Thomy de Ravel

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4005188/publications.pdf

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21	777	623188 14	713013 21
papers	citations	h-index	g-index

22	22	22	1757
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	BCAP31-related syndrome: The first de novo report. European Journal of Medical Genetics, 2020, 63, 103732.	0.7	7
2	The majority of autosomal recessive nanophthalmos and posterior microphthalmia can be attributed to biallelic sequence and structural variants in MFRP and PRSS56. Scientific Reports, 2020, 10, 1289.	1.6	24
3	The clinical relevance of intragenic NRXN1 deletions. Journal of Medical Genetics, 2020, 57, 347-355.	1.5	11
4	Maternal copy-number variations in the DMD gene as secondary findings in noninvasive prenatal screening. Genetics in Medicine, 2019, 21, 2774-2780.	1.1	16
5	Mutation spectrum and clinical investigation of achromatopsia patients with mutations in the GNAT2 gene. Human Mutation, 2019, 40, 1145-1155.	1.1	15
6	Neurofibromatosis type 1â€related pseudarthrosis: Beyond the pseudarthrosis site. Human Mutation, 2019, 40, 1760-1767.	1.1	11
7	Predicting fetoplacental chromosomal mosaicism during nonâ€invasive prenatal testing. Prenatal Diagnosis, 2018, 38, 258-266.	1.1	58
8	Genome-wide haplotyping embryos developing from 0PN and 1PN zygotes increases transferrable embryos in PGT-M. Human Reproduction, 2018, 33, 2302-2311.	0.4	33
9	Detecting AGG Interruptions in Females With a FMR1 Premutation by Long-Read Single-Molecule Sequencing: A 1 Year Clinical Experience. Frontiers in Genetics, 2018, 9, 150.	1.1	26
10	Principles guiding embryo selection following genome-wide haplotyping of preimplantation embryos. Human Reproduction, 2017, 32, 687-697.	0.4	40
11	Massive parallel sequencing identifies RAPSN and PDHA1 mutations causing fetal akinesia deformation sequence. European Journal of Paediatric Neurology, 2017, 21, 745-753.	0.7	15
12	Accuracy and clinical value of maternal incidental findings during noninvasive prenatal testing for fetal aneuploidies. Genetics in Medicine, 2017, 19, 306-313.	1.1	47
13	Mutations in Splicing Factor Genes Are a Major Cause of Autosomal Dominant Retinitis Pigmentosa in Belgian Families. PLoS ONE, 2017, 12, e0170038.	1.1	47
14	Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management. European Journal of Human Genetics, 2015, 23, 1286-1293.	1.4	108
15	Microdeletion of the escape genes KDM5C and IQSEC2 in a girl with severe intellectual disability and autistic features. European Journal of Medical Genetics, 2015, 58, 324-327.	0.7	36
16	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	1.1	45
17	Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. European Journal of Medical Genetics, 2014, 57, 151-156.	0.7	91
18	A prospective study of the clinical utility of prenatal chromosomal microarray analysis in fetuses with ultrasound abnormalities and an exploration of a framework for reporting unclassified variants and risk factors. Genetics in Medicine, 2014, 16, 469-476.	1.1	66

#	Article	IF	Citations
19	Identity-by-descentâ€“guided mutation analysis and exome sequencing in consanguineous families reveals unusual clinical and molecular findings in retinal dystrophy. Genetics in Medicine, 2014, 16, 671-680.	1.1	53
20	Presenting symptoms in adults with the 22q11 deletion syndrome. European Journal of Medical Genetics, 2014, 57, 157-162.	0.7	24
21	â€œOpitz C syndrome and pseudohypoaldosteronismâ€•is caused by a chromosome 4q deletion. American Journal of Medical Genetics, Part A, 2009, 149A, 1315-1316.	0.7	4