Karen L Stals

List of Publications by Year
in descending order

12	376	1040056 9	1125743 13
papers	citations	h-index	g-index

13	13	13	848
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	2.4	8
2	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	2.4	41
3	A hypomorphic allele of SLC35D1 results in Schneckenbecken-like dysplasia. Human Molecular Genetics, 2019, 28, 3543-3551.	2.9	9
4	Risk category system to identify pituitary adenoma patients with AIP mutations. Journal of Medical Genetics, 2018, 55, 254-260.	3.2	35
5	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	6.2	88
6	Phenotype of CNTNAP1: a study of patients demonstrating a specific severe congenital hypomyelinating neuropathy with survival beyond infancy. European Journal of Human Genetics, 2018, 26, 796-807.	2.8	13
7	Diagnosis of lethal or prenatalâ€onset autosomal recessive disorders by parental exome sequencing. Prenatal Diagnosis, 2018, 38, 33-43.	2.3	64
8	In-frame seven amino-acid duplication in AIP arose over the last 3000 years, disrupts protein interaction and stability and is associated with gigantism. European Journal of Endocrinology, 2017, 177, 257-266.	3.7	12
9	Increased Population Risk ofAIP-Related Acromegaly and Gigantism in Ireland. Human Mutation, 2017, 38, 78-85.	2.5	25
10	AIP mutations in young patients with acromegaly and the Tampico Giant: the Mexican experience. Endocrine, 2016, 53, 402-411.	2.3	20
11	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. European Journal of Human Genetics, 2015, 23, 401-404.	2.8	51
12	An unusual case of an ACTH-secreting macroadenoma with a germline variant in the aryl hydrocarbon receptor-interacting protein (AIP) gene. Endocrinology, Diabetes and Metabolism Case Reports, 2015, 2015, 140105.	0.5	9