Luigi Bisceglia

List of Publications by Year
in descending order

55 papers	3,367 citations	257450 24 h-index	182427 51 g-index
55 all docs	55 docs citations	55 times ranked	3618 citing authors

#	Article	IF	Citations
1	The putative forkhead transcription factor FOXL2 is mutated in blepharophimosis/ptosis/epicanthus inversus syndrome. Nature Genetics, 2001, 27, 159-166.	21.4	886
2	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. Nature Genetics, 1999, 23, 52-57.	21.4	280
3	Comparison between SLC3A1 and SLC7A9 Cystinuria Patients and Carriers. Journal of the American Society of Nephrology: JASN, 2002, 13, 2547-2553.	6.1	234
4	MYO6, the Human Homologue of the Gene Responsible for Deafness in Snellâ€™s Waltzer Mice, Is Mutated in Autosomal Dominant Nonsyndromic Hearing Loss. American Journal of Human Genetics, 2001, 69, 635-640.	6.2	212
5	New insights into cystinuria: 40 new mutations, genotype-phenotype correlation, and digenic inheritance causing partial phenotype. Journal of Medical Genetics, 2005, 42, 58-68.	3.2	140
6	Functional analysis of mutations in SLC7A9, and genotypeâ€“phenotype correlation in non-Type I cystinuria. Human Molecular Genetics, 2001, 10, 305-316.	2.9	125
7	Genetic Heterogeneity in Italian Families with IgA Nephropathy: Suggestive Linkage for Two Novel IgA Nephropathy Loci. American Journal of Human Genetics, 2006, 79, 1130-1134.	6.2	111
8	VSX1Mutational Analysis in a Series of Italian Patients Affected by Keratoconus: Detection of a Novel Mutation. , 2005, 46, 39.		109
9	Genetic heterogeneity in cystinuria: the SLC3A1 gene is linked to type I but not to type III cystinuria.. Proceedings of the National Academy of Sciences of the United States of America, 1995, 92, 9667-9671.	7.1	98
10	Linkage Analysis in Keratoconus: Replication of Locus 5q21.2 and Identification of Other Suggestive Loci. , 2009, 50, 1081.		97
11	Mutational screening of VSX1, SPARC, SOD1, LOX, and TIMP3 in keratoconus. Molecular Vision, 2011, 17, 2482-94.	1.1	76
12	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. Journal of Inherited Metabolic Disease, 2013, 36, 43-53.	3.6	70
13	Analysis of the complete coding region of the CFTR gene in a cohort of CF patients from North-Eastern Italy: identification of 90% of the mutations. Human Genetics, 1995, 95, 397-402.	3.8	56
14	Hereditary Hyperferritinemia-Cataract Syndrome Caused by a 29-Base Pair Deletion in the Iron Responsive Element of Ferritin L-Subunit Gene. Blood, 1997, 90, 2084-2088.	1.4	54
15	Expression ofVSX1in Human Corneal Keratocytes during Differentiation into Myofibroblasts in Response to Wound Healing. , 2006, 47, 5243.		53
16	WT1 mutations in nephrotic syndrome revisited. High prevalence in young girls, associations and renal phenotypes. Pediatric Nephrology, 2006, 21, 1393-1398.	1.7	46
17	Molecular analysis of the cystinuria disease gene: identification of four new mutations, one large deletion, and one polymorphism. Human Genetics, 1996, 98, 447-451.	3.8	44
18	Role of interferon-Î³ gene polymorphisms in susceptibility to IgA nephropathy: a family-based association study. European Journal of Human Genetics, 2006, 14, 488-496.	2.8	43

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19	Mitochondrial DNA copy number differentiates the Leberâ€™s hereditary optic neuropathy affected individuals from the unaffected mutation carriers. Brain, 2016, 139, e1-e1.	7.6	39
20	Clinical, biochemical and molecular characterization of Cystinuria in a cohort of 12 patients. Clinical Genetics, 2012, 81, 47-55.	2.0	35
21	Large rearrangements detected by MLPA, point mutations, and survey of the frequency of mutations within the SLC3A1 and SLC7A9 genes in a cohort of 172 cystinuric Italian patients. Molecular Genetics and Metabolism, 2010, 99, 42-52.	1.1	34
22	Molecular Analysis of NPHS2 and ACTN4 Genes in a Series of 33 Italian Patients Affected by Adult-Onset Nonfamilial Focal Segmental Glomerulosclerosis. Nephron Clinical Practice, 2005, 99, c31-c36.	2.3	33
23	Genetic variant of C1GalT1 contributes to the susceptibility to IgA nephropathy. Journal of Nephrology, 2009, 22, 152-9.	2.0	30
24	Development of RNA-SSCP protocols for the identification and screening of CFTR mutations: Identification of two new mutations. Human Mutation, 1994, 4, 136-140.	2.5	27
25	Stability and functional effectiveness of phosphorothioate modified duplex DNA and synthetic â€˜mini-genesâ€™. Nucleic Acids Research, 1995, 23, 4134-4142.	14.5	27
26	A new locus on 3p23â€“p25 for an autosomal-dominant limb-girdle muscular dystrophy, LGMD1H. European Journal of Human Genetics, 2010, 18, 636-641.	2.8	27
27	Expanding the mutation spectrum in 130 probands with ARPKD: identification of 62 novel PKHD1 mutations by sanger sequencing and MLPA analysis. Journal of Human Genetics, 2016, 61, 811-821.	2.3	27
28	Genetic history of cystic fibrosis mutations in Italy. I. Regional distribution. Annals of Human Genetics, 1997, 61, 411-424.	0.8	26
29	Mitochondrial DNA copy number in affected and unaffected LHON mutation carriers. BMC Research Notes, 2018, 11, 911.	1.4	25
30	Detection of two novel large deletions inSLC3A1 by semi-quantitative fluorescent multiplex PCR. Human Mutation, 2000, 15, 373-379.	2.5	24
31	Cystinuria type I: Identification of eight new mutations in SLC3A1. Kidney International, 2001, 59, 1250-1256.	5.2	24
32	The IgA nephropathy Biobank. An important starting point for the genetic dissection of a complex trait. BMC Nephrology, 2005, 6, 14.	1.8	24
33	20 novel point mutations and one large deletion in EXT1 and EXT2 genes: Report of diagnostic screening in a large Italian cohort of patients affected by hereditary multiple exostosis. Gene, 2013, 515, 339-348.	2.2	24
34	Genomic Structure and Organization of the HumanrBATGene (SLC3A1). Genomics, 1996, 37, 249-252.	2.9	22
35	The Missing ApoE Allele. Annals of Human Genetics, 2007, 71, 496-500.	0.8	21
36	Posttransplant Recurrence of Proteinuria in a Case of Focal Segmental Glomerulosclerosis Associated with WT1 Mutation. American Journal of Transplantation, 2006, 6, 2208-2211.	4.7	19

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37	Twenty-Four Novel Mutations Identified in a Cohort of 85 Patients by Direct Sequencing of the SLC3A1 and SLC7A9 Cystinuria Genes. Genetic Testing and Molecular Biomarkers, 2008, 12, 351-355.	1.7	16
38	The motilin gene: subregional localisation, tissue expression, DNA polymorphisms and exclusion as a candidate gene for the HLA-associated immotile cilia syndrome. Human Genetics, 1994, 94, 671-4.	3.8	15
39	Identification and Clinical Characterization of Adult Patients with Multigenerational Diabetes Mellitus. PLoS ONE, 2015, 10, e0135855.	2.5	14
40	The molecular basis of cystinuria: the role of the rBAT gene. Amino Acids, 1996, 11, 225-246.	2.7	13
41	Clinical utility gene card for: Cystinuria. European Journal of Human Genetics, 2012, 20, 3-3.	2.8	12
42	Recombinant Families Locate the Gene for Non-Type I Cystinuria between Markers C13 and D19S587 on Chromosome 19q13.1. Genomics, 1999, 60, 362-365.	2.9	9
43	Detection of dystrophin deletion carriers using FISH analysis. Clinical Genetics, 1997, 52, 17-22.	2.0	9
44	An overview of SLC3A1 and SLC7A9 mutations in Greek cystinuria patients. Molecular Genetics and Metabolism, 2008, 95, 192-193.	1.1	9
45	Molecular analysis in a family presenting with a mild form of late-onset autosomal dominant chronic progressive external ophthalmoplegia. Neuromuscular Disorders, 2009, 19, 423-426.	0.6	8
46	Polymorphism of motilin gene in patients with Crohn's disease. Digestive Diseases and Sciences, 1998, 43, 715-719.	2.3	7
47	Leberâ€™s hereditary optic neuropathy, intellectual disability and epilepsy presenting with variable penetrance associated to the m.3460Gâ€‰>A mutation and a heteroplasmic expansion of the microsatellite in MTRNR1 gene â€“ case report. BMC Medical Genetics, 2018, 19, 129.	2.1	7
48	A YAC Contig Spanning the Blepharophimosis-Ptosis- Epicanthus inversus Syndrome and Propionic Acidemia Loci. European Journal of Human Genetics, 1997, 5, 171-174.	2.8	7
49	Leber's hereditary optic neuropathy (LHON) in an Apulian cohort of subjects. Acta Myologica, 2017, 36, 163-177.	1.5	7
50	Phenotype characterization and prevalence of rBAT M467T mutation in Italian cystinuric patients. Journal of Inherited Metabolic Disease, 1996, 19, 243-245.	3.6	5
51	Molecular screening of genetic defects with RNAâ€“SSCP analysis: the PKU and cystinuria model. Molecular and Cellular Probes, 1995, 9, 201-205.	2.1	3
52	Cellular Retinol Binding Protein 1 (RBP1): A frequent polymorphism, refined map position and exclusion as the Blepharophimosis Ptosis Epicanthus inversus Syndrome gene. Molecular and Cellular Probes, 1998, 12, 255-258.	2.1	2
53	Homozygosity for a novel splice site mutation (2790-2 Aâ†’G) preceding exon 15 of the CFTR gene in a cystic fibrosis patient of North-East Italian descent. Molecular and Cellular Probes, 1995, 9, 139-141.	2.1	1
54	IndirectCFTRMutation Identification by PCR/OLA Anomalous Electropherograms. Genetic Testing and Molecular Biomarkers, 2005, 9, 285-291.	1.7	1

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55	Human Gene Mutations. Human Genetics, 2007, 122, 207-215.	3.8	0