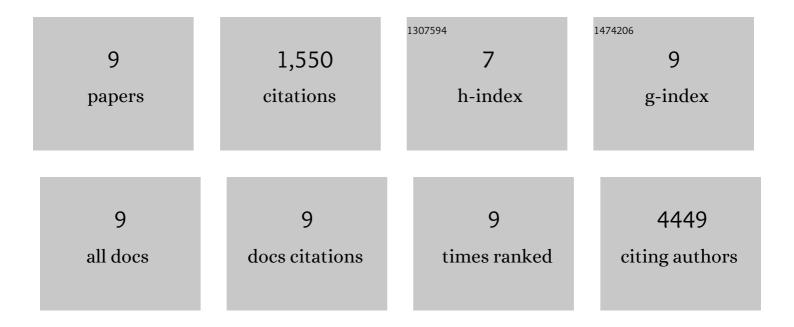
Marta Girdea

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/6542720/publications.pdf Version: 2024-02-01



Μάρτα Οιρπέα

#	Article	IF	CITATIONS
1	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
2	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	2.5	390
3	PhenoTips: Patient Phenotyping Software for Clinical and Research Use. Human Mutation, 2013, 34, 1057-1065.	2.5	207
4	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. Human Mutation, 2015, 36, 931-940.	2.5	107
5	Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. Genetics in Medicine, 2016, 18, 608-617.	2.4	85
6	The Matchmaker Exchange API: Automating Patient Matching Through the Exchange of Structured Phenotypic and Genotypic Profiles. Human Mutation, 2015, 36, 922-927.	2.5	50
7	Prioritizing Clinically Relevant Copy Number Variation from Genetic Interactions and Gene Function Data. PLoS ONE, 2015, 10, e0139656.	2.5	9
8	Distributed Cognition and Process Management Enabling Individualized Translational Research: The NIH Undiagnosed Diseases Program Experience. Frontiers in Medicine, 2016, 3, 39.	2.6	3
9	MG-132â€Diagnostic utility of whole genome sequencing in paediatric medicine. Journal of Medical Genetics, 2015, 52, A12.1-A12.	3.2	1