Brage Storstein Andresen

List of Publications by Citations

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	Clear correlation of genotype with disease phenotype in very-long-chain acyl-CoA dehydrogenase deficiency. *American Journal of Human Genetics*, **1999**, 64, 479-94	11	248
141	ETFDH mutations as a major cause of riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. *Brain*, **2007**, 130, 2045-54	11.2	238
140	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. *American Journal of Human Genetics*, **2001**, 68, 1408-18	11	191
139	Clear relationship between ETF/ETFDH genotype and phenotype in patients with multiple acyl-CoA dehydrogenation deficiency. *Human Mutation*, **2003**, 22, 12-23	4.7	167
138	Mutation analysis in mitochondrial fatty acid oxidation defects: Exemplified by acyl-CoA dehydrogenase deficiencies, with special focus on genotype-phenotype relationship. *Human Mutation*, **2001**, 18, 169-89	4.7	159
137	Protein misfolding and degradation in genetic diseases. *Human Mutation*, **1999**, 14, 186-98	4.7	151
136	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. *American Journal of Human Genetics*, **2007**, 80, 416-32	11	125
135	CUGBP1 and MBNL1 preferentially bind to 3QUTRs and facilitate mRNA decay. *Scientific Reports*, **2012**, 2, 209	4.9	120
134	The molecular basis of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency in compound heterozygous patients: is there correlation between genotype and phenotype?. *Human Molecular Genetics*, **1997**, 6, 695-707	5.6	107
133	Mitochondrial fatty acid oxidation defects--remaining challenges. *Journal of Inherited Metabolic Disease*, **2008**, 31, 643-57	5.4	107
132	Splicing factor 1 modulates dietary restriction and TORC1 pathway longevity in C. elegans. *Nature*, **2017**, 541, 102-106	50.4	102
131	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. *Blood*, **1994**, 84, 4053-4060	2.2	98
130	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. *Human Molecular Genetics*, **1996**, 5, 461-72	5.6	92
129	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 ethylmalonic aciduria. *Human Molecular Genetics*, **1998**, 7, 619-27	5.6	91
128	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. *Human Genetics*, **2008**, 124, 43-56	6.3	84
127	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. *Blood*, **1997**, 90, 323-330	2.2	81
126	Genetic defects in fatty acid beta-oxidation and acyl-CoA dehydrogenases. Molecular pathogenesis and genotype-phenotype relationships. *FEBS Journal*, **2004**, 271, 470-82		77

125	Mutations in the medium chain acyl-CoA dehydrogenase (MCAD) gene. *Human Mutation*, **1992**, 1, 271-9	4.7	77
124	VLCAD deficiency: pitfalls in newborn screening and confirmation of diagnosis by mutation analysis. *Molecular Genetics and Metabolism*, **2006**, 88, 166-70	3.7	75
123	Internalization, lysosomal degradation and new synthesis of surface membrane CD4 in phorbol ester-activated T-lymphocytes and U-937 cells. *Experimental Cell Research*, **1992**, 201, 160-73	4.2	74
122	Ethylmalonic aciduria is associated with an amino acid variant of short chain acyl-coenzyme A dehydrogenase. *Pediatric Research*, **1996**, 39, 1059-66	3.2	73
121	Medium-chain acyl-CoA dehydrogenase deficiency: genotype-biochemical phenotype correlations. *Molecular Genetics and Metabolism*, **2006**, 87, 32-9	3.7	72
120	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. *Clinica Chimica Acta*, **1991**, 203, 23-34	6.2	72
119	Effects of two mutations detected in medium chain acyl-CoA dehydrogenase (MCAD)-deficient patients on folding, oligomer assembly, and stability of MCAD enzyme. *Journal of Biological Chemistry*, **1995**, 270, 10284-90	5.4	69
118	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency: the prevalent mutation G985 (K304E) is subject to a strong founder effect from northwestern Europe. *Human Heredity*, **1993**, 43, 342-50	1.1	65
117	RNA-sequencing of a mouse-model of spinal muscular atrophy reveals tissue-wide changes in splicing of U12-dependent introns. *Nucleic Acids Research*, **2017**, 45, 395-416	20.1	63
116	Diagnostic assessment and long-term follow-up of 13 patients with Very Long-Chain Acyl-Coenzyme A dehydrogenase (VLCAD) deficiency. *Neuromuscular Disorders*, **2009**, 19, 324-9	2.9	62
115	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. *American Journal of Human Genetics*, **2000**, 67, 1095-103	11	61
114	Very long chain acyl-coenzyme A dehydrogenase deficiency with adult onset. *Annals of Neurology*, **1998**, 43, 540-4	9.4	58
113	Genetic basis for correction of very-long-chain acyl-coenzyme A dehydrogenase deficiency by bezafibrate in patient fibroblasts: toward a genotype-based therapy. *American Journal of Human Genetics*, **2007**, 81, 1133-43	11	57
112	Features of 5Qsplice-site efficiency derived from disease-causing mutations and comparative genomics. *Genome Research*, **2008**, 18, 77-87	9.7	56
111	Biochemical, clinical and molecular findings in LCHAD and general mitochondrial trifunctional protein deficiency. *Journal of Inherited Metabolic Disease*, **2005**, 28, 533-44	5.4	55
110	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. *Human Genetics*, **1991**, 86, 545-51	6.3	55
109	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. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **1993**, 1182, 264-74	6.9	54
108	Prospective diagnosis of 2-methylbutyryl-CoA dehydrogenase deficiency in the Hmong population by newborn screening using tandem mass spectrometry. *Pediatrics*, **2003**, 112, 74-8	7.4	53

107	The role of chaperone-assisted folding and quality control in inborn errors of metabolism: protein folding disorders. *Journal of Inherited Metabolic Disease*, **2001**, 24, 189-212	5.4	52
106	Identification of isobutyryl-CoA dehydrogenase and its deficiency in humans. *Molecular Genetics and Metabolism*, **2002**, 77, 68-79	3.7	52
105	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. *Journal of Investigative Dermatology*, **1999**, 112, 184-90	4.3	51
104	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 cblE type of homocystinuria. *Human Mutation*, **2010**, 31, 437-44	4.7	48
103	Defective folding and rapid degradation of mutant proteins is a common disease mechanism in genetic disorders. *Journal of Inherited Metabolic Disease*, **2000**, 23, 441-7	5.4	48
102	Biochemical screening of 504,049 newborns in Denmark, the Faroe Islands and Greenland--experience and development of a routine program for expanded newborn screening. *Molecular Genetics and Metabolism*, **2012**, 107, 281-93	3.7	47
101	Global identification of hnRNP A1 binding sites for SSO-based splicing modulation. *BMC Biology*, **2016**, 14, 54	7.3	41
100	A comprehensive HADHA c.1528G>C frequency study reveals high prevalence of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency in Poland. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S373-7	5.4	40
99	Mutation and biochemical analysis in carnitine palmitoyltransferase type II (CPT II) deficiency. *Journal of Inherited Metabolic Disease*, **2003**, 26, 543-57	5.4	39
98	Human ClpP protease: cDNA sequence, tissue-specific expression and chromosomal assignment of the gene. *FEBS Letters*, **1995**, 377, 249-52	3.8	39
97	Structural organization of the human short-chain acyl-CoA dehydrogenase gene. *Mammalian Genome*, **1997**, 8, 922-6	3.2	38
96	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. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 2933-2941	5.6	37
95	The mutational spectrum in very long-chain acyl-CoA dehydrogenase deficiency. *Journal of Inherited Metabolic Disease*, **1996**, 19, 169-72	5.4	36
94	Human and mouse mitochondrial orthologs of bacterial ClpX. *Mammalian Genome*, **2000**, 11, 899-905	3.2	35
93	Identification of six novel PTH1R mutations in families with a history of primary failure of tooth eruption. *PLoS ONE*, **2013**, 8, e74601	3.7	35
92	2-Methylbutyryl-coenzyme A dehydrogenase deficiency: functional and molecular studies on a defect in isoleucine catabolism. *Molecular Genetics and Metabolism*, **2008**, 93, 30-5	3.7	33
91	Prevalent mutations in fatty acid oxidation disorders: diagnostic considerations. *European Journal of Pediatrics*, **2000**, 159 Suppl 3, S213-8	4.1	32
90	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). *American Journal of Human Genetics*, **1993**, 53, 730-9	11	32

89	Short/branched-chain acyl-CoA dehydrogenase deficiency due to an IVS3+3A>G mutation that causes exon skipping. *Human Genetics*, **2006**, 118, 680-90	6.3	31
88	An intronic variation in SLC52A1 causes exon skipping and transient riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. *Molecular Genetics and Metabolism*, **2017**, 122, 182-188	3.7	30
87	Newborn screening for medium chain acyl-CoA dehydrogenase deficiency in England: prevalence, predictive value and test validity based on 1.5 million screened babies. *Journal of Medical Screening*, **2011**, 18, 173-81	1.4	30
86	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. *Blood*, **1997**, 90, 323-30	2.2	30
85	SMN2 exon 7 splicing is inhibited by binding of hnRNP A1 to a common ESS motif that spans the 3Q splice site. *Human Mutation*, **2011**, 32, 220-30	4.7	29
84	Late-onset form of beta-electron transfer flavoprotein deficiency. *Molecular Genetics and Metabolism*, **2003**, 78, 247-9	3.7	29
83	A common W556S mutation in the LDL receptor gene of Danish patients with familial hypercholesterolemia encodes a transport-defective protein. *Atherosclerosis*, **1997**, 131, 67-72	3.1	28
82	Variations in IBD (ACAD8) in children with elevated C4-carnitine detected by tandem mass spectrometry newborn screening. *Pediatric Research*, **2006**, 60, 315-20	3.2	28
81	2-ethylhydracrylic aciduria in short/branched-chain acyl-CoA dehydrogenase deficiency: application to diagnosis and implications for the R-pathway of isoleucine oxidation. *Clinical Chemistry*, **2005**, 51, 610-7	5.5	28
80	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 α1-Antitrypsin Gene. *Clinical Chemistry*, **1992**, 38, 2100-2107	5.5	28
79	MCAD deficiency in Denmark. *Molecular Genetics and Metabolism*, **2012**, 106, 175-88	3.7	26
78	Mitochondrial trifunctional protein deficiency in human cultured fibroblasts: effects of bezafibrate. *Journal of Inherited Metabolic Disease*, **2016**, 39, 47-58	5.4	24
77	The Y42H mutation in medium-chain acyl-CoA dehydrogenase, which is prevalent in babies identified by MS/MS-based newborn screening, is temperature sensitive. *FEBS Journal*, **2004**, 271, 4053-63		24
76	Characterization of a disease-causing Glu119-Lys mutation in the low-density lipoprotein receptor gene in two Danish families with heterozygous familial hypercholesterolemia. *Human Mutation*, **1994**, 4, 102-13	4.7	24
75	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. *Blood*, **1994**, 84, 4053-60	2.2	24
74	Urgent metabolic service improves survival in long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency detected by symptomatic identification and pilot newborn screening. *Journal of Inherited Metabolic Disease*, **2011**, 34, 185-95	5.4	23
73	Lipid-storage myopathy and respiratory insufficiency due to ETFQO mutations in a patient with late-onset multiple acyl-CoA dehydrogenation deficiency. *Journal of Inherited Metabolic Disease*, **2004**, 27, 671-8	5.4	23
72	Expression of wild-type and mutant medium-chain acyl-CoA dehydrogenase (MCAD) cDNA in eucaryotic cells. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **1992**, 1180, 65-72	6.9	23

71	DeepCLIP: predicting the effect of mutations on protein-RNA binding with deep learning. *Nucleic Acids Research*, **2020**, 48, 7099-7118	20.1	22
70	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. *Human Mutation*, **2014**, 35, 86-95	4.7	22
69	Two mutations in the same low-density lipoprotein receptor allele act in synergy to reduce receptor function in heterozygous familial hypercholesterolemia. *Human Mutation*, **1997**, 9, 437-44	4.7	22
68	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. *Molecular Genetics and Metabolism*, **2004**, 82, 121-9	3.7	22
67	Myopathy in very-long-chain acyl-CoA dehydrogenase deficiency: clinical and biochemical differences with the fatal cardiac phenotype. *Neuromuscular Disorders*, **1999**, 9, 313-9	2.9	22
66	Prenatal diagnosis of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency in a family with a previous fatal case of sudden unexpected death in childhood. *Prenatal Diagnosis*, **1995**, 15, 82-6	3.2	22
65	VLCAD deficiency: Follow-up and outcome of patients diagnosed through newborn screening in Victoria. *Molecular Genetics and Metabolism*, **2016**, 118, 282-7	3.7	21
64	A severe genotype with favourable outcome in very long chain acyl-CoA dehydrogenase deficiency. *Archives of Disease in Childhood*, **2001**, 84, 58-60	2.2	21
63	The most common mutation causing medium-chain acyl-CoA dehydrogenase deficiency is strongly associated with a particular haplotype in the region of the gene. *Human Genetics*, **1991**, 87, 425-8	6.3	21
62	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. *Nucleic Acids Research*, **2015**, 43, 4627-39	20.1	20
61	Adolescent myopathic presentation in two sisters with very long-chain acyl-CoA dehydrogenase deficiency. *Journal of Inherited Metabolic Disease*, **1999**, 22, 802-10	5.4	20
60	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. *Molecular Genetics and Metabolism*, **1999**, 67, 138-47	3.7	20
59	Comparison between medium-chain acyl-CoA dehydrogenase mutant proteins overexpressed in bacterial and mammalian cells. *Human Mutation*, **1995**, 6, 226-31	4.7	20
58	Disease-causing mutations in exon 11 of the medium-chain acyl-CoA dehydrogenase gene. *American Journal of Human Genetics*, **1994**, 54, 975-88	11	20
57	The phenylalanine hydroxylase c.30C>G synonymous variation (p.G10G) creates a common exonic splicing silencer. *Molecular Genetics and Metabolism*, **2010**, 100, 316-23	3.7	19
56	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. *Blood*, **1997**, 90, 323-330	2.2	19
55	A synonymous polymorphic variation in ACADM exon 11 affects splicing efficiency and may affect fatty acid oxidation. *Molecular Genetics and Metabolism*, **2013**, 110, 122-8	3.7	18
54	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. *Journal of Inherited Metabolic Disease*, **2006**, 29, 620-6	5.4	18

53	Surface membrane CD4 turnover in phorbol ester stimulated T-lymphocytes. Evidence of degradation and increased synthesis. *FEBS Letters*, **1990**, 276, 59-62	3.8	18
52	Newborn screening for MCAD deficiency: experience of the first three years in British Columbia, Canada. *Canadian Journal of Public Health*, **2008**, 99, 276-80	3.2	17
51	A pathogenic haplotype, common in Europeans, causes autosomal recessive albinism and uncovers missing heritability in OCA1. *Scientific Reports*, **2019**, 9, 645	4.9	16
50	Splicing of phenylalanine hydroxylase (PAH) exon 11 is vulnerable: molecular pathology of mutations in PAH exon 11. *Molecular Genetics and Metabolism*, **2012**, 106, 403-11	3.7	16
49	Ethnicity of children with homozygous c.985A>G medium-chain acyl-CoA dehydrogenase deficiency: findings from screening approximately 1.1 million newborn infants. *Journal of Medical Screening*, **2008**, 15, 112-7	1.4	16
48	Intronic PAH gene mutations cause a splicing defect by a novel mechanism involving U1snRNP binding downstream of the 5' splice site. *PLoS Genetics*, **2018**, 14, e1007360	6	15
47	2-methylbutyryl-CoA dehydrogenase deficiency associated with autism and mental retardation: a case report. *Journal of Medical Case Reports*, **2007**, 1, 98	1.2	15
46	Amino acid polymorphism (Gly209Ser) in the ACADS gene. *Human Molecular Genetics*, **1994**, 3, 1711	5.6	14
45	Analysis of the Epstein-Barr virus (EBV) latent membrane protein 1 (LMP-1) gene and promoter in Hodgkin's disease isolates: selection against EBV variants with mutations in the LMP-1 promoter ATF-1/CREB-1 binding site. *Journal of Clinical Pathology*, **2000**, 53, 280-8		13
44	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. *Molecular Genetics and Metabolism*, **2016**, 119, 258-269	3.7	13
43	Functional testing of keratin 14 mutant proteins associated with the three major subtypes of epidermolysis bullosa simplex. *Experimental Dermatology*, **2003**, 12, 472-9	4	12
42	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. *Orphanet Journal of Rare Diseases*, **2010**, 5, 26	4.2	11
41	Very-long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency: monitoring of treatment by carnitine/acylcarnitine analysis in blood spots. *Acta Paediatrica, International Journal of Paediatrics*, **2000**, 89, 492-495	3.1	11
40	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. *PLoS Genetics*, **2016**, 12, e1006039	6	11
39	Long-term outcome of isobutyryl-CoA dehydrogenase deficiency diagnosed following an episode of ketotic hypoglycaemia. *Molecular Genetics and Metabolism Reports*, **2017**, 10, 28-30	1.8	10
38	Neurodevelopmental profiles of children with very long chain acyl-CoA dehydrogenase deficiency diagnosed by newborn screening. *Molecular Genetics and Metabolism*, **2014**, 113, 278-82	3.7	10
37	Interstitial deletion of 1p22.2p31.1 and medium-chain acyl-CoA dehydrogenase deficiency in a patient with global developmental delay. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1581-6	2.5	10
36	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. *Biochemical Medicine and Metabolic Biology*, **1994**, 52, 36-44		10

35	Characterization of a disease-causing Lys329 to Glu mutation in 16 patients with medium-chain acyl-CoA dehydrogenase deficiency. *Journal of Inherited Metabolic Disease*, **1991**, 14, 314-6	5.4	10
34	Next generation sequencing of RNA reveals novel targets of resveratrol with possible implications for Canavan disease. *Molecular Genetics and Metabolism*, **2019**, 126, 64-76	3.7	10
33	Impaired folding and subunit assembly as disease mechanism: the example of medium-chain acyl-CoA dehydrogenase deficiency. *Progress in Molecular Biology and Translational Science*, **1998**, 58, 301-37		9
32	DNA-based prenatal diagnosis for severe and variant forms of multiple acyl-CoA dehydrogenation deficiency. *Prenatal Diagnosis*, **2005**, 25, 60-4	3.2	9
31	A silent A to G mutation in exon 11 of the medium-chain acyl-CoA dehydrogenase (MCAD) gene. *Human Molecular Genetics*, **1993**, 2, 488	5.6	9
30	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. *Journal of Inherited Metabolic Disease*, **1994**, 17, 169-84	5.4	9
29	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. *Clinical Chemistry*, **1992**, 38, 2100-7	5.5	9
28	Blocking of an intronic splicing silencer completely rescues IKBKAP exon 20 splicing in familial dysautonomia patient cells. *Nucleic Acids Research*, **2018**, 46, 7938-7952	20.1	9
27	High-resolution melting analysis, a simple and effective method for reliable mutation scanning and frequency studies in the ACADVL gene. *Journal of Inherited Metabolic Disease*, **2010**, 33, 247-60	5.4	8
26	DNA-based prenatal diagnosis for very-long-chain acyl-CoA dehydrogenase deficiency. *Journal of Inherited Metabolic Disease*, **1999**, 22, 281-5	5.4	8
25	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. *JIMD Reports*, **2012**, 3, 11-5	1.9	7
24	Abnormal Newborn Screening in a Healthy Infant of a Mother with Undiagnosed Medium-Chain Acyl-CoA Dehydrogenase Deficiency. *JIMD Reports*, **2015**, 23, 67-70	1.9	6
23	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. *FEBS Journal*, **2005**, 272, 4549-57	5.7	6
22	Very-long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency: monitoring of treatment by carnitine/acylcarnitine analysis in blood spots. *Acta Paediatrica, International Journal of Paediatrics*, **2000**, 89, 492-5	3.1	6
21	Fibroblast Fatty-Acid Oxidation Flux Assays Stratify Risk in Newborns with Presumptive-Positive Results on Screening for Very-Long Chain Acyl-CoA Dehydrogenase Deficiency. *International Journal of Neonatal Screening*, **2017**, 3, 2	2.6	5
20	Characterization of mouse Clpp protease cDNA, gene, and protein. *Mammalian Genome*, **2000**, 11, 275-80	3.2	5
19	Absence of an intron splicing silencer in porcine Smn1 intron 7 confers immunity to the exon skipping mutation in human SMN2. *PLoS ONE*, **2014**, 9, e98841	3.7	3
18	Use of Molecular Genetic Analyses in Danish Routine Newborn Screening. *International Journal of Neonatal Screening*, **2021**, 7,	2.6	3

17	Identification of SRSF10 as a regulator of SMN2 ISS-N1. *Human Mutation*, **2021**, 42, 246-260	4.7	3
16	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
15	Relevance of expanded neonatal screening of medium-chain acyl co-a dehydrogenase deficiency: outcome of a decade in galicia (Spain). *JIMD Reports*, **2011**, 1, 131-6	1.9	2
14	DeepCLIP: Predicting the effect of mutations on protein-RNA binding with Deep Learning		2
13	A Large Intragenic Deletion in the ACADM Gene Can Cause MCAD Deficiency but is not Detected on Routine Sequencing. *JIMD Reports*, **2013**, 11, 13-6	1.9	1
12	Biochemical characterisation of mutations of human medium-chain acyl-CoA dehydrogenase. *Advances in Experimental Medicine and Biology*, **1999**, 466, 387-93	3.6	1
11	Mutations of human medium-chian acyl-CoA dehydrogenase. *Biochemical Society Transactions*, **1998** , 26, S65	5.1	1
10	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency due to heterozygosity for the common mutation and an allele resulting in low levels of MCAD mRNA. *Journal of Inherited Metabolic Disease* , **1994**, 17, 275-8	5.4	1
9	Molecular analysis of medium-chain acyl-CoA dehydrogenase deficiency: a diagnostic approach. *Progress in Clinical and Biological Research*, **1992**, 375, 441-52		1
8	Molecular diagnosis and characterization of medium-chain acyl-CoA dehydrogenase deficiency. *Scandinavian Journal of Clinical and Laboratory Investigation, Supplement*, **1995**, 220, 9-25		1
7	Down-Regulation of CK2□Leads toUp-Regulation of the Cyclin-Dependent Kinase Inhibitor p27 in Conditions Unfavorable for the Growth of Myoblast Cells. *Cellular Physiology and Biochemistry*, **2020**, 54, 1177-1198	3.9	1
6	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
5	Down-regulation of CK2□correlates with decreased expression levels of DNA replication minichromosome maintenance protein complex (MCM) genes. *Scientific Reports*, **2019**, 9, 14581	4.9	0
4	Pseudoexon activation in disease by non-splice site deep intronic sequence variation - wild type pseudoexons constitute high-risk sites in the human genome. *Human Mutation*, **2021**, 43, 103	4.7	0
3	-targeting miR-agshRNAs combine efficacy with specificity and safety for retinal gene therapy.. *Molecular Therapy - Nucleic Acids*, **2022**, 28, 58-76	10.7	0
2	Medium Chain Acyl-CoA Dehydrogenase Deficiency. *Biochemical Society Transactions*, **2000**, 28, A73-A73	5.1	
1	Properties of five rare human mutations causing medium chain acyl-CoA dehydrogenase deficiency. *Biochemical Society Transactions*, **2000**, 28, A160-A160	5.1	