Peter D Turnpenny

List of Publications by Year
in descending order

53 papers	3,114 citations	218677 26 h-index	182427 51 g-index
58 all docs	58 docs citations	58 times ranked	6242 citing authors

#	Article	IF	Citations
1	Further delineation of phenotypic spectrum of <scp>SCN2A</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 867-877.	1.2	3
2	Natural history of NF1 c.2970_2972del p.(Met992del): confirmation of a low risk of complications in a longitudinal study. European Journal of Human Genetics, 2022, 30, 291-297.	2.8	5
3	Understanding the new <scp>BRD4</scp>â€related syndrome: Clinical and genomic delineation with an international cohort study. Clinical Genetics, 2022, 102, 117-122.	2.0	3
4	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	2.4	34
5	Consolidating biallelic SDHD variants as a cause of mitochondrial complex II deficiency. European Journal of Human Genetics, 2021, 29, 1570-1576.	2.8	3
6	Final Exon Frameshift Biallelic PTPN23 Variants Are Associated with Microcephalic Complex Hereditary Spastic Paraplegia. Brain Sciences, 2021, 11, 614.	2.3	5
7	Delineating the <scp>Smithâ€Kingsmore</scp> syndrome phenotype: Investigation of 16 patients with the <scp>MTOR</scp> c.<scp>5395G</scp>Â>â€‰A p.(<scp>Glu1799Lys</scp>) missense variant. American Journal of Medical Genetics, Part A, 2021, 185, 2445-2454.	1.2	4
8	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
9	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	6.2	11
10	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	2.4	53
11	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
12	Ectrodactylyâ€ectodermal dysplasiaâ€clefting syndrome presenting with bilateral choanal atresia and rectal stenosis. American Journal of Medical Genetics, Part A, 2020, 182, 1939-1943.	1.2	2
13	Opposite Modulation of RAC1 by Mutations in TRIO Is Associated with Distinct, Domain-Specific Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 338-355.	6.2	58
14	Primrose syndrome: Characterization of the phenotype in 42 patients. Clinical Genetics, 2020, 97, 890-901.	2.0	18
15	Clinical findings of 21 previously unreported probands with HNRNPUâ€related syndrome and comprehensive literature review. American Journal of Medical Genetics, Part A, 2020, 182, 1637-1654.	1.2	19
16	Amniotic band sequence in paternal halfâ€siblings with vascular Ehlersâ€“Danlos syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 553-556.	1.2	5
17	Diagnosis and management of individuals with Fetal Valproate Spectrum Disorder; a consensus statement from the European Reference Network for Congenital Malformations and Intellectual Disability. Orphanet Journal of Rare Diseases, 2019, 14, 180.	2.7	33
18	Delineation of dominant and recessive forms of LZTR1â€associated Noonan syndrome. Clinical Genetics, 2019, 95, 693-703.	2.0	35

#	Article	IF	Citations
19	Recontacting patients in clinical genetics services: recommendations of the European Society of Human Genetics. European Journal of Human Genetics, 2019, 27, 169-182.	2.8	65
20	27 years of prenatal diagnosis for Huntington disease in the United Kingdom. Genetics in Medicine, 2019, 21, 1639-1643.	2.4	9
21	Extending the clinical and genetic spectrum of ARID2 related intellectual disability. A case series of 7 patients. European Journal of Medical Genetics, 2019, 62, 27-34.	1.3	13
22	Recontacting or not recontacting? A survey of current practices in clinical genetics centres in Europe. European Journal of Human Genetics, 2018, 26, 946-954.	2.8	33
23	Diagnosis of lethal or prenatalâ€onset autosomal recessive disorders by parental exome sequencing. Prenatal Diagnosis, 2018, 38, 33-43.	2.3	64
24	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
25	Dimensions of responsibility in medical genetics: exploring the complexity of the â€œduty to recontactâ€. New Genetics and Society, 2018, 37, 187-206.	1.2	4
26	Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. American Journal of Human Genetics, 2018, 103, 786-793.	6.2	17
27	De novo gainâ€ofâ€function variants in KCNT2 as a novel cause of developmental and epileptic encephalopathy. Annals of Neurology, 2018, 83, 1198-1204.	5.3	41
28	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	1.8	75
29	Recontacting in clinical genetics and genomic medicine? We need to talk about it. European Journal of Human Genetics, 2017, 25, 520-521.	2.8	16
30	A 'joint venture' model of recontacting in clinical genomics: challenges for responsible implementation. European Journal of Medical Genetics, 2017, 60, 403-409.	1.3	36
31	Polycystic Kidney Disease with Hyperinsulinemic Hypoglycemia Caused by a Promoter Mutation in Phosphomannomutase 2. Journal of the American Society of Nephrology: JASN, 2017, 28, 2529-2539.	6.1	99
32	Recontacting in clinical practice: an investigation of the views of healthcare professionals and clinical scientists in the United Kingdom. European Journal of Human Genetics, 2017, 25, 275-279.	2.8	35
33	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
34	Recontacting in clinical practice: the views and expectations of patients in the United Kingdom. European Journal of Human Genetics, 2017, 25, 1106-1112.	2.8	23
35	MYT1L mutations cause intellectual disability and variable obesity by dysregulating gene expression and development of the neuroendocrine hypothalamus. PLoS Genetics, 2017, 13, e1006957.	3.5	60
36	Chromosome 17q12 microdeletions but not intragenic HNF1B mutations link developmental kidney disease and psychiatric disorder. Kidney International, 2016, 90, 203-211.	5.2	64

#	Article	IF	Citations
37	Recontact in clinical practice: a survey of clinical genetics services in the United Kingdom. Genetics in Medicine, 2016, 18, 876-881.	2.4	40
38	Digesting GWAS. Cellular and Molecular Gastroenterology and Hepatology, 2016, 2, 542-543.	4.5	0
39	Clinical and genetic aspects of KBG syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2835-2846.	1.2	87
40	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. European Journal of Human Genetics, 2015, 23, 401-404.	2.8	51
41	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	21.4	133
42	Whole exome sequencing in family trios revealsde novomutations inPURAas a cause of severe neurodevelopmental delay and learning disability. Journal of Medical Genetics, 2014, 51, 806-813.	3.2	73
43	Factors determining penetrance in familial atypical haemolytic uraemic syndrome. Journal of Medical Genetics, 2014, 51, 756-764.	3.2	28
44	From the Ottomans to the present day: 150 years of Scottish medical charity in the Holy Land. BMJ, The, 2013, 347, f6994-f6994.	6.0	0
45	Alagille syndrome: pathogenesis, diagnosis and management. European Journal of Human Genetics, 2012, 20, 251-257.	2.8	319
46	Defective Somitogenesis and Abnormal Vertebral Segmentation in Man. Advances in Experimental Medicine and Biology, 2008, 638, 164-189.	1.6	8
47	Abnormal vertebral segmentation and the notch signaling pathway in man. Developmental Dynamics, 2007, 236, 1456-1474.	1.8	143
48	Of eponyms, acronyms and ... orthonyms. Nature Reviews Genetics, 2003, 4, 152-156.	16.3	8
49	Mutations in the human Delta homologue, DLL3, cause axial skeletal defects in spondylocostal dysostosis. Nature Genetics, 2000, 24, 438-441.	21.4	362
50	Fetal anticonvulsant syndrome and mutation in the maternal MTHFR gene. Clinical Genetics, 1999, 56, 216-220.	2.0	58
51	Delineation of two distinct 6p deletion syndromes. Human Genetics, 1999, 104, 64-72.	3.8	108
52	Bilateral microphthalmia, esophageal atresia, and cryptorchidism: The anophthalmia-esophageal-genital syndrome. , 1997, 70, 171-173.		22
53	Human Genetics and Genetic Enhancement. , 0, , 241-264.		1