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	Crosstalk between cilia and autophagy: implication for human diseases. Autophagy, 2023, 19, 24-43.	9.1	10
2	<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
3	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
4	Micro-RNA in Cholangiocarcinoma: Implications for Diagnosis, Prognosis, and Therapy. Journal of Molecular Pathology, 2022, 3, 88-103.	1.2	9
5	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
6	Linear Skin Defects with Multiple Congenital Anomalies (LSDMCA): An Unconventional Mitochondrial Disorder. Genes, 2021, 12, 263.	2.4	8
7	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
8	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
9	The role of OFD1 in selective autophagy. Molecular and Cellular Oncology, 2021, 8, 1903291.	0.7	4
10	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
11	The TBC1D31/praja2 complex controls primary ciliogenesis through PKAâ€directed OFD1 ubiquitylation. EMBO Journal, 2021, 40, e106503.	7.8	15
12	Histone Deacetylase Sirtuin 1 Promotes Loss of Primary Cilia in Cholangiocarcinoma. Hepatology, 2021, 74, 3235-3248.	7.3	9
13	Regulation of autophagosome biogenesis by OFD1â€mediated selective autophagy. EMBO Journal, 2021, 40, e105120.	7.8	25
14	Mutation-Independent Therapies for Retinal Diseases: Focus on Gene-Based Approaches. Frontiers in Neuroscience, 2020, 14, 588234.	2.8	9
15	Dopamine, Alpha-Synuclein, and Mitochondrial Dysfunctions in Parkinsonian Eyes. Frontiers in Neuroscience, 2020, 14, 567129.	2.8	31
16	The centrosomal/basal body protein OFD1 is required for microtubule organization and cell cycle progression. Tissue and Cell, 2020, 64, 101369.	2.2	17
17	Î±-synuclein overexpression in the retina leads to vision impairment and degeneration of dopaminergic amacrine cells. Scientific Reports, 2020, 10, 9619.	3.3	27
18	Expansion of the phenotype of lateral meningocele syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1259-1262.	1.2	9

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19	The Pervasive Role of the miR-181 Family in Development, Neurodegeneration, and Cancer. International Journal of Molecular Sciences, 2020, 21, 2092.	4.1	93
20	The HOPS complex subunit VPS39 controls ciliogenesis through autophagy. Human Molecular Genetics, 2020, 29, 1018-1029.	2.9	16
21	OFD Type I syndrome: lessons learned from a rare ciliopathy. Biochemical Society Transactions, 2020, 48, 1929-1939.	3.4	13
22	The Autophagy-Cilia Axis: An Intricate Relationship. Cells, 2019, 8, 905.	4.1	27
23	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	2.5	104
24	miRâ€181a/b downregulation exerts a protective action on mitochondrial disease models. EMBO Molecular Medicine, 2019, 11, .	6.9	58
25	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
26	The deubiquitinating enzyme Usp14 controls ciliogenesis and Hedgehog signaling. Human Molecular Genetics, 2019, 28, 764-777.	2.9	25
27	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
28	The centrosomal OFD1 protein interacts with the translation machinery and regulates the synthesis of specific targets. Scientific Reports, 2017, 7, 1224.	3.3	36
29	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
30	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	2.5	54
31	Synthetic long non-coding RNAs [SINEUPs] rescue defective gene expression in vivo. Scientific Reports, 2016, 6, 27315.	3.3	37
32	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. Nature Genetics, 2016, 48, 648-656.	21.4	119
33	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
34	Update on oral-facial-digital syndromes (OFDS). Cilia, 2016, 5, 12.	1.8	68
35	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
36	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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37	Microphthalmia With Linear Skin Lesions (MLS) Syndrome: An Unconventional Mitochondrial Disorder. , 2016, , 1449-1451.		1
38	Metabolic Regulation of the Ultradian Oscillator Hes1 by Reactive Oxygen Species. Journal of Molecular Biology, 2015, 427, 1887-1902.	4.2	11
39	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
40	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	3.8	71
41	The oral-facial-digital syndrome gene C2CD3 encodes a positive regulator of centriole elongation. Nature Genetics, 2014, 46, 905-911.	21.4	121
42	A network-based approach to dissect the cilia/centrosome complex interactome. BMC Genomics, 2014, 15, 658.	2.8	19
43	CNS involvement in OFD1 syndrome: a clinical, molecular, and neuroimaging study. Orphanet Journal of Rare Diseases, 2014, 9, 74.	2.7	34
44	Ciliopathy proteins regulate paracrine signaling by modulating proteasomal degradation of mediators. Journal of Clinical Investigation, 2014, 124, 2059-2070.	8.2	79
45	Autophagy promotes primary ciliogenesis by removing OFD1 from centriolar satellites. Nature, 2013, 502, 254-257.	27.8	363
46	OFD1mutations in males: phenotypic spectrum and ciliary basal body docking impairment. Clinical Genetics, 2013, 84, 86-90.	2.0	32
47	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. European Journal of Medical Genetics, 2013, 56, 301-308.	1.3	17
48	The buccohypophyseal canal is an ancestral vertebrate trait maintained by modulation in sonic hedgehog signaling. BMC Biology, 2013, 11, 27.	3.8	35
49	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
50	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
51	Ofd1 Controls Dorso-Ventral Patterning and Axoneme Elongation during Embryonic Brain Development. PLoS ONE, 2012, 7, e52937.	2.5	28
52	Ofd1 is required in limb bud patterning and endochondral bone development. Developmental Biology, 2011, 349, 179-191.	2.0	56
53	The primary cilium in different tissuesâ€"lessons from patients and animal models. Pediatric Nephrology, 2011, 26, 655-662.	1.7	20
54	Cerebral dysgenesis does not exclude OFD I syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 455-457.	1.2	4

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55	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
56	Renal insufficiency, a frequent complication with age in oralâ€facialâ€digital syndrome type I. Clinical Genetics, 2010, 77, 258-265.	2.0	29
57	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
58	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
59	The molecular basis of oralâ€facialâ€digital syndrome, type 1. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2009, 151C, 318-325.	1.6	90
60	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
61	The dynamic cilium in human diseases. PathoGenetics, 2009, 2, 3.	5.7	85
62	Identification of novel antigens with induced immune response in monoclonal gammopathy of undetermined significance. Blood, 2009, 114, 3276-3284.	1.4	38
63	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
64	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
65	Canonical and Non Canonical Activation of Hedgehog Pathway in Multiple Myeloma. Blood, 2008, 112, 2748-2748.	1.4	0
66	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
67	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
68	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
69	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
70	Oralâ€“facialâ€“digital syndromes: Review and diagnostic guidelines. American Journal of Medical Genetics, Part A, 2007, 143A, 3314-3323.	1.2	134
71	OFD1-Mediated T Cell Responses in MGUS Patients: Implications for Immunotherapy.. Blood, 2007, 110, 1488-1488.	1.4	0
72	Oral-facial digital syndrome type 1. Indian Pediatrics, 2007, 44, 854-6.	0.4	14

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73	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
74	X-inactivation and human disease: X-linked dominant male-lethal disorders. Current Opinion in Genetics and Development, 2006, 16, 254-259.	3.3	74
75	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
76	Oralâ€“facialâ€“digital syndrome type 1, Caroli's disease and cystic renal disease. Nephrology Dialysis Transplantation, 2006, 21, 1705-1709.	0.7	13
77	Twenty-six novelEFNB1 mutations in familial and sporadic craniofrontonasal syndrome (CFNS). Human Mutation, 2005, 26, 113-118.	2.5	61
78	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
79	CDKL5/Stk9 kinase inactivation is associated with neuronal developmental disorders. Human Molecular Genetics, 2005, 14, 3775-3786.	2.9	102
80	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
81	X-linked Opitz syndrome: Novel mutations in theMID1gene and redefinition of the clinical spectrum. , 2003, 120A, 222-228.		91
82	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
83	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
84	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
85	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
86	Identification of the Gene for Oral-Facial-Digital Type I Syndrome. American Journal of Human Genetics, 2001, 68, 569-576.	6.2	308
87	IL1RAPL2 maps to Xq22 and is specifically expressed in the central nervous system. Gene, 2001, 275, 217-221.	2.2	23
88	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
89	Identification of a New EGF-Repeat-Containing Gene from Human Xp22: A Candidate for Developmental Disorders. Genomics, 2000, 65, 16-23.	2.9	30
90	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

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91	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
92	Xp contiguous gene syndromes: from clinical observation to disease gene identification. , 2000, , 225-243.		0
93	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
94	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
95	Identification and Characterization of CDS2, a Mammalian Homolog of theDrosophilaCDP-diacylglycerol Synthase Gene. Genomics, 1999, 55, 68-77.	2.9	40
96	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
97	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
98	Identification and Characterization of AFG3L2, a Novel Paraplegin-Related Gene. Genomics, 1999, 59, 51-58.	2.9	70
99	Characterization of a Novel Chromo Domain Gene in Xp22.3 with Homology to Drosophila msl-3. Genomics, 1999, 59, 77-84.	2.9	28
100	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
101	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
102	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
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	Identification and characterization of a novel member of the dystrobrevin gene family. FEBS Letters, 1998, 425, 7-13.	2.8	24
105	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
106	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
107	HumanFIGF:Cloning, Gene Structure, and Mapping to Chromosome Xp22.1 between thePIGAand theGRPRGenes. Genomics, 1998, 47, 207-216.	2.9	25
108	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

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109	Identification and Characterization of a Novel Serineâ€“ Threonine Kinase Gene from the Xp22 Region. Genomics, 1998, 51, 427-433.	2.9	122
110	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
111	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
112	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
113	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
114	Exclusion of PPEF as the gene causing X-linked juvenile retinoschisis. Human Genetics, 1997, 101, 235-237.	3.8	3
115	X-linked recessive chondrodysplasia punctata due to a new point mutation of the ARSE gene. , 1997, 73, 139-143.		28
116	Construction of a YAC Contig Covering Human Chromosome 6p22. Genomics, 1996, 36, 399-407.	2.9	19
117	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
118	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
119	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
120	Different chromosomal localization of the Clcn4 gene in Mus spretus and C57BL/6J mice. Nature Genetics, 1995, 10, 466-471.	21.4	78
121	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
122	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
123	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
124	A gene from the Xp22.3 region shares homology with voltage-gated chloride channels. Human Molecular Genetics, 1994, 3, 547-552.	2.9	108
125	Intragenic Deletion of theKALIG-1Gene in Kallmann's Syndrome. New England Journal of Medicine, 1992, 326, 1752-1755.	27.0	189
126	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

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127	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
128	Molecular characterization of a patient with del(1)(q23â€“q25). Human Genetics, 1991, 87, 269-277.	3.8	28
129	An MspI RFLP at the D17S258 locus. Nucleic Acids Research, 1991, 19, 5482-5482.	14.5	0
130	TwoMsplRFLPs at the D17S258 locus. Nucleic Acids Research, 1990, 18, 7196-7196.	14.5	4