Giorgio Gimelli

List of Publications by Year in descending order

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Version: 2024-02-01

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	The Prognosis of Elderly Patients with Aortic Stenosis after Transcatheter Aortic Valve Replacement. Internal Medicine, 2021, 60, 517-523.	0.7	4
2	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
3	1p31.1 microdeletion including only NEGR1 gene in two patients. European Journal of Medical Genetics, 2020, 63, 103919.	1.3	5
4	Clinical and molecular characterization of a patient with interstitial 6q21q22.1 deletion. Molecular Cytogenetics, 2015, 8, 31.	0.9	17
5	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
6	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
7	Heterogeneous clinical presentation in ICF syndrome: correlation with underlying gene defects. European Journal of Human Genetics, 2013, 21, 1219-1225.	2.8	115
8	Diagnosis and Treatment of Peripheral Arterial Disease in CKD Patients. Seminars in Dialysis, 2013, 26, 240-251.	1.3	2
9	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
10	Genotype-Phenotype Correlation of 2q37 Deletions Including NPPC Gene Associated with Skeletal Malformations. PLoS ONE, 2013, 8, e66048.	2.5	32
11	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
12	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
13	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
14	Mutations in SOX17 are associated with congenital anomalies of the kidney and the urinary tract. Human Mutation, 2010, 31, 1352-1359.	2.5	54
15	Altered Intra-Nuclear Organisation of Heterochromatin and Genes in ICF Syndrome. PLoS ONE, 2010, 5, e11364.	2.5	25
16	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
17	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
18	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

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19	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509
20	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
21	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
22	Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572.	2.9	173
23	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
24	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
25	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
26	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
27	Direct duplication 12p11.21â€“p13.31 mediated by segmental duplications: a new recurrent rearrangement?. Human Genetics, 2005, 118, 207-213.	3.8	6
28	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
29	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
30	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
31	T-cell apoptosis in ICF syndrome. Journal of Allergy and Clinical Immunology, 2001, 108, 310-312.	2.9	17
32	Olfactory Receptorâ€“Gene Clusters, Genomic-Inversion Polymorphisms, and Common Chromosome Rearrangements. American Journal of Human Genetics, 2001, 68, 874-883.	6.2	338
33	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
34	Late acute thrombosis after coronary brachytherapy: When is the risk over?. Catheterization and Cardiovascular Interventions, 2001, 54, 216-218.	1.7	10
35	Genetic variation in ICF syndrome: Evidence for genetic heterogeneity. Human Mutation, 2000, 16, 509-517.	2.5	92
36	A neocentromere in the DAZ region of the human Y chromosome. Chromosoma, 2000, 109, 318-327.	2.2	21

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37	CENP-G in neocentromeres and inactive centromeres. Chromosoma, 2000, 109, 328-333.	2.2	26
38	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
39	Transmission of a Fully Functional Human Neocentromere through Three Generations. American Journal of Human Genetics, 1999, 64, 1440-1444.	6.2	113
40	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
41	A new biallelic DNA polymorphism of the human COL5A1 gene. Human Genetics, 1995, 95, 599-600.	3.8	4
42	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