Elliott H Sherr

List of Publications by Year in Descending Order

68 papers	3,212 citations	31 h-index	56 g-index
77 ext. papers	4,108 ext. citations	7.7 avg, IF	4.81 L-index

#	Paper	IF	Citations
68	De Novo GLI3 Pathogenic Variants May Cause Hypotonia and a Range of Brain Malformations Without Skeletal Abnormalities.. *Pediatric Neurology*, **2022**, 131, 1-3	2.9	
67	DCC regulates astroglial development essential for telencephalic morphogenesis and corpus callosum formation. *ELife*, **2021**, 10,	8.9	2
66	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. *American Journal of Human Genetics*, **2021**, 108, 951-961	11	4
65	DRAXIN regulates interhemispheric fissure remodelling to influence the extent of corpus callosum formation. *ELife*, **2021**, 10,	8.9	2
64	16p11.2 deletion syndrome. *Current Opinion in Genetics and Development*, **2021**, 68, 49-56	4.9	8
63	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. *American Journal of Human Genetics*, **2021**, 108, 1069-1082	11	4
62	Autism-associated biomarkers: test-retest reliability and relationship to quantitative social trait variation in rhesus monkeys. *Molecular Autism*, **2021**, 12, 50	6.5	1
61	Unique variants in CLCN3, encoding an endosomal anion/proton exchanger, underlie a spectrum of neurodevelopmental disorders. *American Journal of Human Genetics*, **2021**, 108, 1450-1465	11	0
60	Altered structural connectivity networks in a mouse model of complete and partial dysgenesis of the corpus callosum. *NeuroImage*, **2020**, 217, 116868	7.9	4
59	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. *Neuron*, **2020**, 106, 404-420.e8	13.9	49
58	Randomized Clinical Trial of First-Line Genome Sequencing in Pediatric White Matter Disorders. *Annals of Neurology*, **2020**, 88, 264-273	9.4	7
57	Abnormal Auditory Mismatch Fields in Children and Adolescents With 16p11.2 Deletion and 16p11.2 Duplication. *Biological Psychiatry: Cognitive Neuroscience and Neuroimaging*, **2020**, 5, 942-950	3.4	1
56	Overcoming presynaptic effects of VAMP2 mutations with 4-aminopyridine treatment. *Human Mutation*, **2020**, 41, 1999-2011	4.7	3
55	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
54	Integrated stress response inhibition provides sex-dependent protection against noise-induced cochlear synaptopathy. *Scientific Reports*, **2020**, 10, 18063	4.9	3
53	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Collosum, Axon, Cardiac, Ocular, and Genital Defects. *American Journal of Human Genetics*, **2019**, 105, 854-868	11	17
52	Sensorimotor Cortical Oscillations during Movement Preparation in 16p11.2 Deletion Carriers. *Journal of Neuroscience*, **2019**, 39, 7321-7331	6.6	6

51	Genomic and phenotypic delineation of congenital microcephaly. *Genetics in Medicine*, **2019**, 21, 545-552	8.1	55
50	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. *Biological Psychiatry*, **2018**, 84, 253-264	7.9	33
49	Genotype-phenotype correlations in individuals with pathogenic RERE variants. *Human Mutation*, **2018**, 39, 666-675	4.7	19
48	Abnormal Speech Motor Control in Individuals with 16p11.2 Deletions. *Scientific Reports*, **2018**, 8, 1274	4.9	18
47	Mutations in Vps15 perturb neuronal migration in mice and are associated with neurodevelopmental disease in humans. *Nature Neuroscience*, **2018**, 21, 207-217	25.5	24
46	Arginine vasopressin in cerebrospinal fluid is a marker of sociality in nonhuman primates. *Science Translational Medicine*, **2018**, 10,	17.5	34
45	DCC mutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. *Human Mutation*, **2018**, 39, 23-39	4.7	26
44	Brain MR Imaging Findings and Associated Outcomes in Carriers of the Reciprocal Copy Number Variation at 16p11.2. *Radiology*, **2018**, 286, 217-226	20.5	19
43	Burden of de novo mutations and inherited rare single nucleotide variants in children with sensory processing dysfunction. *BMC Medical Genomics*, **2018**, 11, 50	3.7	5
42	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. *American Journal of Human Genetics*, **2018**, 103, 752-768	11	19
41	Cerebrospinal fluid vasopressin and symptom severity in children with autism. *Annals of Neurology*, **2018**, 84, 611-615	9.4	18
40	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. *Nature Genetics*, **2017**, 49, 511-514	36.3	54
39	disease: Phenotype clarification and genotype-phenotype correlation. *Neurology: Genetics*, **2017**, 3, e148	3.8	25
38	Linking tuberous sclerosis complex, excessive mTOR signaling, and age-related neurodegeneration: a new association between TSC1 mutation and frontotemporal dementia. *Acta Neuropathologica*, **2017**, 134, 813-816	14.3	9
37	Clinical phenotype of the recurrent 1q21.1 copy-number variant. *Genetics in Medicine*, **2016**, 18, 341-9	8.1	84
36	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. *Nature Genetics*, **2016**, 48, 1185-92	36.3	74
35	16p11.2 deletion and duplication: Characterizing neurologic phenotypes in a large clinically ascertained cohort. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2943-2955	2.5	86
34	Reciprocal white matter alterations due to 16p11.2 chromosomal deletions versus duplications. *Human Brain Mapping*, **2016**, 37, 2833-48	5.9	25

33	Auditory Evoked M100 Response Latency is Delayed in Children with 16p11.2 Deletion but not 16p11.2 Duplication. *Cerebral Cortex*, **2016**, 26, 1957-64	5.1	25
32	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry*, **2016**, 73, 20-30	14.5	120
31	Early Predictors of Impaired Social Functioning in Male Rhesus Macaques (Macaca mulatta). *PLoS ONE*, **2016**, 11, e0165401	3.7	32
30	Whole exome sequencing reveals de novo pathogenic variants in KAT6A as a cause of a neurodevelopmental disorder. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1791-8	2.5	35
29	Relationship between M100 Auditory Evoked Response and Auditory Radiation Microstructure in 16p11.2 Deletion and Duplication Carriers. *American Journal of Neuroradiology*, **2016**, 37, 1178-84	4.4	14
28	De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. *American Journal of Human Genetics*, **2016**, 98, 963-970	11	42
27	The Contribution of the Corpus Callosum to Language Lateralization. *Journal of Neuroscience*, **2016**, 36, 4522-33	6.6	44
26	Astroglial-Mediated Remodeling of the Interhemispheric Midline Is Required for the Formation of the Corpus Callosum. *Cell Reports*, **2016**, 17, 735-747	10.6	45
25	Reply: ARID1B mutations are the major genetic cause of corpus callosum anomalies in patients with intellectual disability. *Brain*, **2016**, 139, e65	11.2	0
24	De novo mutations in KIF1A cause progressive encephalopathy and brain atrophy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 623-35	5.3	68
23	Individuals with agenesis of the corpus callosum show sensory processing differences as measured by the sensory profile. *Neuropsychology*, **2015**, 29, 751-758	3.8	13
22	Abnormal auditory and language pathways in children with 16p11.2 deletion. *NeuroImage: Clinical*, **2015**, 9, 50-7	5.3	17
21	Corpus callosum in cognitive and sensory processing: insights into autism. *Future Neurology*, **2015**, 10, 147-160	1.5	5
20	White Matter Changes of Neurite Density and Fiber Orientation Dispersion during Human Brain Maturation. *PLoS ONE*, **2015**, 10, e0123656	3.7	89
19	A de novo mutation in PRICKLE1 in fetal agenesis of the corpus callosum and polymicrogyria. *Journal of Neurogenetics*, **2015**, 29, 174-7	1.6	11
18	Aberrant white matter microstructure in children with 16p11.2 deletions. *Journal of Neuroscience*, **2014**, 34, 6214-23	6.6	53
17	Epileptic encephalopathies: new genes and new pathways. *Neurotherapeutics*, **2014**, 11, 796-806	6.4	74
16	Opposing brain differences in 16p11.2 deletion and duplication carriers. *Journal of Neuroscience*, **2014**, 34, 11199-211	6.6	108

15	Mutations in PIEZO2 cause Gordon syndrome, Marden-Walker syndrome, and distal arthrogryposis type 5. *American Journal of Human Genetics*, **2014**, 94, 734-44	11	124
14	Clinical, genetic and imaging findings identify new causes for corpus callosum development syndromes. *Brain*, **2014**, 137, 1579-613	11.2	198
13	Mapk/Erk activation in an animal model of social deficits shows a possible link to autism. *Molecular Autism*, **2014**, 5, 57	6.5	36
12	Neurodevelopmental disorders and genetic testing: current approaches and future advances. *Annals of Neurology*, **2013**, 74, 164-70	9.4	19
11	Autism traits in individuals with agenesis of the corpus callosum. *Journal of Autism and Developmental Disorders*, **2013**, 43, 1106-18	4.6	50
10	Both rare and de novo copy number variants are prevalent in agenesis of the corpus callosum but not in cerebellar hypoplasia or polymicrogyria. *PLoS Genetics*, **2013**, 9, e1003823	6	58
9	Quantitative trait loci for interhemispheric commissure development and social behaviors in the BTBR T+ tf/J mouse model of autism. *PLoS ONE*, **2013**, 8, e61829	3.7	42
8	A 600 kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. *Journal of Medical Genetics*, **2012**, 49, 660-8	5.8	182
7	Genetic and functional analyses identify DISC1 as a novel callosal agenesis candidate gene. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1865-76	2.5	34
6	Hi Tmtc4 Interacts with C3G, Wntless, and Zfhx4: A Yeast Two-Hybrid Trap for Proteins Associated with Temtamy Syndrome. *FASEB Journal*, **2011**, 25, 963.7	0.9	
5	Agenesis of the corpus callosum in California 1983-2003: a population-based study. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2495-500	2.5	136
4	Agenesis of the corpus callosum, optic coloboma, intractable seizures, craniofacial and skeletal dysmorphisms: an autosomal recessive disorder similar to Temtamy syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1900-5	2.5	10
3	Agenesis of the corpus callosum: genetic, developmental and functional aspects of connectivity. *Nature Reviews Neuroscience*, **2007**, 8, 287-99	13.5	536
2	Anomalies of the corpus callosum: an MR analysis of the phenotypic spectrum of associated malformations. *American Journal of Roentgenology*, **2006**, 187, 1343-8	5.4	131
1	The ARX story (epilepsy, mental retardation, autism, and cerebral malformations): one gene leads to many phenotypes. *Current Opinion in Pediatrics*, **2003**, 15, 567-71	3.2	108