Peter P Pramstaller

List of Publications by Citations

202 papers	31,705 citations	71 h-index	178 g-index
222 ext. papers	38,032 ext. citations	11.3 avg, IF	4.97 L-index

#	Paper	IF	Citations
202	Biological, clinical and population relevance of 95 loci for blood lipids. *Nature*, **2010**, 466, 707-13	50.4	2742
201	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
200	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. *Nature Genetics*, **2010**, 42, 937-48	36.3	2267
199	Discovery and refinement of loci associated with lipid levels. *Nature Genetics*, **2013**, 45, 1274-1283	36.3	1904
198	Hundreds of variants clustered in genomic loci and biological pathways affect human height. *Nature*, **2010**, 467, 832-8	50.4	1514
197	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. *Nature Genetics*, **2010**, 42, 579-89	36.3	1449
196	Defining the role of common variation in the genomic and biological architecture of adult human height. *Nature Genetics*, **2014**, 46, 1173-86	36.3	1339
195	New genetic loci link adipose and insulin biology to body fat distribution. *Nature*, **2015**, 518, 187-196	50.4	920
194	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. *Nature Genetics*, **2010**, 42, 949-60	36.3	724
193	Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts. *Nature Genetics*, **2009**, 41, 47-55	36.3	708
192	Large-scale association analyses identify new loci influencing glycemic traits and provide insight into the underlying biological pathways. *Nature Genetics*, **2012**, 44, 991-1005	36.3	621
191	A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycemic traits and insulin resistance. *Nature Genetics*, **2012**, 44, 659-69	36.3	615
190	New loci associated with kidney function and chronic kidney disease. *Nature Genetics*, **2010**, 42, 376-84	36.3	599
189	Common variants associated with plasma triglycerides and risk for coronary artery disease. *Nature Genetics*, **2013**, 45, 1345-52	36.3	597
188	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. *Nature Genetics*, **2013**, 45, 145-54	36.3	505
187	Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. *Nature Genetics*, **2013**, 45, 501-12	36.3	437
186	Meta-analysis of genome-wide association studies in >80 000 subjects identifies multiple loci for C-reactive protein levels. *Circulation*, **2011**, 123, 731-8	16.7	395

185	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. *Nature Genetics*, **2018**, 50, 1412-1425	36.3	386
184	New gene functions in megakaryopoiesis and platelet formation. *Nature*, **2011**, 480, 201-8	50.4	330
183	Novel loci for adiponectin levels and their influence on type 2 diabetes and metabolic traits: a multi-ethnic meta-analysis of 45,891 individuals. *PLoS Genetics*, **2012**, 8, e1002607	6	326
182	The prevalence of metabolic syndrome and metabolically healthy obesity in Europe: a collaborative analysis of ten large cohort studies. *BMC Endocrine Disorders*, **2014**, 14, 9	3.3	311
181	Common variants at ten loci modulate the QT interval duration in the QTSCD Study. *Nature Genetics*, **2009**, 41, 407-14	36.3	308
180	FTO genotype is associated with phenotypic variability of body mass index. *Nature*, **2012**, 490, 267-72	50.4	304
179	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. *Nature Communications*, **2016**, 7, 10023	17.4	295
178	Sex-stratified genome-wide association studies including 270,000 individuals show sexual dimorphism in genetic loci for anthropometric traits. *PLoS Genetics*, **2013**, 9, e1003500	6	277
177	Seventy-five genetic loci influencing the human red blood cell. *Nature*, **2012**, 492, 369-75	50.4	257
176	The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. *Nature Genetics*, **2016**, 48, 1171-1184	36.3	251
175	Common variants in 22 loci are associated with QRS duration and cardiac ventricular conduction. *Nature Genetics*, **2010**, 42, 1068-76	36.3	249
174	Lewy body Parkinson's disease in a large pedigree with 77 Parkin mutation carriers. *Annals of Neurology*, **2005**, 58, 411-22	9.4	225
173	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. *PLoS Genetics*, **2015**, 11, e1005378	6	220
172	Positron emission tomographic analysis of the nigrostriatal dopaminergic system in familial Parkinsonism associated with mutations in the Parkin gene. *Annals of Neurology*, **2001**, 49, 367-376	9.4	220
171	Identification of heart rate-associated loci and their effects on cardiac conduction and rhythm disorders. *Nature Genetics*, **2013**, 45, 621-31	36.3	219
170	A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics*, **2019**, 51, 957-972	36.3	217
169	Parkin deletions in a family with adult-onset, tremor-dominant parkinsonism: Expanding the phenotype. *Annals of Neurology*, **2000**, 48, 65-71	9.4	213
168	Corticobasal degeneration shares a common genetic background with progressive supranuclear palsy. *Annals of Neurology*, **2000**, 47, 374-377	9.4	206

167	NRXN3 is a novel locus for waist circumference: a genome-wide association study from the CHARGE Consortium. *PLoS Genetics*, **2009**, 5, e1000539	6	203
166	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. *Nature Genetics*, **2014**, 46, 826-36	36.3	199
165	Distribution, type, and origin of Parkin mutations: review and case studies. *Movement Disorders*, **2004**, 19, 1146-57	7	194
164	A global in vivo Drosophila RNAi screen identifies NOT3 as a conserved regulator of heart function. *Cell*, **2010**, 141, 142-53	56.2	171
163	CUBN is a gene locus for albuminuria. *Journal of the American Society of Nephrology: JASN*, **2011**, 22, 555-70	12.7	170
162	Mutations in PINK1 and Parkin impair ubiquitination of Mitofusins in human fibroblasts. *PLoS ONE*, **2011**, 6, e16746	3.7	168
161	Stratifying type 2 diabetes cases by BMI identifies genetic risk variants in LAMA1 and enrichment for risk variants in lean compared to obese cases. *PLoS Genetics*, **2012**, 8, e1002741	6	162
160	Genome Analyses of >200,000 Individuals Identify 58 Loci for Chronic Inflammation and Highlight Pathways that Link Inflammation and Complex Disorders. *American Journal of Human Genetics*, **2018**, 103, 691-706	11	151
159	Genetic determinants of circulating sphingolipid concentrations in European populations. *PLoS Genetics*, **2009**, 5, e1000672	6	150
158	Genetic adaptation of fatty-acid metabolism: a human-specific haplotype increasing the biosynthesis of long-chain omega-3 and omega-6 fatty acids. *American Journal of Human Genetics*, **2012**, 90, 809-20	11	148
157	A meta-analysis of thyroid-related traits reveals novel loci and gender-specific differences in the regulation of thyroid function. *PLoS Genetics*, **2013**, 9, e1003266	6	146
156	Genome-wide association study identifies novel loci associated with circulating phospho- and sphingolipid concentrations. *PLoS Genetics*, **2012**, 8, e1002490	6	145
155	Genome-wide association and functional follow-up reveals new loci for kidney function. *PLoS Genetics*, **2012**, 8, e1002584	6	143
154	A genome-wide association search for type 2 diabetes genes in African Americans. *PLoS ONE*, **2012**, 7, e29202	3.7	138
153	Novel mutation in the TOR1A (DYT1) gene in atypical early onset dystonia and polymorphisms in dystonia and early onset parkinsonism. *Neurogenetics*, **2001**, 3, 133-43	3	128
152	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics*, **2019**, 51, 1459-1474	36.3	122
151	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. *Nature Communications*, **2014**, 5, 4926	17.4	121
150	Genome-wide meta-analysis uncovers novel loci influencing circulating leptin levels. *Nature Communications*, **2016**, 7, 10494	17.4	107

149	Large-scale replication and heterogeneity in Parkinson disease genetic loci. *Neurology*, **2012**, 79, 659-67	6.5	106
148	Genome-wide association analysis identifies multiple loci related to resting heart rate. *Human Molecular Genetics*, **2010**, 19, 3885-94	5.6	106
147	Mutant Parkin impairs mitochondrial function and morphology in human fibroblasts. *PLoS ONE*, **2010**, 5, e12962	3.7	104
146	Linkage analysis identifies a novel locus for restless legs syndrome on chromosome 2q in a South Tyrolean population isolate. *American Journal of Human Genetics*, **2006**, 79, 716-23	11	90
145	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. *Hypertension*, **2017**,	8.5	85
144	Plasma phosphatidylcholine and sphingomyelin concentrations are associated with depression and anxiety symptoms in a Dutch family-based lipidomics study. *Journal of Psychiatric Research*, **2013**, 47, 357-62	5.2	84
143	Overexpression of blood microRNAs 103a, 30b, and 29a in L-dopa-treated patients with PD. *Neurology*, **2015**, 84, 645-53	6.5	82
142	Serum iron levels and the risk of Parkinson disease: a Mendelian randomization study. *PLoS Medicine*, **2013**, 10, e1001462	11.6	80
141	Genome-wide meta-analysis associates HLA-DQA1/DRB1 and LPA and lifestyle factors with human longevity. *Nature Communications*, **2017**, 8, 910	17.4	78
140	Genetic determinants of heel bone properties: genome-wide association meta-analysis and replication in the GEFOS/GENOMOS consortium. *Human Molecular Genetics*, **2014**, 23, 3054-68	5.6	78
139	A multi-centre clinico-genetic analysis of the VPS35 gene in Parkinson disease indicates reduced penetrance for disease-associated variants. *Journal of Medical Genetics*, **2012**, 49, 721-6	5.8	78
138	52 Genetic Loci Influencing Myocardial Mass. *Journal of the American College of Cardiology*, **2016**, 68, 1435-1448	15.1	76
137	Localising loci underlying complex trait variation using Regional Genomic Relationship Mapping. *PLoS ONE*, **2012**, 7, e46501	3.7	75
136	Genome-wide analyses identify a role for SLC17A4 and AADAT in thyroid hormone regulation. *Nature Communications*, **2018**, 9, 4455	17.4	75
135	Genetic associations for activated partial thromboplastin time and prothrombin time, their gene expression profiles, and risk of coronary artery disease. *American Journal of Human Genetics*, **2012**, 91, 152-62	11	73
134	The arachidonic acid metabolome serves as a conserved regulator of cholesterol metabolism. *Cell Metabolism*, **2014**, 20, 787-798	24.6	72
133	Genomewide meta-analysis identifies loci associated with IGF-I and IGFBP-3 levels with impact on age-related traits. *Aging Cell*, **2016**, 15, 811-24	9.9	71
132	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. *Scientific Reports*, **2017**, 7, 45040	4.9	70

131	Large-scale genome-wide analysis identifies genetic variants associated with cardiac structure and function. *Journal of Clinical Investigation*, **2017**, 127, 1798-1812	15.9	68
130	Evidence of inbreeding depression on human height. *PLoS Genetics*, **2012**, 8, e1002655	6	62
129	Identification of a common variant in the TFR2 gene implicated in the physiological regulation of serum iron levels. *Human Molecular Genetics*, **2011**, 20, 1232-40	5.6	59
128	Discovery and fine mapping of serum protein loci through transethnic meta-analysis. *American Journal of Human Genetics*, **2012**, 91, 744-53	11	58
127	Genome-wide linkage analysis of serum creatinine in three isolated European populations. *Kidney International*, **2009**, 76, 297-306	9.9	57
126	Suggestive evidence for linkage for restless legs syndrome on chromosome 19p13. *Neurogenetics*, **2008**, 9, 75-82	3	56
125	Integration of genome-wide association studies with biological knowledge identifies six novel genes related to kidney function. *Human Molecular Genetics*, **2012**, 21, 5329-43	5.6	54
124	Novel association to the proprotein convertase PCSK7 gene locus revealed by analysing soluble transferrin receptor (sTfR) levels. *Human Molecular Genetics*, **2011**, 20, 1042-7	5.6	51
123	A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. *Nature Communications*, **2016**, 7, 13357	17.4	46
122	Modeling of environmental effects in genome-wide association studies identifies SLC2A2 and HP as novel loci influencing serum cholesterol levels. *PLoS Genetics*, **2010**, 6, e1000798	6	46
121	A new hypothesis for Parkinson's disease pathogenesis: GTPase-p38 MAPK signaling and autophagy as convergence points of etiology and genomics. *Molecular Neurodegeneration*, **2018**, 13, 40	19	44
120	The genetic study of three population microisolates in South Tyrol (MICROS): study design and epidemiological perspectives. *BMC Medical Genetics*, **2007**, 8, 29	2.1	44
119	The trans-ancestral genomic architecture of glycemic traits. *Nature Genetics*, **2021**, 53, 840-860	36.3	44
118	Genome-wide association meta-analyses and fine-mapping elucidate pathways influencing albuminuria. *Nature Communications*, **2019**, 10, 4130	17.4	43
117	Impaired sense of smell and color discrimination in monogenic and idiopathic Parkinson's disease. *Movement Disorders*, **2010**, 25, 2665-9	7	43
116	2q37 as a susceptibility locus for idiopathic basal ganglia calcification (IBGC) in a large South Tyrolean family. *Journal of Molecular Neuroscience*, **2009**, 39, 346-53	3.3	41
115	GWAtoolbox: an R package for fast quality control and handling of genome-wide association studies meta-analysis data. *Bioinformatics*, **2012**, 28, 444-5	7.2	41
114	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40

113	PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. *Nature Communications*, **2018**, 9, 2904	17.4	39
112	A meta-analysis of genome-wide data from five European isolates reveals an association of COL22A1, SYT1, and GABRR2 with serum creatinine level. *BMC Medical Genetics*, **2010**, 11, 41	2.1	39
111	The Cooperative Health Research in South Tyrol (CHRIS) study: rationale, objectives, and preliminary results. *Journal of Translational Medicine*, **2015**, 13, 348	8.5	38
110	Variation in the uric acid transporter gene SLC2A9 and its association with AAO of Parkinson's disease. *Journal of Molecular Neuroscience*, **2011**, 43, 246-50	3.3	38
109	Structural findings in the basal ganglia in genetically determined and idiopathic Parkinson's disease. *Movement Disorders*, **2009**, 24, 99-103	7	38
108	Heritability analysis of life span in a semi-isolated population followed across four centuries reveals the presence of pleiotropy between life span and reproduction. *Journals of Gerontology - Series A Biological Sciences and Medical Sciences*, **2011**, 66, 26-37	6.4	38
107	Genome-wide association analysis and fine mapping of NT-proBNP level provide novel insight into the role of the MTHFR-CLCN6-NPPA-NPPB gene cluster. *Human Molecular Genetics*, **2011**, 20, 1660-71	5.6	38
106	Methods for meta-analyses of genome-wide association studies: critical assessment of empirical evidence. *American Journal of Epidemiology*, **2012**, 175, 739-49	3.8	38
105	Plasma and White Blood Cells Show Different miRNA Expression Profiles in Parkinson's Disease. *Journal of Molecular Neuroscience*, **2017**, 62, 244-254	3.3	35
104	Structural imaging in the presymptomatic stage of genetically determined parkinsonism. *Neurobiology of Disease*, **2010**, 39, 402-8	7.5	35
103	Co-occurrence of restless legs syndrome and Parkin mutations in two families. *Movement Disorders*, **2006**, 21, 258-63	7	34
102	Frequency of heterozygous Parkin mutations in healthy subjects: need for careful prospective follow-up examination of mutation carriers. *Parkinsonism and Related Disorders*, **2009**, 15, 425-9	3.6	33
101	Clinical and genetic evaluation of a family with a mixed dystonia phenotype from South Tyrol. *Annals of Neurology*, **1998**, 44, 394-8	9.4	33
100	Profiling of Parkin-binding partners using tandem affinity purification. *PLoS ONE*, **2013**, 8, e78648	3.7	32
99	Restless legs syndrome: epidemiological and clinicogenetic study in a South Tyrolean population isolate. *Movement Disorders*, **2006**, 21, 1189-95	7	32
98	SLP-2 interacts with Parkin in mitochondria and prevents mitochondrial dysfunction in Parkin-deficient human iPSC-derived neurons and Drosophila. *Human Molecular Genetics*, **2017**, 26, 2412-2425	5.6	31
97	Modulation of genetic associations with serum urate levels by body-mass-index in humans. *PLoS ONE*, **2015**, 10, e0119752	3.7	31
96	Identification of a set of endogenous reference genes for miRNA expression studies in Parkinson's disease blood samples. *BMC Research Notes*, **2014**, 7, 715	2.3	30

95	Sex-dimorphic genetic effects and novel loci for fasting glucose and insulin variability. *Nature Communications*, **2021**, 12, 24	17.4	30
94	Pre-analytic evaluation of volumetric absorptive microsampling and integration in a mass spectrometry-based metabolomics workflow. *Analytical and Bioanalytical Chemistry*, **2017**, 409, 6263-6276	4.4	28
93	Alginate Formulations: Current Developments in the Race for Hydrogel-Based Cardiac Regeneration. *Frontiers in Bioengineering and Biotechnology*, **2020**, 8, 414	5.8	27
92	Common variants in Mendelian kidney disease genes and their association with renal function. *Journal of the American Society of Nephrology: JASN*, **2013**, 24, 2105-17	12.7	27
91	Nonmotor symptoms in Parkin gene-related parkinsonism. *Movement Disorders*, **2010**, 25, 1279-84	7	27
90	Genome-wide analysis of epistasis in body mass index using multiple human populations. *European Journal of Human Genetics*, **2012**, 20, 857-62	5.3	26
89	Phenotypic variability in a large kindred (Family LA) with deletions in the parkin gene. *Movement Disorders*, **2002**, 17, 424-6	7	26
88	A genome-wide screen for interactions reveals a new locus on 4p15 modifying the effect of waist-to-hip ratio on total cholesterol. *PLoS Genetics*, **2011**, 7, e1002333	6	25
87	The power of genetic diversity in genome-wide association studies of lipids. *Nature*, **2021**,	50.4	24
86	Influence of collection tubes during quantitative targeted metabolomics studies in human blood samples. *Clinica Chimica Acta*, **2018**, 486, 320-328	6.2	24
85	Association between restless legs syndrome and migraine: a population-based study. *European Journal of Neurology*, **2014**, 21, 1205-10	6	21
84	Large-scale assessment of polyglutamine repeat expansions in Parkinson disease. *Neurology*, **2015**, 85, 1283-92	6.5	20
83	Estimating the glomerular filtration rate in the general population using different equations: effects on classification and association. *Nephron Clinical Practice*, **2013**, 123, 102-11		20
82	Copy number variation across European populations. *PLoS ONE*, **2011**, 6, e23087	3.7	20
81	Drawing the history of the Hutterite population on a genetic landscape: inference from Y-chromosome and mtDNA genotypes. *European Journal of Human Genetics*, **2010**, 18, 463-70	5.3	20
80	The Impact of CRISPR/Cas9 Technology on Cardiac Research: From Disease Modelling to Therapeutic Approaches. *Stem Cells International*, **2017**, 2017, 8960236	5	19
79	Genetic architecture of circulating lipid levels. *European Journal of Human Genetics*, **2011**, 19, 813-9	5.3	19
78	Genes predict village of origin in rural Europe. *European Journal of Human Genetics*, **2010**, 18, 1269-70	5.3	19

77	Are Requirements to Deposit Data in Research Repositories Compatible With the European Union's General Data Protection Regulation?. *Annals of Internal Medicine*, **2019**, 170, 332-334	8	19
76	Exome sequencing in a family with restless legs syndrome. *Movement Disorders*, **2012**, 27, 1686-9	7	18
75	Genetic variants in RBFOX3 are associated with sleep latency. *European Journal of Human Genetics*, **2016**, 24, 1488-95	5.3	18
74	Heterogeneous susceptibility for uraemic media calcification and concomitant inflammation within the arterial tree. *Nephrology Dialysis Transplantation*, **2015**, 30, 1995-2005	4.3	17
73	Metabolic Signature of Dietary Iron Overload in a Mouse Model. *Cells*, **2018**, 7,	7.9	17
72	Effects of smoking status, history and intensity on heart rate variability in the general population: The CHRIS study. *PLoS ONE*, **2019**, 14, e0215053	3.7	16
71	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. *Nature Communications*, **2020**, 11, 2542	17.4	16
70	Serum iron level and kidney function: a Mendelian randomization study. *Nephrology Dialysis Transplantation*, **2017**, 32, 273-278	4.3	16
69	HDAC Inhibition Improves the Sarcoendoplasmic Reticulum Ca-ATPase Activity in Cardiac Myocytes. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	16
68	Generation of Induced Pluripotent Stem Cells from Frozen Buffy Coats using Non-integrating Episomal Plasmids. *Journal of Visualized Experiments*, **2015**, e52885	1.6	15
67	A network-based meta-analysis for characterizing the genetic landscape of human aging. *Biogerontology*, **2018**, 19, 81-94	4.5	15
66	Characterisation of genome-wide association epistasis signals for serum uric acid in human population isolates. *PLoS ONE*, **2011**, 6, e23836	3.7	15
65	The PPARGC1A locus and CNS-specific PGC-1 isoforms are associated with Parkinson's Disease. *Neurobiology of Disease*, **2019**, 121, 34-46	7.5	15
64	Importance of different types of prior knowledge in selecting genome-wide findings for follow-up. *Genetic Epidemiology*, **2013**, 37, 205-13	2.6	14
63	Genetic structure in contemporary south Tyrolean isolated populations revealed by analysis of Y-chromosome, mtDNA, and Alu polymorphisms. *Human Biology*, **2006**, 78, 441-64	1.2	14
62	Acetylation mediates Cx43 reduction caused by electrical stimulation. *Journal of Molecular and Cellular Cardiology*, **2015**, 87, 54-64	5.8	13
61	Primary familial brain calcification in the RBGC2Rkindred: All linkage roads lead to SLC20A2. *Movement Disorders*, **2016**, 31, 1901-1904	7	13
60	Effects of Calcium, Magnesium, and Potassium Concentrations on Ventricular Repolarization in Unselected Individuals. *Journal of the American College of Cardiology*, **2019**, 73, 3118-3131	15.1	12

59	Kinase inhibition of G2019S-LRRK2 enhances autolysosome formation and function to reduce endogenous alpha-synuclein intracellular inclusions. *Cell Death Discovery*, **2020**, 6, 45	6.9	11
58	Meta-analysis of genome-wide association studies identifies two loci associated with circulating osteoprotegerin levels. *Human Molecular Genetics*, **2014**, 23, 6684-93	5.6	11
57	SNP prioritization using a Bayesian probability of association. *Genetic Epidemiology*, **2013**, 37, 214-21	2.6	11
56	Abnormal premotor-motor interaction in heterozygous Parkin- and Pink1 mutation carriers. *Clinical Neurophysiology*, **2017**, 128, 275-280	4.3	11
55	Copy number variation and association over T-cell receptor genes--influence of DNA source. *Immunogenetics*, **2010**, 62, 561-7	3.2	11
54	Sequential recruitment of study participants may inflate genetic heritability estimates. *Human Genetics*, **2017**, 136, 743-757	6.3	10
53	The Histone Deacetylase Inhibitor Suberoylanilide Hydroxamic Acid (SAHA) Restores Cardiomyocyte Contractility in a Rat Model of Early Diabetes. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	9
52	The arrhythmogenic cardiomyopathy-specific coding and non-coding transcriptome in human cardiac stromal cells. *BMC Genomics*, **2018**, 19, 491	4.5	9
51	Fine-mapping of restless legs locus 4 (RLS4) identifies a haplotype over the SPATS2L and KCTD18 genes. *Journal of Molecular Neuroscience*, **2013**, 49, 600-5	3.3	9
50	Imaging movement-related activity in medicated Parkin-associated and sporadic Parkinson's disease. *Parkinsonism and Related Disorders*, **2010**, 16, 384-7	3.6	9
49	Exclusion of linkage to chromosome 14q in a large South Tyrolean family with Idiopathic Basal Ganglia Calcification (IBGC). *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 1319-22	3.5	9
48	Structural Consistency of the Pain Sensitivity Questionnaire in the Cooperative Health Research In South Tyrol (CHRIS) Population-Based Study. *Journal of Pain*, **2018**, 19, 1424-1434	5.2	8
47	CADPS2 gene expression is oppositely regulated by LRRK2 and alpha-synuclein. *Biochemical and Biophysical Research Communications*, **2017**, 490, 876-881	3.4	8
46	Higher cardiogenic potential of iPSCs derived from cardiac versus skin stromal cells. *Frontiers in Bioscience - Landmark*, **2016**, 21, 719-43	2.8	8
45	Environmental and Genetic Variables Influencing Mitochondrial Health and Parkinson's Disease Penetrance. *Parkinson's Disease*, **2018**, 2018, 8684906	2.6	8
44	Silencing of CCR4-NOT complex subunits affects heart structure and function. *DMM Disease Models and Mechanisms*, **2020**, 13,	4.1	7
43	Application of CRISPR/Cas9 editing and digital droplet PCR in human iPSCs to generate novel knock-in reporter lines to visualize dopaminergic neurons. *Stem Cell Research*, **2019**, 41, 101656	1.6	7
42	PedVizApi: a Java API for the interactive, visual analysis of extended pedigrees. *Bioinformatics*, **2008**, 24, 279-81	7.2	7

41	Elevated levels of alpha-synuclein blunt cellular signal transduction downstream of Gq protein-coupled receptors. *Cellular Signalling*, **2017**, 30, 82-91	4.9	6
40	Compound heterozygous SZT2 mutations in two siblings with early-onset epilepsy, intellectual disability and macrocephaly. *Seizure: the Journal of the British Epilepsy Association*, **2019**, 66, 81-85	3.2	6
39	Exploring Approaches for Detecting Protein Functional Similarity within an Orthology-based Framework. *Scientific Reports*, **2017**, 7, 381	4.9	5
38	KCND3 potassium channel gene variant confers susceptibility to electrocardiographic early repolarization pattern. *JCI Insight*, **2019**, 4,	9.9	5
37	Parkin deletions in a family with adult-onset, tremor-dominant parkinsonism: Expanding the phenotype **2000**, 48, 65		5
36	Haplotype analysis of the 4p16.3 region in Portuguese families with Huntington's disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2015**, 168B, 135-43	3.5	4
35	Genetic analysis of over one million people identifies 535 novel loci for blood pressure		4
34	Generation of human induced pluripotent stem cells (EURACi001-A, EURACi002-A, EURACi003-A) from peripheral blood mononuclear cells of three patients carrying mutations in the CAV3 gene. *Stem Cell Research*, **2018**, 27, 25-29	1.6	3
33	Lipidomics, Atrial Conduction, and Body Mass Index. *Circulation Genomic and Precision Medicine*, **2019**, 12, e002384	5.2	3
32	Influence of L-dopa on subtle motor signs in heterozygous Parkin- and PINK1 mutation carriers. *Parkinsonism and Related Disorders*, **2017**, 42, 95-99	3.6	3
31	Involvement of proprotein convertase PCSK7 in the regulation of systemic iron homeostasis. *Hepatology*, **2013**, 58, 1860-1	11.2	3
30	Update on the management of restless legs syndrome: existing and emerging treatment options. *Nature and Science of Sleep*, **2010**, 2, 199-212	3.6	3
29	Parkin gene modifies the effect of RLS4 on the age at onset of restless legs syndrome (RLS). *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 350-5	3.5	3
28	Variants in STAT5B associate with serum TC and LDL-C levels. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, E1496-501	5.6	3
27	Linkage and association analysis of hyperthyrotropinaemia in an Alpine population reveal two novel loci on chromosomes 3q28-29 and 6q26-27. *Journal of Medical Genetics*, **2011**, 48, 549-56	5.8	3
26	SLP-2: a potential new target for improving mitochondrial function in Parkinson's disease. *Neural Regeneration Research*, **2017**, 12, 1435-1436	4.5	3
25	Parkin Interacts with Apoptosis-Inducing Factor and Interferes with Its Translocation to the Nucleus in Neuronal Cells. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	3
24	Generation of an induced pluripotent stem cell line (EURACi005-A) from a Parkinson's disease patient carrying a homozygous exon 3 deletion in the PRKNgene. *Stem Cell Research*, **2019**, 41, 101624	1.6	3

23	Microbiota, type 2 diabetes and non-alcoholic fatty liver disease: protocol of an observational study. *Journal of Translational Medicine*, **2019**, 17, 408	8.5	3
22	Prevalence and determinants of serum antibodies to SARS-CoV-2 in the general population of the Gardena valley. *Epidemiology and Infection*, **2021**, 149, e194	4.3	3
21	Frequency of Heterozygous Parkin () Variants and Penetrance of Parkinson Disease Risk Markers in the Population-Based CHRIS Cohort. *Frontiers in Neurology*, **2021**, 12, 706145	4.1	3
20	Generation of hiPSC-Derived Functional Dopaminergic Neurons in Alginate-Based 3D Culture. *Frontiers in Cell and Developmental Biology*, **2021**, 9, 708389	5.7	3
19	Association between non-alcoholic fatty liver disease and impaired cardiac sympathetic/parasympathetic balance in subjects with and without type 2 diabetes-The Cooperative Health Research in South Tyrol (CHRIS)-NAFLD sub-study. *Nutrition, Metabolism and Cardiovascular Diseases*, **2021**, 31, 3464-3473	4.5	3
18	Genome-wide association study to identify common variants associated with brachial circumference: a meta-analysis of 14 cohorts. *PLoS ONE*, **2012**, 7, e31369	3.7	2
17	Highly Elevated Plasma Glutamyltransferase Elevations: A Trait Caused by Glutamyltransferase 1 Transmembrane Mutations. *Hepatology*, **2020**, 71, 1124-1127	11.2	2
16	Derivation of human induced pluripotent stem cell line EURACi004-A from skin fibroblasts of a patient with Arrhythmogenic Cardiomyopathy carrying the heterozygous PKP2 mutation c.2569_3018del50. *Stem Cell Research*, **2018**, 32, 78-82	1.6	2
15	SNP-based linkage analysis in extended pedigrees: comparison between two alternative approaches. *Human Heredity*, **2014**, 78, 27-37	1.1	1
14	Prospective epidemiological, molecular, and genetic characterization of a novel coronavirus disease in the Val Venosta/Vinschgau: the CHRIS COVID-19 study protocol. *Pathogens and Global Health*, **2021**, 1-9	3.1	1
13	RIT2 reduces LRRK2 kinase activity and protects against alpha-synuclein neuropathology		1
12	Kinase inhibition of G2019S-LRRK2 restores autolysosome formation and function to reduce endogenous alpha-synuclein intracellular inclusions		1
11	Task matters - challenging the motor system allows distinguishing unaffected Parkin mutation carriers from mutation-free controls. *Parkinsonism and Related Disorders*, **2021**, 86, 101-104	3.6	1
10	Return of research results (RoRR) to the healthy CHRIS cohort: designing a policy with the participants. *Journal of Community Genetics*, **2021**, 12, 577-592	2.5	1
9	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. *Communications Biology*, **2022**, 5,	6.7	1
8	A multi-omics study of circulating phospholipid markers of blood pressure.. *Scientific Reports*, **2022**, 12, 574	4.9	0
7	Generation and characterization of induced pluripotent stem cell (iPSC) lines of two asymptomatic individuals carrying a heterozygous exon 7 deletion in Parkin (PRKN) and two non-carriers from the same family.. *Stem Cell Research*, **2022**, 60, 102692	1.6	0
6	Balancing scientific interests and the rights of participants in designing a recall by genotype study. *European Journal of Human Genetics*, **2021**, 29, 1146-1157	5.3	0

5	Comparative assessment of different familial aggregation methods in the context of large and unstructured pedigrees. *Bioinformatics*, **2019**, 35, 69-76	7.2	0
4	Exome-wide association study of levodopa-induced dyskinesia in Parkinson's disease. *Scientific Reports*, **2021**, 11, 19582	4.9	0
3	Caenorhabditis elegans Parkin: Regulators of its abundance and role in autophagy-lysosomal dynamics. *Open Research Europe*,2, 23		
2	Generation of an induced pluripotent stem cell line (EURACi014-A) from a Parkinson's disease patient with an A53T mutation in the SNCA gene by an integration-free reprogramming method.. *Stem Cell Research*, **2022**, 60, 102713	1.6	
1	A genome on shaky ground: exploring the impact of mitochondrial DNA integrity on Parkinson's disease by highlighting the use of cybrid models.. *Cellular and Molecular Life Sciences*, **2022**, 79, 283	10.3	