

Sophie Nambot

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/7003629/publications.pdf>

Version: 2024-02-01

19
papers

501
citations

759233

12
h-index

839539

18
g-index

20
all docs

20
docs citations

20
times ranked

1366
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical whole-exome sequencing for the diagnosis of rare disorders with congenital anomalies and/or intellectual disability: substantial interest of prospective annual reanalysis. <i>Genetics in Medicine</i> , 2018, 20, 645-654.	2.4	146
2	De novo loss-of-function KCNMA1 variants are associated with a new multiple malformation syndrome and a broad spectrum of developmental and neurological phenotypes. <i>Human Molecular Genetics</i> , 2019, 28, 2937-2951.	2.9	76
3	Increased diagnostic and new genes identification outcome using research reanalysis of singleton exome sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 1519-1531.	2.8	43
4	Okurâ€Chung neurodevelopmental syndrome: Eight additional cases with implications on phenotype and genotype expansion. <i>Clinical Genetics</i> , 2018, 93, 880-890.	2.0	30
5	Further delineation of the female phenotype with <scp><i>KDM5C</i></scp> disease causing variants: 19 new individuals and review of the literature. <i>Clinical Genetics</i> , 2020, 98, 43-55.	2.0	28
6	Reducing diagnostic turnaround times of exome sequencing for families requiring timely diagnoses. <i>European Journal of Medical Genetics</i> , 2017, 60, 595-604.	1.3	22
7	VariantÂrecurrence in neurodevelopmental disorders: the use of publicly available genomic data identifies clinically relevant pathogenic missense variants. <i>Genetics in Medicine</i> , 2019, 21, 2504-2511.	2.4	21
8	Widening of the genetic and clinical spectrum of Lambâ€Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. <i>Genetics in Medicine</i> , 2020, 22, 524-537.	2.4	21
9	Further delineation of a rare recessive encephalomyopathy linked to mutations in <scp>GFER</scp> thanks to data sharing of whole exome sequencing data. <i>Clinical Genetics</i> , 2017, 92, 188-198.	2.0	20
10	<scp>NextÂgeneration</scp> sequencing approaches and challenges in the diagnosis of developmental anomalies and intellectual disability. <i>Clinical Genetics</i> , 2020, 98, 433-444.	2.0	20
11	Genotype-first in a cohort of 95 fetuses with multiple congenital abnormalities: when exome sequencing reveals unexpected fetal phenotype-genotype correlations. <i>Journal of Medical Genetics</i> , 2021, 58, 400-413.	3.2	18
12	2.5 yearsâ™ experience of GeneMatcher data-sharing: a powerful tool for identifying new genes responsible for rare diseases. <i>Genetics in Medicine</i> , 2019, 21, 1657-1661.	2.4	14
13	9q33.3q34.11 microdeletion: new contiguous gene syndrome encompassing STXBP1, LMX1B and ENG genes assessed using reverse phenotyping. <i>European Journal of Human Genetics</i> , 2016, 24, 830-837.	2.8	13
14	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. <i>European Journal of Human Genetics</i> , 2022, 30, 567-576.	2.8	12
15	Copy number variants calling from WES data through eXome hidden Markov model (XHMM) identifies additional 2.5% pathogenic genomic imbalances smaller than 30Âkb undetected by arrayâ€CGH. <i>Annals of Human Genetics</i> , 2022, 86, 171-180.	0.8	6
16	Variants of human <i>CLDN9</i> cause mild to profound hearing loss. <i>Human Mutation</i> , 2021, 42, 1321-1335.	2.5	5
17	TUMOSPEC: A Nation-Wide Study of Hereditary Breast and Ovarian Cancer Families with a Predicted Pathogenic Variant Identified through Multigene Panel Testing. <i>Cancers</i> , 2021, 13, 3659.	3.7	4
18	Same performance of exome sequencing before and after fetal autopsy for congenital abnormalities: toward a paradigm shift in prenatal diagnosis?. <i>European Journal of Human Genetics</i> , 2022, , .	2.8	1

#	ARTICLE	IF	CITATIONS
19	A novel POLD1 pathogenic variant identified in two families with a cancer spectrum mimicking Lynch syndrome. <i>European Journal of Medical Genetics</i> , 2022, 65, 104409.	1.3	0