

# Stefan J White

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4474495/publications.pdf>

Version: 2024-02-01

18  
papers

764  
citations

759233

12  
h-index

839539

18  
g-index

18  
all docs

18  
docs citations

18  
times ranked

1461  
citing authors

#	ARTICLE	IF	CITATIONS
1	Breakpoint characterization of a rare alpha 0 $\alpha$ -thalassemia deletion using targeted locus amplification on genomic DNA. <i>International Journal of Laboratory Hematology</i> , 2021, 43, 1628-1634.	1.3	3
2	Skewed X-inactivation is common in the general female population. <i>European Journal of Human Genetics</i> , 2019, 27, 455-465.	2.8	119
3	Reduced PRC2 function alters male germline epigenetic programming and paternal inheritance. <i>BMC Biology</i> , 2018, 16, 104.	3.8	17
4	Loss of maternal EED results in postnatal overgrowth. <i>Clinical Epigenetics</i> , 2018, 10, 95.	4.1	34
5	Critical points for an accurate human genome analysis. <i>Human Mutation</i> , 2017, 38, 912-921.	2.5	5
6	Flexible and Scalable Full-Length CYP2D6 Long Amplicon PacBio Sequencing. <i>Human Mutation</i> , 2017, 38, 310-316.	2.5	69
7	Targeted Locus Amplification and Next-Generation Sequencing. <i>Methods in Molecular Biology</i> , 2017, 1492, 185-196.	0.9	34
8	Quantitative DNA Analysis Using Droplet Digital PCR. <i>Methods in Molecular Biology</i> , 2017, 1492, 167-177.	0.9	11
9	Genotyping Multiallelic Copy Number Variation with Multiplex Ligation-Dependent Probe Amplification (MLPA). <i>Methods in Molecular Biology</i> , 2017, 1492, 147-153.	0.9	2
10	Multiallelic copy number variation in the complement component 4A (C4A) gene is associated with late-stage age-related macular degeneration (AMD). <i>Journal of Neuroinflammation</i> , 2016, 13, 81.	7.2	31
11	Glucocorticoid-induced leucine zipper (GILZ) inhibits B cell activation in systemic lupus erythematosus. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 739-747.	0.9	36
12	Counting copy number and calories. <i>Nature Genetics</i> , 2015, 47, 852-853.	21.4	5
13	Identification of <i>Streptococcus parasanguinis</i> DNA contamination in human buccal DNA samples. <i>BMC Research Notes</i> , 2013, 6, 481.	1.4	6
14	Correlating Multiallelic Copy Number Polymorphisms with Disease Susceptibility. <i>Human Mutation</i> , 2013, 34, 1-13.	2.5	49
15	Sarcoglycanopathies and the risk of undetected deletion alleles in diagnosis. <i>Human Mutation</i> , 2005, 26, 59-59.	2.5	13
16	Two-color multiplex ligation-dependent probe amplification: Detecting genomic rearrangements in hereditary multiple exostoses. <i>Human Mutation</i> , 2004, 24, 86-92.	2.5	142
17	Detecting Copy Number Changes in Genomic DNA: MAPH and MLPA. <i>Methods in Cell Biology</i> , 2004, 75, 751-768.	1.1	25
18	Comprehensive Detection of Genomic Duplications and Deletions in the DMD Gene, by Use of Multiplex Amplifiable Probe Hybridization. <i>American Journal of Human Genetics</i> , 2002, 71, 365-374.	6.2	163