Sophie Nambot

List of Publications by Year
in descending order

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

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		759233	839539
19	501	12	18
papers	citations	h-index	g-index

20	20	20	1366
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. European Journal of Human Genetics, 2022, 30, 567-576.	2.8	12
2	A novel POLD1 pathogenic variant identified in two families with a cancer spectrum mimicking Lynch syndrome. European Journal of Medical Genetics, 2022, 65, 104409.	1.3	0
3	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. Annals of Human Genetics, 2022, 86, 171-180.	0.8	6
4	Same performance of exome sequencing before and after fetal autopsy for congenital abnormalities: toward a paradigm shift in prenatal diagnosis?. European Journal of Human Genetics, 2022, , .	2.8	1
5	Genotype-first in a cohort of 95 fetuses with multiple congenital abnormalities: when exome sequencing reveals unexpected fetal phenotype-genotype correlations. Journal of Medical Genetics, 2021, 58, 400-413.	3.2	18
6	TUMOSPEC: A Nation-Wide Study of Hereditary Breast and Ovarian Cancer Families with a Predicted Pathogenic Variant Identified through Multigene Panel Testing. Cancers, 2021, 13, 3659.	3.7	4
7	Variants of human CLDN9 cause mild to profound hearing loss. Human Mutation, 2021, 42, 1321-1335.	2.5	5
8	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
9	<scp>Nextâ€generation</scp> sequencing approaches and challenges in the diagnosis of developmental anomalies and intellectual disability. Clinical Genetics, 2020, 98, 433-444.	2.0	20
10	Further delineation of the female phenotype with <scp>KDM5C</scp> disease causing variants: 19 new individuals and review of the literature. Clinical Genetics, 2020, 98, 43-55.	2.0	28
11	Increased diagnostic and new genes identification outcome using research reanalysis of singleton exome sequencing. European Journal of Human Genetics, 2019, 27, 1519-1531.	2.8	43
12	De novo loss-of-function KCNMA1 variants are associated with a new multiple malformation syndrome and a broad spectrum of developmental and neurological phenotypes. Human Molecular Genetics, 2019, 28, 2937-2951.	2.9	76
13	VariantÂrecurrence in neurodevelopmental disorders: the use of publicly available genomic data identifies clinically relevant pathogenic missense variants. Genetics in Medicine, 2019, 21, 2504-2511.	2.4	21
14	2.5 yearsâ€™ experience of GeneMatcher data-sharing: a powerful tool for identifying new genes responsible for rare diseases. Genetics in Medicine, 2019, 21, 1657-1661.	2.4	14
15	Clinical whole-exome sequencing for the diagnosis of rare disorders with congenital anomalies and/or intellectual disability: substantial interest of prospective annual reanalysis. Genetics in Medicine, 2018, 20, 645-654.	2.4	146
16	Okurâ€Chung neurodevelopmental syndrome: Eight additional cases with implications on phenotype and genotype expansion. Clinical Genetics, 2018, 93, 880-890.	2.0	30
17	Further delineation of a rare recessive encephalomyopathy linked to mutations in <scp>GFER</scp> thanks to data sharing of whole exome sequencing data. Clinical Genetics, 2017, 92, 188-198.	2.0	20
18	Reducing diagnostic turnaround times of exome sequencing for families requiring timely diagnoses. European Journal of Medical Genetics, 2017, 60, 595-604.	1.3	22

#	Article	IF	Citations
19	9q33.3q34.11 microdeletion: new contiguous gene syndrome encompassing STXBP1, LMX1B and ENG genes assessed using reverse phenotyping. European Journal of Human Genetics, 2016, 24, 830-837.	2.8	13