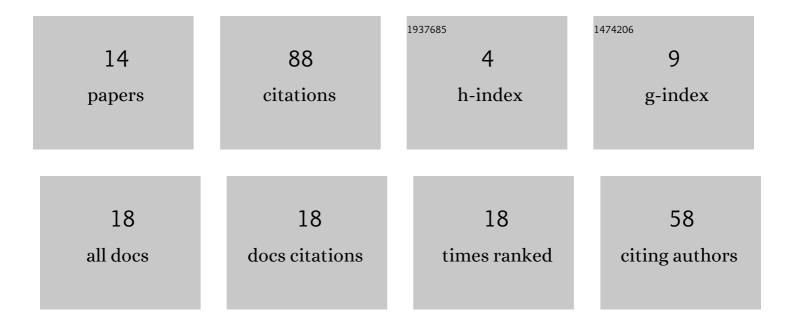
Yunha Choi

List of Publications by Year in descending order

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Ушына Сног

#	Article	IF	CITATIONS
1	Unfavorable clinical outcomes in patients with carbamoyl phosphate synthetase 1 deficiency. Clinica Chimica Acta, 2022, 526, 55-61.	1.1	4
2	Genotype-phenotype correlations and long-term efficacy of pamidronate therapy in patients with osteogenesis imperfecta. Annals of Pediatric Endocrinology and Metabolism, 2022, 27, 22-29.	2.3	2
3	Phenotypic spectrum of patients with mutations in CHD7: clinical implications of endocrinological findings. Endocrine Connections, 2022, 11, .	1.9	4
4	Whole-body MRI evaluation in neurofibromatosis type 1 patients younger than 3 years old and the genetic contribution to disease progression. Orphanet Journal of Rare Diseases, 2022, 17, 24.	2.7	4
5	Diagnostic performance of automated, streamlined, daily updated exome analysis in patients with neurodevelopmental delay. Molecular Medicine, 2022, 28, 38.	4.4	14
6	Metabolic Impacts of Discontinuation and Resumption of Recombinant Human Growth Hormone Treatment during the Transition Period in Patients with Childhood-Onset Growth Hormone Deficiency. Endocrinology and Metabolism, 2022, 37, 359-368.	3.0	3
7	Identification of a novel therapeutic target underlying atypical manifestation of Gaucher disease. Clinical and Translational Medicine, 2022, 12, e862.	4.0	5
8	Clinical and molecular spectra of BRAF-associated RASopathy. Journal of Human Genetics, 2021, 66, 389-399.	2.3	15

Rapidly Progressive Parkinsonism and Dementia with No Insomnia due to the PRNP D178N Mutation.