

# 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  
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95  
docs citations

95  
times ranked

5827  
citing authors

#	ARTICLE	IF	CITATIONS
1	A 20-year Clinical and Genetic Neuromuscular Cohort Analysis in Lebanon: An International Effort. <i>Journal of Neuromuscular Diseases</i> , 2022, 9, 193-210.	2.6	17
2	Identification of novel mutations by targeted NGS in Moroccan families clinically diagnosed with a neuromuscular disorder. <i>Clinica Chimica Acta</i> , 2022, 524, 51-58.	1.1	2
3	Altered action potential waveform and shorter axonal initial segment in hiPSC-derived motor neurons with mutations in VRK1. <i>Neurobiology of Disease</i> , 2022, 164, 105609.	4.4	3
4	Multiallelic rare variants support an oligogenic origin of sudden cardiac death in the young. <i>Herz</i> , 2021, 46, 94-102.	1.1	6
5	FANCA Gene Mutations in North African Fanconi Anemia Patients. <i>Frontiers in Genetics</i> , 2021, 12, 610050.	2.3	1
6	A novel biallelic loss-of-function mutation in <i>STIM1</i> expands the phenotype of <i>STIM1</i> -related diseases. <i>Clinical Genetics</i> , 2021, 100, 84-89.	2.0	3
7	Case Report: Identification of Novel Variants in ERCC4 and DDB2 Genes in Two Tunisian Patients With Atypical Xeroderma Pigmentosum Phenotype. <i>Frontiers in Genetics</i> , 2021, 12, 650639.	2.3	2
8	Clinical and Molecular Update on the Fourth Reported Family with Hamamy Syndrome. <i>Molecular Syndromology</i> , 2021, 12, 342-350.	0.8	1
9	First characterization of congenital myasthenic syndrome type 5 in North Africa. <i>Molecular Biology Reports</i> , 2021, 48, 6999-7006.	2.3	4
10	SOX11-related syndrome: report on a new case and review. <i>Clinical Dysmorphology</i> , 2021, 30, 44-49.	0.3	7
11	Identification of novel pathogenic copy number variations in Charcot-Marie-Tooth disease. <i>Journal of Human Genetics</i> , 2020, 65, 313-323.	2.3	6
12	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. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1865-1872.	1.2	2
13	Identification of a CDH12 potential candidate genetic variant for an autosomal dominant form of transgrediens and progrediens palmoplantar keratoderma in a Tunisian family. <i>Journal of Human Genetics</i> , 2020, 65, 397-410.	2.3	4
14	Recessive marfanoid syndrome with herniation associated with a homozygous mutation in Fibulin-3. <i>European Journal of Medical Genetics</i> , 2020, 63, 103869.	1.3	11
15	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). <i>European Journal of Medical Genetics</i> , 2019, 62, 259-264.	1.3	11
16	Further Delineation of the TRAPPC6B Disorder: Report on a New Family and Review. <i>Journal of Pediatric Genetics</i> , 2019, 08, 252-256.	0.7	2
17	The added value of WES reanalysis in the field of genetic diagnosis: lessons learned from 200 exomes in the Lebanese population. <i>BMC Medical Genomics</i> , 2019, 12, 11.	1.5	39
18	A severe clinical phenotype of Noonan syndrome with neonatal hypertrophic cardiomyopathy in the second case worldwide with <i>RAF1</i> S259Y neomutation. <i>Genetical Research</i> , 2019, 101, e6.	0.9	4

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19	Canine neuropathies: powerful spontaneous models for human hereditary sensory neuropathies. <i>Human Genetics</i> , 2019, 138, 455-466.	3.8	9
20	Loss of Cajal bodies in motor neurons from patients with novel mutations in VRK1. <i>Human Molecular Genetics</i> , 2019, 28, 2378-2394.	2.9	17
21	Identification of a ERCC5 c.2333T>C (L778P) Variant in Two Tunisian Siblings With Mild Xeroderma Pigmentosum Phenotype. <i>Frontiers in Genetics</i> , 2019, 10, 111.	2.3	11
22	A novel PDE6D mutation in a patient with Joubert syndrome type 22 (JBTS22). <i>European Journal of Medical Genetics</i> , 2019, 62, 103576.	1.3	4
23	Rigid spine syndrome associated with sensory-motor axonal neuropathy resembling Charcot-Marie-Tooth disease is characteristic of <i>BCD</i> gene mutations even without cardiac involvement. <i>Muscle and Nerve</i> , 2018, 57, 330-334.	2.2	17
24	COQ8A and MED25 Mutations in a Child with Intellectual Disability, Microcephaly, Seizures, and Spastic Ataxia: Synergistic Effect of Digenic Variants. <i>Molecular Syndromology</i> , 2018, 9, 319-323.	0.8	8
25	Contribution of next generation sequencing in pediatric practice in Lebanon. A Study on 213 cases. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2018, 6, 1041-1052.	1.2	22
26	Molecular diagnosis of inherited peripheral neuropathies by targeted next-generation sequencing: molecular spectrum delineation. <i>BMJ Open</i> , 2018, 8, e021632.	1.9	36
27	Clinical profile of comorbidity of rare diseases in a Tunisian patient: a case report associating incontinentia pigmenti and Noonan syndrome. <i>BMC Pediatrics</i> , 2018, 18, 286.	1.7	3
28	Novel ALPK3 mutation in a Tunisian patient with pediatric cardiomyopathy and facio-thoraco-skeletal features. <i>Journal of Human Genetics</i> , 2018, 63, 1077-1082.	2.3	16
29	Loss of Calmodulin- and Radial-Spoke-Associated Complex Protein CFAP251 Leads to Immotile Spermatozoa Lacking Mitochondria and Infertility in Men. <i>American Journal of Human Genetics</i> , 2018, 103, 413-420.	6.2	74
30	VarAFT: a variant annotation and filtration system for human next generation sequencing data. <i>Nucleic Acids Research</i> , 2018, 46, W545-W553.	14.5	136
31	A recessive ataxia diagnosis algorithm for the next generation sequencing era. <i>Annals of Neurology</i> , 2017, 82, 892-899.	5.3	27
32	Reply: Autosomal recessive cerebellar ataxia caused by a homozygous mutation in <i>PMPCA</i> . <i>Brain</i> , 2016, 139, e20-e20.	7.6	3
33	<i>WDR73</i> Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. <i>Human Mutation</i> , 2015, 36, 1021-1028.	2.5	42
34	<i>PMPCA</i> mutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. <i>Brain</i> , 2015, 138, 1505-1517.	7.6	58
35	Neuroblastoma Amplified Sequence (NBAS) mutation in recurrent acute liver failure: Confirmatory report in a sibship with very early onset, osteoporosis and developmental delay. <i>European Journal of Medical Genetics</i> , 2015, 58, 637-641.	1.3	48
36	The Impairment of MAGMAS Function in Human Is Responsible for a Severe Skeletal Dysplasia. <i>PLoS Genetics</i> , 2014, 10, e1004311.	3.5	34

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37	A Novel Mutation in <i>FGD4/FRABIN</i> Causes Charcot Marie Tooth Disease Type 4H in Patients from a Consanguineous Tunisian Family. <i>Annals of Human Genetics</i> , 2013, 77, 336-343.	0.8	13
38	A novel deletion in <i>ZBTB24</i> in a Lebanese family with immunodeficiency, centromeric instability, and facial anomalies syndrome type 2. <i>Clinical Genetics</i> , 2012, 82, 489-493.	2.0	41
39	Exome sequencing identifies mutations in <i>LZTFL1</i> , a BBSome and smoothed trafficking regulator, in a family with Bardet-Biedl syndrome with situs inversus and insertional polydactyly. <i>Journal of Medical Genetics</i> , 2012, 49, 317-321.	3.2	119
40	Behavioral and Molecular Exploration of the AR-CMT2A Mouse Model <i>Lmna</i> R298C/R298C. <i>NeuroMolecular Medicine</i> , 2012, 14, 40-52.	3.4	30
41	Molecular screening of <i>MECP2</i> gene in a cohort of Lebanese patients suspected with Rett syndrome: report on a mild case with a novel indel mutation. <i>Journal of Intellectual Disability Research</i> , 2012, 56, 415-420.	2.0	6
42	Two novel missense mutations in <i>FGD4/FRABIN</i> cause Charcot-Marie-Tooth type 4H (CMT4H). <i>Journal of the Peripheral Nervous System</i> , 2012, 17, 141-146.	3.1	18
43	Familial Mediterranean fever in a large Lebanese family: Multiple MEFV mutations and evidence for a Founder effect of the p.[M694I] mutation. <i>European Journal of Medical Genetics</i> , 2011, 54, 50-54.	1.3	9
44	A novel MYH7 mutation links congenital fiber type disproportion and myosin storage myopathy. <i>Neuromuscular Disorders</i> , 2011, 21, 254-262.	0.6	47
45	Mutations of <i>POLR3A</i> Encoding a Catalytic Subunit of RNA Polymerase Pol III Cause a Recessive Hypomyelinating Leukodystrophy. <i>American Journal of Human Genetics</i> , 2011, 89, 415-423.	6.2	219
46	A whole-genome scan in a large family with leukodystrophy and oligodontia reveals linkage to 10q22. <i>Neurogenetics</i> , 2011, 12, 73-78.	1.4	13
47	CAMOS, a nonprogressive, autosomal recessive, congenital cerebellar ataxia, is caused by a mutant zinc-finger protein, <i>ZNF592</i> . <i>European Journal of Human Genetics</i> , 2010, 18, 1107-1113.	2.8	26
48	Familial Mediterranean Fever In Lebanon: Founder Effects For Different <i>MEFV</i> Mutations. <i>Annals of Human Genetics</i> , 2008, 72, 41-47.	0.8	23
49	A novel locus for autosomal recessive primary torsion dystonia ( <i>DYT17</i> ) maps to 20p11.22-q13.12. <i>Neurogenetics</i> , 2008, 9, 287-293.	1.4	47
50	Exclusion of chromosome 15q21.1 in autosomal-recessive Weill-Marchesani syndrome in an inbred Lebanese family. <i>Clinical Genetics</i> , 2008, 58, 473-478.	2.0	11
51	Mutations in <i>TREM2</i> lead to pure early-onset dementia without bone cysts. <i>Human Mutation</i> , 2008, 29, E194-E204.	2.5	115
52	Founder Effect and Estimation of the Age of the c.892C>T (p.Arg298Cys) Mutation in <i>LMNA</i> Associated to Charcot-Marie-Tooth Subtype CMT2B1 in Families from North Western Africa. <i>Annals of Human Genetics</i> , 2008, 72, 590-597.	0.8	27
53	G.O.3 Mutations in <i>FGD4</i> encoding the Rho GDP/GTP exchange factor <i>FRABIN</i> cause autosomal recessive Charcot-Marie-Tooth type 4H. <i>Neuromuscular Disorders</i> , 2007, 17, 767.	0.6	0
54	Mutations in <i>FGD4</i> Encoding the Rho GDP/GTP Exchange Factor <i>FRABIN</i> Cause Autosomal Recessive Charcot-Marie-Tooth Type 4H. <i>American Journal of Human Genetics</i> , 2007, 81, 1-16.	6.2	152

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55	Mutation in WNT10A Is Associated with an Autosomal Recessive Ectodermal Dysplasia: The Odonto-onycho-dermal Dysplasia. <i>American Journal of Human Genetics</i> , 2007, 81, 821-828.	6.2	214
56	Escobar Syndrome Is a Prenatal Myasthenia Caused by Disruption of the Acetylcholine Receptor Fetal $\beta$ Subunit. <i>American Journal of Human Genetics</i> , 2006, 79, 303-312.	6.2	146
57	Molecular genetics of autosomal-recessive axonal Charcot-Marie-Tooth neuropathies. <i>NeuroMolecular Medicine</i> , 2006, 8, 87-106.	3.4	36
58	Screening for subtelomeric rearrangements using automated fluorescent genotyping of microsatellite markers: a Lebanese study. <i>European Journal of Medical Genetics</i> , 2006, 49, 117-126.	1.3	2
59	Familial Mediterranean fever in the Syrian population: gene mutation frequencies, carrier rates and phenotype-genotype correlation. <i>European Journal of Medical Genetics</i> , 2006, 49, 481-486.	1.3	68
60	Population Structure in the Mediterranean Basin: A Y Chromosome Perspective. <i>Annals of Human Genetics</i> , 2006, 70, 207-225.	0.8	56
61	A new autosomal recessive oto-facial syndrome with midline malformations. , 2005, 132A, 398-401.		15
62	Molecular study of WISP3 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. <i>American Journal of Medical Genetics, Part A</i> , 2005, 138A, 118-126.	1.2	47
63	Homozygosity mapping of autosomal recessive demyelinating Charcot-Marie-Tooth neuropathy (CMT4H) to a novel locus on chromosome 12p11.21-q13.11. <i>Journal of Medical Genetics</i> , 2005, 42, 260-265.	3.2	42
64	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. <i>Journal of Medical Genetics</i> , 2005, 42, 253-259.	3.2	25
65	Revisiting the craniosynostosis-radial ray hypoplasia association: Baller-Gerold syndrome caused by mutations in the RECQL4 gene. <i>Journal of Medical Genetics</i> , 2005, 43, 148-152.	3.2	179
66	An autosomal recessive cerebellar ataxia syndrome with upward gaze palsy, neuropathy, and seizures. <i>Neurology</i> , 2005, 64, 142-144.	1.1	17
67	Familial Mediterranean fever (FMF) in Lebanon and Jordan: a population genetics study and report of three novel mutations. <i>European Journal of Medical Genetics</i> , 2005, 48, 412-420.	1.3	49
68	Reverse-Hybridization vs. DNA Sequencing in the Molecular Diagnosis of Familial Mediterranean Fever. <i>Genetic Testing and Molecular Biomarkers</i> , 2004, 8, 65-68.	1.7	6
69	Amyloidosis in familial Mediterranean fever patients: correlation with MEFV genotype and SAA1 and MICA polymorphisms effects. <i>BMC Medical Genetics</i> , 2004, 5, 4.	2.1	59
70	Further delineation of the odonto-onycho-dermal dysplasia syndrome. , 2004, 129A, 193-197.		31
71	A new dominant branchiogenic-deafness syndrome with internal auditory canal hypoplasia and abnormal extremities. , 2003, 120A, 276-282.		4
72	Phenotypic and genetic exploration of severe demyelinating and secondary axonal neuropathies resulting from GDAP1 nonsense and splicing mutations. <i>Journal of Medical Genetics</i> , 2003, 40, 87e-87.	3.2	31

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73	Familial Mediterranean fever: the potential for misdiagnosis of E148V using the E148Q usual RFLP detection method. <i>Clinical Genetics</i> , 2002, 61, 71-73.	2.0	11
74	Non-syndromic recessive deafness in Jordan: mapping of a new locus to chromosome 9q34.3 and prevalence of DFNB1 mutations. <i>European Journal of Human Genetics</i> , 2002, 10, 391-394.	2.8	28
75	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. <i>Neurogenetics</i> , 2002, 4, 23-27.	1.4	32
76	Segregation of a mutation in CNGB1 encoding the $\beta$ -subunit of the rod cGMP-gated channel in a family with autosomal recessive retinitis pigmentosa. <i>Human Genetics</i> , 2001, 108, 328-334.	3.8	114
77	New autosomal recessive cerebellar ataxia disorder in a large inbred Lebanese family. <i>American Journal of Medical Genetics Part A</i> , 2001, 101, 135-141.	2.4	20
78	Nonprogressive autosomal recessive ataxia maps to chromosome 9q34-9qter in a large consanguineous lebanese family. <i>Annals of Neurology</i> , 2001, 50, 250-253.	5.3	29
79	Familial Mediterranean fever in Lebanon: mutation spectrum, evidence for cases in Maronites, Greek orthodoxes, Greek catholics, Syrians and Chiites and for an association between amyloidosis and M694V and M694I mutations. <i>European Journal of Human Genetics</i> , 2001, 9, 51-55.	2.8	68
80	Familial Mediterranean Fever: association of elevated IgD plasma levels with specific MEFV mutations. <i>European Journal of Human Genetics</i> , 2001, 9, 849-854.	2.8	34
81	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. <i>Journal of Medical Genetics</i> , 2001, 38, 36e-36.	3.2	53
82	A mutation in periaxin is responsible for CMT4F, an autosomal recessive form of Charcot-Marie-Tooth disease. <i>Human Molecular Genetics</i> , 2001, 10, 415-421.	2.9	188
83	De la souris à l'homme : la p $\alpha$ riaxine responsable d'une forme autosomique r $\alpha$ cessive de la maladie de Charcot-Marie-Tooth.. <i>Medecine/Sciences</i> , 2001, 17, 663.	0.2	2
84	Nonprogressive autosomal recessive ataxia maps to chromosome 9q34-9qter in a large consanguineous Lebanese family. <i>Annals of Neurology</i> , 2001, 50, 250-3.	5.3	5
85	Brown-Vialetto-Van Laere syndrome in a large inbred Lebanese family: Confirmation of autosomal recessive inheritance?. , 2000, 92, 117-121.		50
86	Genetic screening of fourteen mutations in Jordanian familial Mediterranean fever patients. <i>Human Mutation</i> , 2000, 15, 384-384.	2.5	49
87	W179R: A novel missense mutation in the peripherin/RDS gene in a family with autosomal dominant retinitis pigmentosa. <i>Human Mutation</i> , 2000, 15, 583-584.	2.5	8
88	X-linked transposition of the great arteries and incomplete penetrance among males with a nonsense mutation in ZIC3. <i>European Journal of Human Genetics</i> , 2000, 8, 704-708.	2.8	103
89	Mutational Analysis in Lebanese Patients with Congenital Adrenal Hyperplasia due to a Deficit in 21-Hydroxylase. <i>Hormone Research in Paediatrics</i> , 2000, 53, 77-82.	1.8	15
90	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. <i>American Journal of Human Genetics</i> , 2000, 67, 236-243.	6.2	96

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91	Autosomal recessive congenital cerebellar hypoplasia and short stature in a large inbred family. , 1999, 87, 88-90.		12
92	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
93	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