Lilian Bomme Ousager

List of Publications by Year
in descending order

44 papers	2,047 citations	361045 20 h-index	243296 44 g-index
45 all docs	45 docs citations	45 times ranked	4703 citing authors

#	Article	IF	Citations
1	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	1.1	16
2	von Hippel-Lindau disease: Updated guideline for diagnosis and surveillance. European Journal of Medical Genetics, 2022, 65, 104538.	0.7	23
3	Hereditary leiomyomatosis and renal cell carcinoma: a case series and literature review. Orphanet Journal of Rare Diseases, 2021, 16, 34.	1.2	22
4	c.1227_1228dupGG (p.Glu410Glyfs), a frequent variant in Tunisian patients with MUTYH associated polyposis. Cancer Genetics, 2020, 240, 45-53.	0.2	2
5	Low frequency of parental mosaicism in de novo COL4A5 mutations in Xâ€linked Alport syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1452.	0.6	2
6	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
7	Estimating the effect size of the 15Q11.2 BP1â€“BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. Journal of Medical Genetics, 2019, 56, 701-710.	1.5	43
8	Autosomal dominant stapes fixation, syndactyly, and symphalangism in a family with NOG mutation: Long term follow-up on surgical treatment. International Journal of Pediatric Otorhinolaryngology, 2018, 108, 208-212.	0.4	3
9	ENG mutational mosaicism in a family with hereditary hemorrhagic telangiectasia. Molecular Genetics & Genomic Medicine, 2018, 6, 121-125.	0.6	11
10	Pigmentary mosaicism: a review of original literature and recommendations for future handling. Orphanet Journal of Rare Diseases, 2018, 13, 39.	1.2	34
11	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	2.6	48
12	Familial cerebral abscesses caused by hereditary hemorrhagic telangiectasia. Clinical Case Reports (discontinued), 2017, 5, 805-808.	0.2	2
13	De Novo Variants in GRIA4 Lead to Intellectual Disability with or without Seizures and Gait Abnormalities. American Journal of Human Genetics, 2017, 101, 1013-1020.	2.6	53
14	Genetic screening of the FLCN gene identify six novel variants and a Danish founder mutation. Journal of Human Genetics, 2017, 62, 151-157.	1.1	19
15	Novel ELN mutation in a family with supravalvular aortic stenosis and intracranial aneurysm. European Journal of Medical Genetics, 2017, 60, 110-113.	0.7	21
16	Acromelic frontonasal dysostosis andZSWIM6mutation: phenotypic spectrum and mosaicism. Clinical Genetics, 2016, 90, 270-275.	1.0	17
17	<scp>JPâ€“HHT</scp> phenotype in Danish patients with <scp>SMAD4</scp> mutations. Clinical Genetics, 2016, 90, 55-62.	1.0	30
18	Disease pattern in Danish patients with Peutz-Jeghers syndrome. International Journal of Colorectal Disease, 2016, 31, 997-1004.	1.0	13

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19	Germline variants in Hamartomatous Polyposis Syndrome-associated genes from patients with one or few hamartomatous polyps. Scandinavian Journal of Gastroenterology, 2016, 51, 1118-1125.	0.6	8
20	Germline mutations in BMP9 are not identified in a series of Danish and French patients with hereditary hemorrhagic telangiectasia. Gene Reports, 2016, 5, 30-33.	0.4	1
21	Bone structure in two adult subjects with impaired minor spliceosome function resulting from RNU4ATAC mutations causing microcephalic osteodysplastic primordial dwarfism type 1 (MOPD1). Bone, 2016, 92, 145-149.	1.4	8
22	Juvenile Polyps in Denmark From 1995 to 2014. Diseases of the Colon and Rectum, 2016, 59, 751-757.	0.7	18
23	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	4.1	243
24	Two novel mutations in RNU4ATAC in two siblings with an atypical mild phenotype of microcephalic osteodysplastic primordial dwarfism type 1. Clinical Dysmorphology, 2016, 25, 68-72.	0.1	12
25	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	1.1	72
26	Research participants in NGS studies want to know about incidental findings. European Journal of Human Genetics, 2015, 23, 1423-1426.	1.4	28
27	Phenotype and genotype in 103 patients with tricho-rhino-phalangeal syndrome. European Journal of Medical Genetics, 2015, 58, 279-292.	0.7	62
28	Global gene expression profiling of telangiectasial tissue from patients with hereditary hemorrhagic telangiectasia. Microvascular Research, 2015, 99, 118-126.	1.1	13
29	Long Non-Coding RNA Expression Profiles in Hereditary Haemorrhagic Telangiectasia. PLoS ONE, 2014, 9, e90272.	1.1	21
30	On the formation of 7-ketocholesterol from 7-dehydrocholesterol in patients with CTX and SLO. Journal of Lipid Research, 2014, 55, 1165-1172.	2.0	47
31	National mutation study among Danish patients with hereditary haemorrhagic telangiectasia. Clinical Genetics, 2014, 86, 123-133.	1.0	42
32	Mutations in Danish patients with long QT syndrome and the identification of a large founder family with p.F29L in KCNH2. BMC Medical Genetics, 2014, 15, 31.	2.1	14
33	Hamartomatous polyposis syndromes: A review. Orphanet Journal of Rare Diseases, 2014, 9, 101.	1.2	84
34	Histiocytic disorders of the gastrointestinal tract. Human Pathology, 2013, 44, 683-696.	1.1	23
35	Heart defects and other features of the 22q11 distal deletion syndrome. European Journal of Medical Genetics, 2013, 56, 98-107.	0.7	30
36	Distinct mutations in STXBP2 are associated with variable clinical presentations in patients with familial hemophagocytic lymphohistiocytosis type 5 (FHL5). Blood, 2012, 119, 6016-6024.	0.6	137

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37	Duplication of 7q36.3 encompassing the Sonic Hedgehog (SHH) gene is associated with congenital muscular hypertrophy. European Journal of Medical Genetics, 2012, 55, 557-560.	0.7	9
38	Allelic Dropout in the ENG Gene, Affecting the Results of Genetic Testing in Hereditary Hemorrhagic Telangiectasia. Genetic Testing and Molecular Biomarkers, 2012, 16, 1419-1423.	0.3	3
39	Identification of a novel S249C FGFR3 mutation in a keratinocytic epidermal naevus syndrome. British Journal of Dermatology, 2012, 167, 202-204.	1.4	20
40	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. Human Mutation, 2012, 33, 949-959.	1.1	115
41	Mutation update for the PORCN gene. Human Mutation, 2011, 32, 723-728.	1.1	59
42	CNTNAP2 and NRXN1 Are Mutated in Autosomal-Recessive Pitt-Hopkins-like Mental Retardation and Determine the Level of a Common Synaptic Protein in Drosophila. American Journal of Human Genetics, 2009, 85, 655-666.	2.6	573
43	Two siblings with microcephaly, growth retardation, cataract, hearing loss, and unusual appearance. Clinical Dysmorphology, 2009, 18, 181-183.	0.1	3
44	Skin manifestations in a case of trisomy 16 mosaicism. British Journal of Dermatology, 2006, 154, 172-176.	1.4	10