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List of Publications by Year
in descending order

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147 papers	6,605 citations	57719 44 h-index	82499 72 g-index
153 all docs	153 docs citations	153 times ranked	5933 citing authors

#	Article	IF	Citations
1	ETFDH mutations as a major cause of riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. Brain, 2007, 130, 2045-2054.	3.7	292
2	Clear Correlation of Genotype with Disease Phenotype in Veryâ€“Long-Chain Acyl-CoA Dehydrogenase Deficiency. American Journal of Human Genetics, 1999, 64, 479-494.	2.6	285
3	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, 1408-1418.	2.6	219
4	Clear relationship betweenETF/ETFDH genotype and phenotype in patients with multiple acyl-CoA dehydrogenation deficiency. Human Mutation, 2003, 22, 12-23.	1.1	196
5	Protein misfolding and degradation in genetic diseases. , 1999, 14, 186-198.		184
6	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, 169-189.	1.1	178
7	Splicing factor 1 modulates dietary restriction and TORC1 pathway longevity in C. elegans. Nature, 2017, 541, 102-106.	13.7	152
8	CUGBP1 and MBNL1 preferentially bind to 3â€² UTRs and facilitate mRNA decay. Scientific Reports, 2012, 2, 209.	1.6	150
9	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, 416-432.	2.6	140
10	Mitochondrial fatty acid oxidation defectsâ€”remaining challenges. Journal of Inherited Metabolic Disease, 2008, 31, 643-657.	1.7	123
11	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.	1.4	119
12	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.	0.6	112
13	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, 619-627.	1.4	109
14	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 [published erratum appears in Hum Mol Genet 1996 Sep;5(9):1390]. Human Molecular Genetics, 1996, 5, 461-472.	1.4	106
15	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.	1.8	101
16	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, 160-173.	1.2	95
17	VLCAD deficiency: Pitfalls in newborn screening and confirmation of diagnosis by mutation analysis. Molecular Genetics and Metabolism, 2006, 88, 166-170.	0.5	95
18	Ethylmalonic Aciduria Is Associated with an Amino Acid Variant of Short Chain Acyl-Coenzyme A Dehydrogenase. Pediatric Research, 1996, 39, 1059-1066.	1.1	92

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19	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.	6.5	87
20	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.	0.6	86
21	Genetic defects in fatty acid beta-oxidation and acyl-CoA dehydrogenases. Molecular pathogenesis and genotype-phenotype relationships. FEBS Journal, 2004, 271, 470-482.	0.2	86
22	Mutations in the medium chain acyl-CoA dehydrogenase (MCAD) gene. Human Mutation, 1992, 1, 271-279.	1.1	85
23	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.	0.5	83
24	Medium-chain acyl-CoA dehydrogenase deficiency: Genotypeâ€“biochemical phenotype correlations. Molecular Genetics and Metabolism, 2006, 87, 32-39.	0.5	83
25	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, 10284-10290.	1.6	79
26	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, 1095-1103.	2.6	79
27	Diagnostic assessment and long-term follow-up of 13 patients with Very Long-Chain Acyl-Coenzyme A dehydrogenase (VLCAD) deficiency. Neuromuscular Disorders, 2009, 19, 324-329.	0.3	79
28	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, 342-350.	0.4	75
29	Features of 5â€²-splice-site efficiency derived from disease-causing mutations and comparative genomics. Genome Research, 2008, 18, 77-87.	2.4	75
30	Genetic Basis for Correction of Very-Long-Chain Acylâ€“Coenzyme A Dehydrogenase Deficiency by Bezafibrate in Patient Fibroblasts: Toward a Genotype-Based Therapy. American Journal of Human Genetics, 2007, 81, 1133-1143.	2.6	69
31	Prospective Diagnosis of 2-Methylbutyryl-CoA Dehydrogenase Deficiency in the Hmong Population by Newborn Screening Using Tandem Mass Spectrometry. Pediatrics, 2003, 112, 74-78.	1.0	67
32	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.	1.8	66
33	Very long chain acyl-coenzyme A dehydrogenase deficiency with adult onset. Annals of Neurology, 1998, 43, 540-544.	2.8	65
34	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.	1.7	65
35	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, 264-274.	1.8	64
36	Biochemical, clinical and molecular findings in LCHAD and general mitochondrial trifunctional protein deficiency. Journal of Inherited Metabolic Disease, 2005, 28, 533-544.	1.7	63

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37	Global identification of hnRNP A1 binding sites for SSO-based splicing modulation. BMC Biology, 2016, 14, 54.	1.7	62
38	Identification of isobutyryl-CoA dehydrogenase and its deficiency in humans. Molecular Genetics and Metabolism, 2002, 77, 68-79.	0.5	60
39	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, 184-190.	0.3	58
40	Biochemical screening of 504,049 newborns in Denmark, the Faroe Islands and Greenland â€" Experience and development of a routine program for expanded newborn screening. Molecular Genetics and Metabolism, 2012, 107, 281-293.	0.5	55
41	DeepCLIP: predicting the effect of mutations on proteinâ€“RNA binding with deep learning. Nucleic Acids Research, 2020, 48, 7099-7118.	6.5	54
42	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, 437-444.	1.1	53
43	Defective folding and rapid degradation of mutant proteins is a common disease mechanism in genetic disorders. Journal of Inherited Metabolic Disease, 2000, 23, 441-447.	1.7	52
44	Human ClpP protease: cDNA sequence, tissue-specific expression and chromosomal assignment of the gene. FEBS Letters, 1995, 377, 249-252.	1.3	47
45	Mutation and biochemical analysis in carnitine palmitoyltransferase type II (CPT II) deficiency. Journal of Inherited Metabolic Disease, 2003, 26, 543-557.	1.7	47
46	Identification of Six Novel PTH1R Mutations in Families with a History of Primary Failure of Tooth Eruption. PLoS ONE, 2013, 8, e74601.	1.1	47
47	2-Methylbutyryl-coenzyme A dehydrogenase deficiency: Functional and molecular studies on a defect in isoleucine catabolism. Molecular Genetics and Metabolism, 2008, 93, 30-35.	0.5	45
48	A comprehensive HADHA c.1528G>C frequency study reveals high prevalence of longâ€chain 3â€hydroxyacylâ€CoA dehydrogenase deficiency in Poland. Journal of Inherited Metabolic Disease, 2010, 33, 373-377.	1.7	45
49	Structural organization of the human short-chain acyl-CoA dehydrogenase gene. Mammalian Genome, 1997, 8, 922-926.	1.0	42
50	SMN2 exon 7 splicing is inhibited by binding of hnRNP A1 to a common ESS motif that spans the 3â€² splice site. Human Mutation, 2011, 32, 220-230.	1.1	41
51	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.	0.5	41
52	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.	1.8	41
53	The mutational spectrum in very long-chain acyl-CoA dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 1996, 19, 169-172.	1.7	40
54	Short/branched-chain acyl-CoA dehydrogenase deficiency due to an IVS3+3A>G mutation that causes exon skipping. Human Genetics, 2006, 118, 680-690.	1.8	40

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55	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, 173-181.	1.1	40
56	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, 610-617.	1.5	38
57	Prevalent mutations in fatty acid oxidation disorders: diagnostic considerations. European Journal of Pediatrics, 2000, 159, S213-S218.	1.3	37
58	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.	0.6	37
59	Human and mouse mitochondrial orthologs of bacterial ClpX. Mammalian Genome, 2000, 11, 899-905.	1.0	36
60	Late-onset form of Î²-electron transfer flavoprotein deficiency. Molecular Genetics and Metabolism, 2003, 78, 247-249.	0.5	34
61	MCAD deficiency in Denmark. Molecular Genetics and Metabolism, 2012, 106, 175-188.	0.5	33
62	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 Î±1-Antitrypsin Gene. Clinical Chemistry, 1992, 38, 2100-2107.	1.5	32
63	Variations in IBD (ACAD8) in Children with Elevated C4-Carnitine Detected by Tandem Mass Spectrometry Newborn Screening. Pediatric Research, 2006, 60, 315-320.	1.1	32
64	TheETFDHc.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.	1.1	32
65	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.	2.6	32
66	Mitochondrial trifunctional protein deficiency in human cultured fibroblasts: effects of bezafibrate. Journal of Inherited Metabolic Disease, 2016, 39, 47-58.	1.7	31
67	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, 671-678.	1.7	30
68	Urgent metabolic service improves survival in longâ€chain 3â€hydroxyacylâ€CoA dehydrogenase (LCHAD) deficiency detected by symptomatic identification and pilot newborn screening. Journal of Inherited Metabolic Disease, 2011, 34, 185-195.	1.7	30
69	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.	0.6	30
70	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.	0.4	29
71	A severe genotype with favourable outcome in very long chain acyl-CoA dehydrogenase deficiency. Archives of Disease in Childhood, 2001, 84, 58-60.	1.0	29
72	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, 4053-4063.	0.2	29

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73	A pathogenic haplotype, common in Europeans, causes autosomal recessive albinism and uncovers missing heritability in OCA1. Scientific Reports, 2019, 9, 645.	1.6	29
74	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, 4627-4639.	6.5	28
75	VLCAD deficiency: Follow-up and outcome of patients diagnosed through newborn screening in Victoria. Molecular Genetics and Metabolism, 2016, 118, 282-287.	0.5	28
76	2-methylbutyryl-CoA dehydrogenase deficiency associated with autism and mental retardation: a case report. Journal of Medical Case Reports, 2007, 1, 98.	0.4	27
77	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.	1.8	26
78	Intronic PAH gene mutations cause a splicing defect by a novel mechanism involving U1snRNP binding downstream of the 5â€™ splice site. PLoS Genetics, 2018, 14, e1007360.	1.5	26
79	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, 102-113.	1.1	25
80	Myopathy in very-long-chain acyl-CoA dehydrogenase deficiency: clinical and biochemical differences with the fatal cardiac phenotype. Neuromuscular Disorders, 1999, 9, 313-319.	0.3	25
81	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, 121-129.	0.5	25
82	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, 82-86.	1.1	24
83	Splicing of phenylalanine hydroxylase (PAH) exon 11 is vulnerable: Molecular pathology of mutations in PAH exon 11. Molecular Genetics and Metabolism, 2012, 106, 403-411.	0.5	24
84	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, 437-444.	1.1	23
85	The phenylalanine hydroxylase c.30C>G synonymous variation (p.G10G) creates a common exonic splicing silencer. Molecular Genetics and Metabolism, 2010, 100, 316-323.	0.5	23
86	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.	0.5	23
87	Adolescent myopathic presentation in two sisters with very long-chain acyl-CoA dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 1999, 22, 802-810.	1.7	22
88	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, 620-626.	1.7	22
89	A synonymous polymorphic variation in ACADM exon 11 affects splicing efficiency and may affect fatty acid oxidation. Molecular Genetics and Metabolism, 2013, 110, 122-128.	0.5	22
90	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.	0.6	22

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91	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.	1.8	21
92	Comparison between medium-chain acyl-CoA dehydrogenase mutant proteins overexpressed in bacterial and mammalian cells. Human Mutation, 1995, 6, 226-231.	1.1	21
93	A Polymorphic Variant in the Human Electron Transfer Flavoprotein Î±-Chain (Î±-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, 138-147.	0.5	21
94	Newborn Screening for MCAD Deficiency. Canadian Journal of Public Health, 2008, 99, 276-280.	1.1	21
95	Disease-causing mutations in exon 11 of the medium-chain acyl-CoA dehydrogenase gene. American Journal of Human Genetics, 1994, 54, 975-88.	2.6	21
96	Surface membrane CD4 turnover in phorbol ester stimulated T-lymphocytes. FEBS Letters, 1990, 276, 59-62.	1.3	19
97	Amino acid polymorphism (Gly209Ser) in the ACADS gene. Human Molecular Genetics, 1994, 3, 1711-1711.	1.4	19
98	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, 112-117.	1.1	18
99	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. PLoS Genetics, 2016, 12, e1006039.	1.5	18
100	Pseudoexon activation in disease by nonâ€splice site deep intronic sequence variation â€" wild type pseudoexons constitute highâ€risk sites in the human genome. Human Mutation, 2022, 43, 103-127.	1.1	17
101	Next generation sequencing of RNA reveals novel targets of resveratrol with possible implications for Canavan disease. Molecular Genetics and Metabolism, 2019, 126, 64-76.	0.5	16
102	Blocking of an intronic splicing silencer completely rescues IKBKAP exon 20 splicing in familial dysautonomia patient cells. Nucleic Acids Research, 2018, 46, 7938-7952.	6.5	15
103	Identification of SRSF10 as a regulator of SMN2 ISSâ€N1. Human Mutation, 2021, 42, 246-260.	1.1	15
104	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, 280-288.	2.1	14
105	Functional testing of keratin 14 mutant proteins associated with the three major subtypes of epidermolysis bullosa simplex. Experimental Dermatology, 2003, 12, 472-479.	1.4	14
106	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.	1.2	14
107	Long-term outcome of isobutyryl-CoA dehydrogenase deficiency diagnosed following an episode of ketotic hypoglycaemia. Molecular Genetics and Metabolism Reports, 2017, 10, 28-30.	0.4	14
108	Neurodevelopmental profiles of children with very long chain acyl-CoA dehydrogenase deficiency diagnosed by newborn screening. Molecular Genetics and Metabolism, 2014, 113, 278-282.	0.5	13

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109	Mutation analysis in mitochondrial fatty acid oxidation defects: Exemplified by acyl-CoA dehydrogenase deficiencies, with special focus on genotypeâ€“phenotype relationship. Human Mutation, 2001, 18, 169.	1.1	13
110	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.	1.5	13
111	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 Posttranslational Modification of the Enzyme. Biochemical Medicine and Metabolic Biology, 1994, 52, 36-44.	0.7	12
112	Impaired Folding and Subunit Assembly as Disease Mechanism: The Example of Medium-Chain acyl-CoA Dehydrogenase Deficiency. Progress in Molecular Biology and Translational Science, 1997, 58, 301-337.	1.9	12
113	Use of Molecular Genetic Analyses in Danish Routine Newborn Screening. International Journal of Neonatal Screening, 2021, 7, 50.	1.2	12
114	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, 314-316.	1.7	11
115	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, 169-184.	1.7	11
116	Veryâ€longâ€chain acylâ€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.	0.7	11
117	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, 1581-1586.	0.7	11
118	Vulnerable exons, like ACADM exon 5, are highly dependent on maintaining a correct balance between splicing enhancers and silencers. Human Mutation, 2022, 43, 253-265.	1.1	11
119	DNA-based prenatal diagnosis for very-long- chain acyl-CoA dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 1999, 22, 281-285.	1.7	10
120	DNA-based prenatal diagnosis for severe and variant forms of multiple acyl-CoA dehydrogenation deficiency. Prenatal Diagnosis, 2005, 25, 60-64.	1.1	10
121	Highâ€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, 247-260.	1.7	10
122	A silent A to G mutation in exon 11 of the medium-chain acyl-CoA dehydrogenase (MCAD) gene. Human Molecular Genetics, 1993, 2, 488-488.	1.4	9
123	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, 2011, 3, 11-15.	0.7	9
124	Topoisomerase 1 inhibits MYC promoter activity by inducing G-quadruplex formation. Nucleic Acids Research, 2022, 50, 6332-6342.	6.5	9
125	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, 4549-4557.	2.2	7
126	Abnormal Newborn Screening in a Healthy Infant of a Mother with Undiagnosed Medium-Chain Acyl-CoA Dehydrogenase Deficiency. JIMD Reports, 2015, 23, 67-70.	0.7	7

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127	Antisense Oligonucleotide Rescue of Deep-Intronic Variants Activating Pseudoexons in the 6-Pyruvoyl-Tetrahydropterin Synthase Gene. Nucleic Acid Therapeutics, 2022, 32, 378-390.	2.0	7
128	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.	1.2	6
129	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.	0.7	6
130	VEGFA-targeting miR-agshRNAs combine efficacy with specificity and safety for retinal gene therapy. Molecular Therapy - Nucleic Acids, 2022, 28, 58-76.	2.3	6
131	Characterization of mouse Clpp protease cDNA, gene, and protein. Mammalian Genome, 2000, 11, 275-280.	1.0	5
132	Down-regulation of CK2Î± correlates with decreased expression levels of DNA replication minichromosome maintenance protein complex (MCM) genes. Scientific Reports, 2019, 9, 14581.	1.6	5
133	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.	1.1	4
134	Down-Regulation of CK2Î± Leads toUp-Regulation of the Cyclin-Dependent Kinase Inhibitor p27KIP1 in Conditions Unfavorable for the Growth of Myoblast Cells. Cellular Physiology and Biochemistry, 2020, 54, 1177-1198.	1.1	3
135	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, 275-278.	1.7	2
136	Relevance of Expanded Neonatal Screening of Medium-Chain Acyl Co-A Dehydrogenase Deficiency: Outcome of a Decade in Galicia (Spain). JIMD Reports, 2011, 1, 131-136.	0.7	2
137	Essential role of CK2Î± for the interaction and stability of replication fork factors during DNA synthesis and activation of the S-phase checkpoint. Cellular and Molecular Life Sciences, 2022, 79, .	2.4	2
138	77 Mutations of Human Medium-Chain Acyl-CoA Dehydrogenase. Biochemical Society Transactions, 1998, 26, S65-S65.	1.6	1
139	Biochemical Characterisation of Mutations of Human Medium-Chain Acyl-CoA Dehydrogenase. , 1999, 466, 387-393.		1
140	A Large Intragenic Deletion in the ACADM Gene Can Cause MCAD Deficiency but is not Detected on Routine Sequencing. JIMD Reports, 2013, 11, 13-16.	0.7	1
141	Two mutations in the same lowâ€density lipoprotein receptor allele act in synergy to reduce receptor function in heterozygous familial hypercholesterolemia. Human Mutation, 1997, 9, 437-444.	1.1	1
142	Molecular analysis of medium-chain acyl-CoA dehydrogenase deficiency: a diagnostic approach. Progress in Clinical and Biological Research, 1992, 375, 441-52.	0.2	1
143	Molecular diagnosis and characterization of medium-chain acyl-CoA dehydrogenase deficiency. Scandinavian Journal of Clinical and Laboratory Investigation, Supplement, 1995, 220, 9-25.	2.7	1
144	Medium Chain Acyl-CoA Dehydrogenase Deficiency. Biochemical Society Transactions, 2000, 28, A73-A73.	1.6	0

#	Article	IF	Citations
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