Cecilie Bredrup

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3538795/publications.pdf

Version: 2024-02-01

19	646	1040056	940533
papers	citations	9 h-index	16 g-index

20	20	20	1168
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical and molecular response to dasatinib in an adult patient with Penttinen syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 1233-1238.	1.2	1
2	Clinical features and molecular genetics of patients with ABCA4â€retinal dystrophies. Acta Ophthalmologica, 2021, 99, e733-e746.	1.1	6
3	Temperature-dependent autoactivation associated with clinical variability of PDGFRB Asn666 substitutions. Human Molecular Genetics, 2021, 30, 72-77.	2.9	6
4	Positive response to imatinib in <scp>PDGFRB</scp>â€related Kosaki overgrowth syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2597-2601.	1.2	1
5	K⁺ regulates relocation of Pellinoâ€2 to the site of NLRP3 inflammasome activation in macrophages. FEBS Letters, 2021, 595, 2437-2446.	2.8	6
6	Pellinoâ€2 in nonimmune cells: novel interaction partners and intracellular localization. FEBS Letters, 2021, 595, 2909-2921.	2.8	3
7	A tyrosine kinase-activating variant Asn666Ser in PDGFRB causes a progeria-like condition in the severe end of Penttinen syndrome. European Journal of Human Genetics, 2019, 27, 574-581.	2.8	20
8	Recurrent, Activating Variants in the Receptor Tyrosine Kinase DDR2 Cause Warburg-Cinotti Syndrome. American Journal of Human Genetics, 2018, 103, 976-983.	6.2	17
9	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. American Journal of Human Genetics, 2017, 100, 323-333.	6.2	29
10	The intronic <scp>ABCA</scp>4 c.5461â€10T>C variant, frequently seen in patients with Stargardt disease, causes splice defects and reduced <scp>ABCA</scp>4 protein level. Acta Ophthalmologica, 2017, 95, 240-246.	1.1	35
11	Role of Decorin Core Protein in Collagen Organisation in Congenital Stromal Corneal Dystrophy (CSCD). PLoS ONE, 2016, 11, e0147948.	2.5	23
12	Biallelic Mutations in the BEST1 Gene: Additional Families with Autosomal Recessive Bestrophinopathy. Ophthalmic Genetics, 2016, 37, 183-193.	1.2	9
13	Development of Congenital Stromal Corneal Dystrophy Is Dependent on Export and Extracellular Deposition of Truncated Decorin. , 2015, 56, 2909.		15
14	High Myopiaâ€“Excavated Optic Disc Anomaly Associated With a Frameshift Mutation in the MYC-Binding Protein 2 Gene (MYCBP2). American Journal of Ophthalmology, 2015, 159, 973-979.e2.	3.3	13
15	Ciliopathies with Skeletal Anomalies and Renal Insufficiency due to Mutations in the IFT-A Gene WDR19. American Journal of Human Genetics, 2011, 89, 634-643.	6.2	210
16	Decorin Accumulation Contributes to the Stromal Opacities Found in Congenital Stromal Corneal Dystrophy. , 2010, 51, 5578.		40
17	Ophthalmological Aspects of Pierson Syndrome. American Journal of Ophthalmology, 2008, 146, 602-611.e1.	3.3	66
18	Clinical Manifestation of a Novel PAX6 Mutation Arg128Pro. JAMA Ophthalmology, 2008, 126, 428.	2.4	6

#	Article	IF	Citations
19	Congenital Stromal Dystrophy of the Cornea Caused by a Mutation in the Decorin Gene. , 2005, 46, 420.		140