

# Nils Koelling

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/139886/publications.pdf>

Version: 2024-02-01

10  
papers

246  
citations

1307594

7  
h-index

1281871

11  
g-index

11  
all docs

11  
docs citations

11  
times ranked

623  
citing authors

#	ARTICLE	IF	CITATIONS
1	Biallelic <i>GINS2</i> variant p.(Arg114Leu) causes Meier-Gorlin syndrome with craniosynostosis. <i>Journal of Medical Genetics</i> , 2022, 59, 776-780.	3.2	10
2	The developing mouse coronal suture at single-cell resolution. <i>Nature Communications</i> , 2021, 12, 4797.	12.8	48
3	SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. <i>Genetics in Medicine</i> , 2020, 22, 1498-1506.	2.4	31
4	amplimap: a versatile tool to process and analyze targeted NGS data. <i>Bioinformatics</i> , 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. <i>Human Reproduction</i> , 2019, 34, 1404-1415.	0.9	7
6	A de novo substitution in <i>BCL11B</i> leads to loss of interaction with transcriptional complexes and craniosynostosis. <i>Human Molecular Genetics</i> , 2019, 28, 2501-2513.	2.9	23
7	Selfish mutations dysregulating RAS-MAPK signaling are pervasive in aged human testes. <i>Genome Research</i> , 2018, 28, 1779-1790.	5.5	56
8	Disruption of <i>TWIST1</i> translation by 5' UTR variants in Saethre-Chotzen syndrome. <i>Human Mutation</i> , 2018, 39, 1360-1365.	2.5	10
9	De Novo and Inherited Loss-of-Function Variants in <i>TLK2</i> : Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 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. <i>Human Mutation</i> , 2017, 38, 1360-1364.	2.5	14