

# Julie Vogt

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Novel mutations in PIEZO1 cause an autosomal recessive generalized lymphatic dysplasia with non-immune hydrops fetalis. <i>Nature Communications</i> , 2015, 6, 8085.	12.8	247
2	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. <i>Journal of Medical Genetics</i> , 2014, 51, 659-668.	3.2	141
3	Mutation Analysis of CHRNA1, CHRNB1, CHRND, and RAPSN Genes in Multiple Pterygium Syndrome/Fetal Akinesia Patients. <i>American Journal of Human Genetics</i> , 2008, 82, 222-227.	6.2	104
4	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. <i>American Journal of Human Genetics</i> , 2016, 98, 373-381.	6.2	95
5	MYT1L mutations cause intellectual disability and variable obesity by dysregulating gene expression and development of the neuroendocrine hypothalamus. <i>PLoS Genetics</i> , 2017, 13, e1006957.	3.5	60
6	Further delineation of the KAT6B molecular and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2015, 23, 1165-1170.	2.8	56
7	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. <i>American Journal of Human Genetics</i> , 2019, 104, 1210-1222.	6.2	56
8	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 138-150.	6.2	52
9	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. <i>Journal of Medical Genetics</i> , 2016, 53, 62-72.	3.2	48
10	The gain of function <i>SCN1A</i> disorder spectrum: novel epilepsy phenotypes and therapeutic implications. <i>Brain</i> , 2022, 145, 3816-3831.	7.6	43
11	<i>CHRNA1</i> genotype-phenotype correlations in the multiple pterygium syndromes. <i>Journal of Medical Genetics</i> , 2012, 49, 21-26.	3.2	41
12	PLAA Mutations Cause a Lethal Infantile Epileptic Encephalopathy by Disrupting Ubiquitin-Mediated Endolysosomal Degradation of Synaptic Proteins. <i>American Journal of Human Genetics</i> , 2017, 100, 706-724.	6.2	37
13	The clinical, biochemical and genetic features associated with <i>RMND1</i> -related mitochondrial disease. <i>Journal of Medical Genetics</i> , 2016, 53, 768-775.	3.2	35
14	The tale of a nail sign in chromosome 4q34 deletion syndrome. <i>Clinical Dysmorphology</i> , 2006, 15, 127-132.	0.3	34
15	Striking intrafamilial phenotypic variability in Aicardi-Goutières syndrome associated with the recurrent Asian founder mutation in <i>RNASEH2C</i> . <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 338-342.	1.2	28
16	Phenotypic Spectrum in Osteogenesis Imperfecta Due to Mutations in TMEM38B: Unraveling a Complex Cellular Defect. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 2019-2028.	3.6	27
17	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. <i>American Journal of Human Genetics</i> , 2021, 108, 951-961.	6.2	26
18	Germline mutations in RYR1 are associated with foetal akinesia deformation sequence/lethal multiple pterygium syndrome. <i>Acta Neuropathologica Communications</i> , 2014, 2, 148.	5.2	23

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19	PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. <i>Genetics in Medicine</i> , 2019, 21, 2216-2223.	2.4	21
20	A microdeletion encompassing <i>PHF21A</i> in an individual with global developmental delay and craniofacial anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3011-3018.	1.2	16
21	Compound heterozygous <i>RMND1</i> gene variants associated with chronic kidney disease, dilated cardiomyopathy and neurological involvement: a case report. <i>BMC Research Notes</i> , 2016, 9, 325.	1.4	15
22	Expanding the phenotype of <i>ASXL3</i> -related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <i>ASXL3</i> . <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3446-3458.	1.2	12
23	Elucidating the molecular mechanisms associated with <i>TARS2</i> -related mitochondrial disease. <i>Human Molecular Genetics</i> , 2022, 31, 523-534.	2.9	12
24	A human importin- $\beta$ -related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in <i>IPO8</i> . <i>American Journal of Human Genetics</i> , 2021, 108, 1115-1125.	6.2	10
25	Biallelic variants in <i>ZNF142</i> lead to a syndromic neurodevelopmental disorder. <i>Clinical Genetics</i> , 2022, 102, 98-109.	2.0	6
26	A recurrent pathogenic variant in <i>TPM2</i> reveals further phenotypic and genetic heterogeneity in multiple pterygium syndrome-related disorders. <i>Clinical Genetics</i> , 2020, 97, 908-914.	2.0	5
27	Variants in <i>PHF8</i> cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphism. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100102.	1.7	5