CURRICULUM VITAE

Brage Storstein Andresen - Born 7. February 1965 in Oslo, Norway. Married – Three children.
Work address: Dept. of Biochemistry and Molecular Biology, University of Southern Denmark, Campusvej 55, DK-5230 Odense S, Denmark.

Academic degrees

1991	M.Sc. (Cand. Scient., Biology), Aarhus University (AU), Denmark.
1996	Ph.D. Faculty of Health, AU.
2015	FRCPath. (Fellow of The Royal College of Pathologists), UK.

Positions

1995-1997	Assistant professor, Molecular Genetic Laboratory, Aarhus University hospital -Skejby Sygehus and Dept. of Experimental Clinical Research, AU.
1997-2008	Associate professor, Dept. Human Genetics, AU.
2004	Visiting professor, Dept. Pediatrics, Vanderbilt Univ. Medical School, TN, USA.
2008	Visiting Scientist, Cold Spring Harbor Laboratory, NY, USA.
2008 -2014	Professor (WSR) of Eukaryotic Gene Regulation and Metabolism, Dept. of Biochem. & Mol. Biol., University of Southern Denmark (SDU).
2014 -	Professor of Human Molecular Genetics, Dept. of Biochem. & Mol. Biol., SDU.
2018 -	Deputy Head of Department, Dept. of Biochem. & Mol. Biol. (BMB), SDU.
2018 -	Head of Section for Translational Biology, Dept. of Biochem. & Mol. Biol., SDU.

Fellowships

1993-1995	Fellowship. Danish Medical Research Council (now FSS).
1995-1997	Senior fellowship. Danish Medical Research Council (now FSS).

Other education

1996 Pedagogical Training course, Sandbjerg gods, AU.
2009 Research Management Course, Copenhagen School of Business.
2011 SDU's Leadership Development Program.

Academic work 136 peer-reviewed articles + 17 book chapters/Invited articles.
ORCID: 0000-0001-7488-3035 - SCOPUS: H-index = 40 4830 citations. Google Scholar: H-Index = 44 6483 citations I10=111
Presented >50 oral presentations international meetings.

Patent: RAS Exon 2 skipping for cancer treatment - PCT/EP2014/078029 Published: WO 2015/091525. Inventor: Brage Storstein Andresen, SDU.

Recent invited seminars/talks: Cold Spring Harbor Laboratory (2015), 4th International RASopathies Symposium, Seattle, USA (2015), Karolinska Instituttet (2015), Tokuron Special Lecture, Nagoya Medical School, Japan (2016). Splicing 2016, Lisbon, Portugal. 5th Int. RASopathies Symp., Orlando, USA (2017); Danish Institute of Advanced Science (DIAS) – Invited lecture Nov. 2018). Symposium on Precision Medicine in the Region of Southern Denmark (March 21 -2019). Danish Society of Medical Genetics, Annual meeting, Invited Lecture (April 5. 2019).

Peer review (recent examples.): *Nature, Nature Communications, Am. J Hum. Genet., Hum. Mol. Genet., Hum Mut., J. Med. Genet., Mol. Genet. Metab, Hum. Genet., Nucleic Acids Res., Mol. Therapy.*

Evaluator EU Horizon 2020-PHC14; EU Horizon 2020-SC1-PM-08-2017; EU Horizon 2020-FETFLAG-01- and 02- 2018; and EU ERC (2015); Swedish-, Canadian-, Dutch-, Japanese-, Irish, Romanian- Faroese- Estonian- and Swiss- National Research Councils; Prinses Beatrix Fonds (NL) and Institut National du Cancer (France) (2017-20).

Editorial Board *Molecular Genetics and Metabolism,* Elsevier (2007-).

Committees Scientific Advisory Group, UK Collab. Study Newborn Screening MCADD (2004-11).
Workgroup for evaluation of Newborn Screening in DK. Danish National Board of Health (2005-09).
Steering group Newborn Screening in Denmark (2003-).
Chair of teaching, Dept. Hum Genet., AU (2007-08) and BMB, SDU (2015-), Educational board, BMB, SDU (2009-), Studyboard, Faculty of Science, SDU (2018-)
Department Council, BMB, SDU (2013-).
Steering group for Villum Center for Bioanalytical Sciences (2013-19).
Faculty member "Graduate Program for Real World Data Informatics–JUN-KAN" supported by JSPS http://www.jsps.go.jp/english/e-hakasekatei/index.html, Nagoya Univ. Med. School, Japan, 2013-20.
Executive board: Nordic Network Biomedicine, Nordplus/ERASMUS+, OERCompBiomed (2017-).

Current supervision
4 Post. docs, 3 PhD- and 7 Master-students.

Current research profile
The research in my laboratory has for years focused on the molecular genetics and the molecular pathology of human disease with a special emphasis on regulation of normal and aberrant alternative pre-mRNA splicing. In recent years, we

have focused intensely on development of techniques for Next Generation Sequencing (NGS) of RNA, data-analysis pipelines and global mapping and characterization of *in vivo* binding sites for splicing regulatory proteins using iCLIP, tRIP and eCLIP techniques. We employ RNA-affinity purification techniques, binding assays and SPRi to identify and characterize binding of splicing regulatory proteins. We have developed methods for modulation of splicing using splice switching oligonucleotides (SSOs) to target disease-associated splicing regulatory elements for development of new individualized therapies for metabolic- neuromuscular- and cancerous-diseases and tested these in patient cells and mouse models. We have patented SSO technology targeting the RAS oncogenes as a potential new therapeutic approach towards several cancerous diseases. We use RNA-seq to detect disease-causing pseudoexons in human disease genes to develop comprehensive searchable databases to assist in clinical diagnosis in personalized medicine. We have developed SSOs that can block inclusion of disease-causing pseudoexons in diseases, such as Fabry Disease, Homocystinuria, Propionic Acidemia, Afibrinogenemia, Alport disease, Familial Dysautonomia etc. Moreover, we have developed SSOs for therapeutic correction of splicing in PKU. These SSOs may be developed into promising new individualized therapies. We develop software to predict the effect of sequence changes on RNA-binding of splicing regulatory proteins (*DeepClip* - https://deepclip-web.compbio.sdu.dk), and software to pinpoint parts of the transcriptome, which are particularly vulnerable to splicing mutations *VulExMap* - https://vulexmap.compbio.sdu.dk).

My lab consists of: 1 PI, 4 postdoc/AC-TAP, 3 PhD students, 7 MSc students, several project students (Bachelor/ITEK/ERASMUS) and 1 guest Ph.D. student.

Management experience
Head of Research lab with 15-20 people
Deputy Head of Dept., BMB, SDU (2018-) (Responsible for 3 educations/Secretariat: 3 employees)
Member of Department management group, BMB, SDU (2018-).
Head of Section for Translational Biology (4 Labs – 40 people)
Chair of teaching, Dept. Hum Gen, AU (2007-08) and BMB, SDU (2015-)

Current main international collaborators
Prof. and Vice Dean, Kinji Ohno, Nagoya University Graduate School of Medicine, Nagoya, Japan.
Prof. Lourdes Ruiz Desviat, Universidad Autonoma Madrid, Spain.
Prof. Adrian Krainer, Cold Spring Harbor Laboratory, New York, USA.
Assoc. Prof. Michelle Hastings, Rosalind Franklin University of Medicine and Science, Chicago, USA.

Recent major research funding
Danish Medical Research Council (FSS) 2015-19 and 2019-22; Danish Research Council for Natural Science (FNU) 2015-19 and 2020-2023; Novo Nordisk Foundation: 2018-19 and 2020-22; SDU POP-, POC- grants: 2013-15. ODEx: 2017-19; Danish Cancer Society: 2017-18. Lundbeck Foundation (2019-22) (Ph.D. stipend - Jeanne MV Bang). TaNeDS Europe winner (Take a New Challenge for Drug DiScovery) Daichi Sankyo: 2017-2020 (Coapplicant with Prof. Desviat, Univ. Autonoma Madrid - Development of SSO based therapy for PKU).
Societies *The American Society of Human Genetics; European Society of Human Genetics; Society for the Study of Inborn Errors of Metabolism, RNA Society, Danish Society for Medical Genetics, Oligonucleotide Therapeutics Society.*
PUBLICATION LIST
Total 153 articles - Peer review: 136 - Not-peer review: 17
Last five years: First author: 0 – Last author: 17
Patents: 1
ORCID: 0000-0001-7488-3035 - SCOPUS: H-index = 40 4830 citations. Google Scholar: H-Index = 44 6483 citations I10=111
Researcher ID: D-2515-2012.
Scopus Author ID: 35866781200

Full list of publications:
https://www.ncbi.nlm.nih.gov/pubmed/?term=andresen+bs+not+fomsgaard

1. Møller BK, Andresen BS, Christensen EI, Petersen CM. (1990) Surface membrane CD4 turnover in PMA stimula-ted T-Lymphocytes: evidence of degradation and increased synthe-sis. FEBS letters 276:59-62.
2. Gregersen N, Andresen BS, Bross P, Winter V, Rudiger N, Engst S, Christensen E, Kelly D, Strauss A, Kølvraa S, Bolund L and Ghisla S. (1991) 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 protein in E.coli. Human Genetics 86:545-551.
3. Gregersen N, Andresen BS, Bross P, Winter V, Engst S, Blakemore A, Curtis D, Engel P, Kelly D, Strauss AW, Kølvraa S, Bolund L and Ghisla S. (1991) Characterization of a disease-causing Lys329 to Glu mutation in 16 patients with medium-chain acyl-CoA dehydrogenase deficiency. J Inher Metab Dis. 14:314-316.
4. Kølvraa S, Gregersen N, Blakemore A, Schneidermann A, Winter V, Andresen BS, Curtis D, Engel P, Divry P, Rhead W and Bolund L. (1991) The most common mutation causing medium-chain acyl-CoA dehydrogenase (MCAD) deficien-cy probably occurs ex-clusively on chromosomes with a particu-lar haplotype in the region of the gene. Human Genetics 87:425-429.
5. Gregersen N, Blakemore A, Winter V, Andresen BS, Kølvraa S, Bolund L, Curtis D and Engel P. (1991) Specific diagnosis of medium-chain acyl-CoA dehydrogena-se (MCAD) deficiency in dried blood spots by a Polymerase Chain

Reaction (PCR) assay detecting a point-mutation (G985) in the MCAD gene. Clin -Chim Acta 203:23-34.
6. Gregersen N, Winter V, Andresen BS, Kølvraa S, Christensen E, Petersen BN og Bolund L. (1992). Mellem-kædet acyl-CoA dehydrogenase (MCAD) mangel: En livstruende fedtsyreoxidationsdefekt. Ugesk Læger156:483-488.
7. Petersen CM, Christensen EI, Andresen BS, Møller BK. (1992) Internaliza-tion, Lysosomal degradation and new-synthesis of surface membrane CD4 in phorbol ester activated T-lymphocytes and U-937 cells. Exp Cell Res201:160-173.
8. Andresen BS, Knudsen I, Jensen PKA, Rasmussen K and Gregersen N (1992) Two novel non-radioactive PCR-based as-says in which dried blood -spots, genomic DNA or whole cells are used for fast and reli-able detec-tion of the Z and S mutations in the gene for a-1-anti-trypsin. Clinical Chemi-stry 38:2100-2107.
9. Jensen TG, Andresen BS, Bross P, Jensen UB, Holme E, Kølvraa S, Gregersen N and Bolund L (1992) Expression of wildtype and mutant medium-chain acyl-CoA dehydroge-nase (MCAD) cDNA in eucaryotic cells. Biochim Biophys Ac-ta 1180:65-72.
10. Andresen BS, Bross P, Jensen TG, Winter V, Kølvraa S, Jensen UB, Bolund L, Curtis D, Duran M, Kim JJP, Vianey-Saban C, Divry P and Gregersen N. (1993) A rare disease-associated mutation in the gene for Medium Chain Acyl-CoA dehydrogenase (MCAD) chan-ges a conserved arginine residue pre-viously shown to be functionally essential in Short-Chain Acyl-CoA dehydro-genase (SCAD). Am J -Hum Ge-net. 53:730-739.
11. Andresen BS, Kølvraa S, Bross P, Bolund L, Curtis D, Eiberg H, Zhang ZF, Kelly DP, Strauss AW and Greger-sen N. (1993) A silent A to G muta-tion in exon 11 of the Medi-um Chain Acyl-CoA dehy-drogenase (MCAD) gene. Hum Mol Genet 2:488.
12. Gregersen N, Winter V, Curtis D, Deufel T, Mack M, Willems PJ, Ponzone A, P-a-r-r-el-la T, Ponzone R, Ding JH, Zhang W, Chen YT, Kahler S, Roe CR, Kølvraa S, Schneider-man K, An-dresen BS, Bross P and Bolund L. (1993). Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Defi-ciency: The preva-lent muta-tion G985 (K304E) is subject to a strong founder effect from North-western Euro-pe. Human Her-edity 43:342-350.
13. Tanaka K, Yokota I, Coates PM, Strauss AW, Kelly DP, Zhang Z, Gregersen N, Andresen BS, Matsubara Y, Curtis D and Chen YT (1992) Mutations in the Medium-Chain Acyl-CoA Dehydroge-nase (MCAD) gene. Hum Mutat 1:271-279.
14. Bross P, Andresen BS, Winter V, Krautle F, Jensen TG, Kølvraa S, Rasched I, Ghisla S, Bo-lund L and Gregersen N. (1993) Cooverexpression of bacterial Gro-ESL chapero-nins partly overcomes unproductive folding and tetramer assembly in E.coli expressing human medium-chain acyl-Coa dehy-droge-nase (MCAD) carrying a prevalent point mutation A985G found in patients with MCAD deficiency. B-i-o-c-him-- B-i-o-p-h-y-s -A-c-ta 1182:264-274.
15. Andresen BS, Jensen TG, Bross P, Knudsen I, Winter V, Kølvraa S, Bolund L, Ding JH, Chen YT, VanHove JLK, Curtis D, Yokota I, Tanaka K, Kim JJP and Gregersen N. (1994) Disease-causing mutations in exon 11 of the Medium Chain Acyl-CoA dehydrogenase (MCAD) gene. Am J Hum Ge-net 54:975-988.
16. Gregersen N, Winter V, Lyonnet S, Saudubray JM, Wendel U, An-dresen BS, Kølvraa S, Bolund L and Bross P. (1994) Molecular characterization and urinary excretion pattern of metabolites in two families with MCAD deficiency due to compound heterozygosity with a 13 basepair insertion in one allele. J Inher Metab Dis 17:169-184.
17. Andresen BS, Bross P, Knudsen I, Winter V, Kølvraa S, Bolund L, Gregersen N (1994) Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency due to heterozy-gosi-ty for the common mutation and an allele resulting in low levels of MCAD mRNA. J Inher Metab Dis 17:275-278.
18. Bross P, Jensen TG, Andresen BS, Kjeldsen M, Nandy A, Kølvraa S, Ghisla S, Rasched I, Bolund L, Gregersen N (1994) 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. Biochem Med Metab Biol 52:36-44.
19. Jensen HK, Jensen TG, Jensen LG, Hansen PS, Kjeldsen M, Andresen BS, Nielsen V, Meinertz H, Hansen AB, Bolund L, Færgeman O, Gregersen N (1994) Charac-terization of a disease-causing Glu^{119}-Lys mutation in the low density lipoprotein receptor gene in two Danish families with heterozy-gous Familial Hyperchole-sterolemia. Hum Mutat 4:102-113.
20. Kristensen MJK, Bross P, Andresen BS, Kmoch S and Gregersen N. A prevalent amino acid polymorphism at position 209 in the short-chain acyl-CoA dehydrogenase (SCAD) gene. (1994) Hum Mol Genet 3:1711.
21. Gregersen N, Winter V, Jensen PKA, Holmskov A, Kølvraa S, Andresen BS, Christensen E, Bross P, Lundemose J and Gregersen M. (1995) Prenatal diagnosis of medium-chain acyl-coA dehydrogena-se (MCAD) deficiency in a family with a fatal case of sudden unexpected death in childhood. Prenatal diagnosis 15:82-86.
22. Jensen TG, Bross P, Andresen BS, Lund TB, Kristensen TJ, Jensen UB, Winter V, Kølvraa S, Gregersen N and Bolund L (1995) Comparison between medium-chain acyl-CoA dehydroge-nase (MCAD) mutant proteins overexpressed in bacterial and mammalian cells. Hum Mutat 6:226-231.
23. Sandvej K, Peh SC, Andresen BS, Pallesen G (1994) Identification of potential hotspots in the carboxy-terminal part of the EBV-BNLF-1 gene in both malignant and benign EBV associated diseases: High frequency of 30-bp deletion in Malaysian and Danish peripheral T-cell lymphomas. Blood 84:4053-4060.
24. Bross P, Jespersen C, Jensen TG, Andresen BS, Kølvraa S, Bo-lund L and Gregersen N. (1995) Effects of two mutations detected in medium-chain acyl-CoA dehy-droge-nase (MCAD)-deficient patients on folding, oligomer assembly, and stability of MCAD enzyme. J Biol Chem 270:10284-10290.
25. Corydon MJ, Gregersen N, Lehnert W, Ribes A, Rinaldo P, Kmoch S, Christensen E, Kristensen TJ, Andresen BS, Bross P, Winter V, Martinez G, Neve S, Jensen TG, Bolund L and Kølvraa S. (1996) Ethylmalonic aciduria is associated with an a amino acid variant of short-chain acyl-Coenzyme A dehydrogenase. Pediatr Res 39:1-8.
26. Bross P, Andresen BS, Knudsen I, Kruse T, and Gregersen N. (1995) Human ClpP protease: cDNA sequence, tissue-specific expression and chromosomal assignment of the gene. FEBS letters 377:249-252.
27. Andresen BS, Bross P, Vianey-Saban C, Divry P, Zabot MT, Roe CR, Nada MA, Byskov A, Kruse TA, Kristiansen K, Neve S, Corydon MJ, Gregersen N (1996) Cloning and characterization of human very-long-chain acyl-CoA dehydrogenase cDNA, chromosomal assignment of the gene and identification in four patients of 9 different mutations

within this gene. Hum Mol Genet 5:461-472 and 1390.
28. Jensen TG, Andresen BS, Jensen HK, Jensen LG, Heath F, Pedersen S, Nielsen V, Jensen UB, Lund TB, Gregersen N, Kølvraa S and Bolund L (1996) Rapid characterization of mutations in the low density lipoprotein receptor (LDLR) gene by overexpression in COS cells. German Journal of Gastroenterology 34(suppl. 3) 9-11.
29. Andresen BS, Vianey-Saban C, Bross P, Divry P, Roe CR, Nada MA, Knudsen I and Gregersen N (1996) The mutational spectrum in very-long-chain acyl-CoA dehydrogenase (VLCAD) deficiency. J Inher Metab Dis.19:169-172.
30. Jensen HK, Jensen TG, Færgeman O, Jensen LG, Andresen BS, Corydon MJ, Andreasen PH, Hansen PS, Heath F, Bolund L, Gregersen N (1997) Two mutations in the same low density lipoprotein receptor allele act in synergy to reduce receptor function in heterozygous familial hyperchole-sterolemia. Hum Mut 9:437-444.
31. Jensen HK, Holst H, Jensen LG, Jørgensen MM, Andreasen PH, Jensen TG, Andresen BS, Heath F, Hansen PS, Neve S, Kristiansen K, Færgeman O, Kølvraa S, Bolund L, Gregersen N (1997) A common W556S mutation in the LDL receptor gene of Danish patients with familial hyperchole-sterolemia encodes a transport-defective protein. Atherosclerosis 131:67-72.
32. Andresen BS, Bross P, Udvari S, Kirk J, Gray RGF, Kmock S, Chamoles N, Knudsen I, Winter V, Kølvraa S, Bolund L, Wilcken B, Yokota I, Harpey JP, Hart K, Packman S, Saudubray JM, Hale DE, Bolund L, Kølvraa S, Gregersen N (1997) The molecular basis of medium-chain acyl-CoA dehydroge-nase (MCAD) deficiency in compound heterozygous patients - Is there a correlation between genotype and phenotype ? Hum Mol Genet6:695-708.
33. Sandvej K, Gratama JW, Munch M, Zhou XG, Bolhuis RLH, Andresen BS, Gregersen N, Hamilton-Dutoit S (1997) 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 90:323-330.
34. Corydon MJ, An-d-r-e-sen BS, Bross P, Kruse TA, Kjeldsen M, Andreassen PH, Eiberg H, Kølvraa S and Gregersen N. (1997) Structural organization of the human short-chain acyl-CoA dehydrogenase gene. Mammalian Genome8:922-926.
35. Smelt G, Poorthuis BJHM, Onkenhout W, Scholte HR, Andresen BS, Gregersen N, VanDuinen SG, Wintzen AR (1998) Adult onset very-long-chain acyl-Coenzyme A dehydrogena-se deficiency. Clinical, morphological, biochemical and genetic features. Ann. Neurology 43:540-544.
36. Gregersen N, Winter V, Corydon MJ, Corydon TJ, Ribes A, Martinez G, Bennett MJ Vianey-Saban C, Bhala A, Hale DE, Rinaldo P, Lehnert W, Kmoch S, Andresen BS, Bross P, Bolund L and Kølvraa S. (1998) 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 normal population, as was the case for 625G>A, together conferring suceptibility to ethylmalonic aciduria. Hum Mol Genet 7:619-627.
37. Udvari S, Bross P, Andresen BS, Gregersen N, Engel PC. (1999) Biochemical characterisation of mutations of human medium-chain acyl-CoA dehydrogenase. Adv Exp Med Biol. 466:387-93.
38. Udvari S, Bross P, Andresen BS, Gregersen N, Engel PC. (1998) Mutations of human medium-chian acyl-CoA dehydrogenase. Biochem Soc Trans. 26(1):S65.
39. Scholte HR, VanCoster RNA, deJonge PC, Poorthuis BJHM, Jeneson JAL, Andresen BS, Gregersen N, deKlerk JBC and Busch HFM (1999) Myopathy in very-long-chain acyl-Coenzyme A dehydrogena-se deficiency: clinical and biochemical differences with the fatal cardiac phenotype. Neuromuscular Disorders 9:313-319.
40. Sørensen CB, Ladekær-Mikkelsen AS, An-d-r-e-sen BS, Brandrup F, Veien NK, Buus SK, Anton-Lamprecht I, Kruse TA, Jensen PKA, Eiberg H, Bolund L, Gregersen N (1999) Identification of novel and known mutations in the genes for keratin 5 and 14 in Danish patients with Epidermiolysis Bullosa Simplex: Correlation between genotype and phenotype. J Invest Dermatol 112:184-90.
41. Andresen BS, Olpin S, Kvittingen EA, Auguostodes-Savvopoulou P, Lindhout D, Halley DJ, Vianey-Saban C, Wanders RJA, Ijlst L, Schroeder LD, Bolund L, Gregersen N (1999) DNA-based prenatal diagnosis for VLCAD deficiency J Inher Metab Dis 22:281-285.
42. Andresen BS, Olpin S, Poorthuis B, Scholte HR, Vianey-Saban C, Wanders RJA, Ijlst L, Morris A, Pourfazam M, Bartlett K, Bross P, Lund H, Corydon TJ, Schrøder LD, Winter V, Bolund L, Gregersen N (1999) It is possible to correlate genotype with disease phenotype in very-long-chain acyl-CoA dehydrogena-se (VLCAD) deficiency. Am J Hum Genet 64: 479-494.
43. Siggaard C, Rittig S, Corydon TJ, Andreasen PH, Jensen TG, Andresen BS, Robertson GL, Gregersen N, Bolund L, Pedersen EB (1999) 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. J Clin Endocrinol Metab. 84:2933-41.
44. Bross P, Pedersen P, Winter V, Nyholm M, Nagstrup Johansen B, Olsen RKJ, Corydon MJ, Andresen BS, Eiberg H, Kølvraa S, Gregersen N (1999) A polymorphic variant in the human electron transfer flavoprotein a-chain (a-171) displays decreased thermal stability and is overrepresented in very-long-chain acyl-CoA dehydrogenase deficient patients with mild childhood presentation. Molecular Genetics and Metabolism 67:138-147.
45. Brandt-Sørensen C, Ladekjær-Mikkelsen AS, Andresen BS, Brandrup F, Veien NK, Buus SK, Anton-Lamprecht I, Kruse TA, Jensen PKA, Eiberg H, Bolund L, Gregersen N. (1999) Identification of novel and known mutations in the genes for keratin 5 and 14 in Danish patients with epidermolysis Bullosa Simplex: Korrelation mellem genotype og fænotype i danske patienter. Ugeskrift for læger 162:1873-1876.
46. Jensen PKA, Brandt-Sørensen C, Andresen BS, Brandrup F, Veien NK, Buus SK, Gregersen N, Bolund L. (1999) Keratinsygdomme. Ugeskrift for læger 162:1867-1872.
47. Bross P, Corydon TJ, Andresen BS, Jorgensen MM, Bolund L, Gregersen N (1999) Protein misfolding and degradation in genetic diseases. Hum Mutat 14:186-198.
48. Merinero B, Pascual Pascual SI, Perez-Cerda C, Gangoiti J, Castro M, Garcia MJ, Pascua Castroviejo I, Vianey-Saban C, Andresen B, Gregersen N, Ugarte M (1999) Adolescent myopathic presentation in two sisters with very long-

chain acyl-CoA dehydrogenase deficiency. J Inherit Metab Dis 22:802-810.
49. Gregersen N, Bross P, Jørgensen MM, Corydon TJ, Andresen BS (2000) Defective folding and rapid degradation of mutant proteins is a common disease mechanism in genetic disorders. J Inherit Metab Dis23:441-447.
50. Sandvej K, Andresen BS, Zhou XG, Gregersen N, Hamilton-Dutoit S (2000) Analysis of the Epstein-Barr virus (EBV) latent memebrane binding protein 1 (LMP-1) gene and promoter in Hodgkin`s disease isolates: selection against a variant with mutations in the LMP-1 ATF1/CREB-1 binding site. Mol Pathol 53:280-288.
51. Gregersen N, Andresen BS, Corydon MJ, Corydon TJ, Olsen RK, Bolund L, Bross P (2001) Mutation analysis in mitochondrial fatty acid oxidation defects: Exemplified by acyl-CoA dehydrogenase deficiencies, with special focus on genotype-phenotype relationship. Hum Mutat 18:169-89.
52. Gregersen N, Andresen BS, Bross P (2000) Prevalent mutations in fatty acid oxidation disorders: diagnostic considerations. Eur J Pediatr 159 Suppl 3:S213-8.
53. Andresen BS, Corydon, TJ, Wilsbech M, Bross P, Schroeder LD, Hindkær TF, Bolund L, Gregersen N (2000) Characterization of the mouse ClpP protease cDNA, gene and protein. Mammalian Genome 11:275-280.
54. Spiekerkotter U, Schwahn B, Korall H, Trefz FK, Andresen BS, Wendel U (2000) Very-long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency: monitoring of treatment by carnitine/acylcarnitine analysis in blood spots. Acta Paediatr. 89:492-495.
55. Corydon TJ, Jespersgård C, Wilsbech M, Andresen BS, Børglum AD, Gregersen N, Bross P (2000) Human and mouse ClpX orthologues: cDNA sequences, structure of the human gene, tissue-specific expression, chromosomal mapping, and subcellular localisation. Mammalian Genome 11:299-305.
56. Andresen BS, Christensen E, Corydon TJ, Bross P, Pilgaard B, Simonsen H, Knudsen I, Schroeder LD, Gregersen N, Skovby F (2000) Isolated 2-methylbutyrylglycinuria caused by short/branched-chain acyl-CoA dehydrogenase (SBCAD) deficiency: Identification of a new enzyme defect, resolution of its molecular basis and evidence for distinct acyl-CoA dehydrogenases in isoleucine and valine metabolism. Am J Hum Genet 67:1095-1103.
57. Scholte HR, VanCoster RNA, deJonge PC, Poorthuis BJHM, Jeneson JAL, Andresen BS, Gregersen N, deKlerk JBC and Busch HFM (2000) Myopathy in very-long-chain acyl-Coenzyme A dehydrogena-se deficiency. Clinical and biochemical differences with the fatal cardiac phenotype. Neurology (Review series) 2:15-16.
58. Gregersen N, Bross P, Andresen BS, Pedersen CB, Corydon TJ, Bolund L (2001) The role of chaperone-assisted folding and quality control in inborn errors of metabolism: protein folding disorders. J Inherit Metab Dis. 24:189-212.
59. Touma EH, Rashed MS, Vianey-Saban C, Sakr A, Divry P, Gregersen N, Andresen BS (2001) A severe genotype with favourable outcome in very long chain acyl-CoA dehydrogenase deficiency. Archives of Disease in Childhood 84:58-60
60. Merinero B, Pascual SI, Perez-Cerda C, Gangoiti J, Castro M, Garcia MJ, Castroviejo IP, Vianey-Saban C, Andresen B, Gregersen N, Ugarte M (2001) Adolescent myopathic pre-sen-tation in two sisters with very long chain acyl-CoA dehydrogenase deficiency. In: Wahl-gren NG (ed) Review Series: Neurology. Charles Garza, Stockholm, pp 22-26.
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Software and Database submission

1. DeepClip - https://deepclip-web.compbio.sdu.dk
2. VulExMap - https://vulexmap.compbio.sdu.dk

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Teaching Portfolio

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1.Formel pædagogisk uddannelse

•1996 Educational course for assistant professors, Sandbjerg gods, AU.

2.Uddannelsesadministrative opgaver

•Head of teaching, Dept. Human Genetics, AU (2007-2008).
•Educational board, BMB, SDU (2009-present).
•Head of teaching, BMB, SDU (2015-present).
•Studienævnsrepræsentant for BMB, NAT, SDU.
•Medlem af uddannelsesudvalget, NAT, SDU.
•Viceinstitutleder og ansvarlig for uddannelserne i Biomedicin og BMB.
•Member of the Exceutive Executive board: Nordic Network Biomedicine, Nordplus/ERASMUS+, OERCompBiomed (2017-)

3.Erfaring med undervisning, vejledning og eksamen

•Teaching in Medical Genetics for Medical Students and Students in Odontology, Faculty of Health, AU for more than 10 years from 1995-2008. Lectures, examinatoriums and lab course. Together with Ass. Professor Thomas Corydon, I developed and was the responsible teacher for the 3 day laboratory course in Medical genetics at Faculty of Health, AU.

This course had >200 participants every half year.•In 2008 I developed and designed the new course in Medical Genetics for the new study in Molecular Medicine together with 3 other associate professors at AU. I left AU for my position at SDU, when the course started, but it has been running successfully since then. I am now external censor for this course. •I have for more than 15 years been developing and teaching at annual courses (Lab and lectures) in for Molecular Medicine for Medical Science Technologists (Hospitalslaboranter - Dbio). •I have teen teaching and co-designing a 4 day Ph.D. course, at AU in Molecular Medicine (Lab exercises and lectures) for more than 15 years until my assignment in 2008 at SDU. •I have for the last 14 years (2008-2020) given 2-3 annual lectures at Ph.D. courses at AU (”From gene to function”, ”Molecular Medicine” and ”Molecular Genetics in Endocrinology and Cardiology – Translational Medicine”). •I have given numerous lectures at the A-courses in clinical genetics, which is part of the specialty education for MD’s in Clinical Genetics. •I was for 5 years (2009-2013) the responsible teacher for the course in advanced Molecular Biology (BMB508) and BMB809 – 10ECTS (Lectures, Examinatoriums and 3 day lab course). I shared the lectures with Professor Jens Andersen, BMB and carried out the 3 day lab course together with members from my own research group. •Together with Associate Professor Finn Kirpekar I developed “Elitemodulet i Molecular Diagnostics (BMB814 - 25 ECTS – 1 year). This was an interdisciplinary course between Faculty of Health and Faculty of Science, SDU employing teachers from both faculties. I developed and gave >½ the lectures in the module in Medical Genetics (20 Lectures, 18 Examinatoriums and 4 day lab course). This course turned out to be too hard to coordinate, so after 2 years we decided not to offer it anymore.•I developed the course, Human Molecular Medical Genetics – Diagnostics and Pathology (BMB815) – 5 ECTS and this has now been running successfully since 2011 and is again offered in the autumn of 2020. •In the four years, 2010-2014, autumn, I shared the lectures in molecular biology for medical students (Modul 2) with another Professor at BMB. •I have in 2013 developed and has since been the responsible teacher at the 5 ECTS bachelor course “Human diseases” BMB531 for students in biochemistry and molecular biology (BMB) and students in biomedicine at SDU. This course is attended by approx.. 200 students. I share the lectures in this course with Professor, Stephen Douthwaite, BMB and Associate Professor Barbara Guerra, BMB. •Since autum 2014 I have given 4 lectures/year in the course BMB822, “Modern trends and technologies in molecular cell biology”.•In the period up until 2008 I was censuring the written exams from the BMB508 course and before then the course in Genetics from, BMB, SDU.•Since 1991 I have supervised >50 master thesis students (8 last year – 8 this year), numerous bachelor students, ISA and ERASMUS students, who have performed their projects in my laboratory. Additionally I have also been supervisor on several students performing their projects outside my lab. •I have supervised 2 Ph.D. students in Medicine at AU. At SDU I have supervised four Ph.D. students in Molecular Biology who finished in December 2013, May 2015, Oct. 2018 and February 2020. I am currently supervising 3 Ph.D. students in Molecular Biology.•I have supervised one candidate in the pedagogical course for assistant professors at SDU in the Autumn of 2012. •For the past 12 years (2008-2020) I have been censuring Medical Genetics written exam for Medical Students from Faculty of Biomedicine, AU. •I have also censured the written exam in Medical Genetics for Students in Molecular Medicine Students from Faculty of Biomedicine, AU for the last >10 years.•I have for several years censured the written exams in Medical Genetics for medical students at Faculty of Health, KU.•For 13 years (2008-2020) I have censured the written reports from the course in Applied Molecular Biology, Faculty of Science, AU.•I have censured numerous bachelor- and master- of science thesis, gold medal- (KU) or diploma- (AU) assignments etc. from the Faculties of Health and Faculties of Science from AU and KU. •I have been external evaluator of numerous Ph.D. dissertations from AU, SDU and KU, as well as from other countries.

4.Metoder, materialer og redskaber

•As can been seen from the above I have extensive experience with teaching in many contexts spanning from 1 day- 1 week- courses for medical laboratory technologist to MD’s taking their specialty education or Ph.D. courses. I have lectured for big audiences >200 students at the master- bachelor- level in Medicine, Odontology, Biochemistry and Molecular Biology and I also have extensive experience with teaching in smaller audiences of 10-30 people. I also have extensive experience with development, planning and teaching at hands-on lab courses for different types of students. •Depending on the size of the audience I use a mixture of PowerPoint presentation and writing on blackboard/white board. •I have written several lab course manuals for the various lab courses.•I attempt to include questions and small activities during my lectures in order to keep the students focused.•In recent years I have been involved in developing online courses.

5.Uddannelsesudvikling og universitetspædagogisk forskning

•I participated in the ”Future Search” process in the spring of 2011 aiming at overall improvements of the educations at Faculty of Science, SDU.
•In the autumn of 2012 I was a member of the task-force at the Faculty of Science, SDU, who should redesign the common 1 year in Science for all students.
•As chair of the teaching committee and Deputy Head of Department at BMB I am constantly involved in developing and redesigning courses and the overall planning of our educations.

6.Refleksion over egen pædagogisk praksis og fremtidig udvikling, herunder undervisningsevalueringer

•I have always had positive evaluations of my courses.
•I find it important to challenge and engage students and focus on teaching quality.
•Presently and in the next years I will focus on development of online courses, and how to blend in online elements into

traditional theoretical and practical courses.