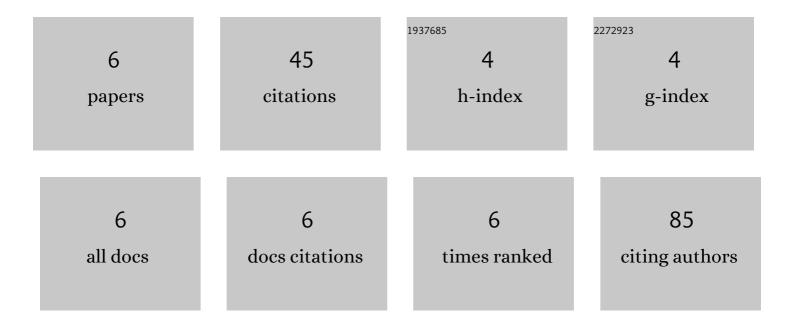
Sofia Frisk

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/3552535/publications.pdf Version: 2024-02-01



SOFIA EDISK

#	Article	IF	CITATIONS
1	Early activating somatic <i>PIK3CA</i> mutations promote ectopic muscle development and upper limb overgrowth. Clinical Genetics, 2019, 96, 118-125.	2.0	14
2	Goltz syndrome in males: A clinical report of a male patient carrying a novel <i><scp>PORCN</scp></i> variant and a review of the literature. Clinical Case Reports (discontinued), 2018, 6, 2103-2110.	0.5	12
3	Detection of germline mosaicism in fathers of children with intellectual disability syndromes caused by de novo variants. Molecular Genetics & Genomic Medicine, 2022, 10, e1880.	1.2	10
4	A complex DICER1 syndrome phenotype associated with a germline pathogenic variant affecting the RNase IIIa domain of DICER1. Journal of Medical Genetics, 2022, 59, 141-146.	3.2	9
5	Dopamine D2 Receptor Antagonist Unbinding Rates Investigated using a Time-Resolved Ion Channel Activation Assay. Biophysical Journal, 2012, 102, 517a.	0.5	0
6	Novel Aspects of the Reversibility of the Antagonism at the Dopamine D2 Receptor by Antipsychotics. Biophysical Journal, 2013, 104, 26a.	0.5	0