David J Bunyan

List of Publications by Year
in descending order

21	617	758635 12	642321 23
papers	citations	h-index	g-index

23	23	23	1252
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Collagen (COL4A) mutations are the most frequent mutations underlying adult focal segmental glomerulosclerosis. Nephrology Dialysis Transplantation, 2016, 31, 961-970.	0.4	199
2	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	1.1	99
3	Identification of a functionally significant tri-allelic genotype in the Tyrosinase gene (TYR) causing hypomorphic oculocutaneous albinism (OCA1B). Scientific Reports, 2017, 7, 4415.	1.6	47
4	Diagnostic screening identifies a wide range of mutations involving the SHOX gene, including a common 47.5â€‰kb deletion 160â€‰kb downstream with a variable phenotypic effect. American Journal of Medical Genetics, Part A, 2013, 161, 1329-1338.	0.7	32
5	Germline and somatic mosaicism in a female carrier of Duchenne muscular dystrophy. Human Genetics, 1994, 93, 541-4.	1.8	31
6	Further defining the phenotypic spectrum of B4GALT7 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 1556-1563.	0.7	31
7	Duplications upstream and downstream of SHOX identified as novel causes of Leriâ€“Weill dyschondrosteosis or idiopathic short stature. American Journal of Medical Genetics, Part A, 2016, 170, 949-957.	0.7	26
8	A novel FOXP3 mutation causing fetal akinesia and recurrent male miscarriages. Clinical Immunology, 2015, 161, 284-285.	1.4	25
9	Homozygosity for a novel deletion downstream of the SHOX gene provides evidence for an additional long range regulatory region with a mild phenotypic effect. American Journal of Medical Genetics, Part A, 2014, 164, 2764-2768.	0.7	18
10	Comparison of Mutation Profiles in the Duchenne Muscular Dystrophy Gene among Populations: Implications for Potential Molecular Therapies. International Journal of Molecular Sciences, 2015, 16, 5334-5346.	1.8	15
11	Exome analysis resolves differential diagnosis of familial kidney disease and uncovers a potential confounding variant. Genetical Research, 2013, 95, 165-173.	0.3	14
12	Screening of a large cohort of blepharophimosis, ptosis, and epicanthus inversus syndrome patients reveals a very strong paternal inheritance bias and a wide spectrum of novel FOXL2 mutations. European Journal of Medical Genetics, 2019, 62, 103668.	0.7	14
13	Screening of a large PAX6 cohort identified many novel variants and emphasises the importance of the paired and homeobox domains. European Journal of Medical Genetics, 2020, 63, 103940.	0.7	13
14	Multiple De Novo Mutations in the MECP2 Gene. Genetic Testing and Molecular Biomarkers, 2008, 12, 373-375.	1.7	12
15	A small intraexonic deletion within the dystrophin gene suggests a possible mechanism of mutagenesis. Human Genetics, 1997, 99, 658-662.	1.8	9
16	Screening of a large Rubinsteinâ€“Taybi cohort identified many novel variants and emphasizes the importance of the CREBBP histone acetyltransferase domain. American Journal of Medical Genetics, Part A, 2020, 182, 2508-2520.	0.7	9
17	Detection of Partial Deletions of Y-chromosome AZFc in Infertile Men Using the Multiplex Ligation-dependent Probe Amplification Assay. Journal of Reproduction and Infertility, 2012, 13, 174-8.	1.0	9
18	<scp>AIFM1</scp>â€associated Xâ€linked spondylometaphyseal dysplasia with cerebral hypomyelination. American Journal of Medical Genetics, Part A, 2021, 185, 1228-1235.	0.7	5

#	Article	IF	Citations
19	Primary Clear Cell Microcystic Adenoma of the Sinonasal Cavity: Pathological or Fortuitous Association?. Case Reports in Pathology, 2017, 2017, 1-5.	0.2	3
20	13q Deletion Size Predicts Disease Progression and Response to Treatment in Patients with Chronic Lymphocytic Leukaemia.. Blood, 2009, 114, 671-671.	0.6	3
21	Rare dosage abnormalities flanking the SHOX gene. Egyptian Journal of Medical Human Genetics, 2021, 22, .	0.5	1