Bruno Reversade

List of Publications by Year
in descending order

108	7,347	57719	62565
papers	citations	44 h-index	80 g-index

138	138	138	11309
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Connective-tissue growth factor (CTGF) modulates cell signalling by BMP and TGF-Î². Nature Cell Biology, 2002, 4, 599-604.	4.6	817
2	ELABELA: A Hormone Essential for Heart Development Signals via the Apelin Receptor. Developmental Cell, 2013, 27, 672-680.	3.1	384
3	Germline NLRP1 Mutations Cause Skin Inflammatory and Cancer Susceptibility Syndromes via Inflammasome Activation. Cell, 2016, 167, 187-202.e17.	13.5	317
4	Regulation of ADMP and BMP2/4/7 at Opposite Embryonic Poles Generates a Self-Regulating Morphogenetic Field. Cell, 2005, 123, 1147-1160.	13.5	232
5	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	9.4	211
6	Embryonic Dorsal-Ventral Signaling: Secreted Frizzled-Related Proteins as Inhibitors of Tolloid Proteinases. Cell, 2006, 124, 147-159.	13.5	199
7	ELABELA deficiency promotes preeclampsia and cardiovascular malformations in mice. Science, 2017, 357, 707-713.	6.0	181
8	Human DPP9 represses NLRP1 inflammasome and protects against autoinflammatory diseases via both peptidase activity and FIIND domain binding. Journal of Biological Chemistry, 2018, 293, 18864-18878.	1.6	172
9	Enteroviral 3C protease activates the human NLRP1 inflammasome in airway epithelia. Science, 2020, 370, .	6.0	151
10	Multiple self-healing squamous epithelioma is caused by a disease-specific spectrum of mutations in TGFBR1. Nature Genetics, 2011, 43, 365-369.	9.4	147
11	RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. Nature, 2018, 557, 564-569.	13.7	141
12	ELABELA Is an Endogenous Growth Factor that Sustains hESC Self-Renewal via the PI3K/AKT Pathway. Cell Stem Cell, 2015, 17, 435-447.	5.2	139
13	Proteolytic cleavage of Chordin as a switch for the dual activities of Twisted gastrulation in BMP signaling. Development (Cambridge), 2001, 128, 4439-4447.	1.2	134
14	Depletion of Bmp2, Bmp4, Bmp7 and Spemann organizer signals induces massive brain formation in Xenopus embryos. Development (Cambridge), 2005, 132, 3381-3392.	1.2	131
15	Chordin Is Required for the Spemann Organizer Transplantation Phenomenon in Xenopus Embryos. Developmental Cell, 2003, 4, 219-230.	3.1	130
16	Default neural induction: neuralization of dissociated Xenopus cells is mediated by Ras/MAPK activation. Genes and Development, 2005, 19, 1022-1027.	2.7	128
17	ELABELA-APJ axis protects from pressure overload heart failure and angiotensin II-induced cardiac damage. Cardiovascular Research, 2017, 113, 760-769.	1.8	111
18	Discovery and Structureâ€“Activity Relationship of a Bioactive Fragment of ELABELA that Modulates Vascular and Cardiac Functions. Journal of Medicinal Chemistry, 2016, 59, 2962-2972.	2.9	100

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19	Genome-wide CRISPR-Cas9 Screen Identifies Leukemia-Specific Dependence on a Pre-mRNA Metabolic Pathway Regulated by DCPS. Cancer Cell, 2018, 33, 386-400.e5.	7.7	99
20	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	2.6	95
21	Homozygous NLRP1 gain-of-function mutation in siblings with a syndromic form of recurrent respiratory papillomatosis. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 19055-19063.	3.3	92
22	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. Neuron, 2014, 84, 1240-1257.	3.8	89
23	Identification of Common Genetic Variants Influencing Spontaneous Dizygotic Twinning and Female Fertility. American Journal of Human Genetics, 2016, 98, 898-908.	2.6	89
24	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255.	9.4	88
25	Alternative Progenitor Cells Compensate to Rebuild the Coronary Vasculature in Elabela- and Apj-Deficient Hearts. Developmental Cell, 2017, 42, 655-666.e3.	3.1	88
26	Mitochondrial peptide BRAWNIN is essential for vertebrate respiratory complex III assembly. Nature Communications, 2020, 11, 1312.	5.8	87
27	The hormonal peptide Elabela guides angioblasts to the midline during vasculogenesis. ELife, 2015, 4, .	2.8	86
28	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	1.5	85
29	Gmnc Is a Master Regulator of the Multiciliated Cell Differentiation Program. Current Biology, 2015, 25, 3267-3273.	1.8	83
30	Loss of CHSY1, a Secreted FRINGE Enzyme, Causes Syndromic Brachydactyly in Humans via Increased NOTCH Signaling. American Journal of Human Genetics, 2010, 87, 768-778.	2.6	82
31	Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. Nature Genetics, 2012, 44, 1272-1276.	9.4	78
32	Sirenomelia in Bmp7 and Tsg compound mutant mice:requirement for Bmp signaling in the development of ventral posterior mesoderm. Development (Cambridge), 2005, 132, 2489-2499.	1.2	75
33	The opposing homeobox genes Goosecoid and Vent1/2 self-regulate Xenopus patterning. EMBO Journal, 2007, 26, 2955-2965.	3.5	73
34	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	1.8	71
35	Metabolic pathway analyses identify proline biosynthesis pathway as a promoter of liver tumorigenesis. Journal of Hepatology, 2020, 72, 725-735.	1.8	71
36	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. American Journal of Human Genetics, 2015, 97, 483-492.	2.6	70

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37	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nature Genetics, 2012, 44, 709-713.	9.4	68
38	The pro-BMP activity of Twisted gastrulation is independent of BMP binding. Development (Cambridge), 2003, 130, 4047-4056.	1.2	65
39	Structural basis of RIP2 activation and signaling. Nature Communications, 2018, 9, 4993.	5.8	65
40	Proteolytic cleavage of Chordin as a switch for the dual activities of Twisted gastrulation in BMP signaling. Development (Cambridge), 2001, 128, 4439-47.	1.2	65
41	Human NLRP1 is a sensor of pathogenic coronavirus 3CL proteases in lung epithelial cells. Molecular Cell, 2022, 82, 2385-2400.e9.	4.5	61
42	Long-Term Culture of Self-renewing Pancreatic Progenitors Derived from Human Pluripotent Stem Cells. Stem Cell Reports, 2017, 8, 1675-1688.	2.3	55
43	Structural basis for distinct inflammasome complex assembly by human NLRP1 and CARD8. Nature Communications, 2021, 12, 188.	5.8	54
44	A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. ELife, 2018, 7, .	2.8	53
45	Gene expression profiles in normal and Otx2-/- early gastrulating mouse embryos. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 14388-14393.	3.3	51
46	Inherited deficiency of stress granule ZNFX1 in patients with monocytosis and mycobacterial disease. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	47
47	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. American Journal of Human Genetics, 2018, 102, 116-132.	2.6	46
48	Somatic genetic rescue of a germline ribosome assembly defect. Nature Communications, 2021, 12, 5044.	5.8	44
49	SMCHD1 is involved inde novomethylation of theDUX4-encoding D4Z4 macrosatellite. Nucleic Acids Research, 2019, 47, 2822-2839.	6.5	39
50	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. Cilia, 2016, 5, 8.	1.8	37
51	IL11 is elevated in systemic sclerosis and IL11-dependent ERK signalling underlies TGFÎ²-mediated activation of dermal fibroblasts. Rheumatology, 2021, 60, 5820-5826.	0.9	36
52	CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. American Journal of Human Genetics, 2017, 101, 391-403.	2.6	35
53	ELABELA antagonizes intrarenal renin-angiotensin system to lower blood pressure and protects against renal injury. American Journal of Physiology - Renal Physiology, 2020, 318, F1122-F1135.	1.3	35
54	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	5.8	35

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55	Direct identification of A-to-I editing sites with nanopore native RNA sequencing. Nature Methods, 2022, 19, 833-844.	9.0	35
56	Quantitative imaging reveals real-time Pou5f3â€“Nanog complexes driving dorsoventral mesendoderm patterning in zebrafish. ELife, 2016, 5, .	2.8	34
57	The Apelin receptor enhances Nodal/TGFÎ² signaling to ensure proper cardiac development. ELife, 2016, 5, .	2.8	34
58	FSHD2- and BAMS-associated mutations confer opposing effects on SMCHD1 function. Journal of Biological Chemistry, 2018, 293, 9841-9853.	1.6	33
59	Loss of the scavenger mRNA decapping enzyme DCPS causes syndromic intellectual disability with neuromuscular defects. Human Molecular Genetics, 2015, 24, 3163-3171.	1.4	31
60	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	5.8	30
61	Loss of PYCR2 Causes Neurodegeneration by Increasing Cerebral Glycine Levels via SHMT2. Neuron, 2020, 107, 82-94.e6.	3.8	30
62	Identical twins carry a persistent epigenetic signature of early genome programming. Nature Communications, 2021, 12, 5618.	5.8	26
63	Katanin p80, NuMA and cytoplasmic dynein cooperate to control microtubule dynamics. Scientific Reports, 2017, 7, 39902.	1.6	25
64	A loss-of-function NUAK2 mutation in humans causes anencephaly due to impaired Hippo-YAP signaling. Journal of Experimental Medicine, 2020, 217, .	4.2	25
65	Nuclear-localized Asunder regulates cytoplasmic dynein localization via its role in the Integrator complex. Molecular Biology of the Cell, 2013, 24, 2954-2965.	0.9	23
66	Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling. American Journal of Human Genetics, 2016, 99, 299-317.	2.6	23
67	Incidentalome from Genomic Sequencing: A Barrier to Personalized Medicine?. EBioMedicine, 2016, 5, 211-216.	2.7	23
68	ENPP1 Mutation Causes Recessive Cole Disease by Altering Melanogenesis. Journal of Investigative Dermatology, 2018, 138, 291-300.	0.3	23
69	Novel mutations in the ciliopathy-associated gene CPLANE1 (C5orf42) cause OFD syndrome type VI rather than Joubert syndrome. European Journal of Medical Genetics, 2018, 61, 585-595.	0.7	22
70	Loss of Iroquois homeobox transcription factors 3 and 5 in osteoblasts disrupts cranial mineralization. Bone Reports, 2016, 5, 86-95.	0.2	21
71	Nextâ€generation sequencing in a series of 80 fetuses with complex cardiac malformations and/or heterotaxy. Human Mutation, 2020, 41, 2167-2178.	1.1	21
72	Multiple Self-Healing Palmoplantar Carcinoma: A Familial Predisposition to Skin Cancer with Primary Palmoplantar and Conjunctival Lesions. Journal of Investigative Dermatology, 2015, 135, 304-308.	0.3	20

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73	R-spondin signalling is essential for the maintenance and differentiation of mouse nephron progenitors. ELife, 2020, 9, .	2.8	20
74	Human Asunder promotes dynein recruitment and centrosomal tethering to the nucleus at mitotic entry. Molecular Biology of the Cell, 2012, 23, 4713-4724.	0.9	19
75	Loss-of-Function Mutations in LGI4, a Secreted Ligand Involved in Schwann Cell Myelination, Are Responsible for Arthrogryposis Multiplex Congenita. American Journal of Human Genetics, 2017, 100, 659-665.	2.6	19
76	R-SPONDIN2 mesenchymal cells form the bud tip progenitor niche during human lung development. Developmental Cell, 2022, 57, 1598-1614.e8.	3.1	19
77	The snRNA-processing complex, Integrator, is required for ciliogenesis and dynein recruitment to the nuclear envelope via distinct mechanisms. Biology Open, 2013, 2, 1390-1396.	0.6	18
78	Discriminative Features in Three Autosomal Recessive Cutis Laxa Syndromes: Cutis Laxa IIA, Cutis Laxa IIB, and Geroderma Osteoplastica. International Journal of Molecular Sciences, 2017, 18, 635.	1.8	18
79	Homozygous Null TBX4 Mutations Lead to Posterior Amelia with Pelvic and Pulmonary Hypoplasia. American Journal of Human Genetics, 2019, 105, 1294-1301.	2.6	17
80	Human model of IRX5 mutations reveals key role for this transcription factor in ventricular conduction. Cardiovascular Research, 2021, 117, 2092-2107.	1.8	17
81	Dominant-negative NFKBIA mutation promotes IL-1Î² production causing hepatic disease with severe immunodeficiency. Journal of Clinical Investigation, 2020, 130, 5817-5832.	3.9	17
82	Discovery of a genetic module essential for assigning leftâ€“right asymmetry in humans and ancestral vertebrates. Nature Genetics, 2022, 54, 62-72.	9.4	16
83	Optimal histone H3 to linker histone H1 chromatin ratio is vital for mesodermal competence in Xenopus. Development (Cambridge), 2013, 140, 853-860.	1.2	13
84	A Micropatterned Humanâ€Specific Neuroepithelial Tissue for Modeling Gene and Drugâ€Induced Neurodevelopmental Defects. Advanced Science, 2021, 8, 2001100.	5.6	13
85	Congenital insensitivity to pain with anhidrosis syndrome: A series from Jordan. Clinical Neurology and Neurosurgery, 2020, 189, 105636.	0.6	12
86	Biallelic <scp>ZNFX1</scp> variants are associated with a spectrum of immunoâ€hematological abnormalities. Clinical Genetics, 2022, 101, 247-254.	1.0	12
87	Heterozygous missense variant in EIF6 gene: A novel form of Shwachmanâ€“Diamond syndrome?. American Journal of Medical Genetics, Part A, 2020, 182, 2010-2020.	0.7	11
88	Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317.	2.6	11
89	Mitchell-Riley syndrome iPSC exhibit reduced pancreatic endoderm differentiation due to an RFX6 mutation. Development (Cambridge), 2020, 147, .	1.2	10
90	Intellectual disability, muscle weakness and characteristic face in three siblings: A newly described recessive syndrome mapping to 3p24.3â€“p25.3. American Journal of Medical Genetics, Part A, 2015, 167, 2508-2515.	0.7	9

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91	Cenani-Lenz syndactyly syndrome - a case report of a family with isolated syndactyly. BMC Medical Genetics, 2018, 19, 125.	2.1	9
92	Novel mutation in HTRA1 in a family with diffuse white matter lesions and inflammatory features. Neurology: Genetics, 2019, 5, e345.	0.9	9
93	Bone matrix hypermineralization associated with low bone turnover in a case of Nasu-Hakola disease. Bone, 2019, 123, 48-55.	1.4	7
94	Definition of the phenotypic spectrum of Temtamy preaxial brachydactyly syndrome associated with autosomal recessive CHYS1 mutations. Middle East Journal of Medical Genetics, 2012, 1, 64-70.	0.0	6
95	ELABELA Is an Endogenous Growth Factor that Sustains hESC Self-Renewal via the PI3K/AKT Pathway. Cell Stem Cell, 2015, 17, 635.	5.2	5
96	Congenital posterior cervical spine malformation due to biallelic c.240â€4T>G RIPPLY2 variant: A discrete entity. American Journal of Medical Genetics, Part A, 2020, 182, 1466-1472.	0.7	5
97	AKT Signaling Modifies the Balance between Cell Proliferation and Migration in Neural Crest Cells from Patients Affected with Bosma Arhinia and Microphthalmia Syndrome. Biomedicines, 2021, 9, 751.	1.4	5
98	HOX epimutations driven by maternal SMCHD1/LRIF1 haploinsufficiency trigger homeotic transformations in genetically wildtype offspring. Nature Communications, 2022, 13, .	5.8	5
99	Generation of three human induced pluripotent stem cell lines with IRX5 knockout and knockin genetic editions using CRISPR-Cas9 system. Stem Cell Research, 2022, 58, 102627.	0.3	4
100	Mutation in TWINKLE in a Large Iranian Family with Progressive External Ophthalmoplegia, Myopathy, Dysphagia and Dysphonia, and Behavior Change. Archives of Iranian Medicine, 2016, 19, 87-91.	0.2	4
101	Leveraging ancestry to improve causal variant identification in exome sequencing for monogenic disorders. European Journal of Human Genetics, 2016, 24, 113-119.	1.4	3
102	A <scp>GLI3</scp> variant leading to polydactyly in heterozygotes and Pallisterâ€Hallâ€like syndrome in a homozygote. Clinical Genetics, 2020, 97, 915-919.	1.0	3
103	Huriez syndrome caused by a large deletion that abrogates the skinâ€specific isoform of SMARCAD1. British Journal of Dermatology, 2021, 184, 1205-1207.	1.4	3
104	Huriez syndrome: Additional pathogenic variants supporting allelism to <scp>SMARCAD</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 1752-1760.	0.7	2
105	Generation of four H1 hESC sublines carrying a hemizygous knock-out/mutant MECP2. Stem Cell Research, 2019, 40, 101533.	0.3	1
106	05-P010 Histone variant H3.3 is required for Xenopus embryonic development. Mechanisms of Development, 2009, 126, S115.	1.7	0
107	06-P037 Dubowitz syndrome maps to chromosome 12 within an interval of 70 genes. Mechanisms of Development, 2009, 126, S130-S131.	1.7	0
108	Editorial overview: Cilia in development and disease. Current Opinion in Genetics and Development, 2019, 56, iii-iv.	1.5	0