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List of Publications by Year in descending order

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687363 552781 1,650 28 13 26 h-index citations g-index papers 29 29 29 4867 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. Nature Genetics, 2013, 45, 145-154.	21.4	675
2	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. Nature Communications, 2016, 7, 10023.	12.8	412
3	A 24-bp duplication in exon 10 of human chitotriosidase gene from the sub-Saharan to the Mediterranean area: role of parasitic diseases and environmental conditions. Genes and Immunity, 2003, 4, 570-574.	4.1	88
4	EDA2R Is Associated with Androgenetic Alopecia. Journal of Investigative Dermatology, 2008, 128, 2268-2270.	0.7	79
5	A Genomewide Search Using an Original Pairwise Sampling Approach for Large Genealogies Identifies a New Locus for Total and Low-Density Lipoprotein Cholesterol in Two Genetically Differentiated Isolates of Sardinia. American Journal of Human Genetics, 2004, 75, 1015-1031.	6.2	48
6	Identification of a founder BRCA2 mutation in Sardinia. British Journal of Cancer, 2000, 82, 553-559.	6.4	42
7	Extent of linkage disequilibrium in a Sardinian sub-isolate: sampling and methodological considerations. Human Molecular Genetics, 2003, 13, 25-33.	2.9	42
8	Not all isolates are equal: linkage disequilibrium analysis on Xq13.3 reveals different patterns in Sardinian sub-populations. Human Genetics, 2002, 111, 9-15.	3.8	39
9	High Differentiation among Eight Villages in a Secluded Area of Sardinia Revealed by Genome-Wide High Density SNPs Analysis. PLoS ONE, 2009, 4, e4654.	2.5	30
10	Bi-allelic Mutations in KLHL7 Cause a Crisponi/CISS1-like Phenotype Associated with Early-Onset Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 236-245.	6.2	28
11	BRCA1 and BRCA2 germline mutations in Sardinian breast cancer families and their implications for genetic counseling. Annals of Oncology, 2002, 13, 1899-1907.	1.2	20
12	Landscape of transcriptome variations uncovering known and novel driver events in colorectal carcinoma. Scientific Reports, 2020, 10, 432.	3.3	16
13	Patterns of Linkage Disequilibrium between SNPs in a Sardinian Population Isolate and the Selection of Markers for Association Studies. Human Heredity, 2008, 65, 9-22.	0.8	14
14	Novel <i>NALCN</i> biallelic truncating mutations in siblings with IHPRF1 syndrome. Clinical Genetics, 2018, 93, 1245-1247.	2.0	14
15	Novel ANKRD11 gene mutation in an individual with a mild phenotype of KBG syndrome associated to a GEFS+ phenotypic spectrum: a case report. BMC Medical Genetics, 2019, 20, 16.	2.1	14
16	Confirmation of a new phenotype in an individual with a variant in the last part of exon 30 of <i>CREBBP</i> . American Journal of Medical Genetics, Part A, 2019, 179, 634-638.	1.2	13
17	Crisponi/coldâ€induced sweating syndrome: Differential diagnosis, pathogenesis and treatment concepts. Clinical Genetics, 2020, 97, 209-221.	2.0	12
18	Infant developmental profile of Crisponi syndrome due to compound heterozygosity for CRLF1 deletion. Clinical Dysmorphology, 2020, 29, 141-143.	0.3	11

#	Article	lF	CITATIONS
19	Application of a New Method for GWAS in a Related Case/Control Sample with Known Pedigree Structure: Identification of New Loci for Nephrolithiasis. PLoS Genetics, 2011, 7, e1001281.	3.5	10
20	Microsatellites and SNPs linkage analysis in a Sardinian genetic isolate confirms several essential hypertension loci previously identified in different populations. BMC Medical Genetics, 2009, 10, 81.	2.1	8
21	A strategy analysis for genetic association studies with known inbreeding. BMC Genetics, 2011, 12, 63.	2.7	8
22	Molecular Characterization of \hat{l}^2 -Thalassemia Mutations in Central Vietnam. Hemoglobin, 2017, 41, 96-99.	0.8	7
23	Exome sequencing in Crisponi/coldâ€induced sweating syndrome–like individuals reveals unpredicted alternative diagnoses. Clinical Genetics, 2019, 95, 607-614.	2.0	7
24	A new case series of Crisponi syndrome in a Turkish family and review of the literature. Clinical Dysmorphology, 2017, 26, 66-72.	0.3	5
25	Molecular basis of openâ€angle glaucoma in Italy. Acta Ophthalmologica, 1998, 76, 16-17.	0.3	2
26	Methylenetetrahydrofolate reductase gene polymorphisms in Burkina Faso. Clinica Chimica Acta, 2005, 360, 199-200.	1.1	0
27	Crisponi/Cold Induced Sweating Syndrome Type 1 With a Private Cytokine Receptor Like Factor 1 (CRLF1) Mutation in an Indian Family. Indian Pediatrics, 2020, 57, 1075-1077.	0.4	0
28	Crisponi/Cold Induced Sweating Syndrome Type 1 With a Private Cytokine Receptor Like Factor 1 (CRLF1) Mutation in an Indian Family. Indian Pediatrics, 2020, 57, 1075-1076.	0.4	0