Sofia Douzgou

List of Publications by Year in Descending Order

83 papers	2,319 citations	23 h-index	47 g-index
103 ext. papers	3,587 ext. citations	7.1 avg, IF	6.1 L-index

#	Paper	IF	Citations
83	Genetic disorders and genetic variants **2022**, 1-5		
82	A standard of care for individuals with PIK3CA-related disorders: An international expert consensus statement. *Clinical Genetics*, **2022**, 101, 32-47	4	3
81	Bi-allelic mutation of CTNNB1 causes a severe form of syndromic microphthalmia, persistent foetal vasculature and vitreoretinal dysplasia.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 110	4.2	0
80	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. *Npj Genomic Medicine*, **2021**, 6, 92	6.2	0
79	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *New England Journal of Medicine*, **2021**, 385, 1868-1880	59.2	34
78	Personalised virtual gene panels reduce interpretation workload and maintain diagnostic rates of proband-only clinical exome sequencing for rare disorders. *Journal of Medical Genetics*, **2021**,	5.8	2
77	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. *Seizure: the Journal of the British Epilepsy Association*, **2021**, 88, 60-72	3.2	3
76	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. *American Journal of Human Genetics*, **2021**, 108, 857-873	11	2
75	The diagnostic utility of clinical exome sequencing in 60 patients with hearing loss disorders: A single-institution experience. *Clinical Otolaryngology*, **2021**, 46, 1257-1262	1.8	0
74	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. *Genetics in Medicine*, **2021**, 23, 2138-2149	8.1	1
73	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	5.3	4
72	Lack of resemblance between Myhre syndrome and other "segmental progeroid" syndromes warrants restraint in applying this classification. *GeroScience*, **2021**, 43, 459-461	8.9	
71	The Role of the European Society of Human Genetics in Delivering Genomic Education. *Frontiers in Genetics*, **2021**, 12, 693952	4.5	2
70	The contribution of X-linked coding variation to severe developmental disorders. *Nature Communications*, **2021**, 12, 627	17.4	5
69	Embryonal sarcoma of the liver in a girl with Cockayne syndrome. *Clinical Genetics*, **2021**,	4	
68	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139
67	SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. *Genetics in Medicine*, **2020**, 22, 1498-1506	8.1	8

66	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. *Genetics in Medicine*, **2020**, 22, 1005-1014	8.1	35
65	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. *Orphanet Journal of Rare Diseases*, **2020**, 15, 103	4.2	14
64	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. *American Journal of Human Genetics*, **2020**, 106, 234-245	11	22
63	Clinical utility of genetic testing in 201 preschool children with inherited eye disorders. *Genetics in Medicine*, **2020**, 22, 745-751	8.1	21
62	Diagnostic yield of panel-based genetic testing in syndromic inherited retinal disease. *European Journal of Human Genetics*, **2020**, 28, 576-586	5.3	11
61	Evidence for 28 genetic disorders discovered by combining healthcare and research data. *Nature*, **2020**, 586, 757-762	50.4	103
60	Modelling the developmental spliceosomal craniofacial disorder Burn-McKeown syndrome using induced pluripotent stem cells. *PLoS ONE*, **2020**, 15, e0233582	3.7	6
59	Congenital cataracts in females caused by BCOR mutations; report of six further families demonstrating clinical variability and diverse genetic mechanisms. *European Journal of Medical Genetics*, **2020**, 63, 103658	2.6	1
58	Modelling the developmental spliceosomal craniofacial disorder Burn-McKeown syndrome using induced pluripotent stem cells **2020**, 15, e0233582		
57	Modelling the developmental spliceosomal craniofacial disorder Burn-McKeown syndrome using induced pluripotent stem cells **2020**, 15, e0233582		
56	Modelling the developmental spliceosomal craniofacial disorder Burn-McKeown syndrome using induced pluripotent stem cells **2020**, 15, e0233582		
55	Modelling the developmental spliceosomal craniofacial disorder Burn-McKeown syndrome using induced pluripotent stem cells **2020**, 15, e0233582		
54	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. *Frontiers in Genetics*, **2019**, 10, 611	4.5	7
53	PEDIA: prioritization of exome data by image analysis. *Genetics in Medicine*, **2019**, 21, 2807-2814	8.1	20
52	Germline selection shapes human mitochondrial DNA diversity. *Science*, **2019**, 364,	33.3	105
51	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
50	The clinical presentation caused by truncating CHD8 variants. *Clinical Genetics*, **2019**, 96, 72-84	4	20
49	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956	11	17

48	The CHD8 overgrowth syndrome: A detailed evaluation of an emerging overgrowth phenotype in 27 patients. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2019**, 181, 557-564	3.1	17
47	Null variants and deletions in BRWD3 cause an X-linked syndrome of mild-moderate intellectual disability, macrocephaly, and obesity: A series of 17 patients. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2019**, 181, 638-643	3.1	3
46	Clinical and genetic variability in children with partial albinism. *Scientific Reports*, **2019**, 9, 16576	4.9	14
45	Chromosome 1q31.2q32.1 deletion in an adult male with intellectual disability, dysmorphic features and obesity. *Clinical Dysmorphology*, **2019**, 28, 131-136	0.9	2
44	A patient with a novel CNTNAP2 homozygous variant: further delineation of the CASPR2 deficiency syndrome and review of the literature. *Clinical Dysmorphology*, **2019**, 28, 66-70	0.9	3
43	Pathogenicity and selective constraint on variation near splice sites. *Genome Research*, **2019**, 29, 159-170	9.7	29
42	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108
41	Hypoglycaemia represents a clinically significant manifestation of PIK3CA- and CCND2-associated segmental overgrowth. *Clinical Genetics*, **2018**, 93, 687-692	4	11
40	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	11	24
39	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. *Journal of Medical Genetics*, **2018**, 55, 104-113	5.8	35
38	A case of diencephalic syndrome presenting with isolated lipodystrophy. *Clinical Dysmorphology*, **2018**, 27, 122-125	0.9	
37	Prevalence and architecture of de novo mutations in developmental disorders. *Nature*, **2017**, 542, 433-438	38.4	765
36	Interrupted/bipartite clavicle as a diagnostic clue in Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1115-1118	2.5	3
35	Diagnosing childhood-onset inborn errors of metabolism by next-generation sequencing. *Archives of Disease in Childhood*, **2017**, 102, 1019-1029	2.2	24
34	The evolving craniofacial phenotype of a patient with Sensenbrenner syndrome caused by IFT140 compound heterozygous mutations. *Clinical Dysmorphology*, **2017**, 26, 247-251	0.9	9
33	Severe intellectual disability in a patient with Burn-McKeown syndrome. *Clinical Dysmorphology*, **2017**, 26, 193-194	0.9	3
32	Panel-Based Clinical Genetic Testing in 85 Children with Inherited Retinal Disease. *Ophthalmology*, **2017**, 124, 985-991	7.3	31
31	Further Clinical Delineation of the MEF2C Haploinsufficiency Syndrome: Report on New Cases and Literature Review of Severe Neurodevelopmental Disorders Presenting with Seizures, Absent Speech, and Involuntary Movements. *Journal of Pediatric Genetics*, **2017**, 6, 129-141	0.7	24

30	Not all epileptic encephalopathies are Dravet syndrome: Early profound Thr226Met phenotype. *Neurology*, **2017**, 89, 1035-1042	6.5	62
29	Diagnosing the cause of bilateral paediatric cataracts: comparison of standard testing with a next-generation sequencing approach. *Eye*, **2016**, 30, 1175-81	4.4	19
28	Dysmorphology services: a snapshot of current practices and a vision for the future. *Clinical Genetics*, **2016**, 89, 27-33	4	4
27	Collaborative Crowdsourcing for the Diagnosis of Rare Genetic Syndromes: The DYSCERNE Experience. *Public Health Genomics*, **2016**, 19, 19-24	1.9	10
26	Severe constipation in a patient with Myhre syndrome: a case report. *Clinical Dysmorphology*, **2016**, 25, 54-7	0.9	4
25	Gain-of-Function Mutations in RARB Cause Intellectual Disability with Progressive Motor Impairment. *Human Mutation*, **2016**, 37, 786-93	4.7	24
24	Two patients with chromosome 22q11.2 deletion presenting with childhood obesity and hyperphagia. *European Journal of Medical Genetics*, **2016**, 59, 401-3	2.6	5
23	35-Year Follow-Up of a Case of Ring Chromosome 2: Array-CGH Analysis and Literature Review of the Ring Syndrome. *Cytogenetic and Genome Research*, **2015**, 145, 6-13	1.9	0
22	Niemann-Pick type C disease: a novel NPC1 mutation segregating in a Greek island. *Clinical Genetics* , **2014**, 85, 543-7	4	9
21	Compound heterozygosity of low-frequency promoter deletions and rare loss-of-function mutations in TXNL4A causes Burn-McKeown syndrome. *American Journal of Human Genetics*, **2014**, 95, 698-707	11	45
20	Dysmorphology at a distance: results of a web-based diagnostic service. *European Journal of Human Genetics*, **2014**, 22, 327-32	5.3	12
19	Neurocognitive effects of methylphenidate on ADHD children with different DAT genotypes: a longitudinal open label trial. *European Journal of Paediatric Neurology*, **2013**, 17, 407-14	3.8	23
18	Haploinsufficiency of SOX5, a member of the SOX (SRY-related HMG-box) family of transcription factors is a cause of intellectual disability. *European Journal of Medical Genetics*, **2013**, 56, 108-13	2.6	22
17	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. *Archives of Disease in Childhood*, **2012**, 97, 812-7	2.2	27
16	Clinical variability of genetic isolates of Cohen syndrome. *Clinical Genetics*, **2011**, 79, 501-6	4	38
15	Generalized pulp stones: report of a case with 6-year follow-up. *International Endodontic Journal*, **2011**, 44, 976-82	5.4	4
14	Mutation spectrum of MLL2 in a cohort of Kabuki syndrome patients. *Orphanet Journal of Rare Diseases*, **2011**, 6, 38	4.2	68
13	Ophthalmic findings in the Greek isolate of Cohen syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 534-9	2.5	10

12	Complex distal 10q rearrangement in a girl with mild intellectual disability: follow up of the patient and review of the literature of non-acrocentric satellited chromosomes. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2841-54	2.5	7
11	Gingival overgrowth, congenital generalized hypertrichosis, mental retardation and epilepsy: case report and overview. *Clinical Dysmorphology*, **2009**, 18, 205-8	0.9	13
10	Epistasis between dopamine regulating genes identifies a nonlinear response of the human hippocampus during memory tasks. *Biological Psychiatry*, **2008**, 64, 226-34	7.9	73
9	Silver-Russell syndrome following in vitro fertilization. *Pediatric and Developmental Pathology*, **2008** , 11, 329-31	2.2	15
8	Compound heterozygosity for GDF5 in Du Pan type chondrodysplasia. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2116-21	2.5	15
7	COMT Val158Met polymorphism predicts negative symptoms response to treatment with olanzapine in schizophrenia. *Schizophrenia Research*, **2007**, 95, 253-5	3.6	46
6	Reassessment of holoprosencephaly-diencephalic hamartoblastoma (HDH) association. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 277-84	2.5	7
5	LGI1 gene mutation screening in sporadic partial epilepsy with auditory features. *Journal of Neurology*, **2005**, 252, 62-6	5.5	10
4	Preliminary data suggest that mutations in the CgRP pathway are not involved in human sporadic cryptorchidism. *Journal of Endocrinological Investigation*, **2004**, 27, 760-4	5.2	12
3	Delineation of the First Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency		1
2	Prevalence, phenotype and architecture of developmental disorders caused by de novo mutation: The Deciphering Developmental Disorders Study		10
1	Pathogenicity and selective constraint on variation near splice sites		2