Jens M Hertz

List of Publications by Year
in descending order

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100 papers	3,675 citations	126907 33 h-index	144013 57 g-index
105 all docs	105 docs citations	105 times ranked	4474 citing authors

#	Article	IF	Citations
1	Detection of DZIP1L mutations by whole-exome sequencing in consanguineous families with polycystic kidney disease. Pediatric Nephrology, 2022, 37, 2657-2665.	1.7	5
2	Guidelines for Genetic Testing and Management of Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 143-154.	4.5	49
3	FC044: Heterozygous Variants in Kinase Domain of NEK8 cause an Autosomal-Dominant Ciliopathy. Nephrology Dialysis Transplantation, 2022, 37, .	0.7	1
4	Skewness of X-chromosome inactivation increases with age and varies across birth cohorts in elderly Danish women. Scientific Reports, 2021, 11, 4326.	3.3	16
5	Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria. European Journal of Human Genetics, 2021, 29, 1186-1197.	2.8	61
6	Biallelic variants in <scp>GLE1</scp> with survival beyond neonatal period. Clinical Genetics, 2020, 98, 622-625.	2.0	1
7	Low frequency of parental mosaicism in de novo COL4A5 mutations in Xâ€linked Alport syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1452.	1.2	2
8	Expanding the cerebrovascular phenotype of the p.R258H variant in ACTA2 related hereditary thoracic aortic disease (HTAD). Journal of the Neurological Sciences, 2020, 415, 116897.	0.6	2
9	Association between periacetabular osteotomy and hip dysplasia among relatives: a cross-sectional study. HIP International, 2019, 29, 424-429.	1.7	4
10	Familial multiple sclerosis patients have a shorter delay in diagnosis than sporadic cases. Multiple Sclerosis and Related Disorders, 2019, 32, 97-102.	2.0	17
11	A novel PDGFRB sequence variant in a family with a mild form of primary familial brain calcification: a case report and a review of the literature. BMC Neurology, 2019, 19, 60.	1.8	8
12	Distribution of disease courses in familial vs sporadic multiple sclerosis. Acta Neurologica Scandinavica, 2019, 139, 231-237.	2.1	8
13	Genetic analysis of Charcot-Marie-Tooth disease in Denmark and the implementation of a next generation sequencing platform. European Journal of Medical Genetics, 2019, 62, 1-8.	1.3	20
14	Hereditary spastic paraplegia type 8: Neuropathological findings. Brain Pathology, 2018, 28, 292-294.	4.1	2
15	Targeted gene sequencing and wholeâ€exome sequencing in autopsied fetuses with prenatally diagnosed kidney anomalies. Clinical Genetics, 2018, 93, 860-869.	2.0	48
16	Anxiety and depression in Klinefelter syndrome: The impact of personality and social engagement. PLoS ONE, 2018, 13, e0206932.	2.5	24
17	Properties and units in the clinical laboratory sciences part XXIV. Properties and units in clinical molecular genetics (IUPAC Technical Report). Pure and Applied Chemistry, 2018, 90, 1199-1220.	1.9	1
18	DNA hypermethylation and differential gene expression associated with Klinefelter syndrome. Scientific Reports, 2018, 8, 13740.	3.3	75

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19	Compound heterozygous mutations in two different domains of ALDH18A1 do not affect the amino acid levels in a patient with hereditary spastic paraplegia. Neurogenetics, 2018, 19, 145-149.	1.4	11
20	Ichthyosis with Confetti Inherited from a Mosaic Father. Acta Dermato-Venereologica, 2018, 98, 130-131.	1.3	3
21	Properties and units in the clinical laboratory sciences part XXIV. Properties and units in clinical molecular genetics (technical report 2017). Clinica Chimica Acta, 2018, 484, 122-131.	1.1	2
22	Moebius sequence â€“a multidisciplinary clinical approach. Orphanet Journal of Rare Diseases, 2017, 12, 4.	2.7	21
23	DOK7 congenital myasthenia may be associated with severe mitral valve insufficiency. Journal of the Neurological Sciences, 2017, 379, 217-218.	0.6	3
24	Chromosomal Aberrations in Monozygotic and Dizygotic Twins Versus Singletons in Denmark During 1968â€“2009. Twin Research and Human Genetics, 2017, 20, 216-225.	0.6	3
25	The Danish <scp>HD</scp> Registryâ€”a nationwide family registry of <scp>HD</scp> families in Denmark. Clinical Genetics, 2017, 92, 338-341.	2.0	7
26	The role of genes, intelligence, personality, and social engagement in cognitive performance in Klinefelter syndrome. Brain and Behavior, 2017, 7, e00645.	2.2	25
27	Severe fluoropyrimidine toxicity due to novel and rare DPYD missense mutations, deletion and genomic amplification affecting DPD activity and mRNA splicing. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 721-730.	3.8	32
28	A Gene Implicated in Activation of Retinoic Acid Receptor Targets Is a Novel Renal Agenesis Gene in Humans. Genetics, 2017, 207, 215-228.	2.9	62
29	The Association between Gender and Familial Prevalence of Hip Dysplasia in Danish Patients. HIP International, 2017, 27, 299-304.	1.7	8
30	X-Linked and Autosomal Recessive Alport Syndrome: Pathogenic Variant Features and Further Genotype-Phenotype Correlations. PLoS ONE, 2016, 11, e0161802.	2.5	75
31	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	2.5	54
32	Epilepsy and cataplexy in Angelman syndrome. Genotype-phenotype correlations. Research in Developmental Disabilities, 2016, 56, 177-182.	2.2	9
33	Re-Examination of Danish Carriers of Balanced Chromosomal Inversions. Cancer Genetics, 2016, 209, 231.	0.4	0
34	The first Danish family reported with an AQP5 mutation presenting diffuse non-epidermolytic palmoplantar keratoderma of Bothnian type, hyperhidrosis and frequent Corynebacterium infections: a case report. BMC Dermatology, 2016, 16, 7.	2.1	13
35	Klinefelter syndrome has increased brain responses to auditory stimuli and motor output, but not to visual stimuli or Stroop adaptation. NeuroImage: Clinical, 2016, 11, 239-251.	2.7	14
36	Odonto-onycho-dermal dysplasia in a patient homozygous for a WNT10A nonsense mutation and mild manifestations of ectodermal dysplasia in carriers of the mutation. BMC Dermatology, 2016, 16, 3.	2.1	10

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37	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	21.4	137
38	Anthropometry in Klinefelter Syndrome - Multifactorial Influences Due to CAG Length, Testosterone Treatment and Possibly Intrauterine Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E508-E517.	3.6	109
39	Novel 31.2â€‰kbÎ±⁰Deletion in a Palestinian Family withÎ±-Thalassemia. Hemoglobin, 2015, 39, 346-349.	0.8	4
40	Clinical utility gene card for: Alport syndrome â€“ update 2014. European Journal of Human Genetics, 2015, 23, 1269-1269.	2.8	47
41	Association of CHRDL1 Mutations and Variants with X-linked Megalocornea, NeuhÃuser Syndrome and Central Corneal Thickness. PLoS ONE, 2014, 9, e104163.	2.5	27
42	A Retrospective Study of Clinical and Mutational Findings in 45 Danish Families with Ectodermal Dysplasia. Acta Dermato-Venereologica, 2014, 94, 531-533.	1.3	10
43	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	2.9	19
44	Eating behavior, prenatal and postnatal growth in Angelman syndrome. Research in Developmental Disabilities, 2014, 35, 2681-2690.	2.2	18
45	Neuropsychology and brain morphology in Klinefelter syndrome â€“ the impact of genetics. Andrology, 2014, 2, 632-640.	3.5	36
46	Neurodevelopmental outcome in Angelman syndrome: Genotypeâ€“phenotype correlations. Research in Developmental Disabilities, 2014, 35, 1742-1747.	2.2	40
47	Neuroanatomical correlates of Klinefelter syndrome studied in relation to the neuropsychological profile. NeuroImage: Clinical, 2014, 4, 1-9.	2.7	59
48	Muscle regeneration and inflammation in patients with facioscapulohumeral muscular dystrophy. Acta Neurologica Scandinavica, 2013, 128, 194-201.	2.1	15
49	Angelman syndrome in Denmark. Birth incidence, genetic findings, and age at diagnosis. American Journal of Medical Genetics, Part A, 2013, 161, 2197-2203.	1.2	76
50	Quaternary Epitopes of Î±345(IV) Collagen Initiate Alport Post-Transplant Anti-GBM Nephritis. Journal of the American Society of Nephrology: JASN, 2013, 24, 889-895.	6.1	7
51	Renal phenotypic investigations of megalin-deficient patients: novel insights into tubular proteinuria and albumin filtration*. Nephrology Dialysis Transplantation, 2013, 28, 585-591.	0.7	71
52	Detailed investigations of proximal tubular function in Imerslund-GrÃsbeck syndrome. BMC Medical Genetics, 2013, 14, 111.	2.1	31
53	Clinical utility gene card for: Alport syndrome. European Journal of Human Genetics, 2012, 20, 713-713.	2.8	37
54	Best practice guidelines and recommendations on the molecular diagnosis of myotonic dystrophy types 1 and 2. European Journal of Human Genetics, 2012, 20, 1203-1208.	2.8	129

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55	Endocrine function in 97 patients with myotonic dystrophy type 1. Journal of Neurology, 2012, 259, 912-920.	3.6	50
56	Limited phenotypic variation of hypocalcified amelogenesis imperfecta in a Danish five-generation family with a novel FAM83H nonsense mutation. International Journal of Paediatric Dentistry, 2011, 21, 407-412.	1.8	16
57	A founder synonymous COL7A1 mutation in three Danish families with dominant dystrophic epidermolysis bullosa pruriginosa identifies exonic regulatory sequences required for exon 87 splicing. British Journal of Dermatology, 2011, 165, 678-682.	1.5	17
58	Genotype and phenotype in Klinefelter syndrome - impact of androgen receptor polymorphism and skewed X inactivation. Journal of Developmental and Physical Disabilities, 2011, 34, e642-e648.	3.6	55
59	Establishment of a pig fibroblast-derived cell line for locus-directed transgene expression in cell cultures and blastocysts. Molecular Biology Reports, 2011, 38, 151-161.	2.3	6
60	A Patient with Cubilin Deficiency. New England Journal of Medicine, 2011, 364, 89-91.	27.0	59
61	Report of a cubilin deficient patient. FASEB Journal, 2011, 25, 1002.12.	0.5	0
62	Aarskogâ€“Scott syndrome: Clinical update and report of nine novel mutations of the FGD1 gene. American Journal of Medical Genetics, Part A, 2010, 152A, 313-318.	1.2	46
63	Skewed X-chromosome inactivation causing diagnostic misinterpretation in congenital nephrogenic diabetes insipidus. Scandinavian Journal of Urology and Nephrology, 2010, 44, 324-330.	1.4	17
64	Pallisterâ€“Killian syndrome in a girl with mild developmental delay and mosaicism for hexasomy 12p. American Journal of Medical Genetics, Part A, 2009, 149A, 510-514.	1.2	16
65	Hypocalcified type of amelogenesis imperfecta in a large family: clinical, radiographic, and histological findings, associated dento-facial anomalies, and resulting treatment load. Acta Odontologica Scandinavica, 2009, 67, 240-247.	1.6	11
66	Alport syndrome. Molecular genetic aspects. Danish Medical Bulletin, 2009, 56, 105-52.	0.3	14
67	Testing for 22q11 microdeletion in 146 fetuses with nuchal translucency above the 99th percentile and a normal karyotype. Acta Obstetricia Et Gynecologica Scandinavica, 2008, 87, 1252-1255.	2.8	18
68	Xâ€linked hypohidrotic ectodermal dysplasia. Genetic and dental findings in 67 Danish patients from 19 families. Clinical Genetics, 2008, 74, 252-259.	2.0	49
69	MLPA and cDNA analysis improves COL4A5 mutation detection in Xâ€linked Alport syndrome. Clinical Genetics, 2008, 74, 522-530.	2.0	11
70	A novel missense mutation (G43S) in the switch I region of Rab27A causing Griscelli syndrome. Molecular Genetics and Metabolism, 2008, 94, 248-254.	1.1	25
71	Non-disjunction of chromosome 13. Human Molecular Genetics, 2007, 16, 2004-2010.	2.9	38
72	The Variant inv(2)(p11.2q13) Is a Genuinely Recurrent Rearrangement but Displays Some Breakpoint Heterogeneity. American Journal of Human Genetics, 2007, 81, 847-856.	6.2	13

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73	Anomalies of tooth formation in hypohidrotic ectodermal dysplasia. International Journal of Paediatric Dentistry, 2007, 17, 10-18.	1.8	87
74	Whole saliva in X-linked hypohidrotic ectodermal dysplasia. International Journal of Paediatric Dentistry, 2007, 17, 155-162.	1.8	30
75	Anthropometric and cephalometric measurements in Xâ€linked hypohidrotic ectodermal dysplasia. Orthodontics and Craniofacial Research, 2007, 10, 203-215.	2.8	27
76	N.P.2 05 A previously unreported non-sense mutation in SMN1 causes spinal muscular atrophy. Neuromuscular Disorders, 2006, 16, 653.	0.6	0
77	Low frequency of Parkin, Tyrosine Hydroxylase, and GTP Cyclohydrolase I gene mutations in a Danish population of early-onset Parkinson's Disease. European Journal of Neurology, 2006, 13, 385-390.	3.3	37
78	Dopa-responsive dystonia and early-onset Parkinson's disease in a patient with GTP cyclohydrolase I deficiency?. Movement Disorders, 2006, 21, 679-682.	3.9	33
79	Trisomy 13 due to rea(13q;13q) is caused by i(13) and not rob(13;13)(q10;q10) in the majority of cases. , 2005, 132A, 310-313.		9
80	Alport syndrome caused by inversion of a 21ÂMb fragment of the long arm of the X-chromosome comprising exon 9 through 51 of the COL4A5 gene. Human Genetics, 2005, 118, 23-28.	3.8	8
81	LGMD2I presenting with a characteristic Duchenne or Becker muscular dystrophy phenotype. Neurology, 2005, 64, 1635-1637.	1.1	60
82	Early onset, non-progressive, mild cerebellar ataxia co-segregating with a familial balanced translocation t(8;20)(p22;q13). Journal of Medical Genetics, 2004, 41, 25e-25.	3.2	5
83	Patients with Goodpasture's disease have two normal COL4A3 alleles encoding the NC1 domain of the type IV collagen Â3 chain. Nephrology Dialysis Transplantation, 2004, 19, 2030-2035.	0.7	16
84	X-Linked Alport Syndrome. Journal of the American Society of Nephrology: JASN, 2003, 14, 2603-2610.	6.1	394
85	A novel mutation (R218Q) at the boundary between the N-terminal and the first transmembrane domain of the glycine receptor in a case of sporadic hyperekplexia. Journal of Medical Genetics, 2003, 40, 71e-71.	3.2	18
86	Detection of mutations in theCOL4A5gene by SSCP in X-linked Alport syndrome. Human Mutation, 2001, 18, 141-148.	2.5	38
87	A new locus for Seckel syndrome on chromosome 18p11.31-q11.2. European Journal of Human Genetics, 2001, 9, 753-757.	2.8	54
88	Cathepsin K gene mutations and 1q21Âhaplotypes in patients with pycnodysostosis in an outbred population. European Journal of Human Genetics, 2000, 8, 431-436.	2.8	24
89	X-linked Alport Syndrome. Journal of the American Society of Nephrology: JASN, 2000, 11, 649-657.	6.1	455
90	Origin of nondisjunction in trisomy 8 and trisomy 8 mosaicism. European Journal of Human Genetics, 1998, 6, 432-438.	2.8	59

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91	Non-disjunction of chromosome 18. Human Molecular Genetics, 1998, 7, 661-669.	2.9	115
92	A novel missense mutation (402C â†' T) in exon 1 in the EDA gene in a family with Xâ€linked hypohidrotic ectodermal dysplasia. Clinical Genetics, 1998, 53, 205-209.	2.0	18
93	Apolipoprotein E alleles in mothers of trisomy 18 conceptuses. Clinical Genetics, 1998, 53, 321-322.	2.0	2
94	Quantification, by solid-phase minisequencing, of the telomeric and centromeric copies of the survival motor neuron gene in families with spinal muscular atrophy. Human Molecular Genetics, 1997, 6, 99-104.	2.9	32
95	A nonsense mutation in the COL4A5 collagen gene in a family with X-linked juvenile Alport syndrome. Kidney International, 1995, 47, 327-332.	5.2	12
96	Mutations in the codon for a conserved arginine-1563 in the COL4A5 collagen gene in Alport syndrome. Kidney International, 1993, 43, 722-729.	5.2	27
97	Genetic analysis of repeated, biparental, diploid, hydatidiform moles. Cancer Genetics and Cytogenetics, 1993, 66, 16-22.	1.0	35
98	Ring chromosome 13: lack of distinct syndromes based on different breakpoints on 13q.. Journal of Medical Genetics, 1992, 29, 704-708.	3.2	23
99	Prenatal cytogenetic diagnosis after transabdominal chorionic villus sampling in the first trimester. Prenatal Diagnosis, 1988, 8, 19-31.	2.3	17
100	Admixture of maternal metaphases in first trimester direct chromosome preparations?. Prenatal Diagnosis, 1986, 6, 383-385.	2.3	4