

John C K Barber

List of Publications by Year in descending order

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Version: 2024-02-01

25
papers

546
citations

623734

14
h-index

642732

23
g-index

28
all docs

28
docs citations

28
times ranked

939
citing authors

#	ARTICLE	IF	CITATIONS
1	8p23.1 duplication syndrome; a novel genomic condition with unexpected complexity revealed by array CGH. <i>European Journal of Human Genetics</i> , 2008, 16, 18-27.	2.8	74
2	15q overgrowth syndrome: A newly recognized phenotype associated with overgrowth, learning difficulties, characteristic facial appearance, renal anomalies and increased dosage of distal chromosome 15q. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 147-154.	1.2	51
3	8p23.1 duplication syndrome; common, confirmed, and novel features in six further patients. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 487-500.	1.2	47
4	Duplications and copy number variants of 8p23.1 are cytogenetically indistinguishable but distinct at the molecular level. <i>European Journal of Human Genetics</i> , 2005, 13, 1131-1136.	2.8	46
5	Centromeric inactivation in a dicentric human Y;21 translocation chromosome. <i>Chromosoma</i> , 1997, 106, 199-206.	2.2	40
6	16p11.2â€”p12.2 duplication syndrome; a genomic condition differentiated from euchromatic variation of 16p11.2. <i>European Journal of Human Genetics</i> , 2013, 21, 182-189.	2.8	32
7	8p23.1 duplication syndrome differentiated from copy number variation of the defensin cluster at prenatal diagnosis in four new families. <i>Molecular Cytogenetics</i> , 2010, 3, 3.	0.9	28
8	Terminal 3p deletions: Phenotypic variability, chromosomal nonâ€”penetrance, or gene modification?. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1899-1901.	1.2	26
9	Small terminal deletions of the long arm of chromosome 2: Two new cases. <i>American Journal of Medical Genetics Part A</i> , 1994, 53, 366-369.	2.4	24
10	Segmental haplosufficiency: transmitted deletions of 2p12 include a pancreatic regeneration gene cluster and have no apparent phenotypic consequences. <i>European Journal of Human Genetics</i> , 2005, 13, 283-291.	2.8	21
11	Inside the 8p23.1 duplication syndrome; eight microduplications of likely or uncertain clinical significance. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2052-2064.	1.2	21
12	Mosaic tetrasomy 8p: Molecular cytogenetic confirmation and measurement of glutathione reductase and tissue plasminogen activator levels. <i>American Journal of Medical Genetics Part A</i> , 1993, 47, 100-105.	2.4	17
13	Unbalanced translocation in a mother and her son in one of two 5;10 translocation families. <i>American Journal of Medical Genetics Part A</i> , 1996, 62, 84-90.	2.4	16
14	Deletions of 2q14 that include the homeobox engrailed 1 (EN1) transcription factor are compatible with a normal phenotype. <i>European Journal of Human Genetics</i> , 2006, 14, 739-743.	2.8	15
15	Inverted duplication of 1q32.1 to 1q44 characterized by array CGH and review of distal 1q partial trisomy. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 793-797.	1.2	15
16	Multipoint FISH: a rapid and reliable way to define cryptic and complex abnormalities. <i>Clinical Genetics</i> , 1999, 56, 192-199.	2.0	13
17	A de novo 4q34 interstitial deletion of at least 9.3â€”Mb with no discernible phenotypic effect. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1764-1769.	1.2	13
18	Expansion of a 12-kb VNTR containing the REXO1L1 gene cluster underlies the microscopically visible euchromatic variant of 8q21.2. <i>European Journal of Human Genetics</i> , 2014, 22, 458-463.	2.8	10

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19	Transmitted deletions of medial 5p and learning difficulties; Does the cadherin cluster only become penetrant when flanking genes are deleted?. American Journal of Medical Genetics, Part A, 2011, 155, 2807-2815.	1.2	9
20	Incomplete penetrance, variable expressivity, or dosage insensitivity in four families with directly transmitted unbalanced chromosome abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 319-329.	1.2	7
21	Further molecular and clinical delineation of the Wisconsin syndrome phenotype associated with interstitial 3q24q25 deletions. American Journal of Medical Genetics, Part A, 2011, 155, 106-112.	1.2	6
22	Transmitted duplication of 12q21.32â€“12q22 includes 48 genes and has no apparent phenotypic consequences. American Journal of Medical Genetics, Part A, 2007, 143A, 615-618.	1.2	4
23	Reassignment of <i>HMX1</i> indicates copy number variation within 4p16.1 may be an alternative cause of oculoauricular phenotypes. American Journal of Medical Genetics, Part A, 2018, 176, 2034-2036.	1.2	3
24	Directly Transmitted 12.3-Mb Deletion with a Consistent Phenotype in the Variable 11q21q22.3 Region. Cytogenetic and Genome Research, 2020, 160, 185-192.	1.1	3
25	Copy number variation of the REXO1L1 gene cluster; euchromatic deletion variant or susceptibility factor?. European Journal of Human Genetics, 2017, 25, 8-9.	2.8	1