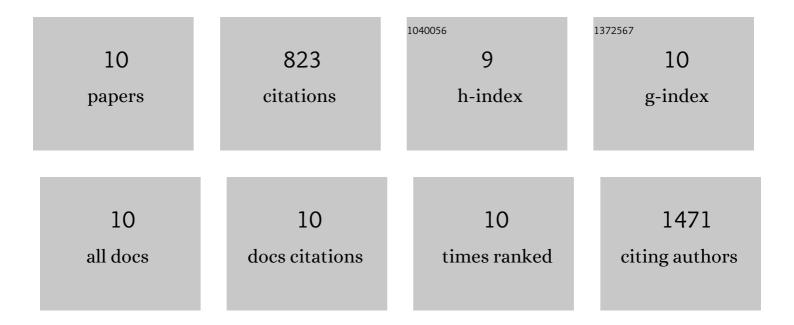
## Sarah B Pierce

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/832527/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	Mutations in the DBP-Deficiency Protein HSD17B4 Cause Ovarian Dysgenesis, Hearing Loss, and Ataxia of Perrault Syndrome. American Journal of Human Genetics, 2010, 87, 282-288.	6.2	231
2	Mutations in mitochondrial histidyl tRNA synthetase <i>HARS2</i> cause ovarian dysgenesis and sensorineural hearing loss of Perrault syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 6543-6548.	7.1	225
3	Mutations in LARS2, Encoding Mitochondrial Leucyl-tRNA Synthetase, Lead to Premature Ovarian Failure and Hearing Loss in Perrault Syndrome. American Journal of Human Genetics, 2013, 92, 614-620.	6.2	176
4	Mutations in Twinkle primase-helicase cause Perrault syndrome with neurologic features. Neurology, 2014, 83, 2054-2061.	1.1	86
5	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
6	De novo mutation in <i>RING1</i> with epigenetic effects on neurodevelopment. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 1558-1563.	7.1	24
7	Mutation of KREMEN1, a modulator of Wnt signaling, is responsible for ectodermal dysplasia including oligodontia in Palestinian families. European Journal of Human Genetics, 2016, 24, 1430-1435.	2.8	20
8	Infantile onset spinocerebellar ataxia caused by compound heterozygosity for Twinkle mutations and modeling of Twinkle mutations causing recessive disease. Journal of Physical Education and Sports Management, 2016, 2, a001107.	1.2	13
9	Garrod's fourth inborn error of metabolism solved by the identification of mutations causing pentosuria. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 18313-18317.	7.1	11
10	Helicase-inactivating <i>BRIP1</i> mutation yields Fanconi anemia with microcephaly and other congenital abnormalities. Journal of Physical Education and Sports Management, 2020, 6, a005652.	1.2	2