Martin Zenker

List of Publications by Year
in descending order

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108	6,379	71102	74163
papers	citations	41 h-index	75 g-index

116	116	116	10105
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Profiling of the Bacterial Microbiota along the Murine Alimentary Tract. International Journal of Molecular Sciences, 2022, 23, 1783.	4.1	6
2	The seventh international <scp>RASopathies</scp> symposium: Pathways to a cureâ€"expanding knowledge, enhancing research, and therapeutic discovery. American Journal of Medical Genetics, Part A, 2022, 188, 1915-1927.	1.2	10
3	Noonan syndrome: improving recognition and diagnosis. Archives of Disease in Childhood, 2022, 107, 1073-1078.	1.9	28
4	Outcomes in growth hormone-treated Noonan syndrome children: impact of PTPN11 mutation status. Endocrine Connections, 2022, 11, .	1.9	2
5	Correlation of PET-MRI, pathology, LOH and surgical success in a case of CHI with atypical large pancreatic focus. Journal of the Endocrine Society, 2022, 6, bvac056.	0.2	2
6	Cutis marmorata telangiectatica congenita being caused by postzygotic GNA11 mutations. European Journal of Medical Genetics, 2022, 65, 104472.	1.3	6
7	Oral HRAS Mutation in Orofacial Nevus Sebaceous Syndrome (Schimmelpenning-Feuerstein-Mims-Syndrome): A Case Report With a Literature Survey. In Vivo, 2022, 36, 274-293.	1.3	3
8	Neurologic and neurodevelopmental complications in cardiofaciocutaneous syndrome are associated with genotype: A multinational cohort study. Genetics in Medicine, 2022, 24, 1556-1566.	2.4	15
9	Gut microbial similarity in twins is driven by shared environment and aging. EBioMedicine, 2022, 79, 104011.	6.1	7
10	WARS1 and SARS1 : Two tRNA synthetases implicated in autosomal recessive microcephaly. Human Mutation, 2022, 43, 1454-1471.	2.5	5
11	Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. European Journal of Human Genetics, 2021, 29, 51-60.	2.8	17
12	Aplasia cutis congenita in a CDC42 â€•related developmental phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 850-855.	1.2	3
13	The clinical significance of A2ML1 variants in Noonan syndrome has to be reconsidered. European Journal of Human Genetics, 2021, 29, 524-527.	2.8	7
14	KRASMutation in an Implant-associated Peripheral Giant Cell Granuloma of the Jaw: Implications of Genetic Analysis of the Lesion for Treatment Concept and Surveillance. In Vivo, 2021, 35, 947-953.	1.3	1
15	Neurofibromatosis Type 1 With Cherubism-like Phenotype, Multiple Osteolytic Bone Lesions of Lower Extremities, and Alagille-syndrome: Case Report With Literature Survey. In Vivo, 2021, 35, 1711-1736.	1.3	3
16	Mosaic <scp>RASopathy</scp> due to <scp>KRAS</scp> variant <scp>G12D</scp> with segmental overgrowth and associated peripheral vascular malformations. American Journal of Medical Genetics, Part A, 2021, 185, 3122-3128.	1.2	11
17	Expanding the clinical phenotype of <scp>RASopathies</scp> in 38 Turkish patients, including the rare <scp>LZTR1</scp>, <scp>RAF1</scp>, <scp>RIT1</scp> variants, and large deletion in <scp>NF1</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3623-3633.	1.2	4
18	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	6.2	23

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19	Activating MRAS mutations cause Noonan syndrome associated with hypertrophic cardiomyopathy. Human Molecular Genetics, 2020, 29, 1772-1783.	2.9	30
20	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
21	MRI Spectrum of Brain Involvement in Sphingosine-1-Phosphate Lyase Insufficiency Syndrome. American Journal of Neuroradiology, 2020, 41, 1943-1948.	2.4	19
22	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	12.8	47
23	Expanding the genotypic and phenotypic spectrum of severe serine biosynthesis disorders. Human Mutation, 2020, 41, 1615-1628.	2.5	14
24	Characteristic dental pattern with hypodontia and short roots in Fraser syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1681-1689.	1.2	7
25	Mosaic Neurofibromatosis Type 1 With Multiple Cutaneous Diffuse and Plexiform Neurofibromas of the Lower Leg. Anticancer Research, 2020, 40, 3423-3427.	1.1	3
26	Genetic testing in inherited endocrine disorders: joint position paper of the European reference network on rare endocrine conditions (Endo-ERN). Orphanet Journal of Rare Diseases, 2020, 15, 144.	2.7	15
27	Long-term Culture of EBV-induced Human Lymphoblastoid Cell Lines Reveals Chromosomal Instability. Journal of Histochemistry and Cytochemistry, 2020, 68, 239-251.	2.5	9
28	Two unrelated families with variable expression of Fraser syndrome due to the same pathogenic variant in the <scp>FRAS1</scp> gene. American Journal of Medical Genetics, Part A, 2020, 182, 773-779.	1.2	5
29	Responsiveness of sphingosine phosphate lyase insufficiency syndrome to vitamin <scp>B6</scp> cofactor supplementation. Journal of Inherited Metabolic Disease, 2020, 43, 1131-1142.	3.6	21
30	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
31	Genetic basis of hypertrophic cardiomyopathy in children. Clinical Research in Cardiology, 2019, 108, 282-289.	3.3	22
32	Adamsâ€“Oliver syndrome caused by mutations of the EOGT gene. American Journal of Medical Genetics, Part A, 2019, 179, 2246-2251.	1.2	17
33	ADA2 deficiency in a patient with Noonan syndromeâ€like disorder with loose anagen hair: The coâ€occurrence of two rare syndromes. American Journal of Medical Genetics, Part A, 2019, 179, 2474-2480.	1.2	5
34	The mosaic hedgehog spectrum: another lesson on the polymorphy of mosaicism. British Journal of Dermatology, 2019, 182, 22-23.	1.5	0
35	Activating Mutations of RRAS2 Are a Rare Cause of Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1223-1232.	6.2	43
36	Hypertrophic Cardiomyopathy in Noonan Syndrome TreatedÂbyÂMEK-Inhibition. Journal of the American College of Cardiology, 2019, 73, 2237-2239.	2.8	96

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37	Expansion of the phenotypic spectrum and description of molecular findings in a cohort of patients with oculocutaneous mosaic RASopathies. Molecular Genetics & Genomic Medicine, 2019, 7, e625.	1.2	39
38	A 2q24.2 microdeletion containing TANK as novel candidate gene for intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 832-836.	1.2	3
39	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. European Journal of Human Genetics, 2019, 27, 1061-1071.	2.8	11
40	Long-term outcomes of childhood onset Noonan compared to sarcomere hypertrophic cardiomyopathy. Cardiovascular Diagnosis and Therapy, 2019, 9, S299-S309.	1.7	16
41	Variants in nuclear factor I genes influence growth and development. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 611-626.	1.6	32
42	Cubilin Single Nucleotide Polymorphism Variants are Associated with Macroangiopathy While a Matrix Metalloproteinase-9 Single Nucleotide Polymorphism Flip-Flop may Indicate Susceptibility of Diabetic Nephropathy in Type-2 Diabetic Patients. Nephron, 2019, 141, 156-165.	1.8	10
43	Identifying facial phenotypes of genetic disorders using deep learning. Nature Medicine, 2019, 25, 60-64.	30.7	449
44	Foodâ€Derived Xenoâ€microRNAs: Influence of Diet and Detectability in Gastrointestinal Tractâ€”Proofâ€ofâ€Principle Study. Molecular Nutrition and Food Research, 2019, 63, e1800076.	3.3	40
45	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	1.8	3
46	Refinement of the critical genomic region for congenital hyperinsulinismÂin the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	1.8	5
47	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
48	ClinGenâ€™s RASopathy Expert Panel consensus methods for variant interpretation. Genetics in Medicine, 2018, 20, 1334-1345.	2.4	126
49	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	6.2	138
50	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	2.4	101
51	bFGF-mediated pluripotency maintenance in human induced pluripotent stem cells is associated with NRAS-MAPK signaling. Cell Communication and Signaling, 2018, 16, 96.	6.5	38
52	Assessing the geneâ€“disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. Human Mutation, 2018, 39, 1485-1493.	2.5	66
53	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	6.2	40
54	Unrecognized juvenile nephropathic cystinosis. Kidney International, 2018, 94, 1027.	5.2	2

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55	A Postzygotic SMO Mutation Caused the Original Case of Happleâ€“Tinschert Syndrome. Acta Dermato-Venereologica, 2018, 98, 534-535.	1.3	11
56	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	2.5	42
57	Generation of functional cardiomyocytes from rat embryonic and induced pluripotent stem cells using feeder-free expansion and differentiation in suspension culture. PLoS ONE, 2018, 13, e0192652.	2.5	5
58	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31
59	Human pluripotent stem cell-derived acinar/ductal organoids generate human pancreas upon orthotopic transplantation and allow disease modelling. Gut, 2017, 66, 473-486.	12.1	174
60	Childhood cancer predisposition syndromesâ€”A concise review and recommendations by the Cancer Predisposition Working Group of the Society for Pediatric Oncology and Hematology. American Journal of Medical Genetics, Part A, 2017, 173, 1017-1037.	1.2	200
61	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	2.8	36
62	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. Scientific Reports, 2017, 7, 12225.	3.3	53
63	CTLA-4-mediated posttranslational modifications direct cytotoxic T-lymphocyte differentiation. Cell Death and Differentiation, 2017, 24, 1739-1749.	11.2	36
64	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. Journal of Clinical Investigation, 2017, 127, 912-928.	8.2	160
65	In Vitro Modeling of Congenital Hypertrophic Cardiomyopathy using Induced Pluripotent Stem Cell-Derived Cardiomyocytes. Thoracic and Cardiovascular Surgeon, 2017, 65, S1-S110.	1.0	1
66	Specific mosaic <scp>KRAS</scp> mutations affecting codon 146 cause oculoectodermal syndrome and encephalocraniocutaneous lipomatosis. Clinical Genetics, 2016, 90, 334-342.	2.0	55
67	Genotype and phenotype in patients with Noonan syndrome and a RIT1 mutation. Genetics in Medicine, 2016, 18, 1226-1234.	2.4	77
68	Dealing with the incidental finding of secondary variants by the example of SRNS patients undergoing targeted next-generation sequencing. Pediatric Nephrology, 2016, 31, 73-81.	1.7	19
69	ABO blood type B and fucosyltransferase 2 non-secretor status as genetic risk factors for chronic pancreatitis. Gut, 2016, 65, 353-354.	12.1	13
70	Surgery in Focal Congenital Hyperinsulinism (CHI) - The "Hyperinsulinism Germany International" Experience in 30 Children. Pediatric Endocrinology Reviews, 2016, 14, 129-137.	1.2	20
71	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	2.5	67
72	Rapidly progressive hypertrophic cardiomyopathy in an infant with Noonan syndrome with multiple lentigines: Palliative treatment with a rapamycin analog. American Journal of Medical Genetics, Part A, 2015, 167, 744-751.	1.2	53

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73	WDR73Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Human Mutation, 2015, 36, 1021-1028.	2.5	42
74	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adamsâ€“Oliver Syndrome With Variable Cardiac Anomalies. Circulation: Cardiovascular Genetics, 2015, 8, 572-581.	5.1	84
75	Synaptic activity controls localization and function of Ct <scp>BP</scp> 1 via binding to <scp>B</scp> assoon and <scp>P</scp> iccolo. EMBO Journal, 2015, 34, 1056-1077.	7.8	51
76	Fucosyltransferase 2 (FUT2) non-secretor status and blood group B are associated with elevated serum lipase activity in asymptomatic subjects, and an increased risk for chronic pancreatitis: a genetic association study. Gut, 2015, 64, 646-656.	12.1	82
77	Cancer spectrum and frequency among children with Noonan, Costello, and cardio-facio-cutaneous syndromes. British Journal of Cancer, 2015, 112, 1392-1397.	6.4	167
78	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	6.2	73
79	A Single-Gene Cause in 29.5% of Cases of Steroid-Resistant Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2015, 26, 1279-1289.	6.1	499
80	Germline PTPN11 and somatic PIK3CA variant in a boy with megalencephaly-capillary malformation syndrome (MCAP) - pure coincidence?. European Journal of Human Genetics, 2015, 23, 409-412.	2.8	17
81	Genetic variation of the RASGRF1 regulatory region affects human hippocampus-dependent memory. Frontiers in Human Neuroscience, 2014, 8, 260.	2.0	22
82	Valenced action/inhibition learning in humans is modulated by a genetic variant linked to dopamine D2 receptor expression. Frontiers in Systems Neuroscience, 2014, 8, 140.	2.5	22
83	Deletions in the 3â€² Part of theNFIXGene Including a Recurrent Alu-Mediated Deletion of Exon 6 and 7 Account for Previously Unexplained Cases of Marshall-Smith Syndrome. Human Mutation, 2014, 35, 1092-1100.	2.5	26
84	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. American Journal of Human Genetics, 2014, 95, 285-293.	6.2	110
85	Juvenile myelomonocytic leukaemia and Noonan syndrome. Journal of Medical Genetics, 2014, 51, 689-697.	3.2	112
86	Clinical and molecular analysis of RASopathies in a group of Turkish patients. Clinical Genetics, 2013, 83, 181-186.	2.0	24
87	Ablepharon macrostomia syndrome: A distinct genetic entity clinically related to the group of FRASâ€“FREM complex disorders. American Journal of Medical Genetics, Part A, 2013, 161, 3012-3017.	1.2	7
88	Mutations inGRIP1cause Fraser syndrome. Journal of Medical Genetics, 2012, 49, 303-306.	3.2	79
89	Noonan syndrome and clinically related disorders. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 161-179.	4.7	303
90	Manitoba-oculo-tricho-anal (MOTA) syndrome is caused by mutations in FREM1. Journal of Medical Genetics, 2011, 48, 375-382.	3.2	60

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91	Clinical manifestations of mutations in RAS and related intracellular signal transduction factors. Current Opinion in Pediatrics, 2011, 23, 443-451.	2.0	81
92	Gain-of-Function Mutations of ARHGAP31, a Cdc42/Rac1 GTPase Regulator, Cause Syndromic Cutis Aplasia and Limb Anomalies. American Journal of Human Genetics, 2011, 88, 574-585.	6.2	100
93	Cardioâ€facioâ€cutaneous syndrome: Does genotype predict phenotype?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 129-135.	1.6	72
94	Phenotype and natural history in Marshallâ€“Smith syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2714-2726.	1.2	44
95	The spectra of clinical phenotypes in aplasia cutis congenita and terminal transverse limb defects. American Journal of Medical Genetics, Part A, 2009, 149A, 1860-1881.	1.2	92
96	Genetics of nephrotic syndrome: new insights into molecules acting at the glomerular filtration barrier. Journal of Molecular Medicine, 2009, 87, 849-857.	3.9	52
97	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. Nature Genetics, 2009, 41, 1022-1026.	21.4	358
98	Genetic and Pathogenetic Aspects of Noonan Syndrome and Related Disorders. Hormone Research, 2009, 72, 57-63.	1.8	51
99	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. Journal of Medical Genetics, 2007, 44, 651-656.	3.2	114
100	BRAF Mutations in Juvenile Myelomonocytic Leukemia.. Blood, 2007, 110, 4602-4602.	1.4	0
101	Genetic Basis and Pancreatic Biology of Johanson-Blizzard Syndrome. Endocrinology and Metabolism Clinics of North America, 2006, 35, 243-253.	3.2	44
102	Genotypeâ€“epigenotypeâ€“phenotype correlations in females with frontometaphyseal dysplasia. American Journal of Medical Genetics, Part A, 2006, 140A, 1069-1073.	1.2	14
103	A variable combination of features of Noonan syndrome and neurofibromatosis type I are caused by mutations in the NF1 gene. American Journal of Medical Genetics, Part A, 2006, 140A, 2749-2756.	1.2	46
104	Expansion of the genotypic and phenotypic spectrum in patients with KRAS germline mutations. Journal of Medical Genetics, 2006, 44, 131-135.	3.2	170
105	Deficiency of UBR1, a ubiquitin ligase of the N-end rule pathway, causes pancreatic dysfunction, malformations and mental retardation (Johanson-Blizzard syndrome). Nature Genetics, 2005, 37, 1345-1350.	21.4	252
106	Novel Germ Line Mutations in the KRAS2 Gene Cause Noonan Syndrome and Deregulate Hematopoietic Cell Growth.. Blood, 2005, 106, 1602-1602.	1.4	0
107	A Dual Phenotype of Periventricular Nodular Heterotopia and Frontometaphyseal Dysplasia in One Patient Caused by a Single FLNA Mutation Leading to Two Functionally Different Aberrant Transcripts. American Journal of Human Genetics, 2004, 74, 731-737.	6.2	55
108	Congenital and Inherited Anomalies. , 0, , 58-68.		1