Julien Thevenon

List of Publications by Year
in descending order

90 papers	3,742 citations	117625 34 h-index	155660 55 g-index
97 all docs	97 docs citations	97 times ranked	7705 citing authors

#	Article	IF	Citations
1	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
2	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Human Mutation, 2022, 43, 582-594.	2.5	6
3	Genome Alert!: A standardized procedure for genomicÂvariant reinterpretation and automated geneâ€“phenotype reassessment in clinical routine. Genetics in Medicine, 2022, 24, 1316-1327.	2.4	5
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	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	2.4	21
7	Phenotype associated with TAF2 biallelic mutations: A clinical description of four individuals and review of the literature. European Journal of Medical Genetics, 2021, 64, 104323.	1.3	5
8	The diagnostic rate of inherited metabolic disorders by exome sequencing in a cohort of 547 individuals with developmental disorders. Molecular Genetics and Metabolism Reports, 2021, 29, 100812.	1.1	2
9	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	3.2	11
10	Neural metabolic imbalance induced by MOF dysfunction triggers pericyte activation and breakdown of vasculature. Nature Cell Biology, 2020, 22, 828-841.	10.3	27
11	A new mutational hotspot in the SKI gene in the context of MFS/TAA molecular diagnosis. Human Genetics, 2020, 139, 461-472.	3.8	8
12	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782.	2.8	27
13	Novel KIAA1033 / WASHC4 mutations in three patients with syndromic intellectual disability and a review of the literature. American Journal of Medical Genetics, Part A, 2020, 182, 792-797.	1.2	12
14	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
15	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
16	Report on three additional patients and genotypeâ€“phenotype correlation in SLC25A22-related disorders group. European Journal of Human Genetics, 2019, 27, 1692-1700.	2.8	10
17	Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. Nature Genetics, 2019, 51, 1438-1441.	21.4	25
18	Exome sequencing in clinical settings: preferences and experiences of parents of children with rare diseases (SEQUAPRE study). European Journal of Human Genetics, 2019, 27, 701-710.	2.8	18

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19	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
20	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
21	Secondary actionable findings identified by exome sequencing: expected impact on the organisation of care from the study of 700 consecutive tests. European Journal of Human Genetics, 2019, 27, 1197-1214.	2.8	18
22	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
23	Diagnostic strategy in segmentation defect of the vertebrae: a retrospective study of 73 patients. Journal of Medical Genetics, 2018, 55, 422.2-429.	3.2	14
24	Unexpected diagnosis of a SHH nonsense variant causing a variable phenotype ranging from familial coloboma and Intellectual disability to isolated microcephaly. Clinical Genetics, 2018, 94, 182-184.	2.0	2
25	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. American Journal of Human Genetics, 2018, 102, 995-1007.	6.2	49
26	Wiedemannâ€Steiner syndrome as a major cause of syndromic intellectual disability: A study of 33 French cases. Clinical Genetics, 2018, 94, 141-152.	2.0	57
27	Further delineation of theMECP2duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. Journal of Medical Genetics, 2018, 55, 359-371.	3.2	45
28	Loss-of-Function Mutations in UNC45A Cause a Syndrome Associating Cholestasis, Diarrhea, Impaired Hearing, and Bone Fragility. American Journal of Human Genetics, 2018, 102, 364-374.	6.2	40
29	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	6.2	88
30	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La RÃ©union Island, in patients with Fryns syndrome. European Journal of Human Genetics, 2018, 26, 340-349.	2.8	27
31	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
32	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
33	The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance. American Journal of Medical Genetics, Part A, 2018, 176, 2740-2750.	1.2	6
34	Further delineation of the clinical spectrum of de novo TRIM8 truncating mutations. American Journal of Medical Genetics, Part A, 2018, 176, 2470-2478.	1.2	19
35	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
36	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	21.4	28

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37	Rettâ€like phenotypes: expanding the genetic heterogeneity to the <scp>KCNA2</scp> gene and first familial case of <scp>CDKL5</scp>â€related disease. Clinical Genetics, 2017, 91, 431-440.	2.0	41
38	Autosomal recessive mutations in <scp>THOC6</scp> cause intellectual disability: syndrome delineation requiring forward and reverse phenotyping. Clinical Genetics, 2017, 91, 92-99.	2.0	28
39	Autosomal recessive truncating <scp>MAB21L1</scp> mutation associated with a syndromic scrotal agenesis. Clinical Genetics, 2017, 91, 333-338.	2.0	15
40	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
41	Intragenic FMR1 disease-causing variants: a significant mutational mechanism leading to Fragile-X syndrome. European Journal of Human Genetics, 2017, 25, 423-431.	2.8	48
42	STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488.	3.2	35
43	PUF60 variants cause a syndrome of ID, short stature, microcephaly, coloboma, craniofacial, cardiac, renal and spinal features. European Journal of Human Genetics, 2017, 25, 552-559.	2.8	42
44	Disruption of the ATXN1â€“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536.	21.4	113
45	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	3.2	85
46	Expanding the clinical spectrum of recessive truncating mutations ofKLHL7to a Bohring-Opitz-like phenotype. Journal of Medical Genetics, 2017, 54, 830-835.	3.2	15
47	Reducing diagnostic turnaround times of exome sequencing for families requiring timely diagnoses. European Journal of Medical Genetics, 2017, 60, 595-604.	1.3	22
48	Clinical delineation of a subtype of frontonasal dysplasia with creased nasal ridge and upper limb anomalies: Report of six unrelated patients. American Journal of Medical Genetics, Part A, 2017, 173, 3136-3142.	1.2	9
49	Dominant variants in the splicing factor PUF60 cause a recognizable syndrome with intellectual disability, heart defects and short stature. European Journal of Human Genetics, 2017, 25, 43-51.	2.8	44
50	Autosomal recessive variations of <scp>TBX6</scp>, from congenital scoliosis to spondylocostal dysostosis. Clinical Genetics, 2017, 91, 908-912.	2.0	38
51	Xq28 duplication including <scp>MECP2</scp> in six unreported affected females: what can we learn for diagnosis and genetic counselling?. Clinical Genetics, 2017, 91, 576-588.	2.0	17
52	Application of wholeâ€exome sequencing to unravel the molecular basis of undiagnosed syndromic congenital neutropenia with intellectual disability. American Journal of Medical Genetics, Part A, 2017, 173, 62-71.	1.2	23
53	A new family with an SLC9A6 mutation expanding the phenotypic spectrum of Christianson syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2103-2110.	1.2	21
54	Homozygous FIBP nonsense variant responsible of syndromic overgrowth, with overgrowth, macrocephaly, retinal coloboma and learning disabilities. Clinical Genetics, 2016, 89, e1-4.	2.0	18

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55	Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea. American Journal of Human Genetics, 2016, 99, 753-761.	6.2	68
56	Large national series of patients with Xq28 duplication involving MECP2: Delineation of brain MRI abnormalities in 30 affected patients. American Journal of Medical Genetics, Part A, 2016, 170, 116-129.	1.2	19
57	Diagnostic odyssey in severe neurodevelopmental disorders: toward clinical wholeâ€exome sequencing as a firstâ€line diagnostic test. Clinical Genetics, 2016, 89, 700-707.	2.0	205
58	Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. American Journal of Human Genetics, 2016, 99, 1368-1376.	6.2	46
59	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	2.5	70
60	Genetic counselling difficulties and ethical implications of incidental findings from arrayâ€CGH: a 7â€year national survey. Clinical Genetics, 2016, 89, 630-635.	2.0	12
61	Autosomal recessive <scp>IFT57</scp> hypomorphic mutation cause ciliary transport defect in unclassified oralâ€“facialâ€“digital syndrome with short stature and brachymesophalangia. Clinical Genetics, 2016, 90, 509-517.	2.0	20
62	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
63	OFIP/KIAA0753 forms a complex with OFD1 and FOR20 at pericentriolar satellites and centrosomes and is mutated in one individual with oral-facial-digital syndrome. Human Molecular Genetics, 2016, 25, 497-513.	2.9	42
64	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
65	Heterozygous deletion of the LRFN2 gene is associated with working memory deficits. European Journal of Human Genetics, 2016, 24, 911-918.	2.8	18
66	A de novo microdeletion of SEMA5A in a boy with autism spectrum disorder and intellectual disability. European Journal of Human Genetics, 2016, 24, 838-843.	2.8	40
67	Compound heterozygous PKHD1 variants cause a wide spectrum of ductal plate malformations. American Journal of Medical Genetics, Part A, 2015, 167, 3046-3053.	1.2	8
68	6q16.3q23.3 duplication associated with Prader-Willi-like syndrome. Molecular Cytogenetics, 2015, 8, 42.	0.9	11
69	RPL10 mutation segregating in a family with Xâ€linked syndromic Intellectual Disability. American Journal of Medical Genetics, Part A, 2015, 167, 1908-1912.	1.2	27
70	Clinical spectrum of eye malformations in four patients with Mowatâ€“Wilson syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1587-1592.	1.2	16
71	Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A. European Journal of Human Genetics, 2015, 23, 1482-1487.	2.8	62
72	Autosomal-recessive SASH1 variants associated with a new genodermatosis with pigmentation defects, palmoplantar keratoderma and skin carcinoma. European Journal of Human Genetics, 2015, 23, 957-962.	2.8	39

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73	Congenital neutropenia with retinopathy, a new phenotype without intellectual deficiency or obesity secondary to VPS13B mutations. American Journal of Medical Genetics, Part A, 2014, 164, 522-527.	1.2	13
74	Further delineation of eye manifestations in homozygous 15q13.3 microdeletions including TRPM1: A differential diagnosis of ceroid lipofuscinosis. American Journal of Medical Genetics, Part A, 2014, 164, 1537-1544.	1.2	22
75	3q27.3 microdeletional syndrome: a recognisable clinical entity associating dysmorphic features, marfanoid habitus, intellectual disability and psychosis with mood disorder. Journal of Medical Genetics, 2014, 51, 21-27.	3.2	12
76	Neuropsychological and neuroimaging phenotype induced by a CAMTA1 mutation. Brain and Development, 2014, 36, 711-715.	1.1	7
77	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	3.8	71
78	The oral-facial-digital syndrome gene C2CD3 encodes a positive regulator of centriole elongation. Nature Genetics, 2014, 46, 905-911.	21.4	121
79	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. Journal of Medical Genetics, 2014, 51, 724-736.	3.2	229
80	Mutations in SLC13A5 Cause Autosomal-Recessive Epileptic Encephalopathy with Seizure Onset in the First Days of Life. American Journal of Human Genetics, 2014, 95, 113-120.	6.2	112
81	20 ans aprÃ¨s: a second mutation in MAOA identified by targeted high-throughput sequencing in a family with altered behavior and cognition. European Journal of Human Genetics, 2014, 22, 776-783.	2.8	75
82	Expanding the clinical phenotype of patients with a ZDHHC9 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 789-795.	1.2	29
83	Delineation of the 3p14.1p13 microdeletion associated with syndromic distal limb contractures. American Journal of Medical Genetics, Part A, 2014, 164, 3027-3034.	1.2	6
84	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. Clinical Genetics, 2013, 84, 507-521.	2.0	23
85	12p13.33 microdeletion including ELKS/ERC1, a new locus associated with childhood apraxia of speech. European Journal of Human Genetics, 2013, 21, 82-88.	2.8	70
86	An Improved Method to Extract DNA from 1 ml of Uncultured Amniotic Fluid from Patients at Less than 16 Weeksâ€™ Gestation. PLoS ONE, 2013, 8, e59956.	2.5	1
87	IntragenicCAMTA1rearrangements cause non-progressive congenital ataxia with or without intellectual disability. Journal of Medical Genetics, 2012, 49, 400-408.	3.2	39
88	TheDYRK1Agene is a cause of syndromic intellectual disability with severe microcephaly and epilepsy. Journal of Medical Genetics, 2012, 49, 731-736.	3.2	103
89	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. American Journal of Human Genetics, 2012, 91, 950-957.	6.2	95
90	De Novo 21q22.1q22.2 deletion including RUNX1 mimicking a congenital infection. American Journal of Medical Genetics, Part A, 2011, 155, 126-129.	1.2	12