3.2	43
50	Whole-exome sequencing improves the diagnosis and care of men with non-obstructive azoospermia. American Journal of Human Genetics, 2022, 109, 508-517.	6.2	41
51	Identification of new FOXP3 mutations and prenatal diagnosis of IPEX syndrome. Prenatal Diagnosis, 2010, 30, 1072-1078.	2.3	39
52	A new mutation identified in SPATA16 in two globozoospermic patients. Journal of Assisted Reproduction and Genetics, 2016, 33, 815-820.	2.5	38
53	Dynamics of Sun5 Localization during Spermatogenesis in Wild Type and Dpy19l2 Knock-Out Mice Indicates That Sun5 Is Not Involved in Acrosome Attachment to the Nuclear Envelope. PLoS ONE, 2015, 10, e0118698.	2.5	37
54	Mutations of the aurora kinase C gene causing macrozoospermia are the most frequent genetic cause of male infertility in Algerian men. Asian Journal of Andrology, 2015, 17, 68.	1.6	37

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55	Can intracytoplasmic morphologically selected sperm injection be used to select normal-sized sperm heads in infertile patients with macrocephalic sperm head syndrome?. Fertility and Sterility, 2010, 93, 1347.e1-1347.e5.	1.0	35
56	Five yearsâ€™ experience of preimplantation genetic diagnosis in the Parisian Center: outcome of the first 441 started cycles. Fertility and Sterility, 2007, 87, 60-73.	1.0	34
57	Single cell quantification of the 8993T>G NARP mitochondrial DNA mutation by fluorescent PCR. Molecular Genetics and Metabolism, 2005, 84, 289-292.	1.1	33
58	Genetics of teratozoospermia: Back to the head. Best Practice and Research in Clinical Endocrinology and Metabolism, 2020, 34, 101473.	4.7	32
59	Preimplantation genetic diagnosis (PGD), a collaborative activity of clinical genetic departments and IVF centres. Prenatal Diagnosis, 2001, 21, 1086-1092.	2.3	31
60	Whole exome sequencing of men with multiple morphological abnormalities of the sperm flagella reveals novel homozygous QRICH2 mutations. Clinical Genetics, 2019, 96, 394-401.	2.0	30
61	Successful preimplantation genetic diagnosis for sex linked Lesch-Nyhan syndrome using specific diagnosis. , 1999, 19, 1237-1241.		27
62	The sodium/proton exchanger <scp>SLC9C1</scp> (<scp>sNHE</scp>) is essential for human sperm motility and fertility. Clinical Genetics, 2021, 99, 684-693.	2.0	26
63	Fine Characterisation of a Recombination Hotspot at the DPY19L2 Locus and Resolution of the Paradoxical Excess of Duplications over Deletions in the General Population. PLoS Genetics, 2013, 9, e1003363.	3.5	25
64	Progesterone-induced Acrosome Exocytosis Requires Sequential Involvement of Calcium-independent Phospholipase A2Î² (iPLA2Î²) and Group X Secreted Phospholipase A2 (sPLA2). Journal of Biological Chemistry, 2016, 291, 3076-3089.	3.4	25
65	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome and recurrent intrauterine fetal death. Lancet, The, 2015, 385, 2120.	13.7	24
66	Creation of knock out and knock in mice by CRISPR/Cas9 to validate candidate genes for human male infertility, interest, difficulties and feasibility. Molecular and Cellular Endocrinology, 2018, 468, 70-80.	3.2	24
67	Genetic analyses of a large cohort of infertile patients with globozoospermia, DPY19L2 still the main actor, GGN confirmed as a guest player. Human Genetics, 2021, 140, 43-57.	3.8	24
68	The sperm-associated antigen 6 interactome and its role in spermatogenesis. Reproduction, 2019, 158, 183-199.	2.6	24
69	CFAP61 is required for sperm flagellum formation and male fertility in human and mouse. Development (Cambridge), 2021, 148, .	2.5	24
70	Bi-allelic truncating variants in CFAP206 cause male infertility in human and mouse. Human Genetics, 2021, 140, 1367-1377.	3.8	23
71	The essential role of intraflagellar transport protein IFT81 in male mice spermiogenesis and fertility. American Journal of Physiology - Cell Physiology, 2020, 318, C1092-C1106.	4.6	20
72	Defect in the nuclear pore membrane glycoprotein 210-like gene is associated with extreme uncondensed sperm nuclear chromatin and male infertility: a case report. Human Reproduction, 2021, 36, 693-701.	0.9	20

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73	A missense mutation in IFT74, encoding for an essential component for intraflagellar transport of Tubulin, causes asthenozoospermia and male infertility without clinical signs of Bardetâ€“Biedl syndrome. Human Genetics, 2021, 140, 1031-1043.	3.8	20
74	Homozygous mutations in CCDC34 cause male infertility with oligoasthenoteratozoospermia in humans and mice. Journal of Medical Genetics, 2022, 59, 710-718.	3.2	20
75	Spermaurin, an La1-like peptide from the venom of the scorpionScorpio maurus palmatus, improves sperm motility and fertilization in different mammalian species. Molecular Human Reproduction, 2016, 23, 116-131.	2.8	18
76	Improved single-cell protocol for preimplantation genetic diagnosis of spinal muscular atrophy. Fertility and Sterility, 2005, 84, 734-739.	1.0	17
77	Clinical applications of fetal sex determination in maternal blood in a preimplantation genetic diagnosis centre. Human Reproduction, 2002, 17, 2183-2186.	0.9	16
78	Snake venoms as a source of compounds modulating sperm physiology: Secreted phospholipases A2 from Oxyuranus scutellatus scutellatus impact sperm motility, acrosome reaction and in vitro fertilization in mice. Biochimie, 2010, 92, 826-836.	2.6	16
79	Group X secreted phospholipase A₂ specifically decreases sperm motility in mice. Journal of Cellular Physiology, 2011, 226, 2601-2609.	4.1	15
80	Denaturing high-performance liquid chromatography (DHPLC)-based prenatal diagnosis for tuberous sclerosis. Prenatal Diagnosis, 2001, 21, 279-283.	2.3	13
81	Microduplication of the ARID1A gene causes intellectual disability with recognizable syndromic features. Genetics in Medicine, 2017, 19, 701-710.	2.4	13
82	AU040320 deficiency leads to disruption of acrosome biogenesis and infertility in homozygous mutant mice. Scientific Reports, 2018, 8, 10379.	3.3	13
83	Genomic duplication in the 19q13.42 imprinted region identified as a new genetic cause of intrauterine growth restriction. Clinical Genetics, 2018, 94, 575-580.	2.0	12
84	Genetic diagnosis, sperm phenotype and ICSI outcome in case of severe asthenozoospermia with multiple morphological abnormalities of the flagellum. Human Reproduction, 2021, 36, 2848-2860.	0.9	12
85	Oligogenic heterozygous inheritance of sperm abnormalities in mouse. ELife, 2022, 11, .	6.0	12
86	Leucine zipper transcription factor-like 1 (LZTFL1), an intraflagellar transporter protein 27 (IFT27) associated protein, is required for normal sperm function and male fertility. Developmental Biology, 2021, 477, 164-176.	2.0	11
87	PCR from Single Cells for Preimplantation Diagnosis. , 1996, 5, 245-258.		9
88	Birth of healthy female twins after preimplantation genetic diagnosis of cystic fibrosis combined with gender determination. Molecular Human Reproduction, 2002, 8, 688-694.	2.8	9
89	The place of social sexing' in medicine and science. Human Reproduction, 2002, 17, 248-249.	0.9	9
90	Biallelic variant in cyclin B3 is associated with failure of maternal meiosis II and recurrent digynic triploidy. Journal of Medical Genetics, 2021, 58, 783-788.	3.2	9

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91	Rapid Proteomic Profiling by MALDIâ€TOF Mass Spectrometry for Better Brain Tumor Classification. Proteomics - Clinical Applications, 2020, 14, 1900116.	1.6	8
92	Commentary on "morphological characteristics and initial genetic study of multiple morphological anomalies of the flagella in China". Asian Journal of Andrology, 2016, 18, 812.	1.6	8
93	The effect of group X secreted phospholipase A2 on fertilization outcome is specific and not mimicked by other secreted phospholipases A2 or progesterone. Biochimie, 2014, 99, 88-95.	2.6	7
94	Sun proteins and Dpy19l2 forming LINC-like links are critical for spermiogenesis. Biology Open, 2016, 5, 535-536.	1.2	7
95	Slo3 K+ channel blocker clofilium extends bull and mouse sperm-fertilizing competence. Reproduction, 2018, 156, 463-476.	2.6	7
96	Microdeletion del(22)(q12.1) excluding the MN1 gene in a patient with craniofacial anomalies. American Journal of Medical Genetics, Part A, 2016, 170, 498-503.	1.2	6
97	Deciphering the Genetics of Male Infertility: Progress and Challenges. Journal of Urology, 2011, 186, 1183-1184.	0.4	5
98	Identification and Characterization of the Most Common Genetic Variant Responsible for Acephalic Spermatozoa Syndrome in Men Originating from North Africa. International Journal of Molecular Sciences, 2021, 22, 2187.	4.1	5
99	A recurrent ZP1 variant is responsible for oocyte maturation defect with degenerated oocytes in infertile females. Clinical Genetics, 2022, 102, 22-29.	2.0	5
100	Sex selection by preimplantation genetic diagnosis: should it be carried out for social purposes?: Is preimplantation genetic diagnosis for 'social sexing' desirable in today's and tomorrow's society?. Human Reproduction, 2003, 18, 463-464.	0.9	4
101	Ethics and genetics of carrier embryos. Human Reproduction, 2006, 21, 2722-2723.	0.9	4
102	Comparative testicular transcriptome of wild type and globozoospermic Dpy19l2 knock out mice. Basic and Clinical Andrology, 2013, 23, 7.	1.9	4
103	From azoospermia to macrozoospermia, a phenotypic continuum due to mutations in the ZMYND15 gene. Asian Journal of Andrology, 2022, 24, 243.	1.6	4
104	The involvement of the nuclear lamina in human and rodent spermiogenesis: a systematic review. Basic and Clinical Andrology, 2018, 28, 7.	1.9	3
105	Evolution of preimplantation genetic diagnosis in France. Reproductive BioMedicine Online, 2001, 3, 226-229.	2.4	2
106	Measure of sperm DNA fragmentation (SDF): how, why and when?. Translational Andrology and Urology, 2017, 6, S588-S589.	1.4	1
107	Enzymatic activity of mouse group X-sPLA2 improves inÂvitro production of preimplantation bovine embryos. Theriogenology, 2019, 131, 113-122.	2.1	1
108	Combined Use of Whole Exome Sequencing and CRISPR/Cas9 to Study the Etiology of Non-Obstructive Azoospermia: Demonstration of the Dispensable Role of the Testis-Specific Genes C1orf185 and CCT6B. Cells, 2022, 11, 118.	4.1	1

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109	Chronology of reported denaturing high performance liquid chromatography (DHPLC)-based prenatal diagnoses. Prenatal Diagnosis, 2003, 23, 81-81.	2.3	0