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	<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
2	Rare dosage abnormalities flanking the SHOX gene. Egyptian Journal of Medical Human Genetics, 2021, 22, .	0.5	1
3	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
4	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
5	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	1.1	99
6	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
7	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
8	Primary Clear Cell Microcystic Adenoma of the Sinonasal Cavity: Pathological or Fortuitous Association?. Case Reports in Pathology, 2017, 2017, 1-5.	0.2	3
9	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
10	Further defining the phenotypic spectrum of B4GALT7 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 1556-1563.	0.7	31
11	Collagen (COL4A) mutations are the most frequent mutations underlying adult focal segmental glomerulosclerosis. Nephrology Dialysis Transplantation, 2016, 31, 961-970.	0.4	199
12	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
13	A novel FOXP3 mutation causing fetal akinesia and recurrent male miscarriages. Clinical Immunology, 2015, 161, 284-285.	1.4	25
14	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
15	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
16	Exome analysis resolves differential diagnosis of familial kidney disease and uncovers a potential confounding variant. Genetical Research, 2013, 95, 165-173.	0.3	14
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	13q Deletion Size Predicts Disease Progression and Response to Treatment in Patients with Chronic Lymphocytic Leukaemia.. Blood, 2009, 114, 671-671.	0.6	3

#	Article	IF	Citations
19	Multiple De Novo Mutations in the MECP2 Gene. Genetic Testing and Molecular Biomarkers, 2008, 12, 373-375.	1.7	12
20	A small intraexonic deletion within the dystrophin gene suggests a possible mechanism of mutagenesis. Human Genetics, 1997, 99, 658-662.	1.8	9
21	Germline and somatic mosaicism in a female carrier of Duchenne muscular dystrophy. Human Genetics, 1994, 93, 541-4.	1.8	31