

# Valentina Guida

## List of Publications by Year in descending order

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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	Neonatal Marfan Syndrome by Inherited Mutation. <i>Indian Journal of Pediatrics</i> , 2021, 88, 176-177.	0.8	1
2	Genome-Wide DNA Methylation Analysis of a Cohort of 41 Patients Affected by Oculo-Auriculo-Vertebral Spectrum (OAVS). <i>International Journal of Molecular Sciences</i> , 2021, 22, 1190.	4.1	16
3	Novel dilated cardiomyopathy associated to <i>Calreticulin</i> and <i>Myo7A</i> gene mutation in Usher syndrome. <i>ESC Heart Failure</i> , 2021, 8, 2310-2315.	3.1	6
4	Copy number variation analysis implicates novel pathways in patients with oculo-auriculo-vertebral spectrum and congenital heart defects. <i>Clinical Genetics</i> , 2021, 100, 268-279.	2.0	9
5	A new case of SMABF2 diagnosed in stillbirth expands the prenatal presentation and mutational spectrum of <i>ASCC1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 508-512.	1.2	10
6	Atrioventricular canal defect as partial expression of heterotaxia in patients with Bardet-Biedl syndrome. <i>Journal of Pediatrics</i> , 2020, 218, 263-264.	1.8	4
7	Common atrium/atrioventricular canal defect and postaxial polydactyly: A mild clinical subtype of Ellis-van Creveld syndrome caused by hypomorphic mutations in the <i>EVC</i> gene. <i>Human Mutation</i> , 2020, 41, 2087-2093.	2.5	7
8	Delineation of MidXq28 duplication syndrome distal to MECP2 and proximal to RAB39B genes. <i>Clinical Genetics</i> , 2019, 96, 246-253.	2.0	6
9	Novel $\beta$ -Actin Gene Mutation p.(Ala21Val) Causing Familial Hypertrophic Cardiomyopathy, Myocardial Noncompaction, and Transmural Crypts. <i>Clinical Pathologic Correlation. Journal of the American Heart Association</i> , 2018, 7, .	3.7	18
10	Heterozygous missense mutations in <i>NFATC1</i> are associated with atrioventricular septal defect. <i>Human Mutation</i> , 2018, 39, 1428-1441.	2.5	15
11	Lack of pathogenic mutations in <i>SOS1</i> gene in phenytoin-induced gingival overgrowth patients. <i>Archives of Oral Biology</i> , 2017, 80, 160-163.	1.8	3
12	GLI1 inactivation is associated with developmental phenotypes overlapping with Ellis-van Creveld syndrome. <i>Human Molecular Genetics</i> , 2017, 26, 4556-4571.	2.9	50
13	Oculodentodigital Dysplasia with Massive Brain Calcification and a New Mutation of <i>GJA1</i> Gene. <i>Journal of Alzheimer's Disease</i> , 2015, 49, 27-30.	2.6	11
14	A de novo proximal 3q29 chromosome microduplication in a patient with oculo auriculo vertebral spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 797-801.	1.2	21
15	<i>JAG1</i> Mutation in a patient with deletion 22q11.2 syndrome and tetralogy of Fallot. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 3133-3136.	1.2	9
16	Novel and recurrent <i>EVC</i> and <i>EVC2</i> mutations in Ellis-van Creveld syndrome and Weyers acrofacial dysostosis. <i>European Journal of Medical Genetics</i> , 2013, 56, 80-87.	1.3	64
17	A variant in the carboxyl-terminus of connexin 40 alters GAP junctions and increases risk for tetralogy of Fallot. <i>European Journal of Human Genetics</i> , 2013, 21, 69-75.	2.8	36
18	Application of MLPA assay to characterize unsolved $\beta$ -globin gene rearrangements. <i>Blood Cells, Molecules, and Diseases</i> , 2011, 46, 139-144.	1.4	39

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19	Ebstein anomaly: Genetic heterogeneity and association with microdeletions 1p36 and 8p23.1. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2196-2202.	1.2	38
20	Design of novel three-phase PCL/TZâ€“HA biomaterials for use in bone regeneration applications. <i>Journal of Materials Science: Materials in Medicine</i> , 2010, 21, 2569-2581.	3.6	30
21	Familial transposition of the great arteries caused by multiple mutations in laterality genes. <i>Heart</i> , 2010, 96, 673-677.	2.9	126
22	Multiplex Ligation-Dependent Probe Amplification Analysis of <i>GATA4</i> Gene Copy Number Variations in Patients with Isolated Congenital Heart Disease. <i>Disease Markers</i> , 2010, 28, 287-292.	1.3	8
23	Analysis of TP53 codon 72 polymorphism in HPV-positive and HPV-negative penile carcinoma. <i>Cancer Letters</i> , 2008, 269, 159-164.	7.2	15
24	A Functional Variant of the Adipocyte Glycerol Channel Aquaporin 7 Gene Is Associated With Obesity and Related Metabolic Abnormalities. <i>Diabetes</i> , 2007, 56, 1468-1474.	0.6	108
25	Sequence-specific modification of a $\hat{A}$ -thalassemia locus by small DNA fragments in human erythroid progenitor cells. <i>Haematologica</i> , 2007, 92, 129-130.	3.5	11
26	Functional analysis of splicing mutations in exon 7 of NF1 gene. <i>BMC Medical Genetics</i> , 2007, 8, 4.	2.1	32
27	Interaction of DIO2 T92A and PPAR $\hat{3}$ 2 P12A Polymorphisms in the Modulation of Metabolic Syndrome**. <i>Obesity</i> , 2007, 15, 2889-2895.	3.0	24
28	Detection of a Rare $\hat{2}$ -Globin Nonsense Mutation [Codon 59 (AAG $\hat{+}$ TAG)] in an Italian Family. <i>Hemoglobin</i> , 2006, 30, 405-407.	0.8	4
29	Screening of mutations in the CFTR gene in 1195 couples entering assisted reproduction technique programs. <i>European Journal of Human Genetics</i> , 2005, 13, 959-964.	2.8	46
30	Denaturing HPLC-Based Assay for Molecular Screening of Nondeletional Mutations Causing $\hat{1}$ -Thalassemias. <i>Clinical Chemistry</i> , 2004, 50, 1242-1245.	3.2	8
31	Letters to the Editor. <i>Clinical Genetics</i> , 2004, 66, 478-480.	2.0	8
32	Phylogeographic Analysis of Haplogroup E3b (E-M215) Y Chromosomes Reveals Multiple Migratory Events Within and Out Of Africa. <i>American Journal of Human Genetics</i> , 2004, 74, 1014-1022.	6.2	197
33	Molecular detection of novel WFS1 mutations in patients with Wolfram syndrome by a DHPLC-based assay. <i>Human Mutation</i> , 2003, 21, 622-629.	2.5	47
34	Validation of dHPLC for Molecular Diagnosis of $\hat{2}$ -Thalassemia in Southern Italy. <i>Genetic Testing and Molecular Biomarkers</i> , 2003, 7, 269-275.	1.7	7
35	Use of DHPLC for Rapid Screening of Recombinant Clones. <i>BioTechniques</i> , 2003, 34, 706-708.	1.8	5
36	Extrachromosomal genes: a powerful tool in gene targeting approaches. <i>Gene Therapy</i> , 2002, 9, 679-682.	4.5	2

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37	Reliability of DHPLC in mutational screening of $\beta$ -globin (HBB) alleles. Human Mutation, 2002, 19, 287-295.	2.5	48
38	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
39	Tracing European Founder Lineages in the Near Eastern mtDNA Pool. American Journal of Human Genetics, 2000, 67, 1251-1276.	6.2	837
40	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