Peter D Turnpenny

List of Publications by Year
in descending order

53 papers	3,114 citations	218592 26 h-index	182361 51 g-index
58 all docs	58 docs citations	58 times ranked	6242 citing authors

#	Article	IF	Citations
1	Mutations in the human Delta homologue, DLL3, cause axial skeletal defects in spondylocostal dysostosis. Nature Genetics, 2000, 24, 438-441.	9.4	362
2	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	13.7	343
3	Alagille syndrome: pathogenesis, diagnosis and management. European Journal of Human Genetics, 2012, 20, 251-257.	1.4	319
4	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	9.4	186
5	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	6.0	158
6	Abnormal vertebral segmentation and the notch signaling pathway in man. Developmental Dynamics, 2007, 236, 1456-1474.	0.8	143
7	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	9.4	133
8	Delineation of two distinct 6p deletion syndromes. Human Genetics, 1999, 104, 64-72.	1.8	108
9	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.	3.0	99
10	Clinical and genetic aspects of KBG syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2835-2846.	0.7	87
11	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	0.9	75
12	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.	1.5	73
13	Recontacting patients in clinical genetics services: recommendations of the European Society of Human Genetics. European Journal of Human Genetics, 2019, 27, 169-182.	1.4	65
14	Chromosome 17q12 microdeletions but not intragenic HNF1B mutations link developmental kidney disease and psychiatric disorder. Kidney International, 2016, 90, 203-211.	2.6	64
15	Diagnosis of lethal or prenatalâ€onset autosomal recessive disorders by parental exome sequencing. Prenatal Diagnosis, 2018, 38, 33-43.	1.1	64
16	MYT1L mutations cause intellectual disability and variable obesity by dysregulating gene expression and development of the neuroendocrine hypothalamus. PLoS Genetics, 2017, 13, e1006957.	1.5	60
17	Fetal anticonvulsant syndrome and mutation in the maternal MTHFR gene. Clinical Genetics, 1999, 56, 216-220.	1.0	58
18	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.	2.6	58

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19	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	1.1	53
20	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. European Journal of Human Genetics, 2015, 23, 401-404.	1.4	51
21	De novo gainâ€ofâ€function variants in KCNT2 as a novel cause of developmental and epileptic encephalopathy. Annals of Neurology, 2018, 83, 1198-1204.	2.8	41
22	Recontact in clinical practice: a survey of clinical genetics services in the United Kingdom. Genetics in Medicine, 2016, 18, 876-881.	1.1	40
23	A 'joint venture' model of recontacting in clinical genomics: challenges for responsible implementation. European Journal of Medical Genetics, 2017, 60, 403-409.	0.7	36
24	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.	1.4	35
25	Delineation of dominant and recessive forms of LZTR1â€associated Noonan syndrome. Clinical Genetics, 2019, 95, 693-703.	1.0	35
26	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
27	Recontacting or not recontacting? A survey of current practices in clinical genetics centres in Europe. European Journal of Human Genetics, 2018, 26, 946-954.	1.4	33
28	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.	1.2	33
29	Factors determining penetrance in familial atypical haemolytic uraemic syndrome. Journal of Medical Genetics, 2014, 51, 756-764.	1.5	28
30	Recontacting in clinical practice: the views and expectations of patients in the United Kingdom. European Journal of Human Genetics, 2017, 25, 1106-1112.	1.4	23
31	Bilateral microphthalmia, esophageal atresia, and cryptorchidism: The anophthalmia-esophageal-genital syndrome. , 1997, 70, 171-173.		22
32	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.	0.7	19
33	Primrose syndrome: Characterization of the phenotype in 42 patients. Clinical Genetics, 2020, 97, 890-901.	1.0	18
34	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.	2.6	17
35	Recontacting in clinical genetics and genomic medicine? We need to talk about it. European Journal of Human Genetics, 2017, 25, 520-521.	1.4	16
36	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16

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37	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.	0.7	13
38	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.	2.6	11
39	27 years of prenatal diagnosis for Huntington disease in the United Kingdom. Genetics in Medicine, 2019, 21, 1639-1643.	1.1	9
40	Of eponyms, acronyms and ... orthonyms. Nature Reviews Genetics, 2003, 4, 152-156.	7.7	8
41	Defective Somitogenesis and Abnormal Vertebral Segmentation in Man. Advances in Experimental Medicine and Biology, 2008, 638, 164-189.	0.8	8
42	Final Exon Frameshift Biallelic PTPN23 Variants Are Associated with Microcephalic Complex Hereditary Spastic Paraplegia. Brain Sciences, 2021, 11, 614.	1.1	5
43	Amniotic band sequence in paternal halfâ€siblings with vascular Ehlersâ€“Danlos syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 553-556.	0.7	5
44	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.	1.4	5
45	Dimensions of responsibility in medical genetics: exploring the complexity of the â€œduty to recontactâ€• New Genetics and Society, 2018, 37, 187-206.	0.7	4
46	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.	0.7	4
47	Consolidating biallelic SDHD variants as a cause of mitochondrial complex II deficiency. European Journal of Human Genetics, 2021, 29, 1570-1576.	1.4	3
48	Further delineation of phenotypic spectrum of <scp>SCN2A</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 867-877.	0.7	3
49	Understanding the new <scp>BRD4</scp>â€related syndrome: Clinical and genomic delineation with an international cohort study. Clinical Genetics, 2022, 102, 117-122.	1.0	3
50	Ectrodactylyâ€ectodermal dysplasiaâ€clefting syndrome presenting with bilateral choanal atresia and rectal stenosis. American Journal of Medical Genetics, Part A, 2020, 182, 1939-1943.	0.7	2
51	Human Genetics and Genetic Enhancement. , 0, , 241-264.		1
52	From the Ottomans to the present day: 150 years of Scottish medical charity in the Holy Land. BMJ, The, 2013, 347, f6994-f6994.	3.0	0
53	Digesting GWAS. Cellular and Molecular Gastroenterology and Hepatology, 2016, 2, 542-543.	2.3	0