John C K Barber

List of Publications by Year
in descending order

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25	546	623734	642732
papers	citations	14 h-index	23 g-index

28	28	28	939
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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
2	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
3	Reassignment of HMX1 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
4	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
5	Inside the 8p23.1 duplication syndrome; eight microduplications of likely or uncertain clinical significance. American Journal of Medical Genetics, Part A, 2015, 167, 2052-2064.	1.2	21
6	Expansion of a 12-kb VNTR containing the REXO1L1 gene cluster underlies the microscopically visible euchromatic variant of 8q21.2. European Journal of Human Genetics, 2014, 22, 458-463.	2.8	10
7	8p23.1 duplication syndrome; common, confirmed, and novel features in six further patients. American Journal of Medical Genetics, Part A, 2013, 161, 487-500.	1.2	47
8	16p11.2â€“p12.2 duplication syndrome; a genomic condition differentiated from euchromatic variation of 16p11.2. European Journal of Human Genetics, 2013, 21, 182-189.	2.8	32
9	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
10	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
11	A de novo 4q34 interstitial deletion of at least 9.3â€‰Mb with no discernible phenotypic effect. American Journal of Medical Genetics, Part A, 2010, 152A, 1764-1769.	1.2	13
12	8p23.1 duplication syndrome differentiated from copy number variation of the defensin cluster at prenatal diagnosis in four new families. Molecular Cytogenetics, 2010, 3, 3.	0.9	28
13	Inverted duplication of 1q32.1 to 1q44 characterized by array CGH and review of distal 1q partial trisomy. American Journal of Medical Genetics, Part A, 2009, 149A, 793-797.	1.2	15
14	15q overgrowth syndrome: A newly recognized phenotype associated with overgrowth, learning difficulties, characteristic facial appearance, renal anomalies and increased dosage of distal chromosome 15q. American Journal of Medical Genetics, Part A, 2009, 149A, 147-154.	1.2	51
15	Terminal 3p deletions: Phenotypic variability, chromosomal nonâ€penetrance, or gene modification?. American Journal of Medical Genetics, Part A, 2008, 146A, 1899-1901.	1.2	26
16	8p23.1 duplication syndrome; a novel genomic condition with unexpected complexity revealed by array CGH. European Journal of Human Genetics, 2008, 16, 18-27.	2.8	74
17	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
18	Deletions of 2q14 that include the homeobox engrailed 1 (EN1) transcription factor are compatible with a normal phenotype. European Journal of Human Genetics, 2006, 14, 739-743.	2.8	15

#	Article	IF	Citations
19	Segmental haplosufficiency: transmitted deletions of 2p12 include a pancreatic regeneration gene cluster and have no apparent phenotypic consequences. European Journal of Human Genetics, 2005, 13, 283-291.	2.8	21
20	Duplications and copy number variants of 8p23.1 are cytogenetically indistinguishable but distinct at the molecular level. European Journal of Human Genetics, 2005, 13, 1131-1136.	2.8	46
21	Multipaint FISH: a rapid and reliable way to define cryptic and complex abnormalities. Clinical Genetics, 1999, 56, 192-199.	2.0	13
22	Centromeric inactivation in a dicentric human Y;21 translocation chromosome. Chromosoma, 1997, 106, 199-206.	2.2	40
23	Unbalanced translocation in a mother and her son in one of two 5;10 translocation families. American Journal of Medical Genetics Part A, 1996, 62, 84-90.	2.4	16
24	Small terminal deletions of the long arm of chromosome 2: Two new cases. American Journal of Medical Genetics Part A, 1994, 53, 366-369.	2.4	24
25	Mosaic tetrasomy 8p: Molecular cytogenetic confirmation and measurement of glutathione reductase and tissue plasminogen activator levels. American Journal of Medical Genetics Part A, 1993, 47, 100-105.	2.4	17