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List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	A bioinformatics pipeline for estimating mitochondrial DNA copy number and heteroplasmy levels from whole genome sequencing data. NAR Genomics and Bioinformatics, 2022, 4, lqac034.	3.2	12
2	Congenital hypopituitarism in two brothers with a duplication of the â€~acrogigantism gene' GPR101: clinical findings and review of the literature. Pituitary, 2021, 24, 229-241.	2.9	2
3	Composition of cutaneous bacterial microbiome in seborrheic dermatitis patients: A cross-sectional study. PLoS ONE, 2021, 16, e0251136.	2.5	12
4	A comparison of genotyping arrays. European Journal of Human Genetics, 2021, 29, 1611-1624.	2.8	43
5	Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. Nature Genetics, 2021, 53, 1300-1310.	21.4	590
6	Unique near-complete deletion of GLI2 in a patient with combined pituitary hormone deficiency and post-axial polydactyly. Growth Hormone and IGF Research, 2020, 50, 35-41.	1.1	7
7	Reduced penetrance of pathogenic ACMG variants in a deeply phenotyped cohort study and evaluation of ClinVar classification over time. Genetics in Medicine, 2020, 22, 1812-1820.	2.4	24
8	Whole-Exome Sequencing in Age-Related Macular Degeneration Identifies Rare Variants in COL8A1, a Component of Bruch's Membrane. Ophthalmology, 2018, 125, 1433-1443.	5.2	35