Giorgio Gimelli

List of Publications by Year in descending order

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42 papers	3,618 citations	279798 23 h-index	265206 42 g-index
42 all docs	42 docs citations	42 times ranked	4826 citing authors

#	Article	IF	Citations
1	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
2	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509
3	Olfactory Receptorâ€“Gene Clusters, Genomic-Inversion Polymorphisms, and Common Chromosome Rearrangements. American Journal of Human Genetics, 2001, 68, 874-883.	6.2	338
4	Immunolocalization of CENP-A suggests a distinct nucleosome structure at the inner kinetochore plate of active centromeres. Current Biology, 1997, 7, 901-904.	3.9	334
5	Heterozygous Submicroscopic Inversions Involving Olfactory Receptorâ€“Gene Clusters Mediate the Recurrent t(4;8)(p16;p23) Translocation. American Journal of Human Genetics, 2002, 71, 276-285.	6.2	185
6	Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572.	2.9	173
7	Mutations in ZBTB24 Are Associated with Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome Type 2. American Journal of Human Genetics, 2011, 88, 796-804.	6.2	158
8	Genomic inversions of human chromosome 15q11-q13 in mothers of Angelman syndrome patients with class II (BP2/3) deletions. Human Molecular Genetics, 2003, 12, 849-858.	2.9	131
9	Overexpression of the C-type natriuretic peptide (CNP) is associated with overgrowth and bone anomalies in an individual with balanced t(2;7) translocation. Human Mutation, 2007, 28, 724-731.	2.5	118
10	Heterogeneous clinical presentation in ICF syndrome: correlation with underlying gene defects. European Journal of Human Genetics, 2013, 21, 1219-1225.	2.8	115
11	Transmission of a Fully Functional Human Neocentromere through Three Generations. American Journal of Human Genetics, 1999, 64, 1440-1444.	6.2	113
12	Genetic variation in ICF syndrome: Evidence for genetic heterogeneity. Human Mutation, 2000, 16, 509-517.	2.5	92
13	Deletion of a 5-cM Region at Chromosome 8p23 Is Associated With a Spectrum of Congenital Heart Defects. Circulation, 2000, 102, 432-437.	1.6	83
14	Detailed phenotypeâ€“genotype study in five patients with chromosome 6q16 deletion: narrowing the critical region for Praderâ€“Willi-like phenotype. European Journal of Human Genetics, 2008, 16, 1443-1449.	2.8	74
15	SatelliteÂ2 methylation patterns in normal and ICF syndrome cells and association of hypomethylation with advanced replication. Human Genetics, 2001, 109, 452-462.	3.8	58
16	Mutations in SOX17 are associated with congenital anomalies of the kidney and the urinary tract. Human Mutation, 2010, 31, 1352-1359.	2.5	54
17	Microarray based analysis of an inherited terminal 3p26.3 deletion, containing only the CHL1 gene, from a normal father to his two affected children. Orphanet Journal of Rare Diseases, 2011, 6, 12.	2.7	42
18	Two classes of low-copy repeats comediate a new recurrent rearrangement consisting of duplication at 8p23.1 and triplication at 8p23.2. Human Mutation, 2007, 28, 459-468.	2.5	41

#	Article	IF	Citations
19	Genotype-Phenotype Correlation of 2q37 Deletions Including NPPC Gene Associated with Skeletal Malformations. PLoS ONE, 2013, 8, e66048.	2.5	32
20	CENP-G in neocentromeres and inactive centromeres. Chromosoma, 2000, 109, 328-333.	2.2	26
21	Complex pathogenesis of Hirschsprung's disease in a patient with hydrocephalus, vesico-ureteral reflux and a balanced translocation t(3;17)(p12;q11). European Journal of Human Genetics, 2009, 17, 483-490.	2.8	26
22	Molecular characterization of a t(2;6) balanced translocation that is associated with a complex phenotype and leads to truncation of theTCBA1gene. Human Mutation, 2005, 26, 426-436.	2.5	25
23	Altered Intra-Nuclear Organisation of Heterochromatin and Genes in ICF Syndrome. PLoS ONE, 2010, 5, e11364.	2.5	25
24	Heterozygous deletion of CHL1 gene: Detailed array-CGH and clinical characterization of a new case and review of the literature. European Journal of Medical Genetics, 2014, 57, 626-629.	1.3	23
25	A neocentromere in the DAZ region of the human Y chromosome. Chromosoma, 2000, 109, 318-327.	2.2	21
26	ASDIN: Arterial Interventions in Arteriovenous Access and Chronic Kidney Disease: A Role for Interventional Nephrologists. Seminars in Dialysis, 2009, 22, 545-556.	1.3	21
27	T-cell apoptosis in ICF syndrome. Journal of Allergy and Clinical Immunology, 2001, 108, 310-312.	2.9	17
28	Clinical and molecular characterization of a patient with interstitial 6q21q22.1 deletion. Molecular Cytogenetics, 2015, 8, 31.	0.9	17
29	Clinical and Molecular Cytogenetic Characterization of a de novo Interstitial 1p31.1p31.3 Deletion in a Boy with Moderate Intellectual Disability and Severe Language Impairment. Cytogenetic and Genome Research, 2015, 146, 39-43.	1.1	16
30	Vertebral Artery Embolic Protection Via Ipsilateral Brachial Approach During Left Subclavian Artery Angioplasty and Stenting. Vascular and Endovascular Surgery, 2006, 40, 235-238.	0.7	12
31	Late acute thrombosis after coronary brachytherapy: When is the risk over?. Catheterization and Cardiovascular Interventions, 2001, 54, 216-218.	1.7	10
32	The impact of increased pulmonary arterial pressure on outcomes after transcatheter aortic valve replacement. Catheterization and Cardiovascular Interventions, 2020, 96, E723-E734.	1.7	10
33	A novel case of immunodeficiency, centromeric instability, and facial anomalies (the ICF syndrome): immunologic and cytogenetic studies. Haematologica, 2002, 87, 329-31.	3.5	10
34	Hemodynamically supported percutaneous coronary revascularization improves left ventricular function in patients with ischemic dilated cardiomyopathy at very high risk for surgery: a single-center experience. Journal of Invasive Cardiology, 2008, 20, 642-6.	0.4	9
35	Parental Imbalances Involving Chromosomes 15q and 22q May Predispose to the Formation of De Novo Pathogenic Microdeletions and Microduplications in the Offspring. PLoS ONE, 2013, 8, e57910.	2.5	7
36	Are the nail-patella syndrome and the autosomal Goltz-like syndrome the phenotypic expressions of different alleles at the COL5A1 locus?. Human Genetics, 1993, 91, 175-7.	3.8	6

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37	Direct duplication 12p11.21â€“p13.31 mediated by segmental duplications: a new recurrent rearrangement?. Human Genetics, 2005, 118, 207-213.	3.8	6
38	1p31.1 microdeletion including only NEGR1 gene in two patients. European Journal of Medical Genetics, 2020, 63, 103919.	1.3	5
39	A new biallelic DNA polymorphism of the human COL5A1 gene. Human Genetics, 1995, 95, 599-600.	3.8	4
40	The Prognosis of Elderly Patients with Aortic Stenosis after Transcatheter Aortic Valve Replacement. Internal Medicine, 2021, 60, 517-523.	0.7	4
41	A de novo 11p12-p15.4 duplication in a patient with pharmacoresistant epilepsy, mental retardation, and dysmorphisms. Brain and Development, 2010, 32, 248-252.	1.1	3
42	Diagnosis and Treatment of Peripheral Arterial Disease in CKD Patients. Seminars in Dialysis, 2013, 26, 240-251.	1.3	2