Charles Coutton

List of Publications by Year
in descending order

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55 papers	3,106 citations	218381 26 h-index	174990 52 g-index
59 all docs	59 docs citations	59 times ranked	2564 citing authors

#	Article	IF	Citations
1	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. Human Genetics, 2022, 141, 65-80.	1.8	14
2	From azoospermia to macrozoospermia, a phenotypic continuum due to mutations in the ZMYND15 gene. Asian Journal of Andrology, 2022, 24, 243.	0.8	4
3	Whole-exome sequencing improves the diagnosis and care of men with non-obstructive azoospermia. American Journal of Human Genetics, 2022, 109, 508-517.	2.6	41
4	Neurodevelopmental phenotype in 36 new patients with 8p inverted duplicationâ€“deletion: Genotypeâ€“phenotype correlation for anomalies of the corpus callosum. Clinical Genetics, 2022, 101, 307-316.	1.0	4
5	Understanding the new <scp>BRD4</scp>â€related syndrome: Clinical and genomic delineation with an international cohort study. Clinical Genetics, 2022, 102, 117-122.	1.0	3
6	Oligogenic heterozygous inheritance of sperm abnormalities in mouse. ELife, 2022, 11, .	2.8	12
7	The genetic architecture of morphological abnormalities of the sperm tail. Human Genetics, 2021, 140, 21-42.	1.8	130
8	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.	1.8	24
9	The sodium/proton exchanger <scp>SLC9C1</scp> (<scp>sNHE</scp>) is essential for human sperm motility and fertility. Clinical Genetics, 2021, 99, 684-693.	1.0	26
10	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.	1.8	5
11	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.	1.8	20
12	Bi-allelic truncating variants in CFAP206 cause male infertility in human and mouse. Human Genetics, 2021, 140, 1367-1377.	1.8	23
13	CFAP61 is required for sperm flagellum formation and male fertility in human and mouse. Development (Cambridge), 2021, 148, .	1.2	24
14	Genetics of teratozoospermia: Back to the head. Best Practice and Research in Clinical Endocrinology and Metabolism, 2020, 34, 101473.	2.2	32
15	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.	1.5	43
16	Whole exome sequencing of men with multiple morphological abnormalities of the sperm flagella reveals novel homozygous QRICH2 mutations. Clinical Genetics, 2019, 96, 394-401.	1.0	30
17	CFAP70 mutations lead to male infertility due to severe astheno-teratozoospermia. A case report. Human Reproduction, 2019, 34, 2071-2079.	0.4	43
18	Mutations in TTC29, Encoding an Evolutionarily Conserved Axonemal Protein, Result in Asthenozoospermia and Male Infertility. American Journal of Human Genetics, 2019, 105, 1148-1167.	2.6	44

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19	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.	2.6	113
20	Genetic causes of male infertility: snapshot on morphological abnormalities of the sperm flagellum. Basic and Clinical Andrology, 2019, 29, 2.	0.8	43
21	Bi-allelic Mutations in TTC21A Induce Asthenoteratospermia in Humans and Mice. American Journal of Human Genetics, 2019, 104, 738-748.	2.6	103
22	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.	1.6	24
23	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.	2.6	121
24	Mutations in CFAP43 and CFAP44 cause male infertility and flagellum defects in Trypanosoma and human. Nature Communications, 2018, 9, 686.	5.8	173
25	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.	1.4	95
26	Genomic duplication in the 19q13.42 imprinted region identified as a new genetic cause of intrauterine growth restriction. Clinical Genetics, 2018, 94, 575-580.	1.0	12
27	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.	0.4	93
28	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.	2.6	81
29	Sun proteins and Dpy19l2 forming LINC-like links are critical for spermiogenesis. Biology Open, 2016, 5, 535-536.	0.6	7
30	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.	0.7	6
31	Endocrinological Abnormalities Are a Main Feature of 17p13.1 Microduplication Syndrome: A New Case and Literature Review. Molecular Syndromology, 2016, 7, 337-343.	0.3	4
32	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.	0.4	85
33	Clinical and molecular findings in 39 patients with KBG syndrome caused by deletion or mutation of ANKRD11. American Journal of Medical Genetics, Part A, 2016, 170, 2847-2859.	0.7	62
34	Whole-exome sequencing of familial cases of multiple morphological abnormalities of the sperm flagella (MMAF) reveals newDNAH1mutations. Human Reproduction, 2016, 31, 2872-2880.	0.4	96
35	Commentary on "morphological characteristics and initial genetic study of multiple morphological anomalies of the flagella in China". Asian Journal of Andrology, 2016, 18, 812.	0.8	8
36	Teratozoospermia: spotlight on the main genetic actors in the human. Human Reproduction Update, 2015, 21, 455-485.	5.2	255

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37	13q31.1 microdeletion: A prenatal case report with macrocephaly and macroglossia. European Journal of Medical Genetics, 2015, 58, 526-530.	0.7	6
38	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.	1.3	61
39	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.	1.3	83
40	Array-CGH in children with mild intellectual disability: a population-based study. European Journal of Pediatrics, 2015, 174, 75-83.	1.3	16
41	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.	0.8	37
42	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.	2.6	328
43	Maternal complex chromosomal rearrangement leads to TCF12 microdeletion in a patient presenting with coronal craniosynostosis and intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 1530-1536.	0.7	11
44	Optimized Generation of Functional Neutrophils and Macrophages from Patient-Specific Induced Pluripotent Stem Cells:Ex VivoModels of X⁰-Linked, AR22⁰- and AR47⁰- Chronic Granulomatous Diseases. BioResearch Open Access, 2014, 3, 311-326.	2.6	30
45	7p22.3 microdeletion disrupting SNX8 in a patient presenting with intellectual disability but no tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2014, 164, 2133-2135.	0.7	7
46	Comparative testicular transcriptome of wild type and globozoospermic Dpy19l2 knock out mice. Basic and Clinical Andrology, 2013, 23, 7.	0.8	4
47	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.	1.5	25
48	Identification of a new recurrent Aurora kinase C mutation in both European and African men with macrozoospermia. Human Reproduction, 2012, 27, 3337-3346.	0.4	52
49	MLPA and sequence analysis of DPY19L2 reveals point mutations causing globozoospermia. Human Reproduction, 2012, 27, 2549-2558.	0.4	62
50	17p13.1 microduplication in a boy with Silverâ€“Russell syndrome features and intellectual disability. American Journal of Medical Genetics, Part A, 2012, 158A, 2564-2570.	0.7	14
51	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.	1.2	144
52	Clinical, Functional and Genetic Analysis of Twenty-Four Patients with Chronic Granulomatous Disease â€“ Identification of Eight Novel Mutations in CYBB and NCF2 Genes. Journal of Clinical Immunology, 2012, 32, 942-958.	2.0	19
53	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.	2.6	165
54	From lowe syndrome to Dent disease: correlations between mutations of the OCRL1 gene and clinical and biochemical phenotypes. Human Mutation, 2011, 32, 379-388.	1.1	120

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55	Development of a multiplex ligation-dependent probe amplification (MLPA) assay for quantification of the OCRL1 gene. Clinical Biochemistry, 2010, 43, 609-614.	0.8	16