Helena GÃ¡sdal Karstensen

List of Publications by Year
in descending order

4 papers	78 citations	1937685 4 h-index	2272923 4 g-index
4 all docs	4 docs citations	4 times ranked	151 citing authors

#	Article	IF	Citations
1	Phenotypic spectrum of the recurrent TRPM3 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