Michael Ludwig

List of Publications by Year
in descending order

105	2,819	159585	214800
papers	citations	30 h-index	47 g-index

106	106	106	2555
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	First genome-wide association study of esophageal atresia identifies three genetic risk loci at CTNNA3, FOXF1/FOXC2/FOXL1, and HNF1B. Human Genetics and Genomics Advances, 2022, 3, 100093.	1.7	4
2	Currarino syndrome: a comprehensive genetic review of a rare congenital disorder. Orphanet Journal of Rare Diseases, 2021, 16, 167.	2.7	10
3	PPAR-Responsive Elements Enriched with Alu Repeats May Contribute to Distinctive PPARÎ³â€“DNMT1 Interactions in the Genome. Cancers, 2021, 13, 3993.	3.7	2
4	SLC20A1 Is Involved in Urinary Tract and Urorectal Development. Frontiers in Cell and Developmental Biology, 2020, 8, 567.	3.7	22
5	Ubiquitin Carboxyl-Terminal Hydrolases (UCHs): Potential Mediators for Cancer and Neurodegeneration. International Journal of Molecular Sciences, 2020, 21, 3910.	4.1	20
6	Human exome and mouse embryonic expression data implicate ZFHX3, TRPS1, and CHD7 in human esophageal atresia. PLoS ONE, 2020, 15, e0234246.	2.5	9
7	Title is missing!. , 2020, 15, e0234246.		0
8	Title is missing!. , 2020, 15, e0234246.		0
9	Title is missing!. , 2020, 15, e0234246.		0
10	Title is missing!. , 2020, 15, e0234246.		0
11	Title is missing!. , 2020, 15, e0234246.		0
12	Title is missing!. , 2020, 15, e0234246.		0
13	Frequent detection of Saffold cardiovirus in adenoids. PLoS ONE, 2019, 14, e0218873.	2.5	3
14	Mutational Landscape of the BAP1 Locus Reveals an Intrinsic Control to Regulate the miRNA Network and the Binding of Protein Complexes in Uveal Melanoma. Cancers, 2019, 11, 1600.	3.7	30
15	Exome chip association study excluded the involvement of rare coding variants with large effect sizes in the etiology of anorectal malformations. PLoS ONE, 2019, 14, e0217477.	2.5	3
16	Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. American Journal of Human Genetics, 2019, 104, 994-1006.	6.2	47
17	HSPA6: A new autosomal recessive candidate gene for the VATER/VACTERL malformation spectrum. Birth Defects Research, 2019, 111, 591-597.	1.5	15
18	A classic twin study of lower urinary tract obstruction: Report of 3 cases and literature review. LUTS: Lower Urinary Tract Symptoms, 2019, 11, O85-O88.	1.3	6

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19	Exome sequencing in syndromic brain malformations identifies novel mutations in ACTB, and SLC9A6, and suggests BAZ1A as a new candidate gene. Birth Defects Research, 2018, 110, 587-597.	1.5	21
20	Towards a Central Role of ISL1 in the Bladder Exstrophyâ€“Epispadias Complex (BEEC): Computational Characterization of Genetic Variants and Structural Modelling. Genes, 2018, 9, 609.	2.4	6
21	A case of Type 1 Dent disease presenting with isolated persistant proteinuria. Turk Pediatri Arsivi, 2018, 55, 72-75.	0.9	3
22	Low maternal folate concentrations and maternal MTHFR C677T polymorphism are associated with an increased risk for neural tube defects in offspring: a case-control study among Pakistani case and control mothers. Asia Pacific Journal of Clinical Nutrition, 2018, 27, 253-260.	0.4	8
23	ISL1 is a major susceptibility gene for classic bladder exstrophy and a regulator of urinary tract development. Scientific Reports, 2017, 7, 42170.	3.3	41
24	Role of the LF-SINEâ€“Derived Distal ISL1 Enhancer in Patients with Classic Bladder Exstrophy. Journal of Pediatric Genetics, 2017, 06, 169-173.	0.7	3
25	Arrayâ€based molecular karyotyping in 115 VATER/VACTERL and VATER/VACTERLâ€like patients identifies diseaseâ€causing copy number variations. Birth Defects Research, 2017, 109, 1063-1069.	1.5	26
26	Targeted Resequencing of Putative Growth-Related Genes Using Whole Exome Sequencing in Patients with Severe Primary IGF-I Deficiency. Hormone Research in Paediatrics, 2017, 88, 408-417.	1.8	9
27	Dent disease in Poland: what we have learned so far?. International Urology and Nephrology, 2017, 49, 2005-2017.	1.4	11
28	Proteinuria in Dent disease: a review of the literature. Pediatric Nephrology, 2017, 32, 1851-1859.	1.7	46
29	Liddle syndrome in a Turkish family with heterogeneous phenotypes. Pediatrics International, 2016, 58, 801-804.	0.5	9
30	Whole-Exome Sequencing in Nine Monozygotic Discordant Twins. Twin Research and Human Genetics, 2016, 19, 60-65.	0.6	24
31	The oculocerebrorenal syndrome of Lowe: an update. Pediatric Nephrology, 2016, 31, 2201-2212.	1.7	106
32	Arrayâ€based molecular karyotyping in fetal brain malformations: Identification of novel candidate genes and chromosomal regions. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 16-26.	1.6	13
33	Long-term renal outcome in children withOCRLmutations: retrospective analysis of a large international cohort. Nephrology Dialysis Transplantation, 2016, 33, gfw350.	0.7	27
34	Copy number variations in 375 patients with oesophageal atresia and/or tracheoesophageal fistula. European Journal of Human Genetics, 2016, 24, 1715-1723.	2.8	27
35	Array-based molecular karyotyping in fetuses with isolated brain malformations identifies disease-causing CNVs. Journal of Neurodevelopmental Disorders, 2016, 8, 11.	3.1	24
36	CNV analysis in 169 patients with bladder exstrophy-epispadias complex. BMC Medical Genetics, 2016, 17, 35.	2.1	15

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37	Underlying genetic factors of the VATER/VACTERL association with special emphasis on the â€œRenalâ€• phenotype. Pediatric Nephrology, 2016, 31, 2025-2033.	1.7	34
38	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
39	Lowe syndrome/Dent-2 disease: A comprehensive review of known and novel aspects. Journal of Pediatric Genetics, 2015, 02, 053-068.	0.7	22
40	Novel OCRL mutations in patients with Dent-2 disease. Journal of Pediatric Genetics, 2015, 01, 015-023.	0.7	29
41	Targeted Resequencing of 29 Candidate Genes and Mouse Expression Studies ImplicateZIC3andFOXF1in Human VATER/VACTERL Association. Human Mutation, 2015, 36, 1150-1154.	2.5	46
42	Mutations in PTF1A are not a common cause for human VATER/VACTERL association or neural tube defects mirroring Danforthâ€™s short tail mouse. Molecular Medicine Reports, 2015, 12, 1579-1583.	2.4	3
43	Investigation of the role of TCF4 rare sequence variants in schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2015, 168, 354-362.	1.7	12
44	Genetics of Bladder-Exstrophy-Epispadias Complex (BEEC): Systematic Elucidation of Mendelian and Multifactorial Phenotypes. Current Genomics, 2015, 17, 4-13.	1.6	36
45	WNT3 involvement in human bladder exstrophy and cloaca development in zebrafish. Human Molecular Genetics, 2015, 24, 5069-5078.	2.9	23
46	Characterization of 28 novel patients expands the mutational and phenotypic spectrum of Lowe syndrome. Pediatric Nephrology, 2015, 30, 931-943.	1.7	35
47	Clinical utility gene card for: Lowe syndrome. European Journal of Human Genetics, 2015, 23, 889-889.	2.8	2
48	Genome-wide Association Study and Meta-Analysis Identify ISL1 as Genome-wide Significant Susceptibility Gene for Bladder Exstrophy. PLoS Genetics, 2015, 11, e1005024.	3.5	41
49	Genome-wide array data and next generation sequencing unravel the etiology of urogenital malformations. Journal of Pediatric Genetics, 2015, 01, 209-216.	0.7	1
50	Genomeâ€wide mapping of copy number variations in patients with both anorectal malformations and central nervous system abnormalities. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 235-242.	1.6	18
51	Clinical utility gene card for: Dent disease (Dent-1 and Dent-2). European Journal of Human Genetics, 2014, 22, 1338-1338.	2.8	9
52	Classic bladder exstrophy: Frequent 22q11.21 duplications and definition of a 414 kb phenocritical region. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 512-517.	1.6	21
53	Genome-wide association study and mouse expression data identify a highly conserved 32 kb intergenic region between WNT3 and WNT9b as possible susceptibility locus for isolated classic exstrophy of the bladder. Human Molecular Genetics, 2014, 23, 5536-5544.	2.9	19
54	Whole-exome resequencing reveals recessive mutations in TRAP1 in individuals with CAKUT and VACTERL association. Kidney International, 2014, 85, 1310-1317.	5.2	106

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55	Novel Hemoglobin UKB demonstrates the importance of using different methods of detection. Clinica Chimica Acta, 2014, 431, 58-59.	1.1	2
56	Heterozygous FGF8 mutations in patients presenting cryptorchidism and multiple VATER/VACTERL features without limb anomalies. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 750-759.	1.6	14
57	De novo microduplications at 1q41, 2q37.3, and 8q24.3 in patients with VATER/VACTERL association. European Journal of Human Genetics, 2013, 21, 1377-1382.	2.8	38
58	Candidate gene association study implicates p63 in the etiology of nonsyndromic bladderâ€exstrophyâ€epispadias complex. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 759-763.	1.6	8
59	De novo 13q deletions in two patients with mild anorectal malformations as part of VATER/VACTERL and VATER/VACTERLâ€like association and analysis of EFNB2 in patients with anorectal malformations. American Journal of Medical Genetics, Part A, 2013, 161, 3035-3041.	1.2	32
60	Isolated bladder exstrophy associated with a de novo 0.9 Mb microduplication on chromosome 19p13.12. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 133-139.	1.6	18
61	CNV Analysis in Monozygotic Twin Pairs Discordant for Urorectal Malformations. Twin Research and Human Genetics, 2013, 16, 802-807.	0.6	9
62	Involvement of the WNT and FGF signaling pathways in non-isolated anorectal malformations: Sequencing analysis of WNT3A, WNT5A, WNT11, DACT1, FGF10, FGFR2 and the T gene. International Journal of Molecular Medicine, 2012, 30, 1459-1464.	4.0	24
63	Familial occurrence of the VATER/VACTERL association. Pediatric Surgery International, 2012, 28, 725-729.	1.4	36
64	Identification of an Alu-mediated 12.2-kb deletion of the complete LPAR6 (P2RY5) gene in a Turkish family with hypotrichosis and woolly hair. Experimental Dermatology, 2012, 21, 469-471.	2.9	3
65	De novo microduplication at 22q11.21 in a patient with VACTERL association. European Journal of Medical Genetics, 2011, 54, 9-13.	1.3	56
66	Phenotype Severity in the Bladder Exstrophy-Epispadias Complex: Analysis of Genetic and Nongenetic Contributing Factors in 441 Families from North America and Europe. Journal of Pediatrics, 2011, 159, 825-831.e1.	1.8	33
67	Effect of growth hormone replacement therapy in a boy with Dent's disease: a case report. Journal of Medical Case Reports, 2011, 5, 400.	0.8	5
68	Novel techniques and newer markers for the evaluation of â€œproximal tubular dysfunctionâ€• International Urology and Nephrology, 2011, 43, 1107-1115.	1.4	8
69	Clinical and laboratory features of Macedonian children with OCRL mutations. Pediatric Nephrology, 2011, 26, 557-562.	1.7	18
70	Autosomal-dominant non-syndromic anal atresia: sequencing of candidate genes, array-based molecular karyotyping, and review of the literature. European Journal of Pediatrics, 2011, 170, 741-746.	2.7	16
71	Maternal de novo triple mosaicism for two single OCRL nucleotide substitutions (c.1736A>T,) Tj ETQq1 1 0.784314 rgBT /Overlock	3.8	19
72	Genome-wide expression profiling of urinary bladder implicates desmosomal and cytoskeletal dysregulation in the bladder exstrophy-epispadias complex. International Journal of Molecular Medicine, 2011, 27, 755-65.	4.0	19

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73	A novel CLCN5 mutation in a boy with Bartter-like syndrome and partial growth hormone deficiency. Pediatric Nephrology, 2010, 25, 2363-2368.	1.7	33
74	Embryonic expression of the cysteine rich protein 61 (CYR61) gene: A candidate for the development of human epispadias. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 546-550.	1.6	7
75	Evidence for linkage of the bladder exstrophyâ€epispadias complex on chromosome 4q31.21â€22 and 19q13.31â€41 from a consanguineous iranian family. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 757-761.	1.6	7
76	Microduplications at 22q11.21 are associated with non-syndromic classic bladder exstrophy. European Journal of Medical Genetics, 2010, 53, 55-60.	1.3	45
77	Disorders of the Renal Proximal Tubule. Nephron Physiology, 2010, 118, p1-p6.	1.2	33
78	Dent-2 Disease: A Mild Variant of Lowe Syndrome. Journal of Pediatrics, 2009, 155, 94-99.	1.8	112
79	Genomeâ€wide linkage scan for bladder exstrophyâ€epispadias complex. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 174-178.	1.6	20
80	Bladder exstrophyâ€epispadias complex. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 509-522.	1.6	53
81	Possible association of Down syndrome and exstrophyâ€“epispadias complex: report of two new cases and review of the literature. European Journal of Pediatrics, 2009, 168, 881-883.	2.7	5
82	VitaminÂA responsive night blindness in Dentâ€™s disease. Pediatric Nephrology, 2009, 24, 1765-1770.	1.7	22
83	The Exstrophy-epispadias complex. Orphanet Journal of Rare Diseases, 2009, 4, 23.	2.7	198
84	Effect of fungicides on the complex of Fusarium species and saprophytic fungi colonizing wheat kernels. European Journal of Plant Pathology, 2008, 120, 157-166.	1.7	89
85	Investigation of FGF10 as a candidate gene in patients with anorectal malformations and exstrophy of the cloaca. Pediatric Surgery International, 2008, 24, 893-897.	1.4	22
86	Successful assisted reproduction in adult males with bladder extrophyâ€“epispadias complex. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2008, 139, 258-259.	1.1	1
87	Renal Phenotype in Lowe Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2008, 3, 1430-1436.	4.5	116
88	Concordance analyses of twins with bladder exstrophyâ€“epispadias complex suggest genetic etiology. American Journal of Medical Genetics, Part A, 2007, 143A, 2751-2756.	1.2	53
89	Genome-wide analysis for micro-aberrations in familial exstrophy of the bladder using array-based comparative genomic hybridization. BJU International, 2007, 100, 646-650.	2.5	12
90	Bladder exstrophyâ€“epispadias complex: Investigation of suppressor of variegation, enhancer of zeste and Trithorax (SET) as a candidate gene in a large cohort of patients. Scandinavian Journal of Urology and Nephrology, 2006, 40, 221-224.	1.4	16

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91	Hypercalciuria in patients with CLCN5 mutations. Pediatric Nephrology, 2006, 21, 1241-1250.	1.7	45
92	MTHFR 677 TT genotype in a mother and her child with Down syndrome, atrioventricular canal and exstrophy of the bladder: implications of a mutual genetic risk factor?. European Journal of Pediatrics, 2006, 165, 566-568.	2.7	13
93	Novel OCRL1 Mutations in Patients With the Phenotype of Dent Disease. American Journal of Kidney Diseases, 2006, 48, 942.e1-942.e14.	1.9	68
94	Bladder exstrophy and Epstein type congenital macrothrombocytopenia: Evidence for a common cause?. American Journal of Medical Genetics, Part A, 2006, 140A, 2251-2253.	1.2	11
95	Family-based association study of theMTHFR polymorphism C677T in the bladder-exstrophy-epispadias-complex. American Journal of Medical Genetics, Part A, 2006, 140A, 2506-2509.	1.2	6
96	Recent advances in understanding the clinical and genetic heterogeneity of Dent's disease. Nephrology Dialysis Transplantation, 2006, 21, 2708-2717.	0.7	51
97	Functional evaluation of Dentâ€™s disease-causing mutations: implications for ClC-5 channel trafficking and internalization. Human Genetics, 2005, 117, 228-237.	3.8	52
98	CLCN5 mutation (R347X) associated with hypokalaemic metabolic alkalosis in a Turkish child: an unusual presentation of Dent's disease. Nephrology Dialysis Transplantation, 2005, 20, 1476-1479.	0.7	37
99	Dent diseaseâ€like phenotype and the chloride channel ClCâ€4 (CLCN4) gene. American Journal of Medical Genetics Part A, 2004, 128A, 434-435.	2.4	13
100	Pseudohypoaldosteronism type 1 and the genes encoding prostasin, alpha-spectrin, and Nedd4. International Journal of Molecular Medicine, 2004, 14, 1101-4.	4.0	4
101	Four Additional CLCN5 Exons Encode a Widely Expressed Novel Long CLC-5 Isoform but Fail to Explain Dentâ€™s Phenotype in Patients without Mutations in the Short Variant. Kidney and Blood Pressure Research, 2003, 26, 176-184.	2.0	15
102	A novel stable polyalanine [poly(A)] expansion in the HOXA13 gene associated with hand-foot-genital syndrome: proper function of poly(A)-harbouring transcription factors depends on a critical repeat length?. Human Genetics, 2002, 110, 488-494.	3.8	75
103	Somatic Mosaicism in Hemophilia A: A Fairly Common Event. American Journal of Human Genetics, 2001, 69, 75-87.	6.2	144
104	Homozygous ?-thalassemia associated with hypospadias: SEA-type deletion does not affect expression of the -14 gene and loss of the ?1-globin gene on 16p13.3 is compensated by its duplicate ?2 on chromosome 10. American Journal of Medical Genetics Part A, 2001, 101, 286-287.	2.4	5
105	Induction of Apoptosis and Necrosis in Human Neuroblastoma Cells by Cholesterol Oxides. Annals of the New York Academy of Sciences, 1999, 893, 379-381.	3.8	12