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	Biallelic <scp>ZNFX1</scp> variants are associated with a spectrum of immunoâ€hematological abnormalities. Clinical Genetics, 2022, 101, 247-254.	1.0	12
2	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
3	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
4	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
5	Human NLRP1 is a sensor of pathogenic coronavirus 3CL proteases in lung epithelial cells. Molecular Cell, 2022, 82, 2385-2400.e9.	4.5	61
6	HOX epimutations driven by maternal SMCHD1/LRIF1 haploinsufficiency trigger homeotic transformations in genetically wildtype offspring. Nature Communications, 2022, 13, .	5.8	5
7	Direct identification of A-to-I editing sites with nanopore native RNA sequencing. Nature Methods, 2022, 19, 833-844.	9.0	35
8	R-SPONDIN2 mesenchymal cells form the bud tip progenitor niche during human lung development. Developmental Cell, 2022, 57, 1598-1614.e8.	3.1	19
9	Human model of IRX5 mutations reveals key role for this transcription factor in ventricular conduction. Cardiovascular Research, 2021, 117, 2092-2107.	1.8	17
10	A Micropatterned Humanâ€Specific Neuroepithelial Tissue for Modeling Gene and Drugâ€Induced Neurodevelopmental Defects. Advanced Science, 2021, 8, 2001100.	5.6	13
11	Structural basis for distinct inflammasome complex assembly by human NLRP1 and CARD8. Nature Communications, 2021, 12, 188.	5.8	54
12	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
13	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
14	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
15	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
16	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
17	Somatic genetic rescue of a germline ribosome assembly defect. Nature Communications, 2021, 12, 5044.	5.8	44
18	Identical twins carry a persistent epigenetic signature of early genome programming. Nature Communications, 2021, 12, 5618.	5.8	26

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19	Metabolic pathway analyses identify proline biosynthesis pathway as a promoter of liver tumorigenesis. Journal of Hepatology, 2020, 72, 725-735.	1.8	71
20	Congenital insensitivity to pain with anhidrosis syndrome: A series from Jordan. Clinical Neurology and Neurosurgery, 2020, 189, 105636.	0.6	12
21	Enteroviral 3C protease activates the human NLRP1 inflammasome in airway epithelia. Science, 2020, 370, .	6.0	151
22	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
23	Mitchell-Riley syndrome iPSC exhibit reduced pancreatic endoderm differentiation due to an RFX6 mutation. Development (Cambridge), 2020, 147, .	1.2	10
24	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
25	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	5.8	30
26	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
27	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
28	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
29	Mitochondrial peptide BRAWNIN is essential for vertebrate respiratory complex III assembly. Nature Communications, 2020, 11, 1312.	5.8	87
30	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
31	Loss of PYCR2 Causes Neurodegeneration by Increasing Cerebral Glycine Levels via SHMT2. Neuron, 2020, 107, 82-94.e6.	3.8	30
32	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	5.8	35
33	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
34	R-spondin signalling is essential for the maintenance and differentiation of mouse nephron progenitors. ELife, 2020, 9, .	2.8	20
35	Generation of four H1 hESC sublines carrying a hemizygous knock-out/mutant MECP2. Stem Cell Research, 2019, 40, 101533.	0.3	1
36	Editorial overview: Cilia in development and disease. Current Opinion in Genetics and Development, 2019, 56, iii-iv.	1.5	0

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37	SMCHD1 is involved inde novomethylation of theDUX4-encoding D4Z4 macrosatellite. Nucleic Acids Research, 2019, 47, 2822-2839.	6.5	39
38	Novel mutation in HTRA1 in a family with diffuse white matter lesions and inflammatory features. Neurology: Genetics, 2019, 5, e345.	0.9	9
39	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
40	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
41	Bone matrix hypermineralization associated with low bone turnover in a case of Nasu-Hakola disease. Bone, 2019, 123, 48-55.	1.4	7
42	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
43	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
44	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
45	ENPP1 Mutation Causes Recessive Cole Disease by Altering Melanogenesis. Journal of Investigative Dermatology, 2018, 138, 291-300.	0.3	23
46	Structural basis of RIP2 activation and signaling. Nature Communications, 2018, 9, 4993.	5.8	65
47	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
48	RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. Nature, 2018, 557, 564-569.	13.7	141
49	A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. ELife, 2018, 7, .	2.8	53
50	FSHD2- and BAMS-associated mutations confer opposing effects on SMCHD1 function. Journal of Biological Chemistry, 2018, 293, 9841-9853.	1.6	33
51	Cenani-Lenz syndactyly syndrome - a case report of a family with isolated syndactyly. BMC Medical Genetics, 2018, 19, 125.	2.1	9
52	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255.	9.4	88
53	ELABELA-APJ axis protects from pressure overload heart failure and angiotensin II-induced cardiac damage. Cardiovascular Research, 2017, 113, 760-769.	1.8	111
54	Katanin p80, NuMA and cytoplasmic dynein cooperate to control microtubule dynamics. Scientific Reports, 2017, 7, 39902.	1.6	25

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55	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
56	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
57	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
58	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
59	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
60	Long-Term Culture of Self-renewing Pancreatic Progenitors Derived from Human Pluripotent Stem Cells. Stem Cell Reports, 2017, 8, 1675-1688.	2.3	55
61	ELABELA deficiency promotes preeclampsia and cardiovascular malformations in mice. Science, 2017, 357, 707-713.	6.0	181
62	Identification of Common Genetic Variants Influencing Spontaneous Dizygotic Twinning and Female Fertility. American Journal of Human Genetics, 2016, 98, 898-908.	2.6	89
63	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
64	Germline NLRP1 Mutations Cause Skin Inflammatory and Cancer Susceptibility Syndromes via Inflammasome Activation. Cell, 2016, 167, 187-202.e17.	13.5	317
65	Loss of Iroquois homeobox transcription factors 3 and 5 in osteoblasts disrupts cranial mineralization. Bone Reports, 2016, 5, 86-95.	0.2	21
66	Incidentalome from Genomic Sequencing: A Barrier to Personalized Medicine?. EBioMedicine, 2016, 5, 211-216.	2.7	23
67	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. Cilia, 2016, 5, 8.	1.8	37
68	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
69	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
70	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
71	Quantitative imaging reveals real-time Pou5f3â€“Nanog complexes driving dorsoventral mesendoderm patterning in zebrafish. ELife, 2016, 5, .	2.8	34
72	The Apelin receptor enhances Nodal/TGFÎ² signaling to ensure proper cardiac development. ELife, 2016, 5, .	2.8	34

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73	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
74	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
75	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
76	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
77	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
78	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
79	Gmnc Is a Master Regulator of the Multiciliated Cell Differentiation Program. Current Biology, 2015, 25, 3267-3273.	1.8	83
80	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
81	The hormonal peptide Elabela guides angioblasts to the midline during vasculogenesis. ELife, 2015, 4, .	2.8	86
82	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. Neuron, 2014, 84, 1240-1257.	3.8	89
83	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	1.8	71
84	ELABELA: A Hormone Essential for Heart Development Signals via the Apelin Receptor. Developmental Cell, 2013, 27, 672-680.	3.1	384
85	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
86	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
87	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
88	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
89	Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. Nature Genetics, 2012, 44, 1272-1276.	9.4	78
90	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

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91	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nature Genetics, 2012, 44, 709-713.	9.4	68
92	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
93	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
94	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	9.4	211
95	05-P010 Histone variant H3.3 is required for Xenopus embryonic development. Mechanisms of Development, 2009, 126, S115.	1.7	0
96	06-P037 Dubowitz syndrome maps to chromosome 12 within an interval of 70 genes. Mechanisms of Development, 2009, 126, S130-S131.	1.7	0
97	The opposing homeobox genes Goosecoid and Vent1/2 self-regulate Xenopus patterning. EMBO Journal, 2007, 26, 2955-2965.	3.5	73
98	Embryonic Dorsal-Ventral Signaling: Secreted Frizzled-Related Proteins as Inhibitors of Tolloid Proteinases. Cell, 2006, 124, 147-159.	13.5	199
99	Default neural induction: neuralization of dissociated Xenopus cells is mediated by Ras/MAPK activation. Genes and Development, 2005, 19, 1022-1027.	2.7	128
100	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
101	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
102	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
103	Chordin Is Required for the Spemann Organizer Transplantation Phenomenon in Xenopus Embryos. Developmental Cell, 2003, 4, 219-230.	3.1	130
104	The pro-BMP activity of Twisted gastrulation is independent of BMP binding. Development (Cambridge), 2003, 130, 4047-4056.	1.2	65
105	Connective-tissue growth factor (CTGF) modulates cell signalling by BMP and TGF-Î². Nature Cell Biology, 2002, 4, 599-604.	4.6	817
106	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
107	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
108	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