SÃ©bastien KÃœry

List of Publications by Year
in descending order

114 papers	7,414 citations	87723 38 h-index	62479 80 g-index
130 all docs	130 docs citations	130 times ranked	13582 citing authors

#	Article	IF	Citations
1	Genome-wide association scan identifies a colorectal cancer susceptibility locus on chromosome 8q24. Nature Genetics, 2007, 39, 989-994.	9.4	676
2	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	13.7	544
3	Identification of SLC39A4, a gene involved in acrodermatitis enteropathica. Nature Genetics, 2002, 31, 239-240.	9.4	486
4	Discovery of common and rare genetic risk variants for colorectal cancer. Nature Genetics, 2019, 51, 76-87.	9.4	377
5	Identification of Genetic Susceptibility Loci for Colorectal Tumors in a Genome-Wide Meta-analysis. Gastroenterology, 2013, 144, 799-807.e24.	0.6	292
6	PNPLA1 mutations cause autosomal recessive congenital ichthyosis in golden retriever dogs and humans. Nature Genetics, 2012, 44, 140-147.	9.4	208
7	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
8	Meta-analysis of new genome-wide association studies of colorectal cancer risk. Human Genetics, 2012, 131, 217-234.	1.8	183
9	Genome-wide association analyses in east Asians identify new susceptibility loci for colorectal cancer. Nature Genetics, 2013, 45, 191-196.	9.4	173
10	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	2.6	171
11	Characterization of Geneâ€“Environment Interactions for Colorectal Cancer Susceptibility Loci. Cancer Research, 2012, 72, 2036-2044.	0.4	140
12	Genome-wide association study of colorectal cancer identifies six new susceptibility loci. Nature Communications, 2015, 6, 7138.	5.8	138
13	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	2.6	136
14	Novel Common Genetic Susceptibility Loci for Colorectal Cancer. Journal of the National Cancer Institute, 2019, 111, 146-157.	3.0	129
15	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142.	2.6	128
16	A Novel Zinc-regulated Human Zinc Transporter, hZTL1, Is Localized to the Enterocyte Apical Membrane. Journal of Biological Chemistry, 2002, 277, 22789-22797.	1.6	123
17	An update on mutations of theSLC39A4gene in acrodermatitis enteropathica. Human Mutation, 2009, 30, 926-933.	1.1	123
18	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	3.8	121

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19	Cumulative Burden of Colorectal Cancerâ€“Associated Genetic Variants Is More Strongly Associated With Early-Onset vs Late-Onset Cancer. Gastroenterology, 2020, 158, 1274-1286.e12.	0.6	110
20	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
21	A large-scale meta-analysis to refine colorectal cancer risk estimates associated with MUTYH variants. British Journal of Cancer, 2010, 103, 1875-1884.	2.9	107
22	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	2.6	102
23	Identification of Susceptibility Loci and Genes for Colorectal Cancer Risk. Gastroenterology, 2016, 150, 1633-1645.	0.6	97
24	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	2.6	90
25	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	2.6	86
26	Low-penetrance alleles predisposing to sporadic colorectal cancers: a French case-controlled genetic association study. BMC Cancer, 2008, 8, 326.	1.1	82
27	Mutations in FAM111B Cause Hereditary Fibrosing Poikiloderma with Tendon Contracture, Myopathy, and Pulmonary Fibrosis. American Journal of Human Genetics, 2013, 93, 1100-1107.	2.6	76
28	Combinations of Cytochrome P450 Gene Polymorphisms Enhancing the Risk for Sporadic Colorectal Cancer Related to Red Meat Consumption. Cancer Epidemiology Biomarkers and Prevention, 2007, 16, 1460-1467.	1.1	74
29	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357.	9.4	73
30	Trans-ethnic genome-wide association study of colorectal cancer identifies a new susceptibility locus in VTI1A. Nature Communications, 2014, 5, 4613.	5.8	72
31	Mutation spectrum of humanSLC39A4in a panel of patients with acrodermatitis enteropathica. Human Mutation, 2003, 22, 337-338.	1.1	70
32	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	2.6	66
33	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	2.6	62
34	Characterization of zinc amino acid complexes for zinc delivery in vitro using Caco-2 cells and enterocytes from hiPSC. BioMetals, 2017, 30, 643-661.	1.8	60
35	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	2.6	59
36	Mendelian randomization study of height and risk of colorectal cancer. International Journal of Epidemiology, 2015, 44, 662-672.	0.9	55

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37	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	2.6	54
38	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	2.6	51
39	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	2.6	45
40	Genetic variant predictors of gene expression provide new insight into risk of colorectal cancer. Human Genetics, 2019, 138, 307-326.	1.8	44
41	Genetic architectures of proximal and distal colorectal cancer are partly distinct. Gut, 2021, 70, 1325-1334.	6.1	44
42	Telomere structure and maintenance gene variants and risk of five cancer types. International Journal of Cancer, 2016, 139, 2655-2670.	2.3	43
43	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
44	Expanding the clinical spectrum of hereditary fibrosing poikiloderma with tendon contractures, myopathy and pulmonary fibrosis due to FAM111B mutations. Orphanet Journal of Rare Diseases, 2015, 10, 135.	1.2	42
45	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	0.7	42
46	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	1.1	41
47	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	1.1	40
48	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. Genetics in Medicine, 2019, 21, 2025-2035.	1.1	40
49	Genome-Wide Interaction Analyses between Genetic Variants and Alcohol Consumption and Smoking for Risk of Colorectal Cancer. PLoS Genetics, 2016, 12, e1006296.	1.5	38
50	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
51	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	2.6	37
52	Pleiotropic effects of genetic risk variants for other cancers on colorectal cancer risk: PAGE, GECCO and CCFR consortia. Gut, 2014, 63, 800-807.	6.1	35
53	Genome-Wide Search for Gene-Gene Interactions in Colorectal Cancer. PLoS ONE, 2012, 7, e52535.	1.1	35
54	Acute cytotoxicity of MIRA-1/NSC19630, a mutant p53-reactivating small molecule, against human normal and cancer cells via a caspase-9-dependent apoptosis. Cancer Letters, 2015, 359, 211-217.	3.2	34

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55	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
56	Identification of shared genetic variants between schizophrenia and lung cancer. Scientific Reports, 2018, 8, 674.	1.6	33
57	Genetic Predictors of Circulating 25-Hydroxyvitamin D and Risk of Colorectal Cancer. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 2037-2046.	1.1	30
58	Okurâ€Chung neurodevelopmental syndrome: Eight additional cases with implications on phenotype and genotype expansion. Clinical Genetics, 2018, 93, 880-890.	1.0	30
59	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
60	A genome-wide association study for colorectal cancer identifies a risk locus in 14q23.1. Human Genetics, 2015, 134, 1249-1262.	1.8	28
61	Identification of a common variant with potential pleiotropic effect on risk of inflammatory bowel disease and colorectal cancer. Carcinogenesis, 2015, 36, 999-1007.	1.3	28
62	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	1.1	28
63	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. American Journal of Human Genetics, 2021, 108, 951-961.	2.6	26
64	The intellectual disability-associated CAMK2G p.Arg292Pro mutation acts as a pathogenic gain-of-function. Human Mutation, 2018, 39, 2008-2024.	1.1	25
65	<scp>ARL6IP1</scp> mutation causes congenital insensitivity to pain, acromutilation and spastic paraplegia. Clinical Genetics, 2018, 93, 169-172.	1.0	24
66	Expression pattern, genomic structure and evaluation of the human SLC30A4 gene as a candidate for acrodermatitis enteropathica. Human Genetics, 2001, 109, 178-185.	1.8	23
67	A dominant vimentin variant causes a rare syndrome with premature aging. European Journal of Human Genetics, 2020, 28, 1218-1230.	1.4	23
68	Disruption of NEUROD2 causes a neurodevelopmental syndrome with autistic features via cell-autonomous defects in forebrain glutamatergic neurons. Molecular Psychiatry, 2021, 26, 6125-6148.	4.1	21
69	FAM111B Mutation Is Associated With Pancreatic Cancer Predisposition. Pancreas, 2019, 48, e41-e42.	0.5	20
70	Risk factors for hepatocellular carcinoma in Caucasian patients with nonâ€viral cirrhosis: the importance of prior obesity. Liver International, 2015, 35, 1872-1876.	1.9	19
71	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
72	Clinical, histopathological and genetic data of ichthyosis in the golden retriever: a prospective study. Journal of Small Animal Practice, 2009, 50, 227-235.	0.5	18

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73	Tropical medicine rounds: Acrodermatitis enteropathica: a review of 29 Tunisian cases. International Journal of Dermatology, 2010, 49, 1038-1044.	0.5	18
74	Colorectal Hamartomatous Polyposis and Ganglioneuromatosis in a Dog. Veterinary Pathology, 2011, 48, 1012-1015.	0.8	17
75	Missense variant contribution to USP9X-female syndrome. Npj Genomic Medicine, 2020, 5, 53.	1.7	17
76	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	1.4	17
77	Clinical utility gene card for: Biotinidase deficiencyâ€"update 2015. European Journal of Human Genetics, 2016, 24, 3-5.	1.4	16
78	The Thorough Screening of the MUTYH Gene in a Large French Cohort of Sporadic Colorectal Cancers. Genetic Testing and Molecular Biomarkers, 2007, 11, 373-380.	1.7	15
79	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	2.6	15
80	Neurodevelopmental Disorders (NDD) Caused by Genomic Alterations of the Ubiquitin-Proteasome System (UPS): the Possible Contribution of Immune Dysregulation to Disease Pathogenesis. Frontiers in Molecular Neuroscience, 2021, 14, 733012.	1.4	15
81	Delineation of the infrequent mosaicism ofKRASmutational status in metastatic colorectal adenocarcinomas. Journal of Clinical Pathology, 2012, 65, 466-469.	1.0	14
82	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
83	Acrodermatitis enteropathica: eine seltene dermatologische Differenzialdiagnose im Kindesalter - Erstbeschreibung zweier neuer Sequenzvarianten. JDDG - Journal of the German Society of Dermatology, 2011, 9, 999-1003.	0.4	13
84	Two novel variants in CNTNAP1 in two siblings presenting with congenital hypotonia and hypomyelinating neuropathy. European Journal of Human Genetics, 2017, 25, 150-152.	1.4	13
85	Acrodermatitis Enteropathica: A NovelSLC39A4Gene Mutation in a Patient with Normal Zinc Levels. Pediatric Dermatology, 2015, 32, e124-e125.	0.5	11
86	Evaluation of the colorectal cancer risk conferred by rareUNC5Calleles. World Journal of Gastroenterology, 2014, 20, 204.	1.4	11
87	Clinical Zinc Deficiency as Early Presentation of Wilson Disease. Journal of Pediatric Gastroenterology and Nutrition, 2015, 60, 457-459.	0.9	10
88	Mutation analysis of the zinc transporter gene SLC30A4 reveals no association with periodic catatonia on chromosome 15q15. Journal of Neural Transmission, 2003, 110, 1329-1332.	1.4	9
89	Transient symptomatic zinc deficiency in a breast-fed infant: Relevance of a genetic study. Nutrition, 2011, 27, 1087-1089.	1.1	9
90	A Zinc Sulphate-Resistant Acrodermatitis Enteropathica Patient with a Novel Mutation in SLC39A4 Gene. JIMD Reports, 2011, 2, 25-28.	0.7	9

#	ARTICLE	IF	CITATIONS
91	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	1.1	9
92	A novel mutation of the SLC39A4 gene causing acrodermatitis enteropathica. British Journal of Dermatology, 2007, 157, 386-387.	1.4	8
93	Three independent mutations in the TSC2 gene in a family with tuberous sclerosis. European Journal of Human Genetics, 2009, 17, 1165-1170.	1.4	8
94	Acrodermatitis enteropathica: an uncommon differential diagnosis in childhood - first description of a new sequence variant. JDDG - Journal of the German Society of Dermatology, 2011, 9, 999-1002.	0.4	8
95	Clinical utility gene card for: acrodermatitis enteropathica. European Journal of Human Genetics, 2012, 20, 3-4.	1.4	8
96	Fine-Mapping of Common Genetic Variants Associated with Colorectal Tumor Risk Identified Potential Functional Variants. PLoS ONE, 2016, 11, e0157521.	1.1	8
97	CUGC for hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP). European Journal of Human Genetics, 2016, 24, 779-779.	1.4	8
98	Dermatological manifestations of hereditary fibrosing poikiloderma with tendon contractures, myopathy and pulmonary fibrosis (<scp>POIKTMP</scp>): a case series of 28 patients. British Journal of Dermatology, 2019, 181, 862-864.	1.4	8
99	Clinical utility gene card for: acrodermatitis enteropathica â€“ update 2015. European Journal of Human Genetics, 2016, 24, 779-779.	1.4	7
100	BAK1 gene variation and abdominal aortic aneurysms-results may have been prematurely overrated. Human Mutation, 2010, 31, 1174-1176.	1.1	6
101	Clinical utility gene card for: Biotinidase deficiency. European Journal of Human Genetics, 2012, 20, 4-4.	1.4	6
102	Transient neonatal zinc deficiency in exclusively breastfed preterm infants. Journal of Paediatrics and Child Health, 2018, 54, 319-322.	0.4	6
103	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	2.6	6
104	SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. Nature Communications, 2022, 13, .	5.8	6
105	Ade novogermlineMLH1mutation in a Lynch syndrome patient with discordant immunohistochemical and molecular biology test results. World Journal of Gastroenterology, 2012, 18, 5635.	1.4	5
106	Rare pathogenic variants in WNK3 cause X-linked intellectual disability. Genetics in Medicine, 2022, 24, 1941-1951.	1.1	5
107	Identification of a Novel Mutation in the SLC39A4 Gene in a Case of Acrodermatitis Enteropathica. Acta Dermato-Venereologica, 2016, 96, 424-425.	0.6	4
108	Reappraisal of the soâ€called â€˜villous tumoursâ€™ of the rectosigmoid, based on histological, immunohistochemical and genotypic features. United European Gastroenterology Journal, 2014, 2, 307-314.	1.6	2

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109	751 Identification of a Common Variant With Potential Pleiotropic Effect on Risk of Inflammatory Bowel Disease and Colorectal Cancer. Gastroenterology, 2013, 144, S-136.	0.6	0
110	G.P.156. Neuromuscular Disorders, 2014, 24, 848.	0.3	0
111	Cover Image, Volume 39, Issue 12. Human Mutation, 2018, 39, i-i.	1.1	0
112	Abstract 2190: Fine-mapping of common genetic variants associated with colorectal tumor risk identified potential functional variants. , 2014, , .		0
113	PoÃ¯kilodermie hÃ©rÃ©ditaire fibrosante, myopathie rÃ©tractile et fibrose pulmonaire (POIKTMP). Les Cahiers De Myologie, 2016, , 12-14.	0.0	0
114	Abstract 5268: Interactions between genetic predictors of gene expression and dietary factors associated with risk of colorectal cancer. , 2018, , .		0