

# Helena GÅ;sdal Karstensen

## List of Publications by Year in descending order

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

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#	ARTICLE	IF	CITATIONS
1	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
2	Successful treatment with dupilumab of an adult with Netherton syndrome. Clinical and Experimental Dermatology, 2020, 45, 915-917.	1.3	29
3	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
4	Phenotypic spectrum of the recurrent <i>TRPM3</i> p.(Val837Met) substitution in seven individuals with global developmental delay and hypotonia. American Journal of Medical Genetics, Part A, 2022, 188, 1667-1675.	1.2	8