Julie Vogt

List of Publications by Year
in descending order

27 papers	1,259 citations	430874 18 h-index	501196 28 g-index
28 all docs	28 docs citations	28 times ranked	3140 citing authors

#	Article	IF	Citations
1	Elucidating the molecular mechanisms associated with TARS2-related mitochondrial disease. Human Molecular Genetics, 2022, 31, 523-534.	2.9	12
2	Variants in PHF8 cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology. Human Genetics and Genomics Advances, 2022, 3, 100102.	1.7	5
3	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
4	The gain of function SCN1A disorder spectrum: novel epilepsy phenotypes and therapeutic implications. Brain, 2022, 145, 3816-3831.	7.6	43
5	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. American Journal of Human Genetics, 2021, 108, 951-961.	6.2	26
6	A human importin-Î²-related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8. American Journal of Human Genetics, 2021, 108, 1115-1125.	6.2	10
7	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
8	A recurrent pathogenic variant in <scp>TPM2</scp> reveals further phenotypic and genetic heterogeneity in multiple pterygium syndromeâ€related disorders. Clinical Genetics, 2020, 97, 908-914.	2.0	5
9	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
10	PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. Genetics in Medicine, 2019, 21, 2216-2223.	2.4	21
11	PLAA Mutations Cause a Lethal Infantile Epileptic Encephalopathy by Disrupting Ubiquitin-Mediated Endolysosomal Degradation of Synaptic Proteins. American Journal of Human Genetics, 2017, 100, 706-724.	6.2	37
12	Phenotypic Spectrum in Osteogenesis Imperfecta Due to Mutations in TMEM38B: Unraveling a Complex Cellular Defect. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 2019-2028.	3.6	27
13	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. American Journal of Human Genetics, 2017, 100, 138-150.	6.2	52
14	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
15	Compound heterozygous RMND1 gene variants associated with chronic kidney disease, dilated cardiomyopathy and neurological involvement: a case report. BMC Research Notes, 2016, 9, 325.	1.4	15
16	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	3.2	35
17	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	6.2	95
18	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. Journal of Medical Genetics, 2016, 53, 62-72.	3.2	48

#	Article	IF	Citations
19	A microdeletion encompassing PHF21A in an individual with global developmental delay and craniofacial anomalies. American Journal of Medical Genetics, Part A, 2015, 167, 3011-3018.	1.2	16
20	Novel mutations in PIEZO1 cause an autosomal recessive generalized lymphatic dysplasia with non-immune hydrops fetalis. Nature Communications, 2015, 6, 8085.	12.8	247
21	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
22	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	3.2	141
23	Germline mutations in RYR1 are associated with foetal akinesia deformation sequence/lethal multiple pterygium syndrome. Acta Neuropathologica Communications, 2014, 2, 148.	5.2	23
24	Striking intrafamilial phenotypic variability in Aicardiâ€“GoutiÃ¨res syndrome associated with the recurrent Asian founder mutation in RNASEH2C. American Journal of Medical Genetics, Part A, 2013, 161, 338-342.	1.2	28
25	CHRNGgenotypeâ€“phenotype correlations in the multiple pterygium syndromes. Journal of Medical Genetics, 2012, 49, 21-26.	3.2	41
26	Mutation Analysis of CHRNA1, CHRNB1, CHRND, and RAPSN Genes in Multiple Pterygium Syndrome/Fetal Akinesia Patients. American Journal of Human Genetics, 2008, 82, 222-227.	6.2	104
27	The tale of a nail sign in chromosome 4q34 deletion syndrome. Clinical Dysmorphology, 2006, 15, 127-132.	0.3	34