Sally Ann Lynch

List of Publications by Year
in descending order

128	7,153	101543	64796
papers	citations	36 h-index	79 g-index

141	141	141	13104
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prevalence and architecture of de novo mutations in developmental disorders. Nature, 2017, 542, 433-438.	27.8	1,211
2	Mutations in the gene encoding the 3â€²-5â€² DNA exonuclease TREX1 cause Aicardi-GoutiÃ¨res syndrome at the AGS1 locus. Nature Genetics, 2006, 38, 917-920.	21.4	752
3	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	6.2	375
4	Mutations in VPS33B, encoding a regulator of SNARE-dependent membrane fusion, cause arthrogryposisâ€“renal dysfunctionâ€“cholestasis (ARC) syndrome. Nature Genetics, 2004, 36, 400-404.	21.4	313
5	A homeobox gene, HLXB9, is the major locus for dominantly inherited sacral agenesis. Nature Genetics, 1998, 20, 358-361.	21.4	287
6	Mutations in the Gene Encoding Filamin A as a Cause for Familial Cardiac Valvular Dystrophy. Circulation, 2007, 115, 40-49.	1.6	257
7	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
8	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
9	Mutations in the TGFÎ² Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromicric and Geleophysic Dysplasias. American Journal of Human Genetics, 2011, 89, 7-14.	6.2	199
10	Mutations in TCF12, encoding a basic helix-loop-helix partner of TWIST1, are a frequent cause of coronal craniosynostosis. Nature Genetics, 2013, 45, 304-307.	21.4	181
11	Unexplained early onset epileptic encephalopathy: Exome screening and phenotype expansion. Epilepsia, 2016, 57, e12-7.	5.1	164
12	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
13	A gene for autosomal dominant sacral agenesis maps to the holoprosencephaly region at 7q36. Nature Genetics, 1995, 11, 93-95.	21.4	150
14	How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. European Journal of Human Genetics, 2012, 20, 381-388.	2.8	142
15	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	6.2	125
16	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2972-2980.	1.2	119
17	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. Human Mutation, 2010, 31, 1142-1154.	2.5	111
18	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108

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19	8p23.1 duplication syndrome; a novel genomic condition with unexpected complexity revealed by array CGH. European Journal of Human Genetics, 2008, 16, 18-27.	2.8	74
20	Adults with Down's syndrome: the prevalence of complications and health care in the community. British Journal of General Practice, 2007, 57, 50-5.	1.4	73
21	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
22	NAA10 mutation causing a novel intellectual disability syndrome with Long QT due to N-terminal acetyltransferase impairment. Scientific Reports, 2015, 5, 16022.	3.3	61
23	Complex Segmental Duplications Mediate a Recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. American Journal of Human Genetics, 2009, 85, 394-400.	6.2	60
24	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59
25	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	6.2	56
26	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
27	Further case of Rubinsteinâ€“Taybi syndrome due to a deletion in EP300. American Journal of Medical Genetics, Part A, 2009, 149A, 997-1000.	1.2	51
28	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48
29	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
30	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
31	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. Journal of Medical Genetics, 2011, 48, 417-421.	3.2	45
32	Towards the identification of a genetic basis for <scp>L</scp>andauâ€<scp>K</scp>leffner <scp>s</scp>yndrome. Epilepsia, 2014, 55, 858-865.	5.1	44
33	Recessive NEK9 mutation causes a lethal skeletal dysplasia with evidence of cell cycle and ciliary defects. Human Molecular Genetics, 2016, 25, 1824-1835.	2.9	44
34	Non-multifactorial neural tube defects. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 135C, 69-76.	1.6	43
35	Clinical and genetic characterisation of infantile liver failure syndrome type 1, due to recessive mutations in LARS. Journal of Inherited Metabolic Disease, 2015, 38, 1085-1092.	3.6	43
36	NRXN1 deletion syndrome; phenotypic and penetrance data from 34 families. European Journal of Medical Genetics, 2019, 62, 204-209.	1.3	43

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37	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	2.5	42
38	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. American Journal of Human Genetics, 2021, 108, 1083-1094.	6.2	42
39	CHRNGgenotypeâ€“phenotype correlations in the multiple pterygium syndromes. Journal of Medical Genetics, 2012, 49, 21-26.	3.2	41
40	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	2.4	41
41	The 12q14 microdeletion syndrome: six new cases confirming the role of HMGA2 in growth. European Journal of Human Genetics, 2011, 19, 534-539.	2.8	39
42	Characterization of a 8q21.11 Microdeletion Syndrome Associated with Intellectual Disability and a Recognizable Phenotype. American Journal of Human Genetics, 2011, 89, 295-301.	6.2	37
43	Incidence and prevalence of mucopolysaccharidosis type 1 in the Irish republic. Archives of Disease in Childhood, 2009, 94, 52-54.	1.9	35
44	Inherited PAX6, NF1 and OTX2 mutations in a child with microphthalmia and aniridia. European Journal of Human Genetics, 2007, 15, 898-901.	2.8	33
45	A Novel Variant of Familial Glucocorticoid Deficiency Prevalent among the Irish Traveler Population. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 2896-2899.	3.6	32
46	Homozygous nonsense and frameshift mutations of the ACTH receptor in children with familial glucocorticoid deficiency (FGD) are not associated with longâ€term mineralocorticoid deficiency. Clinical Endocrinology, 2009, 71, 171-175.	2.4	32
47	The clinical presentation caused by truncating CHD8 variants. Clinical Genetics, 2019, 96, 72-84.	2.0	32
48	Rubinsteinâ€“Taybi syndrome type 2: report of nine new cases that extend the phenotypic and genotypic spectrum. Clinical Dysmorphology, 2016, 25, 135-145.	0.3	31
49	The phenotype of Sotos syndrome in adulthood: A review of 44 individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 502-508.	1.6	31
50	Broadening the phenotype associated with mutations in UPF3B: Two further cases with renal dysplasia and variable developmental delay. European Journal of Medical Genetics, 2012, 55, 476-479.	1.3	30
51	Heterozygous loss-of-function variants of MEIS2 cause a triad of palatal defects, congenital heart defects, and intellectual disability. European Journal of Human Genetics, 2019, 27, 278-290.	2.8	30
52	Barriers and Considerations for Diagnosing Rare Diseases in Indigenous Populations. Frontiers in Pediatrics, 2020, 8, 579924.	1.9	25
53	Unexpected genetic heterogeneity for primary ciliary dyskinesia in the Irish Traveller population. European Journal of Human Genetics, 2015, 23, 210-217.	2.8	24
54	A novel gain-of-function mutation in the ITPR1 suppressor domain causes spinocerebellar ataxia with altered Ca2+ signal patterns. Journal of Neurology, 2017, 264, 1444-1453.	3.6	22

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55	A novel NAA10 p.(R83H) variant with impaired acetyltransferase activity identified in two boys with ID and microcephaly. BMC Medical Genetics, 2019, 20, 101.	2.1	21
56	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
57	Atypical Alstrom syndrome with novel ALMS1 mutations precluded by current diagnostic criteria. European Journal of Medical Genetics, 2014, 57, 55-59.	1.3	20
58	Wide disparity of clinical genetics services and EU rare disease research funding across Europe. Journal of Community Genetics, 2016, 7, 119-126.	1.2	20
59	Catalogue of inherited disorders found among the Irish Traveller population. Journal of Medical Genetics, 2018, 55, 233-239.	3.2	19
60	Stickler's syndrome associated with congenital glaucoma. Ophthalmic Genetics, 1998, 19, 55-58.	1.2	18
61	Novel European SLC1A4 variant: infantile spasms and population ancestry analysis. Journal of Human Genetics, 2016, 61, 761-764.	2.3	18
62	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
63	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
64	A case report of primary ciliary dyskinesia, laterality defects and developmental delay caused by the co-existence of a single gene and chromosome disorder. BMC Medical Genetics, 2015, 16, 45.	2.1	15
65	Chromosomal microarray in unexplained severe early onset epilepsy â€“ A single centre cohort. European Journal of Paediatric Neurology, 2015, 19, 390-394.	1.6	14
66	NRXN1Î±+/- is associated with increased excitability in ASD iPSC-derived neurons. BMC Neuroscience, 2021, 22, 56.	1.9	14
67	A Novel Constellation of Cardiac Findings for Kabuki Syndrome: Hypoplastic Left Heart Syndrome and Partial Anomalous Pulmonary Venous Drainage. Pediatric Cardiology, 2008, 29, 820-822.	1.3	13
68	Intra-familial variability associated with recessive RYR1 mutation diagnosed prenatally by exome sequencing. Prenatal Diagnosis, 2016, 36, 1020-1026.	2.3	13
69	Novel <scp>SMC</scp>1A variant and epilepsy of infancy with migrating focal seizures: Expansion of the phenotype. Epilepsia, 2017, 58, 1301-1302.	5.1	12
70	Beaulieuâ€“Boycottâ€“Innes syndrome: an intellectual disability syndrome with characteristic facies. Clinical Dysmorphology, 2016, 25, 146-151.	0.3	11
71	A retrospective review of the contribution of rare diseases to paediatric mortality in Ireland. Orphanet Journal of Rare Diseases, 2020, 15, 311.	2.7	11
72	The genetic landscape of polycystic kidney disease in Ireland. European Journal of Human Genetics, 2021, 29, 827-838.	2.8	11

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73	Atypical benign partial epilepsy of childhood with acquired neurocognitive, lexical semantic, and autistic spectrum disorder. Epilepsy & Behavior Case Reports, 2016, 6, 42-48.	1.5	10
74	Expanding the clinical spectrum of chromosome 15q26 terminal deletions associated with IGF-1 resistance. European Journal of Pediatrics, 2017, 176, 137-142.	2.7	10
75	Complex rearrangement of the exon 6 genomic region among Opitz G/BBB Syndrome MID1 alterations. European Journal of Medical Genetics, 2013, 56, 404-410.	1.3	9
76	Two new mutations in exon 3 of theNDP gene: S73X and S101F associated with severe and less severe ocular phenotype, respectively. Human Mutation, 1997, 9, 53-56.	2.5	8
77	Diencephalic cachexia of infancy: Russell's syndrome. Clinical Dysmorphology, 2006, 15, 253-254.	0.3	8
78	Predictive Genetic Testing and Alternatives to Face to Face Results Disclosure: A Retrospective Review of Patients Preference for Alternative Modes of BRCA 1 and 2 Results Disclosure in the Republic of Ireland. Journal of Genetic Counseling, 2016, 25, 422-431.	1.6	8
79	A case of persistent pulmonary hypertension in a newborn with Costello syndrome. Clinical Dysmorphology, 2008, 17, 287-288.	0.3	7
80	Communication of Genetic Information by Other Health Professionals: The Role of the Genetic Counsellor in Specialist Clinics. Journal of Genetic Counseling, 2011, 20, 192-203.	1.6	7
81	Status dystonicus in two patients with SOX2â€anophthalmia syndrome and nonsense mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3048-3050.	1.2	7
82	Phenotypic Variability in Leukoencephalopathy with Brain Calcifications and Cysts: Case Report of Siblings from an Irish Traveller Family with a Homozygous SNORD118 Mutation. Journal of Molecular Neuroscience, 2020, 70, 1354-1356.	2.3	7
83	Dying to see you? Deaths on a clinical genetics waiting list in the Republic of Ireland; what are the consequences?. Journal of Community Genetics, 2021, 12, 121-127.	1.2	7
84	De novo missense variants in FBXO11 alter its protein expression and subcellular localization. Human Molecular Genetics, 2022, 31, 440-454.	2.9	7
85	Periventricular Calcification, Abnormal Pterins and Dry Thickened Skin: Expanding the Clinical Spectrum of RMND1?. JIMD Reports, 2015, 26, 13-19.	1.5	6
86	Managing uncertainty in inherited cardiac pathologiesâ€"an international multidisciplinary survey. European Journal of Human Genetics, 2019, 27, 1178-1185.	2.8	6
87	Towards establishing consistency in triage in a tertiary specialty. European Journal of Human Genetics, 2019, 27, 547-555.	2.8	6
88	Integration of genetic and histopathology data in interpretation of kidney disease. Nephrology Dialysis Transplantation, 2020, 35, 1113-1132.	0.7	6
89	The Role of the European Society of Human Genetics in Delivering Genomic Education. Frontiers in Genetics, 2021, 12, 693952.	2.3	6
90	Rare Disease Research Partnership (RAinDRoP): a collaborative approach to identify the top 15 research priorities for rare diseases. HRB Open Research, 2020, 3, 13.	0.6	6

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91	Response to treatment and outcomes of infantile spasms in DownÂsyndrome. Developmental Medicine and Child Neurology, 2022, 64, 780-788.	2.1	6
92	What price a diagnosis?. Developmental Medicine and Child Neurology, 2011, 53, 971-971.	2.1	5
93	Mutations of TCF12, encoding a basic-helix-loop-helix partner of TWIST1, are a frequent cause of coronal craniosynostosis. Lancet, The, 2013, 381, S114.	13.7	5
94	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.5	5
95	Microdeletion 1p35.2: A recognizable facial phenotype with developmental delay. American Journal of Medical Genetics, Part A, 2015, 167, 1916-1920.	1.2	4
96	Friedreich Ataxia in Classical Galactosaemia. JIMD Reports, 2015, 26, 1-5.	1.5	4
97	National Newborn Screening for cystic fibrosis in the Republic of Ireland: genetic data from the first 6.5 years. European Journal of Human Genetics, 2020, 28, 1669-1674.	2.8	4
98	Duplication of referral, a tsunami of paper: how much does it cost the Irish health services?. Irish Journal of Medical Science, 2022, , 1.	1.5	4
99	Vocal cord paralysis in association with 9q34 duplication. Clinical Dysmorphology, 2014, 23, 105-108.	0.3	3
100	One to Watch: A Germ Cell Tumor Arising in an Undescended Testicle in Rubinstein-Taybi Syndrome. Journal of Pediatric Hematology/Oncology, 2016, 38, e191-e192.	0.6	3
101	Congenital radial head dislocation and low immunoglobulin M levels in 6p25.3 deletion. Clinical Dysmorphology, 2017, 26, 181-184.	0.3	3
102	The contribution of 7q33 copy number variations for intellectual disability. Neurogenetics, 2018, 19, 27-40.	1.4	3
103	Cost of exome sequencing in epileptic encephalopathy: is it â€˜worth itâ€™?. Archives of Disease in Childhood, 2018, 103, 304-304.	1.9	3
104	X-linked infantile spinal muscular atrophy (SMAX2) caused by novel c.1681G>A substitution in the UBA1 gene, expanding the phenotype. Neuromuscular Disorders, 2020, 30, 35-37.	0.6	3
105	An estimate of the cumulative paediatric prevalence of rare diseases in Ireland and comment on the literature. European Journal of Human Genetics, 2022, 30, 1211-1215.	2.8	3
106	<scp>FOXN</scp>1 Duplication and Congenital Hypertrichosis. Pediatric Dermatology, 2017, 34, e77-e79.	0.9	2
107	<scp>HK1</scp> haemolytic anaemia in association with a neurological phenotype and coâ€existing <scp>CEP290 Meckelâ€“Gruber</scp> in a Romani family. Clinical Genetics, 2022, 101, 142-143.	2.0	2
108	Nonâ€syndromic bilateral ulnar aplasia with humeroâ€radial synostosis and oligoâ€ectroâ€dactyly. American Journal of Medical Genetics, Part A, 2018, 176, 1180-1183.	1.2	1

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109	A perinatal approach to genetic disorders in Irish Travellers: A review. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2018, 228, 43-47.	1.1	1
110	P496â€…X- linked infantile spinal muscular atrophy (smax2) caused by novel c.1681g>a substitution in the uba1 gene, expanding the phenotype. , 2019, , .		1
111	Derivation of familial iPSC lines from three ASD patients carrying NRXN1Î± and two controls (NUIGi022-A, NUIGi022-B; NUIGi023-A, NUIGi023-B; NUIGi025-A, NUIGi025-B; NUIGi024-A, NUIGi024-B;) Tj ETQq1 1 0.784314 rgBT /Ov		
112	Who supports the support workers? Cross-sectional survey of support workersâ€™ experience and views. European Journal of Human Genetics, 2004, 12, 251-254.	2.8	0
113	Malpuech syndrome: facial features in the absence of clefting. Clinical Dysmorphology, 2006, 15, 243-244.	0.3	0
114	A novel mutation in PIEZO2 in a family presenting with autosomal dominant myopathy, ptosis, external ophthalmoplegia and distal symphalangism. Neuromuscular Disorders, 2015, 25, S276-S277.	0.6	0
115	Bicoronal and metopic craniosynostosis in association with a de novo unbalanced t(2;7) chromosomal translocation. American Journal of Medical Genetics, Part A, 2017, 173, 274-279.	1.2	0
116	Clinical characterisation of neurexin1 deletions and their role in neurodevelopmental disorders. European Neuropsychopharmacology, 2017, 27, S762-S763.	0.7	0
117	DECONSTRUCTING THE NEUREXIN1 DELETION PHENOTYPE: A NEUROPSYCHOLOGICAL, NEUROCOGNITIVE AND NEUROIMAGING PERSPECTIVE. European Neuropsychopharmacology, 2019, 29, S980-S981.	0.7	0
118	GP165â€…Towards estimating the incidence of rare diseases in a paediatric population, born in ireland in the year 2000. , 2019, , .		0
119	OC48â€…Re-interrogation of whole exome sequencing data in developmental epileptic encephalopathies. , 2019, , .		0
120	P493â€…Late and atypical presentation of mecp2 mutation. , 2019, , .		0
121	OC27â€…National newborn screening for cystic fibrosis: genetic data from the first 6 years. , 2019, , .		0
122	GP227â€…Clinical spectrum of classical galactosaemia associated with friedreichâ€™s ataxia in a paediatric cohort in the republic of ireland â€“ an update. , 2019, , .		0
123	OC46â€…The population incidence of childhood gonadoblastoma over 20 years in the republic of ireland. , 2019, , .		0
124	P430â€…Identifying rare disease research priorities through (RAinDRoP) a rare disease research partnership. , 2019, , .		0
125	Derivation of iPSC lines from two patients with autism spectrum disorder carrying NRXN1Î± deletion (NUIGi041-A, NUIG041-B; NUIGi045-A) and one sibling control (NUIGi042-A, NUIGi042-B). Stem Cell Research, 2021, 52, 102222.	0.7	0
126	Derivation of four iPSC lines from a male ASD patient carrying a deletion in the middle coding region of NRXN1Î± gene (NUIGi039-A and NUIGi039-B) and a male sibling control (NUIGi040-A and NUIGi040-B). Stem Cell Research, 2021, 53, 102254.	0.7	0

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127	Fatal fetal abnormality Irish live-born survivalâ€"an observational study. Journal of Community Genetics, 2021, 12, 643-651.	1.2	0
128	The Irish National Rare Disease Office (NRDO): A national step towards improving access to health and care services for individuals and families living with Rare Diseases. International Journal of Integrated Care, 2017, 17, 239.	0.2	0