

# Martin Zenker

## List of Publications by Year in descending order

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108  
papers

6,379  
citations

71102

41  
h-index

74163

75  
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116  
all docs

116  
docs citations

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times ranked

10105  
citing authors

#	ARTICLE	IF	CITATIONS
1	A Single-Gene Cause in 29.5% of Cases of Steroid-Resistant Nephrotic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 1279-1289.	6.1	499
2	Identifying facial phenotypes of genetic disorders using deep learning. <i>Nature Medicine</i> , 2019, 25, 60-64.	30.7	449
3	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. <i>Nature Genetics</i> , 2009, 41, 1022-1026.	21.4	358
4	Noonan syndrome and clinically related disorders. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2011, 25, 161-179.	4.7	303
5	Deficiency of UBR1, a ubiquitin ligase of the N-end rule pathway, causes pancreatic dysfunction, malformations and mental retardation (Johanson-Blizzard syndrome). <i>Nature Genetics</i> , 2005, 37, 1345-1350.	21.4	252
6	Childhood cancer predisposition syndromes – A concise review and recommendations by the Cancer Predisposition Working Group of the Society for Pediatric Oncology and Hematology. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1017-1037.	1.2	200
7	Human pluripotent stem cell-derived acinar/ductal organoids generate human pancreas upon orthotopic transplantation and allow disease modelling. <i>Gut</i> , 2017, 66, 473-486.	12.1	174
8	Expansion of the genotypic and phenotypic spectrum in patients with KRAS germline mutations. <i>Journal of Medical Genetics</i> , 2006, 44, 131-135.	3.2	170
9	Cancer spectrum and frequency among children with Noonan, Costello, and cardio-facio-cutaneous syndromes. <i>British Journal of Cancer</i> , 2015, 112, 1392-1397.	6.4	167
10	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. <i>Journal of Clinical Investigation</i> , 2017, 127, 912-928.	8.2	160
11	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 102, 309-320.	6.2	138
12	The landscape of epilepsy-related GATOR1 variants. <i>Genetics in Medicine</i> , 2019, 21, 398-408.	2.4	137
13	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. <i>Genetics in Medicine</i> , 2018, 20, 1175-1185.	2.4	133
14	ClinGen's RASopathy Expert Panel consensus methods for variant interpretation. <i>Genetics in Medicine</i> , 2018, 20, 1334-1345.	2.4	126
15	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. <i>Journal of Medical Genetics</i> , 2007, 44, 651-656.	3.2	114
16	Juvenile myelomonocytic leukaemia and Noonan syndrome. <i>Journal of Medical Genetics</i> , 2014, 51, 689-697.	3.2	112
17	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. <i>American Journal of Human Genetics</i> , 2014, 95, 285-293.	6.2	110
18	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. <i>Genetics in Medicine</i> , 2018, 20, 630-638.	2.4	101

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19	Gain-of-Function Mutations of ARHGAP31, a Cdc42/Rac1 GTPase Regulator, Cause Syndromic Cutis Aplasia and Limb Anomalies. <i>American Journal of Human Genetics</i> , 2011, 88, 574-585.	6.2	100
20	Hypertrophic Cardiomyopathy in Noonan Syndrome Treated by MEK-Inhibition. <i>Journal of the American College of Cardiology</i> , 2019, 73, 2237-2239.	2.8	96
21	The spectra of clinical phenotypes in aplasia cutis congenita and terminal transverse limb defects. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1860-1881.	1.2	92
22	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adams-Oliver Syndrome With Variable Cardiac Anomalies. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 572-581.	5.1	84
23	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. <i>Gut</i> , 2015, 64, 646-656.	12.1	82
24	Clinical manifestations of mutations in RAS and related intracellular signal transduction factors. <i>Current Opinion in Pediatrics</i> , 2011, 23, 443-451.	2.0	81
25	Mutations in <i>GRIP1</i> cause Fraser syndrome. <i>Journal of Medical Genetics</i> , 2012, 49, 303-306.	3.2	79
26	Genotype and phenotype in patients with Noonan syndrome and a RIT1 mutation. <i>Genetics in Medicine</i> , 2016, 18, 1226-1234.	2.4	77
27	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 475-482.	6.2	73
28	Cardio-facio-cutaneous syndrome: Does genotype predict phenotype?. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2011, 157, 129-135.	1.6	72
29	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. <i>Human Mutation</i> , 2015, 36, 1080-1087.	2.5	67
30	Assessing the gene-disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. <i>Human Mutation</i> , 2018, 39, 1485-1493.	2.5	66
31	Manitoba-oculo-tricho-anal (MOTA) syndrome is caused by mutations in <i>FREM1</i> . <i>Journal of Medical Genetics</i> , 2011, 48, 375-382.	3.2	60
32	A Dual Phenotype of Periventricular Nodular Heterotopia and Frontometaphyseal Dysplasia in One Patient Caused by a Single <i>FLNA</i> Mutation Leading to Two Functionally Different Aberrant Transcripts. <i>American Journal of Human Genetics</i> , 2004, 74, 731-737.	6.2	55
33	Specific mosaic <i>KRAS</i> mutations affecting codon 146 cause oculoectodermal syndrome and encephalocraniocutaneous lipomatosis. <i>Clinical Genetics</i> , 2016, 90, 334-342.	2.0	55
34	Rapidly progressive hypertrophic cardiomyopathy in an infant with Noonan syndrome with multiple lentigines: Palliative treatment with a rapamycin analog. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 744-751.	1.2	53
35	Genetic screening confirms heterozygous mutations in <i>ACAN</i> as a major cause of idiopathic short stature. <i>Scientific Reports</i> , 2017, 7, 12225.	3.3	53
36	Genetics of nephrotic syndrome: new insights into molecules acting at the glomerular filtration barrier. <i>Journal of Molecular Medicine</i> , 2009, 87, 849-857.	3.9	52

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37	Genetic and Pathogenetic Aspects of Noonan Syndrome and Related Disorders. <i>Hormone Research</i> , 2009, 72, 57-63.	1.8	51
38	Synaptic activity controls localization and function of Ct <i>BP</i> 1 via binding to <i>B</i> assoon and <i>P</i> iccolo. <i>EMBO Journal</i> , 2015, 34, 1056-1077.	7.8	51
39	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. <i>American Journal of Human Genetics</i> , 2020, 107, 499-513.	6.2	48
40	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. <i>Nature Communications</i> , 2020, 11, 4625.	12.8	47
41	A variable combination of features of Noonan syndrome and neurofibromatosis type I are caused by mutations in the <i>NF1</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 2749-2756.	1.2	46
42	Genetic Basis and Pancreatic Biology of Johanson-Blizzard Syndrome. <i>Endocrinology and Metabolism Clinics of North America</i> , 2006, 35, 243-253.	3.2	44
43	Phenotype and natural history in Marshall's Smith syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2714-2726.	1.2	44
44	Activating Mutations of <i>RRAS2</i> Are a Rare Cause of Noonan Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 1223-1232.	6.2	43
45	<i>WDR73</i> Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. <i>Human Mutation</i> , 2015, 36, 1021-1028.	2.5	42
46	Further delineation of Malan syndrome. <i>Human Mutation</i> , 2018, 39, 1226-1237.	2.5	42
47	<i>NFIB</i> Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. <i>American Journal of Human Genetics</i> , 2018, 103, 752-768.	6.2	40
48	Food-Derived Xeno-microRNAs: Influence of Diet and Detectability in Gastrointestinal Tract – Proof-of-Principle Study. <i>Molecular Nutrition and Food Research</i> , 2019, 63, e1800076.	3.3	40
49	Expansion of the phenotypic spectrum and description of molecular findings in a cohort of patients with oculocutaneous mosaic RASopathies. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e625.	1.2	39
50	bFGF-mediated pluripotency maintenance in human induced pluripotent stem cells is associated with <i>NRAS</i> -MAPK signaling. <i>Cell Communication and Signaling</i> , 2018, 16, 96.	6.5	38
51	Genotype and phenotype spectrum of <i>NRAS</i> germline variants. <i>European Journal of Human Genetics</i> , 2017, 25, 823-831.	2.8	36
52	CTLA-4-mediated posttranslational modifications direct cytotoxic T-lymphocyte differentiation. <i>Cell Death and Differentiation</i> , 2017, 24, 1739-1749.	11.2	36
53	Variants in nuclear factor I genes influence growth and development. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 611-626.	1.6	32
54	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. <i>Human Mutation</i> , 2018, 39, 1246-1261.	2.5	31

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55	Activating MRAS mutations cause Noonan syndrome associated with hypertrophic cardiomyopathy. <i>Human Molecular Genetics</i> , 2020, 29, 1772-1783.	2.9	30
56	Noonan syndrome: improving recognition and diagnosis. <i>Archives of Disease in Childhood</i> , 2022, 107, 1073-1078.	1.9	28
57	Deletions in the 3' Part of the <i>NFIX</i> Gene Including a Recurrent Alu-Mediated Deletion of Exon 6 and 7 Account for Previously Unexplained Cases of Marshall-Smith Syndrome. <i>Human Mutation</i> , 2014, 35, 1092-1100.	2.5	26
58	Clinical and molecular analysis of RASopathies in a group of Turkish patients. <i>Clinical Genetics</i> , 2013, 83, 181-186.	2.0	24
59	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. <i>American Journal of Human Genetics</i> , 2021, 108, 2112-2129.	6.2	23
60	Genetic variation of the RASGRF1 regulatory region affects human hippocampus-dependent memory. <i>Frontiers in Human Neuroscience</i> , 2014, 8, 260.	2.0	22
61	Valenced action/inhibition learning in humans is modulated by a genetic variant linked to dopamine D2 receptor expression. <i>Frontiers in Systems Neuroscience</i> , 2014, 8, 140.	2.5	22
62	Genetic basis of hypertrophic cardiomyopathy in children. <i>Clinical Research in Cardiology</i> , 2019, 108, 282-289.	3.3	22
63	Responsiveness of sphingosine phosphate lyase insufficiency syndrome to vitamin B6 cofactor supplementation. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1131-1142.	3.6	21
64	Surgery in Focal Congenital Hyperinsulinism (CHI) - The "Hyperinsulinism Germany International" Experience in 30 Children. <i>Pediatric Endocrinology Reviews</i> , 2016, 14, 129-137.	1.2	20
65	Dealing with the incidental finding of secondary variants by the example of SRNS patients undergoing targeted next-generation sequencing. <i>Pediatric Nephrology</i> , 2016, 31, 73-81.	1.7	19
66	MRI Spectrum of Brain Involvement in Sphingosine-1-Phosphate Lyase Insufficiency Syndrome. <i>American Journal of Neuroradiology</i> , 2020, 41, 1943-1948.	2.4	19
67	Germline PTPN11 and somatic PIK3CA variant in a boy with megalencephaly-capillary malformation syndrome (MCAP) - pure coincidence?. <i>European Journal of Human Genetics</i> , 2015, 23, 409-412.	2.8	17
68	Adams' Oliver syndrome caused by mutations of the <i>EOGT</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2246-2251.	1.2	17
69	Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. <i>European Journal of Human Genetics</i> , 2021, 29, 51-60.	2.8	17
70	Long-term outcomes of childhood onset Noonan compared to sarcomere hypertrophic cardiomyopathy. <i>Cardiovascular Diagnosis and Therapy</i> , 2019, 9, S299-S309.	1.7	16
71	Genetic testing in inherited endocrine disorders: joint position paper of the European reference network on rare endocrine conditions (Endo-ERN). <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 144.	2.7	15
72	Neurologic and neurodevelopmental complications in cardiofaciocutaneous syndrome are associated with genotype: A multinational cohort study. <i>Genetics in Medicine</i> , 2022, 24, 1556-1566.	2.4	15

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73	Genotypeâ€“epigenotypeâ€“phenotype correlations in females with frontometaphyseal dysplasia. American Journal of Medical Genetics, Part A, 2006, 140A, 1069-1073.	1.2	14
74	Expanding the genotypic and phenotypic spectrum of severe serine biosynthesis disorders. Human Mutation, 2020, 41, 1615-1628.	2.5	14
75	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
76	A Postzygotic SMO Mutation Caused the Original Case of Happleâ€“Tinschert Syndrome. Acta Dermato-Venereologica, 2018, 98, 534-535.	1.3	11
77	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
78	Mosaic <scp>RASopathy</scp> due to <scp><i>KRAS</i></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
79	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
80	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
81	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
82	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
83	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
84	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
85	Gut microbial similarity in twins is driven by shared environment and aging. EBioMedicine, 2022, 79, 104011.	6.1	7
86	Profiling of the Bacterial Microbiota along the Murine Alimentary Tract. International Journal of Molecular Sciences, 2022, 23, 1783.	4.1	6
87	Cutis marmorata telangiectatica congenita being caused by postzygotic GNA11 mutations. European Journal of Medical Genetics, 2022, 65, 104472.	1.3	6
88	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
89	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
90	Two unrelated families with variable expression of Fraser syndrome due to the same pathogenic variant in the <scp><i>FRAS1</i></scp> gene. American Journal of Medical Genetics, Part A, 2020, 182, 773-779.	1.2	5

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91	Refinement of the critical genomic region for congenital hyperinsulinism in the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	1.8	5
92	<i>WARS1</i> and <i>SARS1</i> : Two tRNA synthetases implicated in autosomal recessive microcephaly. Human Mutation, 2022, 43, 1454-1471.	2.5	5
93	Expanding the clinical phenotype of <i>RASopathies</i> in 38 Turkish patients, including the rare <i>LZTR1</i> , <i>RAF1</i> , <i>RIT1</i> variants, and large deletion in <i>NF1</i> . American Journal of Medical Genetics, Part A, 2021, 185, 3623-3633.	1.2	4
94	A 2q24.2 microdeletion containing <i>TANK</i> as novel candidate gene for intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 832-836.	1.2	3
95	Mosaic Neurofibromatosis Type 1 With Multiple Cutaneous Diffuse and Plexiform Neurofibromas of the Lower Leg. Anticancer Research, 2020, 40, 3423-3427.	1.1	3
96	Aplasia cutis congenita in a CDC42 related developmental phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 850-855.	1.2	3
97	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
98	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	1.8	3
99	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
100	Unrecognized juvenile nephropathic cystinosis. Kidney International, 2018, 94, 1027.	5.2	2
101	Outcomes in growth hormone-treated Noonan syndrome children: impact of PTPN11 mutation status. Endocrine Connections, 2022, 11, .	1.9	2
102	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
103	<i>KRAS</i> Mutation 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
104	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
105	Congenital and Inherited Anomalies. , 0, , 58-68.		1
106	The mosaic hedgehog spectrum: another lesson on the polymorphy of mosaicism. British Journal of Dermatology, 2019, 182, 22-23.	1.5	0
107	Novel Germ Line Mutations in the KRAS2 Gene Cause Noonan Syndrome and Deregulate Hematopoietic Cell Growth.. Blood, 2005, 106, 1602-1602.	1.4	0
108	BRAF Mutations in Juvenile Myelomonocytic Leukemia.. Blood, 2007, 110, 4602-4602.	1.4	0