Thomas MÃ¼ller

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1872795/publications.pdf

Version: 2024-02-01

56 papers	2,410 citations	257450 24 h-index	214800 47 g-index
59 all docs	59 docs citations	59 times ranked	3607 citing authors

#	Article	IF	Citations
1	MYO5B mutations cause microvillus inclusion disease and disrupt epithelial cell polarity. Nature Genetics, 2008, 40, 1163-1165.	21.4	321
2	Faecal calprotectin indicates intestinal inflammation in COVID-19. Gut, 2020, 69, 1543-1544.	12.1	247
3	Loss of Syntaxin 3 Causes Variant Microvillus Inclusion Disease. Gastroenterology, 2014, 147, 65-68.e10.	1.3	151
4	Loss-of-function of MYO5B is the main cause of microvillus inclusion disease: 15 novel mutations and a CaCo-2 RNAi cell model. Human Mutation, 2010, 31, 544-551.	2.5	122
5	Mutations in SPINT2 Cause a Syndromic Form of Congenital Sodium Diarrhea. American Journal of Human Genetics, 2009, 84, 188-196.	6.2	110
6	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	12.8	110
7	Re-evaluation of the penicillamine challenge test in the diagnosis of Wilsonâ€™s disease in children. Journal of Hepatology, 2007, 47, 270-276.	3.7	100
8	Cargo-selective apical exocytosis in epithelial cells is conducted by Myo5B, Slp4a, Vamp7, and Syntaxin 3. Journal of Cell Biology, 2015, 211, 587-604.	5.2	88
9	Intestinal Failure and Aberrant Lipid Metabolism in Patients WithÂDGAT1 Deficiency. Gastroenterology, 2018, 155, 130-143.e15.	1.3	83
10	Loss of dermatan sulfate epimerase (DSE) function results in musculocontractural Ehlersâ€“Danlos syndrome. Human Molecular Genetics, 2013, 22, 3761-3772.	2.9	78
11	Congenital secretory diarrhoea caused by activating germline mutations inGUCY2C. Gut, 2016, 65, 1306-1313.	12.1	74
12	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. American Journal of Human Genetics, 2016, 99, 414-422.	6.2	73
13	Deficiency of the sphingosine-1-phosphate lyase SGPL1 is associated with congenital nephrotic syndrome and congenital adrenal calcifications. Human Mutation, 2017, 38, 365-372.	2.5	71
14	An inducible mouse model for microvillus inclusion disease reveals a role for myosin Vb in apical and basolateral trafficking. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 12408-12413.	7.1	67
15	The phenotype of the musculocontractural type of Ehlersâ€Danlos syndrome due to CHST14 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 103-115.	1.2	53
16	Abnormal Rab11â€Rab8â€vesicles cluster in enterocytes of patients with microvillus inclusion disease. Traffic, 2017, 18, 453-464.	2.7	45
17	Loss of MYO5B Leads to Reductions in Na+ Absorption With Maintenance of CFTR-Dependent Clâ€“ Secretion in Enterocytes. Gastroenterology, 2018, 155, 1883-1897.e10.	1.3	45
18	WDR73Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Human Mutation, 2015, 36, 1021-1028.	2.5	42

#	Article	IF	Citations
19	Disrupted apical exocytosis of cargo vesicles causes enteropathy in FHL5 patients with Munc18-2 mutations. JCI Insight, 2017, 2, .	5.0	41
20	Case of syndromic tufting enteropathy harbors SPINT2 mutation seen in congenital sodium diarrhea. Clinical Dysmorphology, 2010, 19, 48.	0.3	38
21	Autosomal Recessive Keratoderma-Ichthyosis-Deafness (ARKID) Syndrome IsÂCaused by VPS33B Mutations AffectingÂRab Protein Interaction andÂCollagen Modification. Journal of Investigative Dermatology, 2017, 137, 845-854.	0.7	37
22	Vedolizumab use after failure of TNF-Î± antagonists in children and adolescents with inflammatory bowel disease. BMC Gastroenterology, 2018, 18, 140.	2.0	37
23	Homozygous SYNE1 mutation causes congenital onset of muscular weakness with distal arthrogryposis: a genotypeâ€“phenotype correlation. European Journal of Human Genetics, 2017, 25, 262-266.	2.8	34
24	Nuclear FOXO3 predicts adverse clinical outcome and promotes tumor angiogenesis in neuroblastoma. Oncotarget, 2016, 7, 77591-77606.	1.8	31
25	3D bioprinted, vascularized neuroblastoma tumor environment in fluidic chip devices for precision medicine drug testing. Biofabrication, 2022, 14, 035002.	7.1	28
26	Towards understanding microvillus inclusion disease. Molecular and Cellular Pediatrics, 2016, 3, 3.	1.8	26
27	SPINT2 (HAI-2) missense variants identified in congenital sodium diarrhea/tufting enteropathy affect the ability of HAI-2 to inhibit prostasin but not matriptase. Human Molecular Genetics, 2019, 28, 828-841.	2.9	25
28	AP1S1 missense mutations cause a congenital enteropathy via an epithelial barrier defect. Human Genetics, 2020, 139, 1247-1259.	3.8	24
29	Chondroitin SulfateN-acetylgalactosaminyltransferase-1 (CSGalNAcT-1) Deficiency Results in a Mild Skeletal Dysplasia and Joint Laxity. Human Mutation, 2017, 38, 34-38.	2.5	22
30	Congenital Diarrhea and Cholestatic Liver Disease: Phenotypic Spectrum Associated with MYO5B Mutations. Journal of Clinical Medicine, 2021, 10, 481.	2.4	20
31	Severe Deoxyguanosine Kinase Deficiency in Austria. Journal of Pediatric Gastroenterology and Nutrition, 2019, 68, e1-e6.	1.8	13
32	Stereotactic radiofrequency ablation of a variety of liver masses in children. International Journal of Hyperthermia, 2020, 37, 1074-1081.	2.5	13
33	Pathogenic STX3 variants affecting the retinal and intestinal transcripts cause an early-onset severe retinal dystrophy in microvillus inclusion disease subjects. Human Genetics, 2021, 140, 1143-1156.	3.8	13
34	â€œEnhanced acquisition of antibiotic-resistant intestinal E. coli during the first year of life assessed in a prospective cohort studyâ€• Antimicrobial Resistance and Infection Control, 2019, 8, 79.	4.1	12
35	Increased Fecal Neopterin Parallels Gastrointestinal Symptoms in COVID-19. Clinical and Translational Gastroenterology, 2021, 12, e00293.	2.5	12
36	Low sodium status in cystic fibrosisâ€”as assessed by calculating fractional Na + excretionâ€”is associated with decreased growth parameters. Journal of Cystic Fibrosis, 2016, 15, 400-405.	0.7	10

#	Article	IF	Citations
37	Omega-3 Fatty Acids and Their Role in Pediatric Cancer. Nutrients, 2021, 13, 1800.	4.1	10
38	UNC45A deficiency causes microvillus inclusion diseaseâ€“like phenotype by impairing myosin VBâ€“dependent apical trafficking. Journal of Clinical Investigation, 2022, 132, .	8.2	9
39	Reduced NHE3 activity results in congenital diarrhea and can predispose to inflammatory bowel disease. American Journal of Physiology - Regulatory Integrative and Comparative Physiology, 2017, 312, R311-R311.	1.8	7
40	Novel PCNT variants in MOPDII with attenuated growth restriction and pachygyria. Clinical Genetics, 2020, 98, 282-287.	2.0	7
41	Sensitivity and specificity of the antigen-based anterior nasal self-testing programme for detecting SARS-CoV-2 infection in schools, Austria, March 2021. Eurosurveillance, 2021, 26, .	7.0	7
42	The SZT2 Interactome Unravels New Functions of the KICSTOR Complex. Cells, 2021, 10, 2711.	4.1	7
43	Synonymous mutation in adenosine triphosphatase copperâ€transporting beta causes enhanced exon skipping in Wilson disease. Hepatology Communications, 2022, 6, 1611-1619.	4.3	6
44	Co-existence of ABCB11 and DCDC2 disease: Infantile cholestasis requires both next-generation sequencing and clinical-histopathologic correlation. European Journal of Human Genetics, 2020, 28, 840-844.	2.8	5
45	Highly Elevated Plasma Î³â€Glutamyltransferase Elevations: A Trait Caused by Î³â€Glutamyltransferase 1 Transmembrane Mutations. Hepatology, 2020, 71, 1124-1127.	7.3	4
46	Advanced Microscopy for Liver and Gut Ultrastructural Pathology in Patients with MVID and PFIC Caused by MYO5B Mutations. Journal of Clinical Medicine, 2021, 10, 1901.	2.4	4
47	Biallelic variants in VPS50 cause a neurodevelopmental disorder with neonatal cholestasis. Brain, 2021, 144, 3036-3049.	7.6	4
48	Further delineation of putative ACTB lossâ€ofâ€function variants: A 4â€patient series. Human Mutation, 2020, 41, 753-758.	2.5	3
49	Characteristic facial features and cortical blindness distinguish the DOCK7â€related epileptic encephalopathy. Molecular Genetics & Genomic Medicine, 2021, 9, e1607.	1.2	3
50	Influence of early biliary complications on survival rates after pediatric liver transplantationâ€”A positive outlook. Pediatric Transplantation, 2021, 25, e14075.	1.0	3
51	Long-Term Follow-Up of Tufting Enteropathy Caused by EPCAM Mutation p.Asp253Asn and Absent EPCAM Expression. JPGN Reports, 2021, 2, e029.	0.4	3
52	A novel <scp>IKBKG</scp> mutation in a patient with incontinentia pigmenti and features of hepatic ciliopathy. Australasian Journal of Dermatology, 2018, 59, e262-e265.	0.7	2
53	Three Novel EPCAM Variants Causing Tufting Enteropathy in Three Families. Children, 2021, 8, 503.	1.5	2
54	Early onset congenital diarrheas; single center experience. Pediatrics and Neonatology, 2021, 62, 612-619.	0.9	2

#	Article	IF	Citations
55	Isolated choanal and gut atresias: pathogenetic role of serine protease inhibitor type 2 (SPINT2) gene mutations unlikely. European Journal of Medical Research, 2018, 23, 13.	2.2	1
56	The COVIDâ€19 pandemic reduced paediatric emergency department visits but did not significantly increase urgent cases. Acta Paediatrica, International Journal of Paediatrics, 2021, 111, 130.	1.5	1