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	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
2	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
3	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501
4	Mutations in the SHANK2 synaptic scaffolding gene in autism spectrum disorder and mental retardation. Nature Genetics, 2010, 42, 489-491.	21.4	491
5	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	21.4	431
6	Molecular identification of the corticosterone-sensitive extraneuronal catecholamine transporter. Nature Neuroscience, 1998, 1, 349-351.	14.8	359
7	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
8	Targeted Mutation Reveals Essential Functions of the Homeodomain Transcription Factor Shox2 in Sinoatrial and Pacemaking Development. Circulation, 2007, 115, 1830-1838.	1.6	222
9	Genotypes and phenotypes in children with short stature: clinical indicators of SHOX haploinsufficiency. Journal of Medical Genetics, 2007, 44, 306-313.	3.2	206
10	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
11	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
12	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
13	Phenotypic variation and genetic heterogeneity in LÃ©ri-Weill syndrome. European Journal of Human Genetics, 2000, 8, 54-62.	2.8	138
14	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
15	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
16	The pseudoautosomal regions, SHOX and disease. Current Opinion in Genetics and Development, 2006, 16, 233-239.	3.3	123
17	The distinct and overlapping phenotypic spectra of FOXP1 and FOXP2 in cognitive disorders. Human Genetics, 2012, 131, 1687-1698.	3.8	115
18	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

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19	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
20	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
21	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
22	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
23	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
24	A Track Record on SHOX: From Basic Research to Complex Models and Therapy. Endocrine Reviews, 2016, 37, 417-448.	20.1	87
25	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
26	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
27	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
28	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
29	BNP is a transcriptional target of the short stature homeobox gene SHOX. Human Molecular Genetics, 2007, 16, 3081-3087.	2.9	57
30	Correlation of SHOX2 Gene Amplification and DNA Methylation in Lung Cancer Tumors. BMC Cancer, 2011, 11, 102.	2.6	55
31	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
32	Foxp1 Regulates Cortical Radial Migration and Neuronal Morphogenesis in Developing Cerebral Cortex. PLoS ONE, 2015, 10, e0127671.	2.5	52
33	MEGAP impedes cell migration via regulating actin and microtubule dynamics and focal complex formation. Experimental Cell Research, 2006, 312, 2379-2393.	2.6	51
34	Srgap3 ^{â€“/â€“} mice present a neurodevelopmental disorder with schizophreniaâ€related intermediate phenotypes. FASEB Journal, 2012, 26, 4418-4428.	0.5	51
35	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
36	SHOXat a glance: from gene to protein. Archives of Physiology and Biochemistry, 2007, 113, 116-123.	2.1	47

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37	Height mattersâ€"from monogenic disorders to normal variation. Nature Reviews Endocrinology, 2013, 9, 171-177.	9.6	46
38	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
39	Lï¿½ri-Weill syndrome as part of a contiguous gene syndrome at Xp22.3. , 1999, 83, 367-371.		45
40	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
41	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
42	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
43	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
44	FGFR3 is a target of the homeobox transcription factor SHOX in limb development. Human Molecular Genetics, 2011, 20, 1524-1535.	2.9	41
45	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
46	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
47	The cellular function of srGAP3 and its role in neuronal morphogenesis. Mechanisms of Development, 2013, 130, 391-395.	1.7	37
48	Alternative Splicing and Nonsense-Mediated RNA Decay Contribute to the Regulation of SHOX Expression. PLoS ONE, 2011, 6, e18115.	2.5	36
49	Investigation of SHANK3 in schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 390-398.	1.7	34
50	Growth Hormone and Gonadotropin-Releasing Hormone Analog Therapy in Haploinsufficiency of SHOX.. Endocrine Journal, 2001, 48, 317-322.	1.6	33
51	Foxp1 expression is essential for sex-specific murine neonatal ultrasonic vocalization. Human Molecular Genetics, 2017, 26, 1511-1521.	2.9	32
52	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
53	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
54	Inhibition of HDAC6 activity protects dopaminergic neurons from alpha-synuclein toxicity. Scientific Reports, 2020, 10, 6064.	3.3	31

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55	Cytogenetic and molecular characterization of two isodicentric Y chromosomes. American Journal of Medical Genetics Part A, 2001, 101, 20-25.	2.4	30
56	SHOX triggers the lysosomal pathway of apoptosis via oxidative stress. Human Molecular Genetics, 2014, 23, 1619-1630.	2.9	30
57	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
58	Parkinson mice show functional and molecular changes in the gut long before motoric disease onset. Molecular Neurodegeneration, 2021, 16, 34.	10.8	29
59	Sex Hormones Regulate SHANK Expression. Frontiers in Molecular Neuroscience, 2018, 11, 337.	2.9	28
60	High-resolution fluorescence in situ hybridization of human Y-linked genes on released chromatin. Chromosome Research, 1997, 5, 23-30.	2.2	24
61	Retinoic acid catabolizing enzyme CYP 26C1 is a genetic modifier in SHOX deficiency. EMBO Molecular Medicine, 2016, 8, 1455-1469.	6.9	23
62	Emerging evidence for gene mutations driving both brain and gut dysfunction in autism spectrum disorder. Molecular Psychiatry, 2021, 26, 1442-1444.	7.9	22
63	A direct regulatory link between microRNA-137 and SHANK2: implications for neuropsychiatric disorders. Journal of Neurodevelopmental Disorders, 2018, 10, 15.	3.1	21
64	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
65	The humanSHOX mutation database. Human Mutation, 2002, 20, 338-341.	2.5	20
66	The novel humanSHOX allelic variant database. Human Mutation, 2007, 28, 933-938.	2.5	18
67	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
68	SHOX in Short Stature Syndromes. Hormone Research in Paediatrics, 2001, 55, 21-23.	1.8	16
69	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
70	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
71	Identification of Novel SHOX Target Genes in the Developing Limb Using a Transgenic Mouse Model. PLoS ONE, 2014, 9, e98543.	2.5	14
72	Tbx4 interacts with the short stature homeobox gene Shox2 in limb development. Developmental Dynamics, 2014, 243, 629-639.	1.8	14

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73	The Human Serotonin Type 3 Receptor Gene (HTR3A-E) Allelic Variant Database. Human Mutation, 2017, 38, 137-147.	2.5	14
74	Comparative expression analysis of Shox2-deficient embryonic stem cell-derived sinoatrial node-like cells. Stem Cell Research, 2017, 21, 51-57.	0.7	13
75	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
76	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
77	SHANK2 mutations impair apoptosis, proliferation and neurite outgrowth during early neuronal differentiation in SH-SY5Y cells. Scientific Reports, 2021, 11, 2128.	3.3	11
78	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
79	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
80	Two Cases of Recessive Intellectual Disability Caused by NDST1 and METTL23 Variants. Genes, 2020, 11, 1021.	2.4	9
81	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
82	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
83	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
84	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	3.5	7
85	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
86	Comparative expression profiling in the intestine of patients with Giardiaâ€induced postinfectious functional gastrointestinal disorders. Neurogastroenterology and Motility, 2020, 32, e13868.	3.0	5
87	Evidence That Non-Syndromic Familial Tall Stature Has an Oligogenic Origin Including Ciliary Genes. Frontiers in Endocrinology, 2021, 12, 660731.	3.5	5
88	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
89	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
90	Genome-wide UPD screening in patients with intellectual disability. European Journal of Human Genetics, 2014, 22, 1233-1235.	2.8	3

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91	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
92	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
93	Disrupted Mitochondrial Network Drives Deficits of Learning and Memory in a Mouse Model of FOXP1 Haploinsufficiency. Genes, 2022, 13, 127.	2.4	3
94	Postnatal human enteric neurospheres show a remarkable molecular complexity. Neurogastroenterology and Motility, 2019, 31, e13674.	3.0	2
95	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
96	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
97	Identification of ZBTB26 as a Novel Risk Factor for Congenital Hypothyroidism. Genes, 2021, 12, 1862.	2.4	2
98	Short Stature Homeobox-Containing (SHOX) Gene Deficiency: Genetics and Growth Response to Growth Hormone Treatment in Comparison with Turner Syndrome. , 2012, , 2299-2318.		1