Brunella Franco

List of Publications by Year
in descending order

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130 papers	9,117 citations	44069 48 h-index	43889 91 g-index
135 all docs	135 docs citations	135 times ranked	9505 citing authors

#	Article	IF	Citations
1	A gene deleted in Kallmann's syndrome shares homology with neural cell adhesion and axonal path-finding molecules. Nature, 1991, 353, 529-536.	27.8	852
2	Host response to EBV infection in X-linked lymphoproliferative disease results from mutations in an SH2-domain encoding gene. Nature Genetics, 1998, 20, 129-135.	21.4	720
3	Mutations of SURF-1 in Leigh Disease Associated with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 1609-1621.	6.2	504
4	Autophagy promotes primary ciliogenesis by removing OFD1 from centriolar satellites. Nature, 2013, 502, 254-257.	27.8	363
5	Opitz G/BBB syndrome, a defect of midline development, is due to mutations in a new RING finger gene on Xp22. Nature Genetics, 1997, 17, 285-291.	21.4	331
6	Identification of the Gene for Oral-Facial-Digital Type I Syndrome. American Journal of Human Genetics, 2001, 68, 569-576.	6.2	308
7	A cluster of sulfatase genes on Xp22.3: Mutations in chondrodysplasia punctata (CDPX) and implications for warfarin embryopathy. Cell, 1995, 81, 15-25.	28.9	303
8	Oral-facial-digital type I protein is required for primary cilia formation and left-right axis specification. Nature Genetics, 2006, 38, 112-117.	21.4	299
9	Functional Implications of the Spectrum of Mutations Found in 234 Cases With X-linked Juvenile Retinoschisis (XLRS). Human Molecular Genetics, 1998, 7, 1185-1192.	2.9	211
10	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
11	Intragenic Deletion of theKALIG-1Gene in Kallmann's Syndrome. New England Journal of Medicine, 1992, 326, 1752-1755.	27.0	189
12	Activation of Autophagy, Observed in Liver Tissues From Patients With Wilson Disease and From ATP7B-Deficient Animals, Protects Hepatocytes From Copper-Induced Apoptosis. Gastroenterology, 2019, 156, 1173-1189.e5.	1.3	150
13	Oralâ€"facialâ€"digital syndromes: Review and diagnostic guidelines. American Journal of Medical Genetics, Part A, 2007, 143A, 3314-3323.	1.2	134
14	Identification and Characterization of a Novel Serineâ€" Threonine Kinase Gene from the Xp22 Region. Genomics, 1998, 51, 427-433.	2.9	122
15	The oral-facial-digital syndrome gene C2CD3 encodes a positive regulator of centriole elongation. Nature Genetics, 2014, 46, 905-911.	21.4	121
16	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. Nature Genetics, 2016, 48, 648-656.	21.4	119
17	Mutations of the Mitochondrial Holocytochrome câ€"Type Synthase in X-Linked Dominant Microphthalmia with Linear Skin Defects Syndrome. American Journal of Human Genetics, 2006, 79, 878-889.	6.2	110
18	A gene from the Xp22.3 region shares homology with voltage-gated chloride channels. Human Molecular Genetics, 1994, 3, 547-552.	2.9	108

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19	A Member of a Gene Family on Xp22.3, VCX-A, Is Deleted in Patients with X-Linked Nonspecific Mental Retardation. American Journal of Human Genetics, 2000, 67, 563-573.	6.2	104
20	Mutations in COX7B Cause Microphthalmia with Linear Skin Lesions, an Unconventional Mitochondrial Disease. American Journal of Human Genetics, 2012, 91, 942-949.	6.2	104
21	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	2.5	104
22	CDKL5/Stk9 kinase inactivation is associated with neuronal developmental disorders. Human Molecular Genetics, 2005, 14, 3775-3786.	2.9	102
23	The Pervasive Role of the miR-181 Family in Development, Neurodegeneration, and Cancer. International Journal of Molecular Sciences, 2020, 21, 2092.	4.1	93
24	OFD1, the Gene Mutated in Oral-Facial-Digital Syndrome Type 1, Is Expressed in the Metanephros and in Human Embryonic Renal Mesenchymal Cells. Journal of the American Society of Nephrology: JASN, 2003, 14, 680-689.	6.1	92
25	X-linked Opitz syndrome: Novel mutations in theMID1gene and redefinition of the clinical spectrum. , 2003, 120A, 222-228.		91
26	The molecular basis of oralâ€fascialâ€digital syndrome, type 1. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2009, 151C, 318-325.	1.6	90
27	The dynamic cilium in human diseases. PathoGenetics, 2009, 2, 3.	5.7	85
28	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	3.2	85
29	Mutational spectrum of the oral-facial-digital type I syndrome: a study on a large collection of patients. Human Mutation, 2008, 29, 1237-1246.	2.5	82
30	Ciliopathy proteins regulate paracrine signaling by modulating proteasomal degradation of mediators. Journal of Clinical Investigation, 2014, 124, 2059-2070.	8.2	79
31	Different chromosomal localization of the Clcn4 gene in Mus spretus and C57BL/6J mice. Nature Genetics, 1995, 10, 466-471.	21.4	78
32	An integrated physical and genetic map of a 35 Mb region on chromosome Xp22.3â€“Xp21.3. Human Molecular Genetics, 1995, 4, 1821-1827.	2.9	76
33	Functional Characterization of the OFD1 Protein Reveals a Nuclear Localization and Physical Interaction with Subunits of a Chromatin Remodeling Complex. Molecular Biology of the Cell, 2007, 18, 4397-4404.	2.1	75
34	Novel Functional Features of the LIS-H Domain: Role in Protein Dimerization, Half-Life and Cellular Localization. Cell Cycle, 2005, 4, 1632-1640.	2.6	74
35	X-inactivation and human disease: X-linked dominant male-lethal disorders. Current Opinion in Genetics and Development, 2006, 16, 254-259.	3.3	74
36	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	3.8	71

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37	Identification and Characterization of AFG3L2, a Novel Paraplegin-Related Gene. Genomics, 1999, 59, 51-58.	2.9	70
38	Update on oral-facial-digital syndromes (OFDS). Cilia, 2016, 5, 12.	1.8	68
39	Opitz G/BBB Syndrome in Xp22: Mutations in the MID1 Gene Cluster in the Carboxy-Terminal Domain. American Journal of Human Genetics, 1998, 63, 703-710.	6.2	63
40	Twenty-six novelEFNB1 mutations in familial and sporadic craniofrontonasal syndrome (CFNS). Human Mutation, 2005, 26, 113-118.	2.5	61
41	miRâ€181a/b downregulation exerts a protective action on mitochondrial disease models. EMBO Molecular Medicine, 2019, 11, .	6.9	58
42	MID2, a Homologue of the Opitz Syndrome Gene MID1: Similarities in Subcellular Localization and Differences in Expression During Development. Human Molecular Genetics, 1999, 8, 1397-1407.	2.9	57
43	Characterization ofCxorf5(71-7A), a Novel Human cDNA Mapping to Xp22 and Encoding a Protein Containing Coiled-Coil Î±-Helical Domains. Genomics, 1998, 51, 243-250.	2.9	56
44	Ofd1 is required in limb bud patterning and endochondral bone development. Developmental Biology, 2011, 349, 179-191.	2.0	56
45	Characterization of a cluster of sulfatase genes on Xp22.3 suggests gene duplications in an ancestral pseudoautosomal region. Human Molecular Genetics, 1996, 5, 423-431.	2.9	55
46	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	2.5	54
47	Variable penetrance of hypogonadism in a sibship with Kallmann syndrome due to a deletion of the KAL gene. American Journal of Medical Genetics Part A, 1995, 57, 476-478.	2.4	50
48	A novel human serine-threonine phosphatase related to the Drosophila retinal degeneration C (rdgC) gene is selectively expressed in sensory neurons of neural crest origin. Human Molecular Genetics, 1997, 6, 1137-1145.	2.9	50
49	Dosage compensation of the mammalian X chromosome influences the phenotypic variability of X-linked dominant male-lethal disorders. Journal of Medical Genetics, 2008, 45, 401-408.	3.2	50
50	Terminal Osseous Dysplasia Is Caused by a Single Recurrent Mutation in the FLNA Gene. American Journal of Human Genetics, 2010, 87, 146-153.	6.2	50
51	Kidney-specific inactivation of Ofd1 leads to renal cystic disease associated with upregulation of the mTOR pathway. Human Molecular Genetics, 2010, 19, 2792-2803.	2.9	46
52	Somatic cell hybrids, sequence-tagged sites, simple repeat polymorphisms, and yeast artificial chromosomes for physical and genetic mapping of proximal 17p. Genomics, 1992, 13, 551-559.	2.9	44
53	Identification and Characterization of a Highly Conserved Protein Absent in the Alport Syndrome (A), Mental Retardation (M), Midface Hypoplasia (M), and Elliptocytosis (E) Contiguous Gene Deletion Syndrome (AMME). Genomics, 1999, 55, 335-340.	2.9	44
54	OFIP/KIAA0753 forms a complex with OFD1 and FOR20 at pericentriolar satellites and centrosomes and is mutated in one individual with oral-facial-digital syndrome. Human Molecular Genetics, 2016, 25, 497-513.	2.9	42

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55	Identification and Characterization of CDS2, a Mammalian Homolog of theDrosophilaCDP-diacylglycerol Synthase Gene. Genomics, 1999, 55, 68-77.	2.9	40
56	Characterization of the OFD1/Ofd1 genes on the human and mouse sex chromosomes and exclusion of Ofd1 for the Xpl mouse mutantâ˜†. Genomics, 2003, 81, 560-569.	2.9	40
57	Identification of SCML2, a Second Human Gene Homologous to theDrosophila Sex comb on midleg(Scm): A New Gene Cluster on Xp22. Genomics, 1999, 58, 65-72.	2.9	39
58	Identification of novel antigens with induced immune response in monoclonal gammopathy of undetermined significance. Blood, 2009, 114, 3276-3284.	1.4	38
59	Identification by Shotgun Sequencing, Genomic Organization, and Functional Analysis of a Fourth Arylsulfatase Gene (ARSF) from the Xp22.3 Region. Genomics, 1997, 42, 192-199.	2.9	37
60	Synthetic long non-coding RNAs [SINEUPs] rescue defective gene expression in vivo. Scientific Reports, 2016, 6, 27315.	3.3	37
61	The centrosomal OFD1 protein interacts with the translation machinery and regulates the synthesis of specific targets. Scientific Reports, 2017, 7, 1224.	3.3	36
62	The buccohypophyseal canal is an ancestral vertebrate trait maintained by modulation in sonic hedgehog signaling. BMC Biology, 2013, 11, 27.	3.8	35
63	CNS involvement in OFD1 syndrome: a clinical, molecular, and neuroimaging study. Orphanet Journal of Rare Diseases, 2014, 9, 74.	2.7	34
64	Identification of a Novel Homolog of the Drosophila staufen Protein in the Chromosome 8q13â€“q21.1 Region. Genomics, 1999, 62, 113-118.	2.9	33
65	The impairment of HCCS leads to MLS syndrome by activating a nonâ€canonical cell death pathway in the brain and eyes. EMBO Molecular Medicine, 2013, 5, 280-293.	6.9	33
66	Microphthalmia with linear skin defects (MLS) syndrome: Clinical, cytogenetic, and molecular characterization of 11 cases. American Journal of Medical Genetics, Part A, 2005, 137A, 190-198.	1.2	32
67	OFD1mutations in males: phenotypic spectrum and ciliary basal body docking impairment. Clinical Genetics, 2013, 84, 86-90.	2.0	32
68	Dopamine, Alpha-Synuclein, and Mitochondrial Dysfunctions in Parkinsonian Eyes. Frontiers in Neuroscience, 2020, 14, 567129.	2.8	31
69	Identification of a New EGF-Repeat-Containing Gene from Human Xp22: A Candidate for Developmental Disorders. Genomics, 2000, 65, 16-23.	2.9	30
70	Renal insufficiency, a frequent complication with age in oralâ€facialâ€digital syndrome type I. Clinical Genetics, 2010, 77, 258-265.	2.0	29
71	Molecular characterization of a patient with del(1)(q23â€“q25). Human Genetics, 1991, 87, 269-277.	3.8	28
72	X-linked recessive chondrodysplasia punctata due to a new point mutation of the ARSE gene. , 1997, 73, 139-143.		28

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73	Characterization of a Novel Chromo Domain Gene in Xp22.3 with Homology to Drosophila msl-3. Genomics, 1999, 59, 77-84.	2.9	28
74	Ofd1 Controls Dorso-Ventral Patterning and Axoneme Elongation during Embryonic Brain Development. PLoS ONE, 2012, 7, e52937.	2.5	28
75	Genomic deletions ofOFD1account for 23% of oral-facial-digital type 1 syndrome after negative DNA sequencing. Human Mutation, 2009, 30, E320-E329.	2.5	27
76	The Autophagy-Cilia Axis: An Intricate Relationship. Cells, 2019, 8, 905.	4.1	27
77	Î±-synuclein overexpression in the retina leads to vision impairment and degeneration of dopaminergic amacrine cells. Scientific Reports, 2020, 10, 9619.	3.3	27
78	HumanFIGF:Cloning, Gene Structure, and Mapping to Chromosome Xp22.1 between thePIGAand theGRPRGenes. Genomics, 1998, 47, 207-216.	2.9	25
79	The deubiquitinating enzyme Usp14 controls ciliogenesis and Hedgehog signaling. Human Molecular Genetics, 2019, 28, 764-777.	2.9	25
80	Regulation of autophagosome biogenesis by OFD1â€mediated selective autophagy. EMBO Journal, 2021, 40, e105120.	7.8	25
81	Identification and characterization of a novel member of the dystrobrevin gene family. FEBS Letters, 1998, 425, 7-13.	2.8	24
82	IL1RAPL2 maps to Xq22 and is specifically expressed in the central nervous system. Gene, 2001, 275, 217-221.	2.2	23
83	Refinement of the NHS locus on chromosome Xp22.13 and analysis of five candidate genes. European Journal of Human Genetics, 2002, 10, 516-520.	2.8	23
84	Mental retardation in a boy with an interstitial deletion at Xp22.3 involving STS, KAL1, and OA1: Implication for the MRX locus. , 1996, 64, 583-587.		21
85	The primary cilium in different tissuesâ€"lessons from patients and animal models. Pediatric Nephrology, 2011, 26, 655-662.	1.7	20
86	Autosomal recessive ‹scp›IFT57‹/scp› hypomorphic mutation cause ciliary transport defect in unclassified oralâ€"facialâ€"digital syndrome with short stature and brachymesophalangia. Clinical Genetics, 2016, 90, 509-517.	2.0	20
87	Integrated Genomics Identifies miR-181/TFAM Pathway as a Critical Driver of Drug Resistance in Melanoma. International Journal of Molecular Sciences, 2021, 22, 1801.	4.1	20
88	Disruption of the IQSEC2 transcript in a female with X;autosome translocation t(X;20)(p11.2;q11.2) and a phenotype resembling X-linked infantile spasms (ISSX) syndrome. Molecular Medicine Reports, 2008, 1, 33-9.	2.4	20
89	Construction of a YAC Contig Covering Human Chromosome 6p22. Genomics, 1996, 36, 399-407.	2.9	19
90	MAEG, an EGF-repeat containing gene, is a new marker associated with dermatome specification and morphogenesis of its derivatives. Mechanisms of Development, 2000, 98, 179-182.	1.7	19

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91	A network-based approach to dissect the cilia/centrosome complex interactome. BMC Genomics, 2014, 15, 658.	2.8	19
92	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. European Journal of Medical Genetics, 2013, 56, 301-308.	1.3	17
93	The centrosomal/basal body protein OFD1 is required for microtubule organization and cell cycle progression. Tissue and Cell, 2020, 64, 101369.	2.2	17
94	<scp>OFD1</scp>: One gene, several disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 57-71.	1.6	17
95	The HOPS complex subunit VPS39 controls ciliogenesis through autophagy. Human Molecular Genetics, 2020, 29, 1018-1029.	2.9	16
96	The TBC1D31/praja2 complex controls primary ciliogenesis through PKAâ€directed OFD1 ubiquitylation. EMBO Journal, 2021, 40, e106503.	7.8	15
97	Oral-facial-digital syndrome type I cells exhibit impaired DNA repair; unanticipated consequences of defective OFD1 outside of the cilia network. Human Molecular Genetics, 2017, 26, ddw364.	2.9	14
98	Oral-facial digital syndrome type 1. Indian Pediatrics, 2007, 44, 854-6.	0.4	14
99	Linkage mapping of a new syndromic form of X-linked mental retardation, MRXS7, associated with obesity. European Journal of Human Genetics, 1999, 7, 828-832.	2.8	13
100	Oralâ€“facialâ€“digital syndrome type 1, Caroli's disease and cystic renal disease. Nephrology Dialysis Transplantation, 2006, 21, 1705-1709.	0.7	13
101	Buccal Anomalies, Cephalometric Analysis and Genetic Study of Two Sisters with Orofaciodigital Syndrome Type I. Cleft Palate-Craniofacial Journal, 2007, 44, 660-666.	0.9	13
102	OFD Type I syndrome: lessons learned from a rare ciliopathy. Biochemical Society Transactions, 2020, 48, 1929-1939.	3.4	13
103	A new candidate region for the positional cloning of the XLP gene. European Journal of Human Genetics, 1998, 6, 509-517.	2.8	11
104	Terminal osseous dysplasia with pigmentary defects: Clinical description of a new family. American Journal of Medical Genetics, Part A, 2007, 143A, 51-57.	1.2	11
105	Metabolic Regulation of the Ultradian Oscillator Hes1 by Reactive Oxygen Species. Journal of Molecular Biology, 2015, 427, 1887-1902.	4.2	11
106	A ZFYVE19 gene mutation associated with neonatal cholestasis and cilia dysfunction: case report with a novel pathogenic variant. Orphanet Journal of Rare Diseases, 2021, 16, 179.	2.7	11
107	Crosstalk between cilia and autophagy: implication for human diseases. Autophagy, 2023, 19, 24-43.	9.1	10
108	Mutation-Independent Therapies for Retinal Diseases: Focus on Gene-Based Approaches. Frontiers in Neuroscience, 2020, 14, 588234.	2.8	9

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109	Expansion of the phenotype of lateral meningocele syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1259-1262.	1.2	9
110	Histone Deacetylase Sirtuin 1 Promotes Loss of Primary Cilia in Cholangiocarcinoma. Hepatology, 2021, 74, 3235-3248.	7.3	9
111	Micro-RNA in Cholangiocarcinoma: Implications for Diagnosis, Prognosis, and Therapy. Journal of Molecular Pathology, 2022, 3, 88-103.	1.2	9
112	High-density physical mapping of a 3-Mb region in Xp22.3 and refined localization of the gene for X-linked recessive chondrodysplasia punctata (CDPX1). Genomics, 1995, 26, 229-238.	2.9	8
113	Oral, facial, digital, vertebral anomalies with psychomotor delay: A mild form of OFD type Gabrielli?. American Journal of Medical Genetics Part A, 2002, 113, 291-294.	2.4	8
114	Linear Skin Defects with Multiple Congenital Anomalies (LSDMCA): An Unconventional Mitochondrial Disorder. Genes, 2021, 12, 263.	2.4	8
115	Search for genomic imbalances in a cohort of 20 patients with oralâ€“facialâ€“digital syndromes negative for mutations and large rearrangements in the OFD1 gene. American Journal of Medical Genetics, Part A, 2009, 149A, 1846-1849.	1.2	6
116	The OFD1 protein is a novel player in selective autophagy: another tile to the cilia/autophagy puzzle. Cell Stress, 2021, 5, 33-36.	3.2	6
117	TwoMsplRFLPs at the D17S258 locus. Nucleic Acids Research, 1990, 18, 7196-7196.	14.5	4
118	Linkage mapping of a nonspecific form of X-linked mental retardation (MRX53) in a large Pakistani family. American Journal of Medical Genetics Part A, 2001, 100, 62-65.	2.4	4
119	Cerebral dysgenesis does not exclude OFD I syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 455-457.	1.2	4
120	The role of OFD1 in selective autophagy. Molecular and Cellular Oncology, 2021, 8, 1903291.	0.7	4
121	Generation and Characterization of a Tumor Stromal Microenvironment and Analysis of Its Interplay with Breast Cancer Cells: An In Vitro Model to Study Breast Cancer-Associated Fibroblast Inactivation. International Journal of Molecular Sciences, 2022, 23, 6875.	4.1	4
122	Exclusion of PPEF as the gene causing X-linked juvenile retinoschisis. Human Genetics, 1997, 101, 235-237.	3.8	3
123	Biallelic variants in <scp> CENPF </scp> causing a phenotype distinct from StrÃ¸mme syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, , .	1.6	3
124	Generation of a transcription map of a 1 Mbase region containing the HFE gene (6p22). European Journal of Human Genetics, 1998, 6, 105-113.	2.8	2
125	Microphthalmia With Linear Skin Lesions (MLS) Syndrome: An Unconventional Mitochondrial Disorder. , 2016, , 1449-1451.		1
126	An Mspl RFLP at the D17S258 locus. Nucleic Acids Research, 1991, 19, 5482-5482.	14.5	0

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127	The impairment of HCCS leads to MLS syndrome by activating a nonâ€canonical cell death pathway in the brain and eyes. EMBO Molecular Medicine, 2014, 6, 849-849.	6.9	0
128	Xp contiguous gene syndromes: from clinical observation to disease gene identification. , 2000, , 225-243.		0
129	OFD1-Mediated T Cell Responses in MGUS Patients: Implications for Immunotherapy.. Blood, 2007, 110, 1488-1488.	1.4	0
130	Canonical and Non Canonical Activation of Hedgehog Pathway in Multiple Myeloma. Blood, 2008, 112, 2748-2748.	1.4	0