Alessandro De Luca

List of Publications by Year
in descending order

115 papers	4,883 citations	101543 36 h-index	106344 65 g-index
116 all docs	116 docs citations	116 times ranked	7782 citing authors

#	Article	IF	Citations
1	â€œSpazio Huntingtonâ€: Tracing the Early Motor, Cognitive and Behavioral Profiles of Kids with Proven Pediatric Huntington Disease and Expanded Mutations > 80 CAG Repeats. Journal of Personalized Medicine, 2022, 12, 120.	2.5	1
2	Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. European Journal of Human Genetics, 2021, 29, 51-60.	2.8	17
3	The clinical significance of A2ML1 variants in Noonan syndrome has to be reconsidered. European Journal of Human Genetics, 2021, 29, 524-527.	2.8	7
4	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
5	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. Genetics in Medicine, 2021, 23, 1116-1124.	2.4	17
6	A Novel Triplet-Primed PCR Assay to Detect the Full Range of Trinucleotide CAG Repeats in the Huntingtin Gene (HTT). International Journal of Molecular Sciences, 2021, 22, 1689.	4.1	8
7	Enlarged spinal nerve roots in RASopathies: Report of two cases. European Journal of Medical Genetics, 2021, 64, 104187.	1.3	2
8	Novel dilated cardiomyopathy associated to Calreticulin and Myo7A gene mutation in Usher syndrome. ESC Heart Failure, 2021, 8, 2310-2315.	3.1	6
9	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
10	Cardiac Defects and Genetic Syndromes: Old Uncertainties and New Insights. Genes, 2021, 12, 1047.	2.4	12
11	Clinical variability of neurofibromatosis 1: A modifying role of cooccurring <scp>PTPN11</scp> variants and atypical brain <scp>MRI</scp> findings. Clinical Genetics, 2021, 100, 563-572.	2.0	6
12	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	6.2	23
13	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
14	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
15	Atrioventricular canal defect as partial expression of heterotaxia in patients with Bardet-Biedl syndrome. Journal of Pediatrics, 2020, 218, 263-264.	1.8	4
16	Tracing the mutated HTT and haplotype of the African ancestor who spread Huntington disease into the Middle East. Genetics in Medicine, 2020, 22, 1903-1908.	2.4	8
17	The Global Emergency of Novel Coronavirus (SARS-CoV-2): An Update of the Current Status and Forecasting. International Journal of Environmental Research and Public Health, 2020, 17, 5648.	2.6	49
18	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48

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19	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
20	Low-Grade Gliomas in Patients with Noonan Syndrome: Case-Based Review of the Literature. Diagnostics, 2020, 10, 582.	2.6	21
21	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	2.4	28
22	Martsolf syndrome with novel mutation in the TBC1D20 gene in a family from Iran. American Journal of Medical Genetics, Part A, 2020, 182, 957-961.	1.2	6
23	Genetic factors in rotator cuff pathology: potential influence of col 5A1 polymorphism in outcomes of rotator cuff repair. BMC Medical Genetics, 2020, 21, 82.	2.1	3
24	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. American Journal of Human Genetics, 2019, 105, 640-657.	6.2	31
25	Isoform-specific NF1 mRNA levels correlate with disease severity in Neurofibromatosis type 1. Orphanet Journal of Rare Diseases, 2019, 14, 261.	2.7	15
26	Prevalence, Type, and Molecular Spectrum of NF1 Mutations in Patients with Neurofibromatosis Type 1 and Congenital Heart Disease. Genes, 2019, 10, 675.	2.4	13
27	Generation of induced pluripotent stem cell line CSSi008-A (4698) from a patient affected by advanced stage of Dentato-Rubral-Pallidoluysian atrophy (DRPLA). Stem Cell Research, 2019, 40, 101551.	0.7	8
28	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508.	6.2	48
29	Clinical and functional characterization of a novel RASopathyâ€causingSHOC2mutation associated with prenatalâ€onset hypertrophic cardiomyopathy. Human Mutation, 2019, 40, 1046-1056.	2.5	18
30	Transplantation of clinical-grade human neural stem cells reduces neuroinflammation, prolongs survival and delays disease progression in the SOD1 rats. Cell Death and Disease, 2019, 10, 345.	6.3	28
31	Mutational spectrum and clinical signatures in 114 families with hereditary multiple osteochondromas: insights into molecular properties of selected exostosin variants. Human Molecular Genetics, 2019, 28, 2133-2142.	2.9	12
32	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. European Journal of Human Genetics, 2019, 27, 1061-1071.	2.8	11
33	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
34	Atrioventricular canal defect and genetic syndromes: The unifying role of sonic hedgehog. Clinical Genetics, 2019, 95, 268-276.	2.0	20
35	Shagreen-patch in a NF1 patient with a new missense mutation. Giornale Italiano Di Dermatologia E Venereologia, 2019, 154, 223-224.	0.8	0
36	Generation of induced pluripotent stem cell line, CSSi004-A (2962), from a patient diagnosed with Huntington's disease at the presymptomatic stage. Stem Cell Research, 2018, 28, 145-148.	0.7	7

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37	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
38	Generation of induced pluripotent stem cell line, CSSi002-A (2851), from a patient with juvenile Huntington Disease. Stem Cell Research, 2018, 27, 86-89.	0.7	5
39	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87.	6.2	144
40	Generation of the induced pluripotent stem cell line CSSi006-A (3681) from a patient affected by advanced-stage Juvenile Onset Huntington's Disease. Stem Cell Research, 2018, 29, 174-178.	0.7	1
41	Biallelic mutations in <scp>DYNC2LI1</scp> are a rare cause of Ellisâ€van Creveld syndrome. Clinical Genetics, 2018, 93, 632-639.	2.0	23
42	Genetics of rotator cuff tears: no association of col5a1 gene in a case-control study. BMC Medical Genetics, 2018, 19, 217.	2.1	20
43	Biological and clinical manifestations of juvenile Huntington's disease: a retrospective analysis. Lancet Neurology, The, 2018, 17, 986-993.	10.2	115
44	Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect. Human Mutation, 2018, 39, 1428-1441.	2.5	15
45	Paroxysmal Dyskinesias in a Mutation Carrier. Tremor and Other Hyperkinetic Movements, 2018, 8, 616.	2.0	2
46	Structural, Functional, and Clinical Characterization of a NovelPTPN11Mutation Cluster Underlying Noonan Syndrome. Human Mutation, 2017, 38, 451-459.	2.5	39
47	Lack of pathogenic mutations in SOS1 gene in phenytoin-induced gingival overgrowth patients. Archives of Oral Biology, 2017, 80, 160-163.	1.8	3
48	GLI1 inactivation is associated with developmental phenotypes overlapping with Ellisâ€“van Creveld syndrome. Human Molecular Genetics, 2017, 26, 4556-4571.	2.9	50
49	Identification of a second <scp>HOXA2</scp> nonsense mutation in a family with autosomal dominant nonâ€syndromic microtia and distinctive ear morphology. Clinical Genetics, 2017, 91, 774-779.	2.0	28
50	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
51	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	2.5	67
52	Noonan syndromeâ€like disorder with loose anagen hair: A second case with neuroblastoma. American Journal of Medical Genetics, Part A, 2015, 167, 1902-1907.	1.2	14
53	Myoclonic status and central fever in Angelman syndrome due to paternal uniparental disomy. Journal of Neurogenetics, 2015, 29, 178-182.	1.4	1
54	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

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55	Specific variants in WDR35 cause a distinctive form of Ellis-van Creveld syndrome by disrupting the recruitment of the EvC complex and SMO into the cilium. Human Molecular Genetics, 2015, 24, 4126-4137.	2.9	42
56	Molecular Diversity and Associated Phenotypic Spectrum of GermlineCBLMutations. Human Mutation, 2015, 36, 787-796.	2.5	36
57	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. European Journal of Human Genetics, 2015, 23, 1068-1071.	2.8	113
58	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. Human Molecular Genetics, 2014, 23, 4315-4327.	2.9	114
59	Novel homozygous mutation in exon 5 of <scp>WFS1</scp> gene in an Apulian family with mild phenotypic expression of Wolfram syndrome. Clinical Genetics, 2014, 86, 197-198.	2.0	3
60	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
61	Congenital heart defects in recurrent reciprocal 1q21.1 deletion and duplication syndromes: Rare association with pulmonary valve stenosis. European Journal of Medical Genetics, 2013, 56, 144-149.	1.3	33
62	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
63	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
64	Loss of <scp>CBL</scp> E3â€ligase activity in Bâ€lineage childhood acute lymphoblastic leukaemia. British Journal of Haematology, 2012, 159, 115-119.	2.5	6
65	Pentalogy of Cantrell with Complete Ectopia Cordis in a Fetus with Asplenia. Pediatric and Developmental Pathology, 2012, 15, 495-498.	1.0	8
66	Search of somatic GATA4 and NKX2.5 gene mutations in sporadic septal heart defects. European Journal of Medical Genetics, 2011, 54, 306-309.	1.3	35
67	Novel and recurrent JAG1 mutations in patients with tetralogy of Fallot. Clinical Genetics, 2011, 80, 591-594.	2.0	11
68	New mutations inZFPM2/FOG2gene in tetralogy of Fallot and double outlet right ventricle. Clinical Genetics, 2011, 80, 184-190.	2.0	69
69	Familial spinal neurofibromatosis due to a multiexonic NF1 gene deletion. Neurogenetics, 2011, 12, 233-240.	1.4	8
70	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
71	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. Human Mutation, 2011, 32, 760-772.	2.5	97
72	Heterozygous Germline Mutations in the CBL Tumor-Suppressor Gene Cause a Noonan Syndrome-like Phenotype. American Journal of Human Genetics, 2010, 87, 250-257.	6.2	221

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73	Germline mosaicism in neurofibromatosis type 1 due to a paternally derived multiâ€exon deletion. American Journal of Medical Genetics, Part A, 2010, 152A, 1467-1473.	1.2	19
74	Duplication of Glu37 in the switch I region of HRAS impairs effector/GAP binding and underlies Costello syndrome by promoting enhanced growth factor-dependent MAPK and AKT activation. Human Molecular Genetics, 2010, 19, 790-802.	2.9	38
75	RASopathies: Clinical Diagnosis in the First Year of Life. Molecular Syndromology, 2010, 1, 282-289.	0.8	73
76	Familial transposition of the great arteries caused by multiple mutations in laterality genes. Heart, 2010, 96, 673-677.	2.9	126
77	Exclusion of Cx43 gene mutation as a major cause of criss-cross heart anomaly in man. International Journal of Cardiology, 2010, 144, 300-302.	1.7	4
78	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
79	Multiple granular cell tumors are an associated feature of LEOPARD syndrome caused by mutation in PTPN11. Clinical Genetics, 2009, 75, 185-189.	2.0	71
80	Founder Effects for ATM Gene Mutations in Italian Ataxia Telangiectasia Families. Annals of Human Genetics, 2009, 73, 532-539.	0.8	17
81	Xenografts of primary human gynecological tumors grown under the renal capsule of NOD/SCID mice show genetic stability during serial transplantation and respond to cytotoxic chemotherapy. Gynecologic Oncology, 2008, 110, 256-264.	1.4	59
82	Ovarian carcinomas with genetic and epigenetic BRCA1 loss have distinct molecular abnormalities. BMC Cancer, 2008, 8, 17.	2.6	254
83	Loss of Heterozygosity at the BRCA2 Locus Detected by Multiplex Ligation-Dependent Probe Amplification is Common in Prostate Cancers from Men with a Germline BRCA2 Mutation. Clinical Cancer Research, 2008, 14, 2953-2961.	7.0	37
84	Deletions of NF1 gene and exons detected by multiplex ligation-dependent probe amplification. Journal of Medical Genetics, 2007, 44, 800-808.	3.2	64
85	Founder and Recurrent CDH1 Mutations in Families With Hereditary Diffuse Gastric Cancer. JAMA - Journal of the American Medical Association, 2007, 297, 2360.	7.4	394
86	Frequency of the TMPRSS2:ERG gene fusion is increased in moderate to poorly differentiated prostate cancers. Journal of Clinical Pathology, 2007, 60, 1238-1243.	2.0	157
87	Clinical lumping and molecular splitting of LEOPARD and NF1/NF1-Noonan syndromes. American Journal of Medical Genetics, Part A, 2007, 143A, 1009-1011.	1.2	13
88	Functional analysis of splicing mutations in exon 7 of NF1gene. BMC Medical Genetics, 2007, 8, 4.	2.1	32
89	DHPLC Screening ofATMGene in Italian Patients Affected by Ataxia-Telangiectasia: Fourteen NovelATMMutations. Disease Markers, 2006, 22, 257-264.	1.3	15
90	MDM2 protein expression is a negative prognostic marker in breast carcinoma. Modern Pathology, 2006, 19, 69-74.	5.5	62

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91	A landscape effect in tenosynovial giant-cell tumor from activation of CSF1 expression by a translocation in a minority of tumor cells. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 690-695.	7.1	474
92	Combinatorial Sequencing-by-Hybridization: Analysis of theNF1Gene. Genetic Testing and Molecular Biomarkers, 2006, 10, 8-17.	1.7	10
93	Novel EXT1 and EXT2 mutations identified by DHPLC in Italian patients with multiple osteochondromas. Human Mutation, 2005, 26, 280-280.	2.5	43
94	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101.	6.2	139
95	Identification of Type A, B, E, and F Botulinum Neurotoxin Genes and of Botulinum Neurotoxigenic Clostridia by Denaturing High-Performance Liquid Chromatography. Applied and Environmental Microbiology, 2004, 70, 4170-4176.	3.1	10
96	Fluorescence in situ hybridization analysis of allelic losses involving the long arm of chromosome 17 in NF1-associated neurofibromas. Cancer Genetics and Cytogenetics, 2004, 150, 168-172.	1.0	12
97	Variations in the NMDA receptor subunit 2B gene (GRIN2B) and schizophrenia: A caseâ€control study. American Journal of Medical Genetics Part A, 2004, 128B, 27-29.	2.4	33
98	Novel and recurrent mutations in theNF1 gene in Italian patients with neurofibromatosis type 1. Human Mutation, 2004, 23, 629-629.	2.5	42
99	Mutations of the Nogo-66 receptor (RTN4R) gene in schizophrenia. Human Mutation, 2004, 24, 534-535.	2.5	77
100	Mutational analysis of parkin gene by denaturing high-performance liquid chromatography (DHPLC) in essential tremor. Parkinsonism and Related Disorders, 2004, 10, 357-362.	2.2	20
101	Association of dopamine D4 receptor (DRD4) exon III repeat polymorphism with temperament in 3-year-old infants. Neurogenetics, 2003, 4, 207-212.	1.4	40
102	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
103	NF1 gene analysis based on DHPLC. Human Mutation, 2003, 21, 171-172.	2.5	41
104	Association study between CAG trinucleotide repeats in the PCQAP gene (PC2) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 227 Td (glutamine 2003, 116B, 32-35.	2.4	15
105	Validation of dHPLC for Molecular Diagnosis of Î²-Thalassemia in Southern Italy. Genetic Testing and Molecular Biomarkers, 2003, 7, 269-275.	1.7	7
106	Rapid scanning of myotubularin (MTM1) gene by denaturing high-performance liquid chromatography (DHPLC). Neuromuscular Disorders, 2002, 12, 501-505.	0.6	22
107	Reliability of DHPLC in mutational screening of ?-globin (HBB) alleles. Human Mutation, 2002, 19, 287-295.	2.5	48
108	Ten novel mutations in the human neurofibromatosis type 1 (NF1) gene in Italian patients. Human Mutation, 2002, 20, 74-75.	2.5	21

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109	Three novel mutations causing a truncated protein within the RP2 gene in Italian families with X-linked retinitis pigmentosa. Mutation Research - Mutation Research Genomics, 2001, 432, 79-82.	1.1	1
110	Dopamine D4 receptor (DRD4) polymorphism and adaptability trait during infancy: a longitudinal study in 1- to 5-month-old neonates. Neurogenetics, 2001, 3, 79-82.	1.4	33
111	Association study of a promoter polymorphism of UFD1L gene with schizophrenia. American Journal of Medical Genetics Part A, 2001, 105, 529-533.	2.4	37
112	Evidence for an Association between the SRD5A2 (Type II Steroid 5Î±-Reductase) Locus and Prostate Cancer in Italian Patients. Disease Markers, 2000, 16, 147-150.	1.3	32
113	A Novel Mutation (R271X) in the Myotubularin Gene Causes a Severe Miotubular Myopathy. Human Heredity, 1999, 49, 59-60.	0.8	8
114	A single-nucleotide polymorphism in the human bone morphogenetic protein-4 (BMP 4) gene. Journal of Human Genetics, 1999, 44, 76-77.	2.3	19
115	Localization of a Gene for Familial Patella Aplasia-Hypoplasia (PTLAH) to Chromosome 17q21â€“22. American Journal of Human Genetics, 1999, 65, 441-447.	6.2	27