Valerie Delague

List of Publications by Year
in descending order

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93 papers	3,657 citations	126907 33 h-index	144013 57 g-index
95 all docs	95 docs citations	95 times ranked	5827 citing authors

#	Article	IF	Citations
1	Mutations of POLR3A Encoding a Catalytic Subunit of RNA Polymerase Pol III Cause a Recessive Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2011, 89, 415-423.	6.2	219
2	Mutation in WNT10A Is Associated with an Autosomal Recessive Ectodermal Dysplasia: The Odonto-onycho-dermal Dysplasia. American Journal of Human Genetics, 2007, 81, 821-828.	6.2	214
3	A mutation in periaxin is responsible for CMT4F, an autosomal recessive form of Charcot-Marie-Tooth disease. Human Molecular Genetics, 2001, 10, 415-421.	2.9	188
4	Revisiting the craniosynostosis-radial ray hypoplasia association: Baller-Gerold syndrome caused by mutations in the RECQL4 gene. Journal of Medical Genetics, 2005, 43, 148-152.	3.2	179
5	Mutations in FGD4 Encoding the Rho GDP/GTP Exchange Factor FRABIN Cause Autosomal Recessive Charcot-Marie-Tooth Type 4H. American Journal of Human Genetics, 2007, 81, 1-16.	6.2	152
6	Escobar Syndrome Is a Prenatal Myasthenia Caused by Disruption of the Acetylcholine Receptor Fetal Î³ Subunit. American Journal of Human Genetics, 2006, 79, 303-312.	6.2	146
7	VarAFT: a variant annotation and filtration system for human next generation sequencing data. Nucleic Acids Research, 2018, 46, W545-W553.	14.5	136
8	Exome sequencing identifies mutations inLZTFL1, a BBSome and smoothened trafficking regulator, in a family with Bardetâ€“Biedl syndrome with situs inversus and insertional polydactyly. Journal of Medical Genetics, 2012, 49, 317-321.	3.2	119
9	Mutations inTREM2lead to pure early-onset dementia without bone cysts. Human Mutation, 2008, 29, E194-E204.	2.5	115
10	Segregation of a mutation in CNGB1 encoding the Î²-subunit of the rod cGMP-gated channel in a family with autosomal recessive retinitis pigmentosa. Human Genetics, 2001, 108, 328-334.	3.8	114
11	Influence of Pasteurization and Fat Composition of Milk on the Volatile Compounds and Flavor Characteristics of a Semi-hard Cheese. Journal of Dairy Science, 1998, 81, 3097-3108.	3.4	104
12	X-linked transposition of the great arteries and incomplete penetrance among males with a nonsense mutation in ZIC3. European Journal of Human Genetics, 2000, 8, 704-708.	2.8	103
13	Mapping of a New Locus for Autosomal Recessive Demyelinating Charcot-Marie-Tooth Disease to 19q13.1-13.3 in a Large Consanguineous Lebanese Family: Exclusion of MAG as a Candidate Gene. American Journal of Human Genetics, 2000, 67, 236-243.	6.2	96
14	Loss of Calmodulin- and Radial-Spoke-Associated Complex Protein CFAP251 Leads to Immotile Spermatozoa Lacking Mitochondria and Infertility in Men. American Journal of Human Genetics, 2018, 103, 413-420.	6.2	74
15	Familial Mediterranean fever in Lebanon: mutation spectrum, evidence for cases in Maronites, Greek orthodoxes, Greek catholics, Syriacs and Chiites and for an association between amyloidosis and M694V and M694I mutations. European Journal of Human Genetics, 2001, 9, 51-55.	2.8	68
16	Familial Mediterranean fever inÂtheÂSyrian population: gene mutation frequencies, carrier rates andÂphenotypeâ€“genotype correlation. European Journal of Medical Genetics, 2006, 49, 481-486.	1.3	68
17	Amyloidosis in familial Mediterranean fever patients: correlation with MEFV genotype and SAA1 and MICA polymorphisms effects. BMC Medical Genetics, 2004, 5, 4.	2.1	59
18	PMPCAmutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. Brain, 2015, 138, 1505-1517.	7.6	58

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19	Population Structure in the Mediterranean Basin: A Y Chromosome Perspective. Annals of Human Genetics, 2006, 70, 207-225.	0.8	56
20	Autosomal recessive non-syndromic hearing loss in the Lebanese population: prevalence of the 30delG mutation and report of two novel mutations in the connexin 26 (GJB2) gene. Journal of Medical Genetics, 2001, 38, 36e-36.	3.2	53
21	Brown-Vialetto-Van Laere syndrome in a large inbred Lebanese family: Confirmation of autosomal recessive inheritance?. , 2000, 92, 117-121.		50
22	Genetic screening of fourteen mutations in Jordanian familial Mediterranean fever patients. Human Mutation, 2000, 15, 384-384.	2.5	49
23	Familial Mediterranean fever (FMF) in Lebanon and Jordan: a population genetics study and report of three novel mutations. European Journal of Medical Genetics, 2005, 48, 412-420.	1.3	49
24	Neuroblastoma Amplified Sequence (NBAS) mutation in recurrent acute liver failure: Confirmatory report in a sibship with very early onset, osteoporosis and developmental delay. European Journal of Medical Genetics, 2015, 58, 637-641.	1.3	48
25	Molecular study ofWISP3 in nine families originating from the Middle-East and presenting with progressive pseudorheumatoid dysplasia: Identification of two novel mutations, and description of a founder effect. American Journal of Medical Genetics, Part A, 2005, 138A, 118-126.	1.2	47
26	A novel locus for autosomal recessive primary torsion dystonia (DYT17) maps to 20p11.22â€“q13.12. Neurogenetics, 2008, 9, 287-293.	1.4	47
27	A novel MYH7 mutation links congenital fiber type disproportion and myosin storage myopathy. Neuromuscular Disorders, 2011, 21, 254-262.	0.6	47
28	Homozygosity mapping of autosomal recessive demyelinating Charcot-Marie-Tooth neuropathy (CMT4H) to a novel locus on chromosome 12p11.21-q13.11. Journal of Medical Genetics, 2005, 42, 260-265.	3.2	42
29	WDR73Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Human Mutation, 2015, 36, 1021-1028.	2.5	42
30	A novel deletion in ZBTB24 in a Lebanese family with immunodeficiency, centromeric instability, and facial anomalies syndrome type 2. Clinical Genetics, 2012, 82, 489-493.	2.0	41
31	The added value of WES reanalysis in the field of genetic diagnosis: lessons learned from 200 exomes in the Lebanese population. BMC Medical Genomics, 2019, 12, 11.	1.5	39
32	Molecular genetics of autosomal-recessive axonal Charcot-Marie-Tooth neuropathies. NeuroMolecular Medicine, 2006, 8, 87-106.	3.4	36
33	Molecular diagnosis of inherited peripheral neuropathies by targeted next-generation sequencing: molecular spectrum delineation. BMJ Open, 2018, 8, e021632.	1.9	36
34	Familial Mediterranean Fever: association of elevated IgD plasma levels with specific MEFV mutations. European Journal of Human Genetics, 2001, 9, 849-854.	2.8	34
35	The Impairment of MAGMAS Function in Human Is Responsible for a Severe Skeletal Dysplasia. PLoS Genetics, 2014, 10, e1004311.	3.5	34
36	A new autosomal recessive non-progressive congenital cerebellar ataxia associated with mental retardation, optic atrophy, and skin abnormalities (CAMOS) maps to chromosome 15q24-q26 in a large consanguineous Lebanese Druze family. Neurogenetics, 2002, 4, 23-27.	1.4	32

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37	Phenotypic and genetic exploration of severe demyelinating and secondary axonal neuropathies resulting from GDAP1 nonsense and splicing mutations. Journal of Medical Genetics, 2003, 40, 87e-87.	3.2	31
38	Further delineation of the odonto-onycho-dermal dysplasia syndrome. , 2004, 129A, 193-197.		31
39	Behavioral and Molecular Exploration of the AR-CMT2A Mouse Model Lmna R298C/R298C. NeuroMolecular Medicine, 2012, 14, 40-52.	3.4	30
40	Nonprogressive autosomal recessive ataxia maps to chromosome 9q34-9qter in a large consanguineous lebanese family. Annals of Neurology, 2001, 50, 250-253.	5.3	29
41	Non-syndromic recessive deafness in Jordan: mapping of a new locus to chromosome 9q34.3 and prevalence of DFNB1 mutations. European Journal of Human Genetics, 2002, 10, 391-394.	2.8	28
42	Founder Effect and Estimation of the Age of the c.892C>T (p.Arg298Cys) Mutation in LMNA Associated to Charcotâ€Marieâ€Tooth Subtype CMT2B1 in Families from North Western Africa. Annals of Human Genetics, 2008, 72, 590-597.	0.8	27
43	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27
44	CAMOS, a nonprogressive, autosomal recessive, congenital cerebellar ataxia, is caused by a mutant zinc-finger protein, ZNF592. European Journal of Human Genetics, 2010, 18, 1107-1113.	2.8	26
45	Assignment of a new congenital fibrosis of extraocular muscles type 3 (CFEOM3) locus, FEOM4, based on a balanced translocation t(2;13) (q37.3;q12.11) and identification of candidate genes. Journal of Medical Genetics, 2005, 42, 253-259.	3.2	25
46	Familial Mediterranean Fever In Lebanon: Founder Effects For DifferentMEFVMutations. Annals of Human Genetics, 2008, 72, 41-47.	0.8	23
47	Contribution of next generation sequencing in pediatric practice in Lebanon. A Study on 213 cases. Molecular Genetics & Genomic Medicine, 2018, 6, 1041-1052.	1.2	22
48	New autosomal recessive cerebellar ataxia disorder in a large inbred Lebanese family. American Journal of Medical Genetics Part A, 2001, 101, 135-141.	2.4	20
49	Two novel missense mutations in FGD4/FRABIN cause Charcotâ€Marieâ€Tooth type 4H (CMT4H). Journal of the Peripheral Nervous System, 2012, 17, 141-146.	3.1	18
50	An autosomal recessive cerebellar ataxia syndrome with upward gaze palsy, neuropathy, and seizures. Neurology, 2005, 64, 142-144.	1.1	17
51	Rigid spine syndrome associated with sensoryâ€motor axonal neuropathy resembling Charcotâ€“Marieâ€Tooth disease is characteristic of Bclâ€2â€associated athanogeneâ€3 gene mutations even without cardiac involvement. Muscle and Nerve, 2018, 57, 330-334.	2.2	17
52	Loss of Cajal bodies in motor neurons from patients with novel mutations in VRK1. Human Molecular Genetics, 2019, 28, 2378-2394.	2.9	17
53	A 20-year Clinical and Genetic Neuromuscular Cohort Analysis in Lebanon: An International Effort. Journal of Neuromuscular Diseases, 2022, 9, 193-210.	2.6	17
54	Novel ALPK3 mutation in a Tunisian patient with pediatric cardiomyopathy and facio-thoraco-skeletal features. Journal of Human Genetics, 2018, 63, 1077-1082.	2.3	16

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55	Mutational Analysis in Lebanese Patients with Congenital Adrenal Hyperplasia due to a Deficit in 21-Hydroxylase. Hormone Research in Paediatrics, 2000, 53, 77-82.	1.8	15
56	A new autosomal recessive oto-facial syndrome with midline malformations. , 2005, 132A, 398-401.		15
57	A whole-genome scan in a large family with leukodystrophy and oligodontia reveals linkage to 10q22. Neurogenetics, 2011, 12, 73-78.	1.4	13
58	A Novel Mutation in FGD4/FRABIN Causes Charcot Marie Tooth Disease Type 4H in Patients from a Consanguineous Tunisian Family. Annals of Human Genetics, 2013, 77, 336-343.	0.8	13
59	Autosomal recessive congenital cerebellar hypoplasia and short stature in a large inbred family. , 1999, 87, 88-90.		12
60	Familial Mediterranean fever: the potential for misdiagnosis of E148V using the E148Q usual RFLP detection method. Clinical Genetics, 2002, 61, 71-73.	2.0	11
61	Exclusion of chromosome 15q21.1 in autosomal-recessive Weill-Marchesani syndrome in an inbred Lebanese family. Clinical Genetics, 2008, 58, 473-478.	2.0	11
62	A novel EXT2 mutation in a consanguineous family with severe developmental delay, microcephaly, seizures, feeding difficulties, and osteopenia extends the phenotypic spectrum of autosomal recessive EXT2-related syndrome (AREXT2). European Journal of Medical Genetics, 2019, 62, 259-264.	1.3	11
63	Identification of a ERCC5 c.2333T>C (L778P) Variant in Two Tunisian Siblings With Mild Xeroderma Pigmentosum Phenotype. Frontiers in Genetics, 2019, 10, 111.	2.3	11
64	Recessive marfanoid syndrome with herniation associated with a homozygous mutation in Fibulin-3. European Journal of Medical Genetics, 2020, 63, 103869.	1.3	11
65	Familial Mediterranean fever in a large Lebanese family: Multiple MEFV mutations and evidence for a Founder effect of the p.[M694I] mutation. European Journal of Medical Genetics, 2011, 54, 50-54.	1.3	9
66	Canine neuropathies: powerful spontaneous models for human hereditary sensory neuropathies. Human Genetics, 2019, 138, 455-466.	3.8	9
67	W179R: A novel missense mutation in the peripherin/RDS gene in a family with autosomal dominant retinitis pigmentosa. Human Mutation, 2000, 15, 583-584.	2.5	8
68	COQ8A and MED25 Mutations in a Child with Intellectual Disability, Microcephaly, Seizures, and Spastic Ataxia: Synergistic Effect of Digenic Variants. Molecular Syndromology, 2018, 9, 319-323.	0.8	8
69	SOX11-related syndrome: report on a new case and review. Clinical Dysmorphology, 2021, 30, 44-49.	0.3	7
70	Reverse-Hybridization vs. DNA Sequencing in the Molecular Diagnosis of Familial Mediterranean Fever. Genetic Testing and Molecular Biomarkers, 2004, 8, 65-68.	1.7	6
71	Molecular screening of MECP2 gene in a cohort of Lebanese patients suspected with Rett syndrome: report on a mild case with a novel indel mutation. Journal of Intellectual Disability Research, 2012, 56, 415-420.	2.0	6
72	Identification of novel pathogenic copy number variations in Charcot-Marie-Tooth disease. Journal of Human Genetics, 2020, 65, 313-323.	2.3	6

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73	Multiallelic rare variants support an oligogenic origin of sudden cardiac death in the young. Herz, 2021, 46, 94-102.	1.1	6
74	Nonprogressive autosomal recessive ataxia maps to chromosome 9q34-9qter in a large consanguineous Lebanese family. Annals of Neurology, 2001, 50, 250-3.	5.3	5
75	A new dominant branchiogenic-deafness syndrome with internal auditory canal hypoplasia and abnormal extremities. , 2003, 120A, 276-282.		4
76	A severe clinical phenotype of Noonan syndrome with neonatal hypertrophic cardiomyopathy in the second case worldwide with RAF1 S259Y neomutation. Genetical Research, 2019, 101, e6.	0.9	4
77	A novel PDE6D mutation in a patient with Joubert syndrome type 22 (JBTS22). European Journal of Medical Genetics, 2019, 62, 103576.	1.3	4
78	Identification of a CDH12 potential candidate genetic variant for an autosomal dominant form of transgrediens and progrediens palmoplantar keratoderma in a Tunisian family. Journal of Human Genetics, 2020, 65, 397-410.	2.3	4
79	First characterization of congenital myasthenic syndrome type 5 in North Africa. Molecular Biology Reports, 2021, 48, 6999-7006.	2.3	4
80	Reply: Autosomal recessive cerebellar ataxia caused by a homozygous mutation inPMPCA. Brain, 2016, 139, e20-e20.	7.6	3
81	Clinical profile of comorbidity of rare diseases in a Tunisian patient: a case report associating incontinentia pigmenti and Noonan syndrome. BMC Pediatrics, 2018, 18, 286.	1.7	3
82	A novel biâ€allelic lossâ€ofâ€function mutation in <scp>STIM1</scp> expands the phenotype of <scp>STIM1</scp>â€related diseases. Clinical Genetics, 2021, 100, 84-89.	2.0	3
83	Altered action potential waveform and shorter axonal initial segment in hiPSC-derived motor neurons with mutations in VRK1. Neurobiology of Disease, 2022, 164, 105609.	4.4	3
84	Autosomal recessive congenital cerebellar hypoplasia and short stature in a large inbred family. American Journal of Medical Genetics Part A, 1999, 87, 88-90.	2.4	3
85	Screening for subtelomeric rearrangements using automated fluorescent genotyping of microsatellite markers: a Lebanese study. European Journal of Medical Genetics, 2006, 49, 117-126.	1.3	2
86	Further Delineation of the TRAPPC6B Disorder: Report on a New Family and Review. Journal of Pediatric Genetics, 2019, 08, 252-256.	0.7	2
87	Developmental delay, intellectual disability, short stature, subglottic stenosis, hearing impairment, onychodysplasia of the index fingers, and distinctive facial features: A newly reported autosomal recessive syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1865-1872.	1.2	2
88	Case Report: Identification of Novel Variants in ERCC4 and DDB2 Genes in Two Tunisian Patients With Atypical Xeroderma Pigmentosum Phenotype. Frontiers in Genetics, 2021, 12, 650639.	2.3	2
89	De la souris Ã l'homme : la pÃ©riaxine responsable d'une forme autosomique rÃ©cessive de la maladie de Charcot-Marie-Tooth.. Medecine/Sciences, 2001, 17, 663.	0.2	2
90	Identification of novel mutations by targeted NGS in Moroccan families clinically diagnosed with a neuromuscular disorder. Clinica Chimica Acta, 2022, 524, 51-58.	1.1	2

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91	FANCA Gene Mutations in North African Fanconi Anemia Patients. Frontiers in Genetics, 2021, 12, 610050.	2.3	1
92	Clinical and Molecular Update on the Fourth Reported Family with Hamamy Syndrome. Molecular Syndromology, 2021, 12, 342-350.	0.8	1
93	G.O.3 Mutations in FGD4 encoding the Rho GDP/GTP exchange factor FRABIN cause autosomal recessive Charcot-Marie-Tooth type 4H. Neuromuscular Disorders, 2007, 17, 767.	0.6	0