

Brage Storstein Andresen

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

142
papers

5,393
citations

41
h-index

67
g-index

153
ext. papers

5,997
ext. citations

5.2
avg, IF

4.75
L-index

#	Paper	IF	Citations
142	-targeting miR-agshRNAs combine efficacy with specificity and safety for retinal gene therapy.. <i>Molecular Therapy - Nucleic Acids</i> , 2022 , 28, 58-76	10.7	0
141	Pseudoexon activation in disease by non-splice site deep intronic sequence variation - wild type pseudoexons constitute high-risk sites in the human genome. <i>Human Mutation</i> , 2021 , 43, 103	4.7	0
140	Use of Molecular Genetic Analyses in Danish Routine Newborn Screening. <i>International Journal of Neonatal Screening</i> , 2021 , 7,	2.6	3
139	Identification of SRSF10 as a regulator of SMN2 ISS-N1. <i>Human Mutation</i> , 2021 , 42, 246-260	4.7	3
138	DeepCLIP: predicting the effect of mutations on protein-RNA binding with deep learning. <i>Nucleic Acids Research</i> , 2020 , 48, 7099-7118	20.1	22
137	Down-Regulation of CK2 β Leads to Up-Regulation of the Cyclin-Dependent Kinase Inhibitor p27 in Conditions Unfavorable for the Growth of Myoblast Cells. <i>Cellular Physiology and Biochemistry</i> , 2020 , 54, 1177-1198	3.9	1
136	A pathogenic haplotype, common in Europeans, causes autosomal recessive albinism and uncovers missing heritability in OCA1. <i>Scientific Reports</i> , 2019 , 9, 645	4.9	16
135	Down-regulation of CK2 β correlates with decreased expression levels of DNA replication minichromosome maintenance protein complex (MCM) genes. <i>Scientific Reports</i> , 2019 , 9, 14581	4.9	0
134	Next generation sequencing of RNA reveals novel targets of resveratrol with possible implications for Canavan disease. <i>Molecular Genetics and Metabolism</i> , 2019 , 126, 64-76	3.7	10
133	Intronic PAH gene mutations cause a splicing defect by a novel mechanism involving U1snRNP binding downstream of the 5' splice site. <i>PLoS Genetics</i> , 2018 , 14, e1007360	6	15
132	Blocking of an intronic splicing silencer completely rescues IKBKAP exon 20 splicing in familial dysautonomia patient cells. <i>Nucleic Acids Research</i> , 2018 , 46, 7938-7952	20.1	9
131	Long-term outcome of isobutyryl-CoA dehydrogenase deficiency diagnosed following an episode of ketotic hypoglycaemia. <i>Molecular Genetics and Metabolism Reports</i> , 2017 , 10, 28-30	1.8	10
130	Splicing factor 1 modulates dietary restriction and TORC1 pathway longevity in <i>C. elegans</i> . <i>Nature</i> , 2017 , 541, 102-106	50.4	102
129	An intronic variation in SLC52A1 causes exon skipping and transient riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. <i>Molecular Genetics and Metabolism</i> , 2017 , 122, 182-188	3.7	30
128	RNA-sequencing of a mouse-model of spinal muscular atrophy reveals tissue-wide changes in splicing of U12-dependent introns. <i>Nucleic Acids Research</i> , 2017 , 45, 395-416	20.1	63
127	Fibroblast Fatty-Acid Oxidation Flux Assays Stratify Risk in Newborns with Presumptive-Positive Results on Screening for Very-Long Chain Acyl-CoA Dehydrogenase Deficiency. <i>International Journal of Neonatal Screening</i> , 2017 , 3, 2	2.6	5
126	Mitochondrial trifunctional protein deficiency in human cultured fibroblasts: effects of bezafibrate. <i>Journal of Inherited Metabolic Disease</i> , 2016 , 39, 47-58	5.4	24

125	VLCAD deficiency: Follow-up and outcome of patients diagnosed through newborn screening in Victoria. <i>Molecular Genetics and Metabolism</i> , 2016 , 118, 282-7	3.7	21
124	Global identification of hnRNP A1 binding sites for SSO-based splicing modulation. <i>BMC Biology</i> , 2016 , 14, 54	7.3	41
123	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. <i>PLoS Genetics</i> , 2016 , 12, e1006039	6	11
122	The prevalent deep intronic c. 639+919 G>A GLA mutation causes pseudoexon activation and Fabry disease by abolishing the binding of hnRNPA1 and hnRNP A2/B1 to a splicing silencer. <i>Molecular Genetics and Metabolism</i> , 2016 , 119, 258-269	3.7	13
121	Abnormal Newborn Screening in a Healthy Infant of a Mother with Undiagnosed Medium-Chain Acyl-CoA Dehydrogenase Deficiency. <i>JIMD Reports</i> , 2015 , 23, 67-70	1.9	6
120	Splice-shifting oligonucleotide (SSO) mediated blocking of an exonic splicing enhancer (ESE) created by the prevalent c.903+469T>C MTRR mutation corrects splicing and restores enzyme activity in patient cells. <i>Nucleic Acids Research</i> , 2015 , 43, 4627-39	20.1	20
119	Absence of an intron splicing silencer in porcine Smn1 intron 7 confers immunity to the exon skipping mutation in human SMN2. <i>PLoS ONE</i> , 2014 , 9, e98841	3.7	3
118	The ETFDH c.158A>G variation disrupts the balanced interplay of ESE- and ESS-binding proteins thereby causing missplicing and multiple Acyl-CoA dehydrogenation deficiency. <i>Human Mutation</i> , 2014 , 35, 86-95	4.7	22
117	Neurodevelopmental profiles of children with very long chain acyl-CoA dehydrogenase deficiency diagnosed by newborn screening. <i>Molecular Genetics and Metabolism</i> , 2014 , 113, 278-82	3.7	10
116	A synonymous polymorphic variation in ACADM exon 11 affects splicing efficiency and may affect fatty acid oxidation. <i>Molecular Genetics and Metabolism</i> , 2013 , 110, 122-8	3.7	18
115	A Large Intragenic Deletion in the ACADM Gene Can Cause MCAD Deficiency but is not Detected on Routine Sequencing. <i>JIMD Reports</i> , 2013 , 11, 13-6	1.9	1
114	Identification of six novel PTH1R mutations in families with a history of primary failure of tooth eruption. <i>PLoS ONE</i> , 2013 , 8, e74601	3.7	35
113	Biochemical screening of 504,049 newborns in Denmark, the Faroe Islands and Greenland--experience and development of a routine program for expanded newborn screening. <i>Molecular Genetics and Metabolism</i> , 2012 , 107, 281-93	3.7	47
112	Splicing of phenylalanine hydroxylase (PAH) exon 11 is vulnerable: molecular pathology of mutations in PAH exon 11. <i>Molecular Genetics and Metabolism</i> , 2012 , 106, 403-11	3.7	16
111	MCAD deficiency in Denmark. <i>Molecular Genetics and Metabolism</i> , 2012 , 106, 175-88	3.7	26
110	Normal Levels of Plasma Free Carnitine and Acylcarnitines in Follow-Up Samples from a Presymptomatic Case of Carnitine Palmitoyl Transferase 1 (CPT1) Deficiency Detected Through Newborn Screening in Denmark. <i>JIMD Reports</i> , 2012 , 3, 11-5	1.9	7
109	CUGBP1 and MBNL1 preferentially bind to 3QTRs and facilitate mRNA decay. <i>Scientific Reports</i> , 2012 , 2, 209	4.9	120
108	Urgent metabolic service improves survival in long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency detected by symptomatic identification and pilot newborn screening. <i>Journal of Inherited Metabolic Disease</i> , 2011 , 34, 185-95	5.4	23

107	SMN2 exon 7 splicing is inhibited by binding of hnRNP A1 to a common ESS motif that spans the 3Q splice site. <i>Human Mutation</i> , 2011 , 32, 220-30	4.7	29
106	Relevance of expanded neonatal screening of medium-chain acyl co-a dehydrogenase deficiency: outcome of a decade in galicia (Spain). <i>JIMD Reports</i> , 2011 , 1, 131-6	1.9	2
105	Newborn screening for medium chain acyl-CoA dehydrogenase deficiency in England: prevalence, predictive value and test validity based on 1.5 million screened babies. <i>Journal of Medical Screening</i> , 2011 , 18, 173-81	1.4	30
104	A novel mutation of the ACADM gene (c.145C>G) associated with the common c.985A>G mutation on the other ACADM allele causes mild MCAD deficiency: a case report. <i>Orphanet Journal of Rare Diseases</i> , 2010 , 5, 26	4.2	11
103	A comprehensive HADHA c.1528G>C frequency study reveals high prevalence of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency in Poland. <i>Journal of Inherited Metabolic Disease</i> , 2010 , 33 Suppl 3, S373-7	5.4	40
102	The phenylalanine hydroxylase c.30C>G synonymous variation (p.G10G) creates a common exonic splicing silencer. <i>Molecular Genetics and Metabolism</i> , 2010 , 100, 316-23	3.7	19
101	High-resolution melting analysis, a simple and effective method for reliable mutation scanning and frequency studies in the ACADVL gene. <i>Journal of Inherited Metabolic Disease</i> , 2010 , 33, 247-60	5.4	8
100	The deep intronic c.903+469T>C mutation in the MTRR gene creates an SF2/ASF binding exonic splicing enhancer, which leads to pseudoexon activation and causes the cbIE type of homocystinuria. <i>Human Mutation</i> , 2010 , 31, 437-44	4.7	48
99	Diagnostic assessment and long-term follow-up of 13 patients with Very Long-Chain Acyl-Coenzyme A dehydrogenase (VLCAD) deficiency. <i>Neuromuscular Disorders</i> , 2009 , 19, 324-9	2.9	62
98	2-Methylbutyryl-coenzyme A dehydrogenase deficiency: functional and molecular studies on a defect in isoleucine catabolism. <i>Molecular Genetics and Metabolism</i> , 2008 , 93, 30-5	3.7	33
97	Ethnicity of children with homozygous c.985A>G medium-chain acyl-CoA dehydrogenase deficiency: findings from screening approximately 1.1 million newborn infants. <i>Journal of Medical Screening</i> , 2008 , 15, 112-7	1.4	16
96	Newborn screening for MCAD deficiency: experience of the first three years in British Columbia, Canada. <i>Canadian Journal of Public Health</i> , 2008 , 99, 276-80	3.2	17
95	Mitochondrial fatty acid oxidation defects--remaining challenges. <i>Journal of Inherited Metabolic Disease</i> , 2008 , 31, 643-57	5.4	107
94	The ACADS gene variation spectrum in 114 patients with short-chain acyl-CoA dehydrogenase (SCAD) deficiency is dominated by missense variations leading to protein misfolding at the cellular level. <i>Human Genetics</i> , 2008 , 124, 43-56	6.3	84
93	Interstitial deletion of 1p22.2p31.1 and medium-chain acyl-CoA dehydrogenase deficiency in a patient with global developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 1581-6	2.5	10
92	Features of 5Q splice-site efficiency derived from disease-causing mutations and comparative genomics. <i>Genome Research</i> , 2008 , 18, 77-87	9.7	56
91	ETFDH mutations as a major cause of riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. <i>Brain</i> , 2007 , 130, 2045-54	11.2	238
90	Seemingly neutral polymorphic variants may confer immunity to splicing-inactivating mutations: a synonymous SNP in exon 5 of MCAD protects from deleterious mutations in a flanking exonic splicing enhancer. <i>American Journal of Human Genetics</i> , 2007 , 80, 416-32	11	125

89	Genetic basis for correction of very-long-chain acyl-coenzyme A dehydrogenase deficiency by bezafibrate in patient fibroblasts: toward a genotype-based therapy. <i>American Journal of Human Genetics</i> , 2007 , 81, 1133-43	11	57
88	2-methylbutyryl-CoA dehydrogenase deficiency associated with autism and mental retardation: a case report. <i>Journal of Medical Case Reports</i> , 2007 , 1, 98	1.2	15
87	Variations in IBD (ACAD8) in children with elevated C4-carnitine detected by tandem mass spectrometry newborn screening. <i>Pediatric Research</i> , 2006 , 60, 315-20	3.2	28
86	Medium-chain acyl-CoA dehydrogenase deficiency: genotype-biochemical phenotype correlations. <i>Molecular Genetics and Metabolism</i> , 2006 , 87, 32-9	3.7	72
85	VLCAD deficiency: pitfalls in newborn screening and confirmation of diagnosis by mutation analysis. <i>Molecular Genetics and Metabolism</i> , 2006 , 88, 166-70	3.7	75
84	TAT gene mutation analysis in three Palestinian kindreds with oculocutaneous tyrosinaemia type II; characterization of a silent exonic transversion that causes complete missplicing by exon 11 skipping. <i>Journal of Inherited Metabolic Disease</i> , 2006 , 29, 620-6	5.4	18
83	Short/branched-chain acyl-CoA dehydrogenase deficiency due to an IVS3+3A>G mutation that causes exon skipping. <i>Human Genetics</i> , 2006 , 118, 680-90	6.3	31
82	Two novel variants of human medium chain acyl-CoA dehydrogenase (MCAD). K364R, a folding mutation, and R256T, a catalytic-site mutation resulting in a well-folded but totally inactive protein. <i>FEBS Journal</i> , 2005 , 272, 4549-57	5.7	6
81	Biochemical, clinical and molecular findings in LCHAD and general mitochondrial trifunctional protein deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2005 , 28, 533-44	5.4	55
80	DNA-based prenatal diagnosis for severe and variant forms of multiple acyl-CoA dehydrogenation deficiency. <i>Prenatal Diagnosis</i> , 2005 , 25, 60-4	3.2	9
79	2-ethylhydracrylic aciduria in short/branched-chain acyl-CoA dehydrogenase deficiency: application to diagnosis and implications for the R-pathway of isoleucine oxidation. <i>Clinical Chemistry</i> , 2005 , 51, 610-7 ⁵	5.5	28
78	The Y42H mutation in medium-chain acyl-CoA dehydrogenase, which is prevalent in babies identified by MS/MS-based newborn screening, is temperature sensitive. <i>FEBS Journal</i> , 2004 , 271, 4053-63		24
77	Lipid-storage myopathy and respiratory insufficiency due to ETFQO mutations in a patient with late-onset multiple acyl-CoA dehydrogenation deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2004 , 27, 671-8	5.4	23
76	Genetic defects in fatty acid beta-oxidation and acyl-CoA dehydrogenases. Molecular pathogenesis and genotype-phenotype relationships. <i>FEBS Journal</i> , 2004 , 271, 470-82		77
75	Homozygosity for a severe novel medium-chain acyl-CoA dehydrogenase (MCAD) mutation IVS3-1G > C that leads to introduction of a premature termination codon by complete missplicing of the MCAD mRNA and is associated with phenotypic diversity ranging from sudden neonatal death to asymptomatic status. <i>Molecular Genetics and Metabolism</i> , 2004 , 82, 121-9	3.7	22
74	Prospective diagnosis of 2-methylbutyryl-CoA dehydrogenase deficiency in the Hmong population by newborn screening using tandem mass spectrometry. <i>Pediatrics</i> , 2003 , 112, 74-8	7.4	53
73	Mutation and biochemical analysis in carnitine palmitoyltransferase type II (CPT II) deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2003 , 26, 543-57	5.4	39
72	Clear relationship between ETF/ETFHDH genotype and phenotype in patients with multiple acyl-CoA dehydrogenation deficiency. <i>Human Mutation</i> , 2003 , 22, 12-23	4.7	167

71	Functional testing of keratin 14 mutant proteins associated with the three major subtypes of epidermolysis bullosa simplex. <i>Experimental Dermatology</i> , 2003 , 12, 472-9	4	12
70	Late-onset form of beta-electron transfer flavoprotein deficiency. <i>Molecular Genetics and Metabolism</i> , 2003 , 78, 247-9	3-7	29
69	Identification of isobutyryl-CoA dehydrogenase and its deficiency in humans. <i>Molecular Genetics and Metabolism</i> , 2002 , 77, 68-79	3-7	52
68	Mutation analysis in mitochondrial fatty acid oxidation defects: Exemplified by acyl-CoA dehydrogenase deficiencies, with special focus on genotype-phenotype relationship. <i>Human Mutation</i> , 2001 , 18, 169-89	4-7	159
67	The role of chaperone-assisted folding and quality control in inborn errors of metabolism: protein folding disorders. <i>Journal of Inherited Metabolic Disease</i> , 2001 , 24, 189-212	5-4	52
66	A severe genotype with favourable outcome in very long chain acyl-CoA dehydrogenase deficiency. <i>Archives of Disease in Childhood</i> , 2001 , 84, 58-60	2-2	21
65	Medium-chain acyl-CoA dehydrogenase (MCAD) mutations identified by MS/MS-based prospective screening of newborns differ from those observed in patients with clinical symptoms: identification and characterization of a new, prevalent mutation that results in mild MCAD deficiency. <i>American Journal of Human Genetics</i> , 2001 , 68, 1408-18	11	191
64	Mutation analysis in mitochondrial fatty acid oxidation defects: Exemplified by acyl-CoA dehydrogenase deficiencies, with special focus on genotype-phenotype relationship 2001 , 18, 169		3
63	Medium Chain Acyl-CoA Dehydrogenase Deficiency. <i>Biochemical Society Transactions</i> , 2000 , 28, A73-A73	5-1	
62	Properties of five rare human mutations causing medium chain acyl-CoA dehydrogenase deficiency. <i>Biochemical Society Transactions</i> , 2000 , 28, A160-A160	5-1	
61	Analysis of the Epstein-Barr virus (EBV) latent membrane protein 1 (LMP-1) gene and promoter in Hodgkin disease isolates: selection against EBV variants with mutations in the LMP-1 promoter ATF-1/CREB-1 binding site. <i>Journal of Clinical Pathology</i> , 2000 , 53, 280-8		13
60	Very-long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency: monitoring of treatment by carnitine/acylcarnitine analysis in blood spots. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2000 , 89, 492-495	3-1	11
59	Defective folding and rapid degradation of mutant proteins is a common disease mechanism in genetic disorders. <i>Journal of Inherited Metabolic Disease</i> , 2000 , 23, 441-7	5-4	48
58	Characterization of mouse Clpp protease cDNA, gene, and protein. <i>Mammalian Genome</i> , 2000 , 11, 275-89	2	5
57	Human and mouse mitochondrial orthologs of bacterial ClpX. <i>Mammalian Genome</i> , 2000 , 11, 899-905	3-2	35
56	Isolated 2-methylbutyrylglycinuria caused by short/branched-chain acyl-CoA dehydrogenase deficiency: identification of a new enzyme defect, resolution of its molecular basis, and evidence for distinct acyl-CoA dehydrogenases in isoleucine and valine metabolism. <i>American Journal of Human Genetics</i> , 2000 , 67, 1095-103	11	61
55	Prevalent mutations in fatty acid oxidation disorders: diagnostic considerations. <i>European Journal of Pediatrics</i> , 2000 , 159 Suppl 3, S213-8	4-1	32
54	Very-long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency: monitoring of treatment by carnitine/acylcarnitine analysis in blood spots. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2000 , 89, 492-5	3-1	6

53	Biochemical characterisation of mutations of human medium-chain acyl-CoA dehydrogenase. <i>Advances in Experimental Medicine and Biology</i> , 1999 , 466, 387-93	3.6	1
52	Identification of novel and known mutations in the genes for keratin 5 and 14 in Danish patients with epidermolysis bullosa simplex: correlation between genotype and phenotype. <i>Journal of Investigative Dermatology</i> , 1999 , 112, 184-90	4.3	51
51	Adolescent myopathic presentation in two sisters with very long-chain acyl-CoA dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 1999 , 22, 802-10	5.4	20
50	DNA-based prenatal diagnosis for very-long-chain acyl-CoA dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 1999 , 22, 281-5	5.4	8
49	Protein misfolding and degradation in genetic diseases. <i>Human Mutation</i> , 1999 , 14, 186-98	4.7	151
48	Clear correlation of genotype with disease phenotype in very-long-chain acyl-CoA dehydrogenase deficiency. <i>American Journal of Human Genetics</i> , 1999 , 64, 479-94	11	248
47	Myopathy in very-long-chain acyl-CoA dehydrogenase deficiency: clinical and biochemical differences with the fatal cardiac phenotype. <i>Neuromuscular Disorders</i> , 1999 , 9, 313-9	2.9	22
46	A polymorphic variant in the human electron transfer flavoprotein alpha-chain (alpha-T171) displays decreased thermal stability and is overrepresented in very-long-chain acyl-CoA dehydrogenase-deficient patients with mild childhood presentation. <i>Molecular Genetics and Metabolism</i> , 1999 , 67, 188-17	3.7	20
45	Clinical and Molecular Evidence of Abnormal Processing and Trafficking of the Vasopressin Preprohormone in a Large Kindred with Familial Neurohypophyseal Diabetes Insipidus due to A Signal Peptide Mutation. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999 , 84, 2933-2941	5.6	37
44	Impaired folding and subunit assembly as disease mechanism: the example of medium-chain acyl-CoA dehydrogenase deficiency. <i>Progress in Molecular Biology and Translational Science</i> , 1998 , 58, 301-37		9
43	Very long chain acyl-coenzyme A dehydrogenase deficiency with adult onset. <i>Annals of Neurology</i> , 1998 , 43, 540-4	9.4	58
42	Identification of four new mutations in the short-chain acyl-CoA dehydrogenase (SCAD) gene in two patients: one of the variant alleles, 511C-->T, is present at an unexpectedly high frequency in the general population, as was the case for 625G-->A, together conferring susceptibility to childhood myopathy. <i>Human Molecular Genetics</i> , 1998 , 7, 418-27	5.6	91
41	Mutations of human medium-chain acyl-CoA dehydrogenase. <i>Biochemical Society Transactions</i> , 1998 , 26, S65	5.1	1
40	The molecular basis of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency in compound heterozygous patients: is there correlation between genotype and phenotype?. <i>Human Molecular Genetics</i> , 1997 , 6, 695-707	5.6	107
39	A common W556S mutation in the LDL receptor gene of Danish patients with familial hypercholesterolemia encodes a transport-defective protein. <i>Atherosclerosis</i> , 1997 , 131, 67-72	3.1	28
38	Sequence Analysis of the Epstein-Barr Virus (EBV) Latent Membrane Protein-1 Gene and Promoter Region: Identification of Four Variants Among Wild-Type EBV Isolates. <i>Blood</i> , 1997 , 90, 323-330	2.2	81
37	Structural organization of the human short-chain acyl-CoA dehydrogenase gene. <i>Mammalian Genome</i> , 1997 , 8, 922-6	3.2	38
36	Two mutations in the same low-density lipoprotein receptor allele act in synergy to reduce receptor function in heterozygous familial hypercholesterolemia. <i>Human Mutation</i> , 1997 , 9, 437-44	4.7	22

35	Sequence Analysis of the Epstein-Barr Virus (EBV) Latent Membrane Protein-1 Gene and Promoter Region: Identification of Four Variants Among Wild-Type EBV Isolates. <i>Blood</i> , 1997 , 90, 323-330	2.2	19
34	Two mutations in the same low-density lipoprotein receptor allele act in synergy to reduce receptor function in heterozygous familial hypercholesterolemia 1997 , 9, 437		1
33	Sequence analysis of the Epstein-Barr virus (EBV) latent membrane protein-1 gene and promoter region: identification of four variants among wild-type EBV isolates. <i>Blood</i> , 1997 , 90, 323-30	2.2	30
32	The mutational spectrum in very long-chain acyl-CoA dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 1996 , 19, 169-72	5.4	36
31	Cloning and characterization of human very-long-chain acyl-CoA dehydrogenase cDNA, chromosomal assignment of the gene and identification in four patients of nine different mutations within the VLCAD gene. <i>Human Molecular Genetics</i> , 1996 , 5, 461-72	5.6	92
30	Ethylmalonic aciduria is associated with an amino acid variant of short chain acyl-coenzyme A dehydrogenase. <i>Pediatric Research</i> , 1996 , 39, 1059-66	3.2	73
29	Prenatal diagnosis of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency in a family with a previous fatal case of sudden unexpected death in childhood. <i>Prenatal Diagnosis</i> , 1995 , 15, 82-6	3.2	22
28	Effects of two mutations detected in medium chain acyl-CoA dehydrogenase (MCAD)-deficient patients on folding, oligomer assembly, and stability of MCAD enzyme. <i>Journal of Biological Chemistry</i> , 1995 , 270, 10284-90	5.4	69
27	Human ClpP protease: cDNA sequence, tissue-specific expression and chromosomal assignment of the gene. <i>FEBS Letters</i> , 1995 , 377, 249-52	3.8	39
26	Comparison between medium-chain acyl-CoA dehydrogenase mutant proteins overexpressed in bacterial and mammalian cells. <i>Human Mutation</i> , 1995 , 6, 226-31	4.7	20
25	Molecular diagnosis and characterization of medium-chain acyl-CoA dehydrogenase deficiency. <i>Scandinavian Journal of Clinical and Laboratory Investigation, Supplement</i> , 1995 , 220, 9-25		1
24	Identification of potential hot spots in the carboxy-terminal part of the Epstein-Barr virus (EBV) BNLF-1 gene in both malignant and benign EBV-associated diseases: high frequency of a 30-bp deletion in Malaysian and Danish peripheral T-cell lymphomas. <i>Blood</i> , 1994 , 84, 4053-4060	2.2	98
23	Amino acid polymorphism (Gly209Ser) in the ACADS gene. <i>Human Molecular Genetics</i> , 1994 , 3, 1711	5.6	14
22	Molecular genetic characterization and urinary excretion pattern of metabolites in two families with MCAD deficiency due to compound heterozygosity with a 13 base pair insertion in one allele. <i>Journal of Inherited Metabolic Disease</i> , 1994 , 17, 169-84	5.4	9
21	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency due to heterozygosity for the common mutation and an allele resulting in low levels of MCAD mRNA. <i>Journal of Inherited Metabolic Disease</i> , 1994 , 17, 275-8	5.4	1
20	Characterization of a disease-causing Glu119-Lys mutation in the low-density lipoprotein receptor gene in two Danish families with heterozygous familial hypercholesterolemia. <i>Human Mutation</i> , 1994 , 4, 102-13	4.7	24
19	Characterization of wild-type human medium-chain acyl-CoA dehydrogenase (MCAD) and mutant enzymes present in MCAD-deficient patients by two-dimensional gel electrophoresis: evidence for post-translational modification of the enzyme. <i>Biochemical Medicine and Metabolic Biology</i> , 1994 , 52, 36-44		10
18	Identification of potential hot spots in the carboxy-terminal part of the Epstein-Barr virus (EBV) BNLF-1 gene in both malignant and benign EBV-associated diseases: high frequency of a 30-bp deletion in Malaysian and Danish peripheral T-cell lymphomas. <i>Blood</i> , 1994 , 84, 4053-60	2.2	24

17	Disease-causing mutations in exon 11 of the medium-chain acyl-CoA dehydrogenase gene. <i>American Journal of Human Genetics</i> , 1994 , 54, 975-88	11	20
16	Co-overexpression of bacterial GroESL chaperonins partly overcomes non-productive folding and tetramer assembly of E. coli-expressed human medium-chain acyl-CoA dehydrogenase (MCAD) carrying the prevalent disease-causing K304E mutation. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1993 , 1182, 264-74	6.9	54
15	A silent A to G mutation in exon 11 of the medium-chain acyl-CoA dehydrogenase (MCAD) gene. <i>Human Molecular Genetics</i> , 1993 , 2, 488	5.6	9
14	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency: the prevalent mutation G985 (K304E) is subject to a strong founder effect from northwestern Europe. <i>Human Heredity</i> , 1993 , 43, 342-50	1.1	65
13	A rare disease-associated mutation in the medium-chain acyl-CoA dehydrogenase (MCAD) gene changes a conserved arginine, previously shown to be functionally essential in short-chain acyl-CoA dehydrogenase (SCAD). <i>American Journal of Human Genetics</i> , 1993 , 53, 730-9	11	32
12	Expression of wild-type and mutant medium-chain acyl-CoA dehydrogenase (MCAD) cDNA in eucaryotic cells. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1992 , 1180, 65-72	6.9	23
11	Internalization, lysosomal degradation and new synthesis of surface membrane CD4 in phorbol ester-activated T-lymphocytes and U-937 cells. <i>Experimental Cell Research</i> , 1992 , 201, 160-73	4.2	74
10	Two Novel Nonradioactive Polymerase Chain Reaction-Based Assays of Dried Blood Spots, Genomic DNA, or Whole Cells for Fast, Reliable Detection of Z and S Mutations in the α -Antitrypsin Gene. <i>Clinical Chemistry</i> , 1992 , 38, 2100-2107	5.5	28
9	Mutations in the medium chain acyl-CoA dehydrogenase (MCAD) gene. <i>Human Mutation</i> , 1992 , 1, 271-9	4.7	77
8	Two novel nonradioactive polymerase chain reaction-based assays of dried blood spots, genomic DNA, or whole cells for fast, reliable detection of Z and S mutations in the alpha 1-antitrypsin gene. <i>Clinical Chemistry</i> , 1992 , 38, 2100-7	5.5	9
7	Molecular analysis of medium-chain acyl-CoA dehydrogenase deficiency: a diagnostic approach. <i>Progress in Clinical and Biological Research</i> , 1992 , 375, 441-52		1
6	Characterization of a disease-causing Lys329 to Glu mutation in 16 patients with medium-chain acyl-CoA dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 1991 , 14, 314-6	5.4	10
5	The most common mutation causing medium-chain acyl-CoA dehydrogenase deficiency is strongly associated with a particular haplotype in the region of the gene. <i>Human Genetics</i> , 1991 , 87, 425-8	6.3	21
4	Molecular characterization of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency: identification of a lys329 to glu mutation in the MCAD gene, and expression of inactive mutant enzyme protein in E. coli. <i>Human Genetics</i> , 1991 , 86, 545-51	6.3	55
3	Specific diagnosis of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency in dried blood spots by a polymerase chain reaction (PCR) assay detecting a point-mutation (G985) in the MCAD gene. <i>Clinica Chimica Acta</i> , 1991 , 203, 23-34	6.2	72
2	Surface membrane CD4 turnover in phorbol ester stimulated T-lymphocytes. Evidence of degradation and increased synthesis. <i>FEBS Letters</i> , 1990 , 276, 59-62	3.8	18
1	DeepCLIP: Predicting the effect of mutations on protein-RNA binding with Deep Learning		2