Valentina Guida

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/9070568/publications.pdf

Version: 2024-02-01

40 papers	2,660 citations	430874 18 h-index	289244 40 g-index
40 all docs	40 docs citations	40 times ranked	3269 citing authors

#	Article	IF	Citations
1	Tracing European Founder Lineages in the Near Eastern mtDNA Pool. American Journal of Human Genetics, 2000, 67, 1251-1276.	6.2	837
2	The Emerging Tree of West Eurasian mtDNAs: A Synthesis of Control-Region Sequences and RFLPs. American Journal of Human Genetics, 1999, 64, 232-249.	6.2	549
3	Phylogeographic Analysis of Haplogroup E3b (E-M215) Y Chromosomes Reveals Multiple Migratory Events Within and Out Of Africa. American Journal of Human Genetics, 2004, 74, 1014-1022.	6.2	197
4	Do the Four Clades of the mtDNA Haplogroup L2 Evolve at Different Rates?. American Journal of Human Genetics, 2001, 69, 1348-1356.	6.2	185
5	Familial transposition of the great arteries caused by multiple mutations in laterality genes. Heart, 2010, 96, 673-677.	2.9	126
6	A Functional Variant of the Adipocyte Glycerol Channel Aquaporin 7 Gene Is Associated With Obesity and Related Metabolic Abnormalities. Diabetes, 2007, 56, 1468-1474.	0.6	108
7	Novel and recurrent EVC and EVC2 mutations in Ellis-van Creveld syndrome and Weyers acrofacial dyostosis. European Journal of Medical Genetics, 2013, 56, 80-87.	1.3	64
8	GLI1 inactivation is associated with developmental phenotypes overlapping with Ellisâ€“van Creveld syndrome. Human Molecular Genetics, 2017, 26, 4556-4571.	2.9	50
9	Reliability of DHPLC in mutational screening of ?-globin (HBB) alleles. Human Mutation, 2002, 19, 287-295.	2.5	48
10	Molecular detection of novel WFS1 mutations in patients with Wolfram syndrome by a DHPLC-based assay. Human Mutation, 2003, 21, 622-629.	2.5	47
11	Screening of mutations in the CFTR gene in 1195 couples entering assisted reproduction technique programs. European Journal of Human Genetics, 2005, 13, 959-964.	2.8	46
12	Application of MLPA assay to characterize unsolved Î±-globin gene rearrangements. Blood Cells, Molecules, and Diseases, 2011, 46, 139-144.	1.4	39
13	Ebstein anomaly: Genetic heterogeneity and association with microdeletions 1p36 and 8p23.1. American Journal of Medical Genetics, Part A, 2011, 155, 2196-2202.	1.2	38
14	A variant in the carboxyl-terminus of connexin 40 alters GAP junctions and increases risk for tetralogy of Fallot. European Journal of Human Genetics, 2013, 21, 69-75.	2.8	36
15	Functional analysis of splicing mutations in exon 7 of NF1gene. BMC Medical Genetics, 2007, 8, 4.	2.1	32
16	Design of novel three-phase PCL/TZâ€“HA biomaterials for use in bone regeneration applications. Journal of Materials Science: Materials in Medicine, 2010, 21, 2569-2581.	3.6	30
17	Interaction of DIO2 T92A and PPARÎ³2 P12A Polymorphisms in the Modulation of Metabolic Syndrome**. Obesity, 2007, 15, 2889-2895.	3.0	24
18	A de novo proximal 3q29 chromosome microduplication in a patient with oculo auriculo vertebral spectrum. American Journal of Medical Genetics, Part A, 2015, 167, 797-801.	1.2	21

#	Article	IF	Citations
19	Novel Î±â€Actin Gene Mutation p.(Ala21Val) Causing Familial Hypertrophic Cardiomyopathy, Myocardial Noncompaction, and Transmural Crypts. Clinicalâ€Pathologic Correlation. Journal of the American Heart Association, 2018, 7, .	3.7	18
20	Genome-Wide DNA Methylation Analysis of a Cohort of 41 Patients Affected by Oculo-Auriculo-Vertebral Spectrum (OAVS). International Journal of Molecular Sciences, 2021, 22, 1190.	4.1	16
21	Analysis of TP53 codon 72 polymorphism in HPV-positive and HPV-negative penile carcinoma. Cancer Letters, 2008, 269, 159-164.	7.2	15
22	Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect. Human Mutation, 2018, 39, 1428-1441.	2.5	15
23	Sequence-specific modification of a Â-thalassemia locus by small DNA fragments in human erythroid progenitor cells. Haematologica, 2007, 92, 129-130.	3.5	11
24	Oculodentodigital Dysplasia with Massive Brain Calcification and a New Mutation of GJA1 Gene. Journal of Alzheimer's Disease, 2015, 49, 27-30.	2.6	11
25	A new case of SMABF2 diagnosed in stillbirth expands the prenatal presentation and mutational spectrum of ASCC1. American Journal of Medical Genetics, Part A, 2020, 182, 508-512.	1.2	10
26	JAG1 Mutation in a patient with deletion 22q11.2 syndrome and tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2013, 161, 3133-3136.	1.2	9
27	Copy number variation analysis implicates novel pathways in patients with oculoâ€auriculoâ€vertebralâ€spectrum and congenital heart defects. Clinical Genetics, 2021, 100, 268-279.	2.0	9
28	Denaturing HPLC-Based Assay for Molecular Screening of Nondeletional Mutations Causing Î±-Thalassemias. Clinical Chemistry, 2004, 50, 1242-1245.	3.2	8
29	Letters to the Editor. Clinical Genetics, 2004, 66, 478-480.	2.0	8
30	Multiplex Ligation-Dependent Probe Amplification Analysis ofGATA4Gene Copy Number Variations in Patients with Isolated Congenital Heart Disease. Disease Markers, 2010, 28, 287-292.	1.3	8
31	Validation of dHPLC for Molecular Diagnosis of Î²-Thalassemia in Southern Italy. Genetic Testing and Molecular Biomarkers, 2003, 7, 269-275.	1.7	7
32	Common atrium/atrioventricular canal defect and postaxial polydactyly: A mild clinical subtype of Ellisâ€van Creveld syndrome caused by hypomorphic mutations in the EVC gene. Human Mutation, 2020, 41, 2087-2093.	2.5	7
33	Delineation of MidXq28â€duplication syndrome distal to MECP2 and proximal to RAB39B genes. Clinical Genetics, 2019, 96, 246-253.	2.0	6
34	Novel dilated cardiomyopathy associated to Calreticulin and Myo7A gene mutation in Usher syndrome. ESC Heart Failure, 2021, 8, 2310-2315.	3.1	6
35	Use of DHPLC for Rapid Screening of Recombinant Clones. BioTechniques, 2003, 34, 706-708.	1.8	5
36	Detection of a Rare Î²-Globin Nonsense Mutation [Codon 59 (AAGâ†'TAG)] in an Italian Family. Hemoglobin, 2006, 30, 405-407.	0.8	4

#	Article	IF	Citations
37	Atrioventricular canal defect as partial expression of heterotaxia in patients with Bardet-Biedl syndrome. Journal of Pediatrics, 2020, 218, 263-264.	1.8	4
38	Lack of pathogenic mutations in SOS1 gene in phenytoin-induced gingival overgrowth patients. Archives of Oral Biology, 2017, 80, 160-163.	1.8	3
39	Extrachromosomal genes: a powerful tool in gene targeting approaches. Gene Therapy, 2002, 9, 679-682.	4.5	2
40	Neonatal Marfan Syndrome by Inherited Mutation. Indian Journal of Pediatrics, 2021, 88, 176-177.	0.8	1