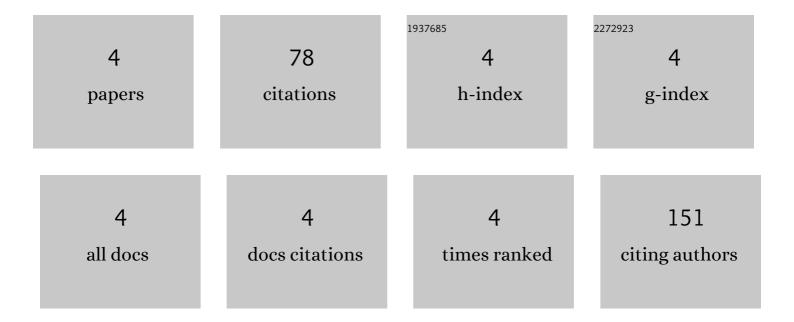
Helena GÃ;sdal Karstensen

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/4249650/publications.pdf

Version: 2024-02-01



| # | Article | IF | CITATIONS |
|---|---|-----|-----------|
| 1 | Phenotypic spectrum of the recurrent <i>TRPM3</i> p.(<scp>Val837Met</scp>) substitution in seven individuals with global developmental delay and hypotonia. American Journal of Medical Genetics, Part A, 2022, 188, 1667-1675. | 1.2 | 8 |
| 2 | Novel HARS2 missense variants identified in individuals with sensorineural hearing impairment and Perrault syndrome. European Journal of Medical Genetics, 2020, 63, 103733. | 1.3 | 9 |
| 3 | De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324. | 6.2 | 32 |
| 4 | Successful treatment with dupilumab of an adult with Netherton syndrome. Clinical and Experimental Dermatology, 2020, 45, 915-917. | 1.3 | 29 |