Gudrun A Rappold

List of Publications by Year
in descending order

98	8,098	76326 40	49909 87
papers	citations	h-index	g-index

104	104	104	10887
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Disrupted Mitochondrial Network Drives Deficits of Learning and Memory in a Mouse Model of FOXP1 Haploinsufficiency. Genes, 2022, 13, 127.	2.4	3
2	Mitochondrial dysfunction and oxidative stress contribute to cognitive and motor impairment in FOXP1 syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	12
3	Emerging evidence for gene mutations driving both brain and gut dysfunction in autism spectrum disorder. Molecular Psychiatry, 2021, 26, 1442-1444.	7.9	22
4	Expression Profiling of Rectal Biopsies Suggests Altered Enteric Neuropathological Traits in Parkinsonâ€™s Disease Patients. Journal of Parkinson's Disease, 2021, 11, 171-176.	2.8	7
5	SHANK2 mutations impair apoptosis, proliferation and neurite outgrowth during early neuronal differentiation in SH-SY5Y cells. Scientific Reports, 2021, 11, 2128.	3.3	11
6	Identification and Tissue-Specific Characterization of Novel SHOX-Regulated Genes in Zebrafish Highlights SOX Family Members Among Other Genes. Frontiers in Genetics, 2021, 12, 688808.	2.3	3
7	Imbalanced post- and extrasynaptic SHANK2A functions during development affect social behavior in SHANK2-mediated neuropsychiatric disorders. Molecular Psychiatry, 2021, 26, 6482-6504.	7.9	8
8	Parkinson mice show functional and molecular changes in the gut long before motoric disease onset. Molecular Neurodegeneration, 2021, 16, 34.	10.8	29
9	Evidence That Non-Syndromic Familial Tall Stature Has an Oligogenic Origin Including Ciliary Genes. Frontiers in Endocrinology, 2021, 12, 660731.	3.5	5
10	The alternative serotonin transporter promoter P2 impacts gene function in females with irritable bowel syndrome. Journal of Cellular and Molecular Medicine, 2021, 25, 8047-8061.	3.6	5
11	Identification of ZBTB26 as a Novel Risk Factor for Congenital Hypothyroidism. Genes, 2021, 12, 1862.	2.4	2
12	Precise Correction of Heterozygous SHOX2 Mutations in hiPSCs Derived from Patients with Atrial Fibrillation via Genome Editing and Sib Selection. Stem Cell Reports, 2020, 15, 999-1013.	4.8	6
13	Generation of two hiPSC lines from a patient with autism spectrum disorder harboring a 120Âkb deletion in SHANK2 and two control lines from each parent. Stem Cell Research, 2020, 49, 102004.	0.7	2
14	Two Cases of Recessive Intellectual Disability Caused by NDST1 and METTL23 Variants. Genes, 2020, 11, 1021.	2.4	9
15	Identification of <i>Transient Receptor Potential Channel 4-Associated Protein</i> as a Novel Candidate Gene Causing Congenital Primary Hypothyroidism. Hormone Research in Paediatrics, 2020, 93, 16-29.	1.8	7
16	Comparative expression profiling in the intestine of patients with Giardiaâ€induced postinfectious functional gastrointestinal disorders. Neurogastroenterology and Motility, 2020, 32, e13868.	3.0	5
17	Inhibition of HDAC6 activity protects dopaminergic neurons from alpha-synuclein toxicity. Scientific Reports, 2020, 10, 6064.	3.3	31
18	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	3.5	7

#	Article	IF	Citations
19	Molecular Characterization of Embryonic Stem Cell-Derived Cardiac Neural Crest-Like Cells Revealed a Spatiotemporal Expression of an Mlc-3 Isoform. International Journal of Stem Cells, 2020, 13, 65-79.	1.8	2
20	Postnatal human enteric neurospheres show a remarkable molecular complexity. Neurogastroenterology and Motility, 2019, 31, e13674.	3.0	2
21	Functional Characterization of Rare Variants in the SHOX2 Gene Identified in Sinus Node Dysfunction and Atrial Fibrillation. Frontiers in Genetics, 2019, 10, 648.	2.3	21
22	Gastrointestinal dysfunction in autism displayed by altered motility and achalasia in Foxp1 ^{+/â^'} mice. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 22237-22245.	7.1	31
23	A direct regulatory link between microRNA-137 and SHANK2: implications for neuropsychiatric disorders. Journal of Neurodevelopmental Disorders, 2018, 10, 15.	3.1	21
24	Functional missense and splicing variants in the retinoic acid catabolizing enzyme CYP26C1 in idiopathic short stature. European Journal of Human Genetics, 2018, 26, 1113-1120.	2.8	10
25	Identification of SLC20A1 and SLC15A4 among other genes as potential risk factors for combined pituitary hormone deficiency. Genetics in Medicine, 2018, 20, 728-736.	2.4	18
26	Sex Hormones Regulate SHANK Expression. Frontiers in Molecular Neuroscience, 2018, 11, 337.	2.9	28
27	Distinct Phenotypes of Shank2 Mouse Models Reflect Neuropsychiatric Spectrum Disorders of Human Patients With SHANK2 Variants. Frontiers in Molecular Neuroscience, 2018, 11, 240.	2.9	48
28	miR-16 and miR-125b are involved in barrier function dysregulation through the modulation of claudin-2 and cingulin expression in the jejunum in IBS with diarrhoea. Gut, 2017, 66, 1537.1-1538.	12.1	105
29	Foxp1 expression is essential for sex-specific murine neonatal ultrasonic vocalization. Human Molecular Genetics, 2017, 26, 1511-1521.	2.9	32
30	Investigation of SHANK3 in schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 390-398.	1.7	34
31	Comparative expression analysis of Shox2-deficient embryonic stem cell-derived sinoatrial node-like cells. Stem Cell Research, 2017, 21, 51-57.	0.7	13
32	miR-16 and miR-103 impact 5-HT4 receptor signalling and correlate with symptom profile in irritable bowel syndrome. Scientific Reports, 2017, 7, 14680.	3.3	46
33	Murine transgenic embryonic stem cell lines for the investigation of sinoatrial node-related molecular pathways. Stem Cell Research, 2017, 25, 278-282.	0.7	3
34	A novel homozygous ARL13B variant in patients with Joubert syndrome impairs its guanine nucleotide-exchange factor activity. European Journal of Human Genetics, 2017, 25, 1324-1334.	2.8	9
35	The Human Serotonin Type 3 Receptor Gene (HTR3A-E) Allelic Variant Database. Human Mutation, 2017, 38, 137-147.	2.5	14
36	Coding and non-coding variants in the SHOX2 gene in patients with early-onset atrial fibrillation. Basic Research in Cardiology, 2016, 111, 36.	5.9	45

#	Article	IF	Citations
37	Retinoic acid catabolizing enzyme CYP 26C1 is a genetic modifier in SHOX deficiency. EMBO Molecular Medicine, 2016, 8, 1455-1469.	6.9	23
38	A Track Record on SHOX: From Basic Research to Complex Models and Therapy. Endocrine Reviews, 2016, 37, 417-448.	20.1	87
39	Identification and functional characterization ofde novo FOXP1variants provides novel insights into the etiology of neurodevelopmental disorder. Human Molecular Genetics, 2016, 25, 546-557.	2.9	69
40	Homozygous missense mutation in theLMAN2Lgene segregates with intellectual disability in a large consanguineous Pakistani family. Journal of Medical Genetics, 2016, 53, 138-144.	3.2	16
41	Exploring the genetics of irritable bowel syndrome: a GWA study in the general population and replication in multinational case-control cohorts. Gut, 2015, 64, 1774-1782.	12.1	97
42	Foxp1 Regulates Cortical Radial Migration and Neuronal Morphogenesis in Developing Cerebral Cortex. PLoS ONE, 2015, 10, e0127671.	2.5	52
43	Identification of Novel SHOX Target Genes in the Developing Limb Using a Transgenic Mouse Model. PLoS ONE, 2014, 9, e98543.	2.5	14
44	SHOX triggers the lysosomal pathway of apoptosis via oxidative stress. Human Molecular Genetics, 2014, 23, 1619-1630.	2.9	30
45	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501
46	Tbx4 interacts with the short stature homeobox gene Shox2 in limb development. Developmental Dynamics, 2014, 243, 629-639.	1.8	14
47	Genome-wide UPD screening in patients with intellectual disability. European Journal of Human Genetics, 2014, 22, 1233-1235.	2.8	3
48	Islet1 is a direct transcriptional target of the homeodomain transcription factor Shox2 and rescues the Shox2-mediated bradycardia. Basic Research in Cardiology, 2013, 108, 339.	5.9	69
49	The cellular function of srGAP3 and its role in neuronal morphogenesis. Mechanisms of Development, 2013, 130, 391-395.	1.7	37
50	Height mattersâ€"from monogenic disorders to normal variation. Nature Reviews Endocrinology, 2013, 9, 171-177.	9.6	46
51	Srgap3 ^{â€"/â€"} mice present a neurodevelopmental disorder with schizophreniaâ€related intermediate phenotypes. FASEB Journal, 2012, 26, 4418-4428.	0.5	51
52	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	13.7	940
53	The distinct and overlapping phenotypic spectra of FOXP1 and FOXP2 in cognitive disorders. Human Genetics, 2012, 131, 1687-1698.	3.8	115
54	Short Stature Homeobox-Containing (SHOX) Gene Deficiency: Genetics and Growth Response to Growth Hormone Treatment in Comparison with Turner Syndrome. , 2012, , 2299-2318.		1

#	Article	IF	Citations
55	The Homeobox Transcription Factor HOXA9 Is a Regulator of SHOX in U2OS Cells and Chicken Micromass Cultures. PLoS ONE, 2012, 7, e45369.	2.5	4
56	Haploinsufficiency of ARID1B, a Member of the SWI/SNF-A Chromatin-Remodeling Complex, Is a Frequent Cause of Intellectual Disability. American Journal of Human Genetics, 2012, 90, 565-572.	6.2	225
57	The HTR3A Polymorphism c. -42C>T Is Associated With Amygdala Responsiveness in Patients With Irritable Bowel Syndrome. Gastroenterology, 2011, 140, 1943-1951.	1.3	73
58	Correlation of SHOX2 Gene Amplification and DNA Methylation in Lung Cancer Tumors. BMC Cancer, 2011, 11, 102.	2.6	55
59	The Jumping SHOX Geneâ€"Crossover in the Pseudoautosomal Region Resulting in Unusual Inheritance of Leri-Weill Dyschondrosteosis. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E356-E359.	3.6	14
60	SrGAP3 interacts with lamellipodin at the cell membrane and regulates Rac-dependent cellular protrusions. Journal of Cell Science, 2011, 124, 3941-3955.	2.0	42
61	FGFR3 is a target of the homeobox transcription factor SHOX in limb development. Human Molecular Genetics, 2011, 20, 1524-1535.	2.9	41
62	Alternative Splicing and Nonsense-Mediated RNA Decay Contribute to the Regulation of SHOX Expression. PLoS ONE, 2011, 6, e18115.	2.5	36
63	Evidence for a Role of srGAP3 in the Positioning of Commissural Axons within the Ventrolateral Funiculus of the Mouse Spinal Cord. PLoS ONE, 2011, 6, e19887.	2.5	13
64	Identification of FOXP1 deletions in three unrelated patients with mental retardation and significant speech and language deficits. Human Mutation, 2010, 31, E1851-E1860.	2.5	130
65	Enhancer elements upstream of the SHOX gene are active in the developing limb. European Journal of Human Genetics, 2010, 18, 527-532.	2.8	43
66	Mutations in the SHANK2 synaptic scaffolding gene in autism spectrum disorder and mental retardation. Nature Genetics, 2010, 42, 489-491.	21.4	491
67	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	21.4	431
68	Shox2 mediates Tbx5 activity by regulating Bmp4 in the pacemaker region of the developing heart. Human Molecular Genetics, 2010, 19, 4625-4633.	2.9	106
69	RIC-3 Exclusively Enhances the Surface Expression of Human Homomeric 5-Hydroxytryptamine Type 3A (5-HT3A) Receptors Despite Direct Interactions with 5-HT3A, -C, -D, and -E Subunits. Journal of Biological Chemistry, 2010, 285, 26956-26965.	3.4	31
70	Dynamic expression of the Slitâ€Robo GTPase activating protein genes during development of the murine nervous system. Journal of Comparative Neurology, 2009, 513, 224-236.	1.6	54
71	First evidence for an association of a functional variant in the microRNA-510 target site of the serotonin receptor-type 3E gene with diarrhea predominant irritable bowel syndrome. Human Molecular Genetics, 2008, 17, 2967-2977.	2.9	173
72	Growth Hormone Is Effective in Treatment of Short Stature Associated with Short Stature Homeobox-Containing Gene Deficiency: Two-Year Results of a Randomized, Controlled, Multicenter Trial. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 219-228.	3.6	107

#	Article	IF	Citations
73	SHOXat a glance: from gene to protein. Archives of Physiology and Biochemistry, 2007, 113, 116-123.	2.1	47
74	Targeted Mutation Reveals Essential Functions of the Homeodomain Transcription Factor Shox2 in Sinoatrial and Pacemaking Development. Circulation, 2007, 115, 1830-1838.	1.6	222
75	Genotypes and phenotypes in children with short stature: clinical indicators of SHOX haploinsufficiency. Journal of Medical Genetics, 2007, 44, 306-313.	3.2	206
76	BNP is a transcriptional target of the short stature homeobox gene SHOX. Human Molecular Genetics, 2007, 16, 3081-3087.	2.9	57
77	The novel humanSHOX allelic variant database. Human Mutation, 2007, 28, 933-938.	2.5	18
78	MEGAP impedes cell migration via regulating actin and microtubule dynamics and focal complex formation. Experimental Cell Research, 2006, 312, 2379-2393.	2.6	51
79	The pseudoautosomal regions, SHOX and disease. Current Opinion in Genetics and Development, 2006, 16, 233-239.	3.3	123
80	Expression of the short stature homeobox gene Shox is restricted by proximal and distal signals in chick limb buds and affects the length of skeletal elements. Developmental Biology, 2006, 298, 585-596.	2.0	44
81	Alteration of DNA binding, dimerization, and nuclear translocation of SHOX homeodomain mutations identified in idiopathic short stature and Leri-Weill dyschondrosteosis. Human Mutation, 2005, 26, 44-52.	2.5	38
82	A Novel Class of Pseudoautosomal Region 1 Deletions Downstream of SHOX Is Associated with LÃ©ri-Weill Dyschondrosteosis. American Journal of Human Genetics, 2005, 77, 533-544.	6.2	125
83	The Short Stature Homeodomain Protein SHOX Induces Cellular Growth Arrest and Apoptosis and Is Expressed in Human Growth Plate Chondrocytes. Journal of Biological Chemistry, 2004, 279, 37103-37114.	3.4	94
84	The novel Rho-GTPase activating gene MEGAP/ srGAP3 has a putative role in severe mental retardation. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 11754-11759.	7.1	202
85	Deletions of the Homeobox GeneSHOX(Short Stature Homeobox) Are an Important Cause of Growth Failure in Children with Short Stature. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 1402-1406.	3.6	188
86	The humanSHOX mutation database. Human Mutation, 2002, 20, 338-341.	2.5	20
87	Growth Hormone and Gonadotropin-Releasing Hormone Analog Therapy in Haploinsufficiency of SHOX.. Endocrine Journal, 2001, 48, 317-322.	1.6	33
88	Cytogenetic and molecular characterization of two isodicentric Y chromosomes. American Journal of Medical Genetics Part A, 2001, 101, 20-25.	2.4	30
89	Phenotypic findings due to trisomy 7p15.3-pter including theTWIST locus. American Journal of Medical Genetics Part A, 2001, 103, 56-62.	2.4	29
90	SHOX in Short Stature Syndromes. Hormone Research in Paediatrics, 2001, 55, 21-23.	1.8	16

#	Article	IF	Citations
91	Phenotypic variation and genetic heterogeneity in LÃ©ri-Weill syndrome. European Journal of Human Genetics, 2000, 8, 54-62.	2.8	138
92	Evidence for Heterogeneity in Recombination in the Human Pseudoautosomal Region: High Resolution Analysis by Sperm Typing and Radiation-Hybrid Mapping. American Journal of Human Genetics, 2000, 66, 557-566.	6.2	108
93	Lï¿½ri-Weill syndrome as part of a contiguous gene syndrome at Xp22.3. , 1999, 83, 367-371.		45
94	Molecular identification of the corticosterone-sensitive extraneuronal catecholamine transporter. Nature Neuroscience, 1998, 1, 349-351.	14.8	359
95	A novel pseudoautosomal gene encoding a putative GTP-binding protein resides in the vicinity of the Xp/Yp telomere. Human Molecular Genetics, 1998, 7, 407-414.	2.9	37
96	Pseudoautosomal deletions encompassing a novel homeobox gene cause growth failure in idiopathic short stature and Turner syndrome. Nature Genetics, 1997, 16, 54-63.	21.4	867
97	High-resolution fluorescence in situ hybridization of human Y-linked genes on released chromatin. Chromosome Research, 1997, 5, 23-30.	2.2	24
98	A human pseudoautosomal gene, ADP/ATP translocase, escapes Xâ€“inactivation whereas a homologue on Xq is subject to Xâ€“inactivation. Nature Genetics, 1993, 3, 82-87.	21.4	82