Diana Baralle

List of Publications by Year
in descending order

96 papers	7,163 citations	101496 36 h-index	66879 78 g-index
109 all docs	109 docs citations	109 times ranked	12492 citing authors

#	Article	IF	Citations
1	A severe case of Boschâ€“<scp>Boonstraâ€“Schaaf</scp> optic atrophy syndrome with a novel description of coloboma and septoâ€optic dysplasia, owing to a start codon variant in the <scp>NR2F1</scp> gene. American Journal of Medical Genetics, Part A, 2022, 188, 900-906.	0.7	5
2	Evidence that the Ser192Tyr/Arg402Gln in cis Tyrosinase gene haplotype is a disease-causing allele in oculocutaneous albinism type 1B (OCA1B). Npj Genomic Medicine, 2022, 7, 2.	1.7	9
3	MRSD: A quantitative approach for assessing suitability of RNA-seq in the investigation of mis-splicing in Mendelian disease. American Journal of Human Genetics, 2022, 109, 210-222.	2.6	12
4	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	2.6	13
5	Short amplicon reverse transcriptionâ€polymerase chain reaction detects aberrant splicing in genes with low expression in blood missed by ribonucleic acid sequencing analysis for clinical diagnosis. Human Mutation, 2022, 43, 963-970.	1.1	4
6	A gene-to-patient approach uplifts novel disease gene discovery and identifies 18 putative novel disease genes. Genetics in Medicine, 2022, 24, 1697-1707.	1.1	14
7	CI-SpliceAIâ€”Improving machine learning predictions of disease causing splicing variants using curated alternative splice sites. PLoS ONE, 2022, 17, e0269159.	1.1	15
8	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	3.6	65
9	Essentiality-specific pathogenicity prioritization gene score to improve filtering of disease sequence data. Briefings in Bioinformatics, 2021, 22, 1782-1789.	3.2	3
10	â€˜Next Generation Sequencingâ€™ as a diagnostic tool in paediatrics. Archives of Disease in Childhood, 2021, 106, 1-2.	1.0	9
11	Pathogenic variants causing ABL1 malformation syndrome cluster in a myristoyl-binding pocket and increase tyrosine kinase activity. European Journal of Human Genetics, 2021, 29, 593-603.	1.4	7
12	A novel ACE2 isoform is expressed in human respiratory epithelia and is upregulated in response to interferons and RNA respiratory virus infection. Nature Genetics, 2021, 53, 205-214.	9.4	125
13	Biallelic variants in COPB1 cause a novel, severe intellectual disability syndrome with cataracts and variable microcephaly. Genome Medicine, 2021, 13, 34.	3.6	18
14	Translating RNA Splicing Analysis into Diagnosis and Therapy. , 2021, 5, .		1
15	Splicing in the Diagnosis of Rare Disease: Advances and Challenges. Frontiers in Genetics, 2021, 12, 689892.	1.1	41
16	Altered regulation of BRCA1 exon 11 splicing is associated with breast cancer risk in carriers of BRCA1 pathogenic variants. Human Mutation, 2021, 42, 1488-1502.	1.1	7
17	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607.	1.6	37
18	Exome sequencing in patients with antiepileptic drug exposure and complex phenotypes. Archives of Disease in Childhood, 2020, 105, 384-389.	1.0	3

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19	Cancer Variant Interpretation Group UK (CanVIG-UK): an exemplar national subspecialty multidisciplinary network. Journal of Medical Genetics, 2020, 57, 829-834.	1.5	30
20	Opposite Modulation of RAC1 by Mutations in TRIO Is Associated with Distinct, Domain-Specific Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 338-355.	2.6	58
21	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human Molecular Genetics, 2020, 29, 1900-1921.	1.4	21
22	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	1.1	99
23	Mutation spectrum of PRPF31, genotype-phenotype correlation in retinitis pigmentosa, and opportunities for therapy. Experimental Eye Research, 2020, 192, 107950.	1.2	36
24	Splicing in the pathogenesis, diagnosis and treatment of ciliopathies. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2019, 1862, 194433.	0.9	25
25	Null variants and deletions in BRWD3 cause an Xâ€linked syndrome of mildâ€“moderate intellectual disability, macrocephaly, and obesity: A series of 17 patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 638-643.	0.7	8
26	A small gene sequencing panel realises a high diagnostic rate in patients with congenital nystagmus following basic phenotyping. Scientific Reports, 2019, 9, 13229.	1.6	9
27	Deleterious de novo variants of Xâ€linked ZC4H2 in females cause a variable phenotype with neurogenic arthrogryposis multiplex congenita. Human Mutation, 2019, 40, 2270-2285.	1.1	29
28	Phenotypic and biochemical analysis of an international cohort of individuals with variants in NAA10 and NAA15. Human Molecular Genetics, 2019, 28, 2900-2919.	1.4	46
29	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066.	2.4	38
30	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
31	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. American Journal of Human Genetics, 2019, 104, 596-610.	2.6	32
32	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	2.6	45
33	Exploring the RNA Gap for Improving Diagnostic Yield in Primary Immunodeficiencies. Frontiers in Genetics, 2019, 10, 1204.	1.1	3
34	Machine Learning Approaches for the Prioritization of Genomic Variants Impacting Pre-mRNA Splicing. Cells, 2019, 8, 1513.	1.8	41
35	RNA splicing analysis in genomic medicine. International Journal of Biochemistry and Cell Biology, 2019, 108, 61-71.	1.2	21
36	Gene-specific metrics to facilitate identification of disease genes for molecular diagnosis in patient genomes: a systematic review. Briefings in Functional Genomics, 2019, 18, 23-29.	1.3	6

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37	Breast cancer risk in neurofibromatosis type 1 is a function of the type of NF1 gene mutation: a new genotype-phenotype correlation. Journal of Medical Genetics, 2019, 56, 209-219.	1.5	26
38	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204
39	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Journal of Medical Genetics, 2018, 55, 104-113.	1.5	59
40	Expanding the neurodevelopmental phenotype of PURA syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 56-67.	0.7	26
41	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	2.6	37
42	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	0.9	75
43	Identification and functional analysis of a novel oculocerebrorenal syndrome of Lowe () gene variant in two pedigrees with varying phenotypes including isolated congenital cataract. Molecular Vision, 2018, 24, 847-852.	1.1	3
44	RNA splicing in human disease and in the clinic. Clinical Science, 2017, 131, 355-368.	1.8	78
45	Novel spliceâ€switching oligonucleotide promotes BRCA1 aberrant splicing and susceptibility to PARP inhibitor action. International Journal of Cancer, 2017, 140, 1564-1570.	2.3	19
46	Identification of a functionally significant tri-allelic genotype in the Tyrosinase gene (TYR) causing hypomorphic oculocutaneous albinism (OCA1B). Scientific Reports, 2017, 7, 4415.	1.6	47
47	A study of splicing mutations in disorders of sex development. Scientific Reports, 2017, 7, 16202.	1.6	5
48	Expanding the ocular phenotype of 14q terminal deletions: A novel presentation of microphthalmia and coloboma in ring 14 syndrome with associated 14q32.31 deletion and review of the literature. American Journal of Medical Genetics, Part A, 2016, 170, 1017-1022.	0.7	6
49	Mutations specific to the Rac-GEF domain ofTRIOcause intellectual disability and microcephaly. Journal of Medical Genetics, 2016, 53, 735-742.	1.5	80
50	Naturally occurringBRCA2alternative mRNA splicing events in clinically relevant samples. Journal of Medical Genetics, 2016, 53, 548-558.	1.5	69
51	Combined genetic and splicing analysis of BRCA1 c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. Human Molecular Genetics, 2016, 25, 2256-2268.	1.4	106
52	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	2.6	230
53	Pallister-Killian syndrome: a study of 22 British patients. Journal of Medical Genetics, 2015, 52, 454-464.	1.5	45
54	Large-scale discovery of novel genetic causes of developmental disorders. Nature, 2015, 519, 223-228.	13.7	998

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55	BRCA1 Exon 11, a CERES (Composite Regulatory Element of Splicing) Element Involved in Splice Regulation. International Journal of Molecular Sciences, 2014, 15, 13045-13059.	1.8	36
56	Whole exome sequencing in family trios revealsde novomutations inPURAas a cause of severe neurodevelopmental delay and learning disability. Journal of Medical Genetics, 2014, 51, 806-813.	1.5	73
57	RBFOX2 protein domains and cellular activities. Biochemical Society Transactions, 2014, 42, 1180-1183.	1.6	19
58	Comprehensive annotation of splice junctions supports pervasive alternative splicing at the BRCA1 locus: a report from the ENIGMA consortium. Human Molecular Genetics, 2014, 23, 3666-3680.	1.4	96
59	Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. Nature Genetics, 2014, 46, 385-388.	9.4	280
60	BRCA1 exon 11 a model of long exon splicing regulation. RNA Biology, 2014, 11, 351-359.	1.5	23
61	Comparison of mRNA Splicing Assay Protocols across Multiple Laboratories: Recommendations for Best Practice in Standardized Clinical Testing. Clinical Chemistry, 2014, 60, 341-352.	1.5	95
62	Role of Pseudoexons and Pseudointrons in Human Cancer. International Journal of Cell Biology, 2013, 2013, 1-16.	1.0	20
63	BRCA1 exon 11 alternative splicing, multiple functions and the association with cancer. Biochemical Society Transactions, 2012, 40, 768-772.	1.6	40
64	Systematic screening of FBN1 gene unclassified missense variants for splice abnormalities. Clinical Genetics, 2012, 82, 223-231.	1.0	12
65	Exon Skipping Mutations in Neurofibromatosis. Methods in Molecular Biology, 2012, 867, 65-76.	0.4	5
66	Evolutionary Constraint Helps Unmask a Splicing Regulatory Region in BRCA1 Exon 11. PLoS ONE, 2012, 7, e37255.	1.1	17
67	Splicing Mechanisms and Mutations in the NF1 Gene. , 2012, , 135-150.		0
68	Kingâ€“Denborough syndrome with and without mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2011, 21, 420-427.	0.3	97
69	An intronic mutation in MLH1 associated with familial colon and breast cancer. Familial Cancer, 2011, 10, 27-35.	0.9	19
70	Prediction of singleâ€nucleotide substitutions that result in exon skipping: identification of a splicing silencer inBRCA1exon 6. Human Mutation, 2011, 32, 436-444.	1.1	120
71	Anophthalmia in frontoâ€“facialâ€“nasal dysplasia. Clinical Dysmorphology, 2011, 20, 73-74.	0.1	3
72	Novel aspects of alternative splicing. FEBS Journal, 2010, 277, 835-835.	2.2	1

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73	Alternative splicing: good and bad effects of translationally silent substitutions. FEBS Journal, 2010, 277, 836-840.	2.2	34
74	Expanding the tuberous sclerosis phenotype: mild disease caused by a TSC1 splicing mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 350-352.	0.9	4
75	Novel roles of U1 snRNP in alternative splicing regulation. RNA Biology, 2010, 7, 412-419.	1.5	36
76	Identification of a de novo BRCA1 mutation in a woman with early onset bilateral breast cancer. Familial Cancer, 2009, 8, 479-482.	0.9	16
77	Missed threads. EMBO Reports, 2009, 10, 810-816.	2.0	107
78	Low U1â€ƒsnRNP dependence at the NF1 exonâ€ƒ29 donor splice site. FEBS Journal, 2009, 276, 2060-2073	2.2	19
79	Polypyrimidine tract binding protein regulates alternative splicing of an aberrant pseudoexon in NF1. FEBS Journal, 2008, 275, 6101-6108.	2.2	14
80	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
81	Can donor splice site recognition occur without the involvement of U1 snRNP?. Biochemical Society Transactions, 2008, 36, 548-550.	1.6	9
82	An Absence of Cutaneous Neurofibromas Associated with a 3-bp Inframe Deletion in Exon 17 of the NF1 Gene (c.2970-2972 delAAT): Evidence of a Clinically Significant NF1 Genotype-Phenotype Correlation. American Journal of Human Genetics, 2007, 80, 140-151.	2.6	335
83	NF1mRNA biogenesis: Effect of the genomic milieu in splicing regulation of theNF1exon 37 region. FEBS Letters, 2006, 580, 4449-4456.	1.3	64
84	A prospective study of neurofibromatosis type 1 cancer incidence in the UK. British Journal of Cancer, 2006, 95, 233-238.	2.9	200
85	Functional splicing assay shows a pathogenic intronic mutation in neurofibromatosis type 1 (NF1) due to intronic sequence exonization. Human Mutation, 2006, 27, 294-295.	1.1	41
86	PMS2 Mutations in Childhood Cancer. Journal of the National Cancer Institute, 2006, 98, 358-361.	3.0	85
87	A centrosomal mechanism involving CDK5RAP2 and CENPJ controls brain size. Nature Genetics, 2005, 37, 353-355.	9.4	520
88	Splicing in action: assessing disease causing sequence changes. Journal of Medical Genetics, 2005, 42, 737-748.	1.5	397
89	Mutations ofVMD2Splicing Regulators Cause Nanophthalmos and Autosomal Dominant Vitreoretinochoroidopathy (ADVIRC). , 2004, 45, 3683.		205
90	hnRNP H binding at the 5' splice site correlates with the pathological effect of two intronic mutations in the NF-1 and TSHÂ genes. Nucleic Acids Research, 2004, 32, 4224-4236.	6.5	71

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91	Automated comparative sequence analysis identifies mutations in 89% of NF1 patients and confirms a mutation cluster in exons 11-17 distinct from the GAP related domain. Journal of Medical Genetics, 2004, 41, e48-e48.	1.5	62
92	Different mutations in theNF1 gene are associated with Neurofibromatosis-Noonan syndrome (NFNS). American Journal of Medical Genetics Part A, 2003, 119A, 1-8.	2.4	64
93	Identification of a mutation that perturbs NF1 agene splicing using genomic DNA samples and a minigene assay. Journal of Medical Genetics, 2003, 40, 220-222.	1.5	72
94	Chromosomal aberrations, subtelomeric defects, and mental retardation. Lancet, The, 2001, 358, 7-8.	6.3	18
95	A case of the new overgrowth syndrome - macrocephaly with cutis marmorata, haemangioma and syndactyly. Clinical Dysmorphology, 2000, 9, 209-211.	0.1	12
96	Craniomicromelic syndrome: Report of a third case. , 1999, 87, 360-361.		3