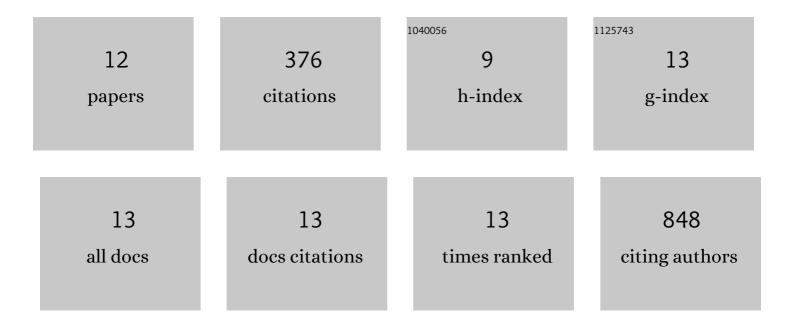
Karen L Stals

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/3954379/publications.pdf Version: 2024-02-01



KADENI STAIS

#	Article	IF	CITATIONS
1	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	6.2	88
2	Diagnosis of lethal or prenatalâ€onset autosomal recessive disorders by parental exome sequencing. Prenatal Diagnosis, 2018, 38, 33-43.	2.3	64
3	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. European Journal of Human Genetics, 2015, 23, 401-404.	2.8	51
4	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
5	Risk category system to identify pituitary adenoma patients with <i>AIP</i> mutations. Journal of Medical Genetics, 2018, 55, 254-260.	3.2	35
6	Increased Population Risk of <i>AIP</i> -Related Acromegaly and Gigantism in Ireland. Human Mutation, 2017, 38, 78-85.	2.5	25
7	AIP mutations in young patients with acromegaly and the Tampico Giant: the Mexican experience. Endocrine, 2016, 53, 402-411.	2.3	20
8	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
9	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
10	A hypomorphic allele of SLC35D1 results in Schneckenbecken-like dysplasia. Human Molecular Genetics, 2019, 28, 3543-3551.	2.9	9
11	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
12	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	2.4	8