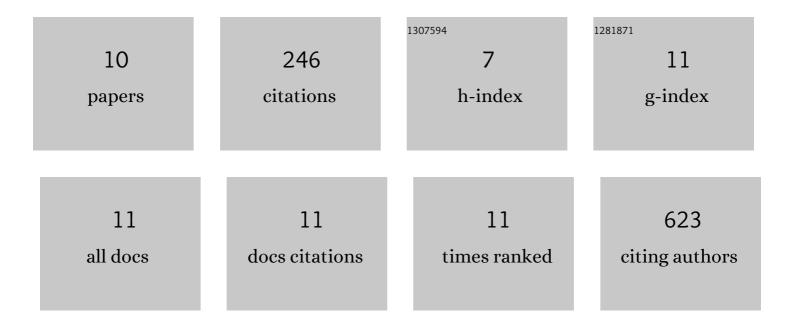
Nils Koelling

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/139886/publications.pdf Version: 2024-02-01



NUS KOFLUNC

#	Article	IF	CITATIONS
1	Biallelic <i>GINS2</i> variant p.(Arg114Leu) causes Meier-Gorlin syndrome with craniosynostosis. Journal of Medical Genetics, 2022, 59, 776-780.	3.2	10
2	The developing mouse coronal suture at single-cell resolution. Nature Communications, 2021, 12, 4797.	12.8	48
3	SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. Genetics in Medicine, 2020, 22, 1498-1506.	2.4	31
4	amplimap: a versatile tool to process and analyze targeted NGS data. Bioinformatics, 2019, 35, 5349-5350.	4.1	9
5	The impact of chemo- and radiotherapy treatments on selfish de novo FGFR2 mutations in sperm of cancer survivors. Human Reproduction, 2019, 34, 1404-1415.	0.9	7
6	A de novo substitution in BCL11B leads to loss of interaction with transcriptional complexes and craniosynostosis. Human Molecular Genetics, 2019, 28, 2501-2513.	2.9	23
7	Selfish mutations dysregulating RAS-MAPK signaling are pervasive in aged human testes. Genome Research, 2018, 28, 1779-1790.	5.5	56
8	Disruption of <i>TWIST1</i> translation by 5′ UTR variants in Saethre-Chotzen syndrome. Human Mutation, 2018, 39, 1360-1365.	2.5	10
9	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	6.2	37
10	Quantification of transmission risk in a male patient with a <i>FLNB</i> mosaic mutation causing Larsen syndrome: Implications for genetic counseling in postzygotic mosaicism cases. Human Mutation, 2017, 38, 1360-1364.	2.5	14