

Thomy de Ravel

List of Publications by Year in descending order

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Version: 2024-02-01

21
papers

777
citations

623188

14
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713013

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all docs

22
docs citations

22
times ranked

1757
citing authors

#	ARTICLE	IF	CITATIONS
1	Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management. <i>European Journal of Human Genetics</i> , 2015, 23, 1286-1293.	1.4	108
2	Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. <i>European Journal of Medical Genetics</i> , 2014, 57, 151-156.	0.7	91
3	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. <i>Genetics in Medicine</i> , 2014, 16, 469-476.	1.1	66
4	Predicting fetoplacental chromosomal mosaicism during noninvasive prenatal testing. <i>Prenatal Diagnosis</i> , 2018, 38, 258-266.	1.1	58
5	Identity-by-descent-guided mutation analysis and exome sequencing in consanguineous families reveals unusual clinical and molecular findings in retinal dystrophy. <i>Genetics in Medicine</i> , 2014, 16, 671-680.	1.1	53
6	Accuracy and clinical value of maternal incidental findings during noninvasive prenatal testing for fetal aneuploidies. <i>Genetics in Medicine</i> , 2017, 19, 306-313.	1.1	47
7	Mutations in Splicing Factor Genes Are a Major Cause of Autosomal Dominant Retinitis Pigmentosa in Belgian Families. <i>PLoS ONE</i> , 2017, 12, e0170038.	1.1	47
8	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. <i>Genetics in Medicine</i> , 2015, 17, 460-466.	1.1	45
9	Principles guiding embryo selection following genome-wide haplotyping of preimplantation embryos. <i>Human Reproduction</i> , 2017, 32, 687-697.	0.4	40
10	Microdeletion of the escape genes KDM5C and IQSEC2 in a girl with severe intellectual disability and autistic features. <i>European Journal of Medical Genetics</i> , 2015, 58, 324-327.	0.7	36
11	Genome-wide haplotyping embryos developing from OPN and 1PN zygotes increases transferrable embryos in PGT-M. <i>Human Reproduction</i> , 2018, 33, 2302-2311.	0.4	33
12	Detecting AGG Interruptions in Females With a FMR1 Premutation by Long-Read Single-Molecule Sequencing: A 1 Year Clinical Experience. <i>Frontiers in Genetics</i> , 2018, 9, 150.	1.1	26
13	Presenting symptoms in adults with the 22q11 deletion syndrome. <i>European Journal of Medical Genetics</i> , 2014, 57, 157-162.	0.7	24
14	The majority of autosomal recessive nanophthalmos and posterior microphthalmia can be attributed to biallelic sequence and structural variants in MFRP and PRSS56. <i>Scientific Reports</i> , 2020, 10, 1289.	1.6	24
15	Maternal copy-number variations in the DMD gene as secondary findings in noninvasive prenatal screening. <i>Genetics in Medicine</i> , 2019, 21, 2774-2780.	1.1	16
16	Massive parallel sequencing identifies RAPSN and PDHA1 mutations causing fetal akinesia deformation sequence. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 745-753.	0.7	15
17	Mutation spectrum and clinical investigation of achromatopsia patients with mutations in the <i>GNAT2</i> gene. <i>Human Mutation</i> , 2019, 40, 1145-1155.	1.1	15
18	Neurofibromatosis type 1-related pseudarthrosis: Beyond the pseudarthrosis site. <i>Human Mutation</i> , 2019, 40, 1760-1767.	1.1	11

#	ARTICLE	IF	CITATIONS
19	The clinical relevance of intragenic NRXN1 deletions. Journal of Medical Genetics, 2020, 57, 347-355.	1.5	11
20	BCAP31-related syndrome: The first de novo report. European Journal of Medical Genetics, 2020, 63, 103732.	0.7	7
21	“Opitz C syndrome and pseudohypoaldosteronism” is caused by a chromosome 4q deletion. American Journal of Medical Genetics, Part A, 2009, 149A, 1315-1316.	0.7	4