# 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 | 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 AIP mutations. Journal of Medical Genetics, 2018, 55, 254-260. | 3.2 | 35 |
| 6 | Increased Population Risk ofAIP-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 |