Elena Cellini

List of Publications by Year in Descending Order

63 papers	1,934 citations	26 h-index	41 g-index
65 ext. papers	2,164 ext. citations	4.1 avg, IF	3.59 L-index

#	Paper	IF	Citations
63	Distinct epilepsy phenotypes and response to drugs in KCNA1 gain- and loss-of function variants. *Epilepsia*, **2021**,	6.4	3
62	Mirror syndromes regarding AKT3 mutations: Loss of function variant leading to microcephaly. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2800-2802	2.5	
61	Multiple genomic copy number variants associated with periventricular nodular heterotopia indicate extreme genetic heterogeneity. *European Journal of Human Genetics*, **2019**, 27, 909-918	5.3	12
60	Diagnostic implications of genetic copy number variation in epilepsy plus. *Epilepsia*, **2019**, 60, 689-706	6.4	37
59	Severe 5,10-methylenetetrahydrofolate reductase deficiency: a rare, treatable cause of complicated hereditary spastic paraplegia. *European Journal of Neurology*, **2018**, 25, 602-605	6	10
58	De novo KCNA1 variants in the PVP motif cause infantile epileptic encephalopathy and cognitive impairment similar to recurrent KCNA2 variants. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1748-1752	2.5	20
57	Recessive mutations in SLC35A3 cause early onset epileptic encephalopathy with skeletal defects. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1119-1123	2.5	10
56	Pyridoxine responsiveness in pyridox(am)ine-5-phosphate oxidase deficiency: The importance of early treatment. *Clinical Neurology and Neurosurgery*, **2017**, 163, 90-93	2	1
55	Clinical features and outcome of 6 new patients carrying de novo gene mutations. *Neurology: Genetics*, **2017**, 3, e206	3.8	30
54	Diagnostic Targeted Resequencing in 349 Patients with Drug-Resistant Pediatric Epilepsies Identifies Causative Mutations in 30 Different Genes. *Human Mutation*, **2017**, 38, 216-225	4.7	102
53	The hyperkinetic movement disorder of FOXG1-related epileptic-dyskinetic encephalopathy. *Developmental Medicine and Child Neurology*, **2016**, 58, 93-7	3.3	23
52	Recurrent drop attacks in early childhood as presenting symptom of benign hereditary chorea caused by TITF1 gene mutations. *Developmental Medicine and Child Neurology*, **2015**, 57, 777-9	3.3	11
51	Low social interactions in eating disorder patients in childhood and adulthood: a multi-centre European case control study. *Journal of Health Psychology*, **2013**, 18, 26-37	3.1	59
50	Fat mass and obesity-associated gene (FTO) in eating disorders: evidence for association of the rs9939609 obesity risk allele with bulimia nervosa and anorexia nervosa. *Obesity Facts*, **2012**, 5, 408-19	5.1	37
49	Periventricular heterotopia with white matter abnormalities associated with 6p25 deletion. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1793-7	2.5	20
48	Membrane cholesterol enrichment prevents A□nduced oxidative stress in Alzheimer□s fibroblasts. *Neurobiology of Aging*, **2011**, 32, 210-22	5.6	34
47	Factors of risk and maintenance for eating disorders: psychometric exploration of the cross-cultural questionnaire (CCQ) across five European countries. *Clinical Psychology and Psychotherapy*, **2011**, 18, 535-52	2.9	10

46	Meta-analysis of the association between variants in SORL1 and Alzheimer disease. *Archives of Neurology*, **2011**, 68, 99-106		135
45	Immunoproteasome LMP2 60HH variant alters MBP epitope generation and reduces the risk to develop multiple sclerosis in Italian female population. *PLoS ONE*, **2010**, 5, e9287	3.7	51
44	Glucocorticoid receptor gene polymorphisms in Italian patients with eating disorders and obesity. *Psychiatric Genetics*, **2010**, 20, 282-8	2.9	26
43	Lack of implication for CALHM1 P86L common variation in Italian patients with early and late onset Alzheimer's disease. *Journal of Alzheimer's Disease*, **2010**, 20, 37-41	4.3	14
42	Role of the neurotrophin network in eating disorders' subphenotypes: body mass index and age at onset of the disease. *Journal of Psychiatric Research*, **2010**, 44, 834-40	5.2	7
41	Variable epilepsy phenotypes associated with a familial intragenic deletion of the SCN1A gene. *Epilepsia*, **2010**, 51, 2474-7	6.4	43
40	Implication of sex and SORL1 variants in italian patients with Alzheimer disease. *Archives of Neurology*, **2009**, 66, 1260-6		34
39	Associations of individual and family eating patterns during childhood and early adolescence: a multicentre European study of associated eating disorder factors. *British Journal of Nutrition*, **2009**, 101, 909-18	3.6	17
38	Implication of GAB2 gene polymorphism in Italian patients with Alzheimer's disease. *Journal of Alzheimer's Disease*, **2009**, 16, 513-5	4.3	19
37	Codon 129 polymorphism of prion protein gene in sporadic Alzheimer's disease. *European Journal of Neurology*, **2008**, 15, 173-8	6	11
36	KIBRA gene variants are associated with episodic memory performance in subjective memory complaints. *Neuroscience Letters*, **2008**, 436, 145-7	3.3	43
35	Lack of association between TNF-alpha polymorphisms and Alzheimer's disease in an Italian cohort. *Neuroscience Letters*, **2008**, 446, 139-42	3.3	18
34	Present and lifetime comorbidity of tobacco, alcohol and drug use in eating disorders: a European multicenter study. *Drug and Alcohol Dependence*, **2008**, 97, 169-79	4.9	57
33	Association of NTRK3 and its interaction with NGF suggest an altered cross-regulation of the neurotrophin signaling pathway in eating disorders. *Human Molecular Genetics*, **2008**, 17, 1234-44	5.6	42
32	Mutational screening analysis of DHCR24/seladin-1 gene in Italian familial Alzheimer's disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 117-9	3.5	9
31	Angiotensin converting enzyme insertion/deletion polymorphism in sporadic and familial Alzheimer's disease and longevity. *Archives of Gerontology and Geriatrics*, **2007**, 45, 201-6	4	21
30	Testing for linkage and association across the dihydrolipoyl dehydrogenase gene region with Alzheimer's disease in three sample populations. *Neurochemical Research*, **2007**, 32, 857-69	4.6	10
29	No association between the LRRK2 G2019S mutation and Alzheimer's disease in Italy. *Cellular and Molecular Neurobiology*, **2007**, 27, 877-81	4.6	12

28	Fragile X Syndrome vs Fragile X-Associated Tremor/Ataxia Syndrome: Reply. *Archives of Neurology*, **2007**, 64, 289		2
27	Association of IL10 promoter polymorphism in Italian Alzheimer's disease. *Neuroscience Letters*, **2007**, 418, 262-5	3.3	34
26	Cystatin C and apoe polymorphisms in Italian Alzheimer's disease. *Neuroscience Letters*, **2006**, 392, 110-3	3.3	7
25	Association analysis of the paraoxonase-1 gene with Alzheimer's disease. *Neuroscience Letters*, **2006**, 408, 199-202	3.3	27
24	Lack of association between the CYP46 gene polymorphism and Italian late-onset sporadic Alzheimer's disease. *Neurobiology of Aging*, **2006**, 27, 773.e1-773.e3	5.6	20
23	Fragile X premutation with atypical symptoms at onset. *Archives of Neurology*, **2006**, 63, 1135-8		22
22	Case-control and combined family trios analysis of three polymorphisms in the ghrelin gene in European patients with anorexia and bulimia nervosa. *Psychiatric Genetics*, **2006**, 16, 51-2	2.9	37
21	Insulin degrading enzyme and alpha-3 catenin polymorphisms in Italian patients with Alzheimer disease. *Alzheimer Disease and Associated Disorders*, **2005**, 19, 246-7	2.5	16
20	Association of BDNF with restricting anorexia nervosa and minimum body mass index: a family-based association study of eight European populations. *European Journal of Human Genetics*, **2005**, 13, 428-34	5.3	115
19	Cholesteryl ester transfer protein (CETP) I405V polymorphism and longevity in Italian centenarians. *Mechanisms of Ageing and Development*, **2005**, 126, 826-8	5.6	51
18	The urokinase-plasminogen activator (PLAU) gene is not associated with late onset Alzheimer's disease. *Neurogenetics*, **2005**, 6, 53-4	3	12
17	Alzheimer's disease: role of size and location of white matter changes in determining cognitive deficits. *Dementia and Geriatric Cognitive Disorders*, **2005**, 20, 358-66	2.6	40
16	Association of BDNF with anorexia, bulimia and age of onset of weight loss in six European populations. *Human Molecular Genetics*, **2004**, 13, 1205-12	5.6	168
15	Brain-derived neurotrophic factor genetic variants are not susceptibility factors to Alzheimer's disease in Italy. *Annals of Neurology*, **2004**, 55, 447-8	9.4	39
14	Psychopathological traits and 5-HT2A receptor promoter polymorphism (-1438 G/A) in patients suffering from Anorexia Nervosa and Bulimia Nervosa. *Neuroscience Letters*, **2004**, 365, 92-6	3.3	31
13	Brain-derived neurotrophic factor, apolipoprotein E genetic variants and cognitive performance in Alzheimer's disease. *Neuroscience Letters*, **2004**, 367, 379-83	3.3	48
12	Identification of new presenilin gene mutations in early-onset familial Alzheimer disease. *Archives of Neurology*, **2003**, 60, 1541-4		37
11	Brain metabolic differences between sporadic and familial Alzheimer's disease. *Neurology*, **2003**, 61, 1138-40	6.5	20

10	Lack of association between NOS3 poly morphism and Italian sporadic and familial Alzheimer's disease. *Journal of Neurology*, **2002**, 249, 110-1	5.5	14
9	A family with spinocerebellar ataxia type 8 expansion and vitamin E deficiency ataxia. *Archives of Neurology*, **2002**, 59, 1952-3		13
8	5-HT2A receptor gene polymorphism and eating disorders. *Neuroscience Letters*, **2002**, 323, 105-8	3.3	69
7	Cathepsin D polymorphism in Italian sporadic and familial Alzheimer's disease. *Neuroscience Letters*, **2002**, 328, 273-6	3.3	22
6	Clinical and genetic analysis of an Italian family with Machado-Joseph disease. *Journal of Neurology*, **2001**, 248, 717-9	5.5	2
5	Genetic risk factors in familial Alzheimer's disease. *Mechanisms of Ageing and Development*, **2001**, 122, 1951-60	5.6	22
4	Genetic and clinical analysis of spinocerebellar ataxia type 8 repeat expansion in Italy. *Archives of Neurology*, **2001**, 58, 1856-9		32
3	Clinical and genetic analysis of hereditary and sporadic ataxia in central Italy. *Brain Research Bulletin* , **2001**, 56, 363-6	3.9	6
2	Alpha2-macroglobulin polymorphisms in Italian sporadic and familial Alzheimer's disease. *Neuroscience Letters*, **2001**, 299, 9-12	3.3	15
1	Acylphosphatase expression during macrophage differentiation and activation of U-937 cell line. *Biochimie*, **1999**, 81, 1031-5	4.6	25