

# Elmo Christian Saarentaus

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6451341/publications.pdf>

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#	ARTICLE	IF	CITATIONS
1	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. American Journal of Human Genetics, 2018, 102, 1204-1211.	2.6	102
2	Impact of constitutional TET2 haploinsufficiency on molecular and clinical phenotype in humans. Nature Communications, 2019, 10, 1252.	5.8	67
3	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
4	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17,458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
5	Contribution of rare and common variants to intellectual disability in a sub-isolate of Northern Finland. Nature Communications, 2019, 10, 410.	5.8	32
6	Genome-wide association meta-analysis identifies 48 risk variants and highlights the role of the stria vascularis in hearing loss. American Journal of Human Genetics, 2022, 109, 1077-1091.	2.6	27
7	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. Journal of Medical Genetics, 2017, 54, 598-606.	1.5	22
8	Polygenic burden has broader impact on health, cognition, and socioeconomic outcomes than most rare and high-risk copy number variants. Molecular Psychiatry, 2021, 26, 4884-4895.	4.1	8
9	Duplications at 19q13.33 in patients with neurodevelopmental disorders. Neurology: Genetics, 2018, 4, e210.	0.9	4
10	Heterozygous TYROBP deletion (PLOSFIN) is not a strong risk factor for cognitive impairment. Neurobiology of Aging, 2018, 64, 159.e1-159.e4.	1.5	3