Alexander P A Stegmann

List of Publications by Year in Descending Order

37 papers	1,022 citations	18 h-index	31 g-index
43 ext. papers	1,550 ext. citations	8.4 avg, IF	3.17 L-index

#	Paper	IF	Citations
37	Distinguishing Marshall from Stickler syndrome: a clinical and genetic challenge. *Clinical Dysmorphology*, **2021**, 30, 58-61	0.9	1
36	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. *American Journal of Human Genetics*, **2021**, 108, 502-516	11	12
35	Blount disease and familial inheritance in Ghana, area cross-sectional study. *BMJ Paediatrics Open*, **2021**, 5, e001052	2.4	0
34	Genotype-phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 90	14.4	2
33	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. *American Journal of Human Genetics*, **2021**, 108, 346-356	11	7
32	Tibia hemimelia in a patient with CHARGE syndrome: A rare but recurrent phenomenon. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	
31	Acute encephalopathy after head trauma in a patient with a RHOBTB2 mutation. *Neurology: Genetics*, **2020**, 6, e418	3.8	5
30	Genetic analysis of spinal dysraphism with a hamartomatous growth (appendix) of the spinal cord: a case series. *BMC Neurology*, **2020**, 20, 121	3.1	0
29	De novo variants in MPP5 cause global developmental delay and behavioral changes. *Human Molecular Genetics*, **2020**, 29, 3388-3401	5.6	2
28	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
27	Germline AGO2 mutations impair RNA interference and human neurological development. *Nature Communications*, **2020**, 11, 5797	17.4	14
26	The adult phenotype of Schaaf-Yang syndrome. *Orphanet Journal of Rare Diseases*, **2020**, 15, 294	4.2	4
25	Mutations in RPSA and NKX2-3 link development of the spleen and intestinal vasculature. *Human Mutation*, **2020**, 41, 196-202	4.7	8
24	Disruptive variants of associate with autism and interfere with neuronal development and synaptic transmission. *Science Advances*, **2019**, 5, eaax2166	14.3	16
23	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. *European Journal of Human Genetics*, **2019**, 27, 738-746	5.3	11
22	PRRT2-related phenotypes in patients with a 16p11.2 deletion. *European Journal of Medical Genetics*, **2019**, 62, 265-269	2.6	12
21	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679	17.4	21

20	Diagnostic exome sequencing in 100 consecutive patients with both epilepsy and intellectual disability. *Epilepsia*, **2019**, 60, 155-164	6.4	36
19	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. *American Journal of Human Genetics*, **2019**, 104, 203-212	11	18
18	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. *Genetics in Medicine*, **2019**, 21, 1295-1307	8.1	36
17	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in Drosophila. *American Journal of Human Genetics*, **2018**, 102, 44-57	11	34
16	Haploinsufficiency of CUX1 Causes Nonsyndromic Global Developmental Delay With Possible Catch-up Development. *Annals of Neurology*, **2018**, 84, 200-207	9.4	6
15	Loss-of-function zinc finger mutation in the gene associated with erythrocytosis. *Blood*, **2018**, 132, 1455-1458	21	10
14	SLC10A7 mutations cause a skeletal dysplasia with amelogenesis imperfecta mediated by GAG biosynthesis defects. *Nature Communications*, **2018**, 9, 3087	17.4	21
13	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2018**, 102, 1195-1203	11	24
12	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. *Journal of Medical Genetics*, **2018**, 55, 104-113	5.8	35
11	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. *Nature Communications*, **2018**, 9, 4619	17.4	39
10	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. *Annals of Neurology*, **2018**, 84, 788-795	9.4	18
9	Heterozygous HNRNPU variants cause early onset epilepsy and severe intellectual disability. *Human Genetics*, **2017**, 136, 821-834	6.3	39
8	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. *American Journal of Human Genetics*, **2017**, 100, 91-104	11	43
7	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2017**, 101, 716-724	11	38
6	De novo, deleterious sequence variants that alter the transcriptional activity of the homeoprotein PBX1 are associated with intellectual disability and pleiotropic developmental defects. *Human Molecular Genetics*, **2017**, 26, 4849-4860	5.6	26
5	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. *PLoS Genetics*, **2017**, 13, e1006864	6	67
4	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. *Genetics in Medicine*, **2017**, 19, 667-675	8.1	98
3	Biallelicframeshift mutation in RIN2 in a patient with intellectual disability and cataract, without RIN2 syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 3238-3240	2.5	

2	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. *American Journal of Human Genetics*, **2016**, 99, 711-719	11	44
1	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. *Nature Neuroscience*, **2016**, 19, 1194-6	25.5	258