Research unit of Clinical Genetics, SDU
Department of Clinical Genetics, OUH
Department of Clinical Research, SDU
Clinical Genome Center (CGC)

ANNUAL REPORT 2024

OUH
Odense
University Hospital

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Clinical Genome Center

SDU

WELCOME

Here, in my first year back at the Department of Clinical Genetics and my first year as Head of Research, it is my privilege to present the annual report from The Research unit Clinical Genetics at the Department of Clinical Genetics, Odense University Hospital (OUH), and the Department of Clinical Research at the University of Southern Denmark (SDU).

Our research focuses on rare and complex hereditary diseases, cancer and fetal genetics.

The Department of Clinical Genetics houses the Clinical Genome Center (CGC), led by Professor Mads Thomassen; a resource center offering core facilities and support to research projects at OUH and SDU as well as collaborating with other institutions.

The department moreover houses the Center of Rare and Complex Diseases (CAKS), that supports the multidisciplinary care of patients with rare genetic diseases, as well as the OUH ERN Secretariat. The department is head of one of the 9 European Reference Networks at OUH, ERN-ITHACA, the European Reference Network for Rare Malformation Syndromes, Intellectual and Other Neurodevelopmental Disorders, led by Professor Lilian Bomme Ousager. The department participates in two other networks, ERN-VASCERN, the ERN for Rare, Multisystemic Vascular Diseases, and ERN-SKIN, the ERN for Rare and Complex Skin Diseases.

In addition, we are delighted to have a close collaboration with Genome Biology Research at the Department of Molecular Medicine, SDU, led by Professor Vijay Tiwari, and with the Danish Twin Research Center, Department of Clinical Research, SDU, led by Professor Kaare Christensen.

This year, we warmly welcomed Professor Kirsten Ohm Kyvik to our department, employed as physician and professor.

I would like to thank all the researchers, employees, collaborators, patient representatives and all the patients and healthy volunteers participating in our research projects for all of your dedicated work and engagement. Finally, I would also like to thank all the foundations that support our work.

For our activities and publications – I hope you will take a look at the following pages.

Thank you

Head of Research Anja Lisbeth Frederiksen
MD, PhD, Professor

CONTENTS

ORGANISATION

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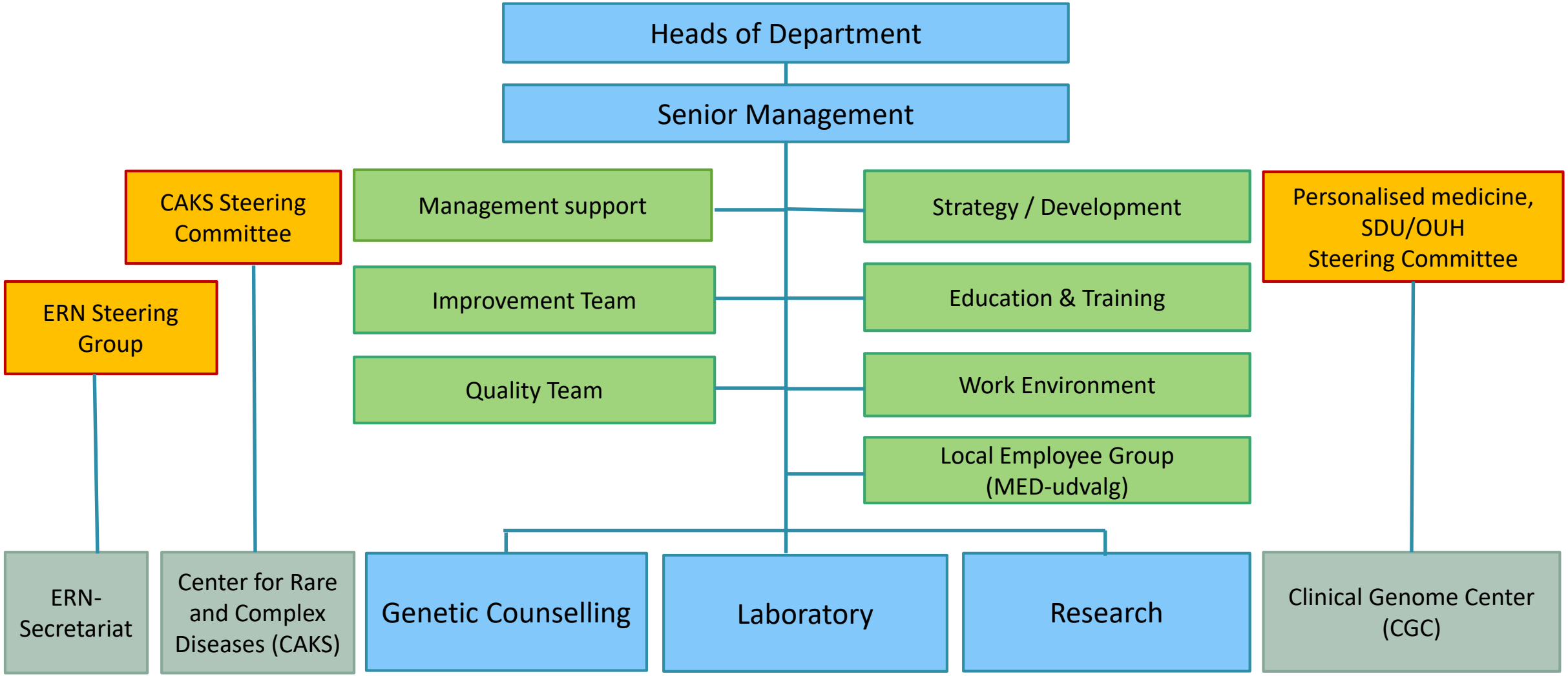
Department of Clinical Genetics

and associated centres



Upcoming PhD defences (1)

Nhu Do, PhD student

Supervisors: **Professor Mads Thomassen, Professor Torben Kruse,** Department of Clinical Genetics, OUH.

Project title: ***Integrated genomic analysis of primary breast tumors. A possible tool towards reduction of overtreatment.***
PhD defence: Fall 2025

The PhD project is comprised of three studies related to prediction of clinical outcome in systemically untreated breast cancer (BC) patients. The ultimate goal of the project is to reduce overtreatment for these patients as more than 90% of all current BC patients receive adjuvant systemic treatment, despite the fact that up to 40% might not benefit from it.

Study I highlights the relative ability of mRNA and long non-coding RNA (lncRNA)-based signatures to predict recurrence in low-risk BC patients. We furthermore compared the prognostic utility of the two RNA molecules using two different classification strategies.

In the second study, we proposed that an integration of mRNA, lncRNA and miRNA gene expression data would improve patient classification performance. The applied classification approach had been shown to be most favorable in the first study and ensured a sensitivity of ≥90%, while maximizing specificity.

In Study III, we performed whole exome sequencing on the majority of our patient cohort. Somatic variant calling of single nucleotide variants (SNVs) and insertion-deletion mutations (indels) was completed and further analyses were performed to obtain more profound biological and functional understanding of the somatic genetic basis of low-risk BC.

Upcoming PhD defences (2)

Project title: ***Predicting response to medical treatment of inflammatory bowel disease (IBD) using transcriptomics analysis on intestinal biopsies and blood: a prospective cohort study of personalised medicine*.**
PhD hand-in: 18 January 2025.

Zainab's PhD research aimed to identify predictive biomarkers for anti-TNF treatment response.

- Study 1 conducted a multi-omics analysis of intestinal biopsies from the BELIEVE cohort to identify potential biomarkers.
- In Study 2, 33 candidate biomarker genes were evaluated in RNA data from biopsies of inflammatory bowel disease patients.
- In Study 3, using single-cell RNA sequencing on biopsies from two ulcerative colitis patients, cell signatures pre- and post-treatment were analysed.

In 2023, Zainab received a grant of DKK 857,000 from the Region of Southern Denmark's pool for Independent and Strategic Research. The grant was for operations, including running single-cell RNA sequencing, spatial transcriptomics, and an automated cell counter.

Zainab Hikmat, PhD student

Main supervisor: **Professor Vibeke Andersen**, Sygehus Sønderjylland and Department of Regional Health Research.
Supervisors: **Professor Mads Thomassen**, Department of Clinical Genetics, OUH, **Maja Dembic, PhD**, CGC, and **Associate Professor Tue Bjerge Bennike**, University of Aalborg.

Ongoing PhD studies (1)

Alexander Venzel Rudbeck, PhD student
Project title: '*Whole genome sequencing of cell-free DNA from metastatic breast cancer patients: A study of accuracy and response*'
Supervisors: Professor Mads Thomassen, Professor Torben Kruse, Department of Clinical Genetics, OUH.

Astrid Skov Midtiby, PhD student
Project title: '*Identification of the underlying genetic mechanisms in patients with skeletal dysplasias to improve diagnosis, genetic counseling and patient follow-up*'
Supervisor: Professor Lilian Bomme Ousager, Department of Clinical Genetics. Co-supervisors: Professor Jens Michael Hertz, Department of Clinical Research, SDU, and Professor Zeynep Tümer and Chief senior consultant Hanne Buciek Hove, Rigshospitalet.

Emilie Boye Lester, PhD student
Project title: '*Long-Read Whole Genome Sequencing - 3rd Generation Nanopore Sequencing In Clinical Genetic Diagnostic*'
Supervisor: Associate Professor Martin Jakob Larsen**,** Department of Clinical Genetics, OUH.

Louise Adel Jensen, PhD student
Project title: '*The Molecular Characterization of Familial Breast Cancer with no Confident Genetic Explanation - Identification of Subgroups and Classification of Genetic Variants*'
Supervisor: Professor Mads Thomassen , Department of Clinical Genetics. Co-Supervisor: Professor Thomas van Overeem Hansen, Department of Clinical Genetics, Rigshospitalet

Ongoing PhD Studies (2)

Maria Lissel Isaksson, PhD student
Project title: '*Genetic causes of short stature in children*'
Supervisor: Professor Lilian Bomme Ousager. Co-supervisors: Associate Professor Martin Larsen, PhD Stine Bjørn Gram, Department of Clinical Genetics, and Associate Professor Dorte Hansen, H. C. Andersen Children's Hospital.

Mikkel Møller Henriksen, PhD student
Project title: '*Characterization of tumor load and mutational profile in patients with gastroesophageal cancer using circulating tumor DNA*'
Supervisors: Professor Mads Thomassen, Professor Torben Kruse, Department of Clinical Genetics, OUH.

Nhu Do, PhD student
Project title: '*Integrated genomic analysis of primary breast tumors. A possible tool towards reduction of overtreatment*'
Supervisors: Professor Mads Thomassen, Professor Torben Kruse, Department of Clinical Genetics, OUH.

Zainab Hikmat, PhD student
Project title: '*Predicting response to medical treatment of inflammatory bowel disease (IBD) using transcriptomics analysis on intestinal biopsies and blood: a prospective cohort study of personalised medicine*'
Supervisors: Professor Vibeke Andersen, Sygehus Sønderjylland and Department of Regional Health Research, and Professor Mads Thomassen, Department of Clinical Genetics, OUH.

Completed PhD studies (1)

Stine Bjørn Gram, PhD

Supervisor: **Professor Lilian Bomme Ousager.**
Co-supervisors: **Professor Klaus Brusgaard** and **Professor Anette Bygum.**

Title: ***Clinical and genetic characterisation of palmoplantar keratoderma***.
PhD defence: 26 April 2024.

The PhD project was about the rare and hereditary disease palmoplantar keratoderma (PPK), a disease characterised by hard and thickened skin on the palms and soles, but complicated by a very heterogeneous nature including several different clinical subtypes and numerous different genetic types from isolated skin disease to syndromes with risk of other diseases. The project consisted of three studies (I-III):

In Study I, a large cohort of 142 patients (78 families) with PPK was established (making it the largest cohort of its kind at the time of publication). The study describes the clinical characteristics and results of systematic genetic testing. A genetic diagnosis was found in 80 percent, which testified to the relevance of genetic testing for patients with PPK.

Study II investigated a frequent variant (*AAGAB*, c.370C>T) in the study population with punctate PPK in the Region of Southern Denmark. The variant was found to be a founder variant with an estimated age of 12.1 generations. Based on these results, we recommended testing for the variant as initial screening in our region and potentially for all Danish patients presenting punctate PPK.

Study III was a systematic review evaluating the literature on the suggested risk of malignancy for patients with punctate PPK.

Completed PhD studies, in collaboration with other departments (1)

Annette Rønholt Larsen, PhD

Title: ***Advanced Genetic Investigations of Unexplained Hyperinsulinemic Hypoglycemia in Children with Atypical Pancreatic Histology or NI-PHHS***
PhD defence: 3 May 2024

The PhD thesis was based on four studies on somatic genetic alterations in pancreatic beta cells that lead to overproduction of insulin.

Opponents at the PhD defence were Professor Klaus Mohnike and Professor Jens Høiriis Nielsen. Chair of the assessment committee was Associate Professor Charlotte Brasch Andersen, Department of Clinical Genetics.

Supervisor: **Professor Henrik Thybo Christesen,** H.C. Andersen Children's Hospital.
Co-supervisors: **Professor Klaus Brusgaard,** Department of Clinical Genetics, **Professor Sönke Detlefsen,** Department of Clinical Pathology, and **Professor Ditte Caroline Andersen,** Research Unit for Clinical Biochemistry, OUH.

Completed PhD studies, in collaboration with other departments (2)

Kirstine Øster Andersen, PhD

Project title: ***Genetic alterations in insulinomas with and without multiple endocrine neoplasia type 1: Understanding tumourigenesis in beta-cells*.**
PhD defence: 8 March 2024

Kirstine's project focused on DNA, RNA, protein and epigenetic markers in insulinoma tissue. Insulinomas are rare, benign tumours that arise in the beta cells of the pancreas. The increased cell count leads to an overproduction of insulin, causing blood sugar levels to drop and can lead to a condition called hyperinsulinaemic hypoglycaemia. The only way to cure insulinomas is through surgery.

Supervisor: **Professor Henrik Christesen**, H.C. Andersen Childrens' Hospital.
Co-supervisor: **Professor Klaus Brusgaard**, Department of Clinical Genetics, OUH.

Ongoing Master studies

Anne Kulmback Munch, Medical student. Project title:
'Classification of gene variants in a Danish population suspected of predisposition to hereditary breast cancer and/or ovarian cancer'
Supervisors: Associate Professor Susanne Eriksen Boonen, Professor Mads Thomassen and PhD Qin Hao.

Emilie Kofoed Hansen, Medical student. Project title:
'NGC hereditary heart disease: Diagnostic yield and genotype-phenotype correlation'
Supervisors: Professor Lilian Bomme Ousager (main), PhD Qin Hao, and PhD Thomas Morris Hey.

Frederik Møller Larsen, Student of Biomedicine. Project title:
'Mutational profiles of gastroesophageal cancers during treatment and identification of emerging treatment-resistance mutation'
Supervisors: Professor Mads Thomassen, PhD student Mikkel Møller Larsen and PhD student Alexander Venzel Rudbeck.

Jeppe Hannibal Niemann, Health IT stud. Project title: '*Language Model for Genetic Diagnostics*'
Oliver Rønholt Grimm, Health IT stud. Project title: '*Language Models for Generating Summaries of Clinically Important EMR-notes in the Genetic Analysis Process*'
Jeppe's and Oliver's projects both involve using large language models (à la ChatGPT) to find and extract relevant clinical information, symptoms and diagnoses from the patient's medical record to help with genome interpretation.
Supervisors: Associate Professor Martin Larsen and PhD Pernille Tørring.

Completed master studies

Naja Slemming-Adamsen, MSc in Medicine
Thesis: '*Splice prediction of pathogenic variants and variants of unknown significance in clinical relevant genes*'.
Main supervisor: Associate Professor Susanne Eriksen Boonen. Co-supervisors: Professor Mads Thomassen, PhD Qin Hao and MSc Caroline Hey Bækgaard

Mette Lykke Jørgensen, MSc in Medicine
Thesis: '*Description of genotypes and clinical phenotypes in Danish families with autosomal dominant palmoplantar keratoderma (PPK) caused by heterozygotic DSG1 variants'*
Supervisors: Professor Lilian Bomme Ousager and PhD Stine Bjørn Gram.

Jens Christian Tabori Kraft, MSc in Medicine
Thesis: '*Analysis of the genetic background for dyslexia in a large Danish family'*
Supervisor: Associate professor Charlotte Brasch Andersen.

Watfa Moussa Chibli, MSc in Computational Biomedicine
Thesis: '*Haplotype analysis of the BRCA1 and BRCA2 genes'*
Supervisors: Professor Mads Thomassen, PhD student Mikkel Møller Henriksen, PhD student Alexander Venzel Rudbeck.

Researchers in the Research unit Clinical Genetics and the Department of Clinical Research, SDU

(In alphabetical order by first name)

- **Anette Bygum**, MD, DMSci, Professor
- **Anja Lisbeth Frederiksen**, Head of Research, MD, PhD, Professor
- **Anna Julie Aavild Ploug**, MD
- **Bjørk Ditlev Marcher Larsen,** MSc, PhD
- **Britta Schlott Kristiansen,** MD
- **Charlotte Brasch Andersen**, MSc, PhD, Associate Professor
- **Christina Fagerberg**, MD, Associate Professor
- **Jens Michael Hertz**, MD, DMSci, Professor
- **Jonas Mengel-From**, MSc, PhD
- **Kirsten Ohm Kyvik**, MD, PhD, MPM, Professor
- **Klaus Brusgaard,** MSc, PhD, Professor
- **Kaare Christensen**, MD, PhD, Professor
- **Lilian Bomme Ousager**, Head of Department, MD, PhD, Professor
- **Mads Thomassen**, Head of CGC, MSc, PhD, Professor
- **Marianne Nygaard**, MSc, Phd, Associate Professor
- **Maja Dembic**, MSc, PhD
- **Mark Burton**, MSc, PhD, Associate Professor
- **Martin J. Larsen**, MSc, PhD, Associate Professor
- **Morten Buch Engelund**, MSc, PhD
- **Pernille M. Tørring**, MD, PhD
- **Qin Hao**, PhD
- **Sepideh Sadegh**, MSc, PhD
- **Sofie Fredberg Jørgensen**, MD
- **Steffen Møller Bøttger,** MSc, PhD
- **Stine Bjørn Gram**, MD, PhD
- **Susanne Eriksen Boonen**, MD, PhD, Associate Professor
- **Torben Kruse,** MSc, PhD, Professor
- **Trine Maxel Juul**, MD, PhD
- **Qihua Tan**, PhD, Professor

+ PhD students

Researchers in Clinical Genome Center

- **Mads Thomassen**, Head of CGC, MSc, PhD, Professor
- **Maja Dembic**, MSc in Molecular Biology, PhD
- **Mark Burton**, MSc in Bioinformatics, PhD, Associate Professor
- **Sepideh Sadegh**, MSc in Computational Biology, PhD
- **Steffen Møller Bøttger,** MSc in Molecular Biology, PhD
- **Torben Kruse,** MSc in Molecular Biology, PhD, Professor
- **Vijay Tiwari,** MSc, PhD, Professor

The next pages present examples of researchers and their research fields.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (1)

Charlotte Brasch Andersen, Associate professor, MSc and PhD, Clinical Laboratory Geneticist, ErCLG

Charlotte is involved in a number of national and international research projects.
The main research area is rare diseases which spans many different diseases and disease mechanisms. The aim of the studies is to understand the disease mechanisms and clinical symptoms behind the identified rare genetic variants.

Currently, Charlotte is involved in a project that aims to identify genes in dyslexia, combining data from whole genome sequencing and linkage analysis, and a project that aims to identify genes involved in vitamin B12 deficiency in families with severe B12 deficiency symptoms.

Charlotte is also part of multiple groups where she does DNA profiling of iPSC cell lines.

Furthermore, Charlotte is very involved in developing clinical postgraduate education:
She is part of the scientific committee for the Master's programme for Personalized Medicine, a collaboration between 5 universities in Denmark, in which she is the appointed representative for the University of Southern Denmark. She is moreover the national chairperson for the Clinical Laboratory Geneticist education and one of the main drivers behind a new course for Clinical Scientists (kliniske akademikere) in Denmark.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (2)

Louise Adel Jensen, PhD student

Title of project: ***The Molecular Characterization of Familial Breast Cancer with no Confident Genetic Explanation - Identification of Subgroups and Classification of Genetic Variants***

Louises PhD project focuses on hereditary breast cancer, utilizing whole-genome sequencing to assess tumor DNA repair ability through Homologous Recombination Proficiency/Deficiency (HRP/D). Additionally, Louise aims to identify mutational signatures to classify molecular subgroups of hereditary breast cancer and improve the interpretation of variants of unknown significance (VUS). The project encompasses several comparative studies:

Short-read (Illumina) versus long-read (Oxford Nanopore) whole-genome sequencing for detecting variants and mutational signatures in fresh-frozen breast cancer tissue.
Algorithms for HRD detection, contrasting deep sequencing (HRDetect) with shallow sequencing (ShallowHRD).
Long-read sequencing protocols to evaluate their effectiveness in evaluating splicing variants in BRCA1 and BRCA2 genes in collaboration with international partners.
These studies aim to refine variant classification and guide personalized counseling and treatment strategies for breast cancer patients.

Main Supervisor: **Professor Mads Thomassen**, Department of Clinical Genetics, OUH.
Co-Supervisor: **Professor Thomas van Overeem Hansen**, Department of Clinical Genetics, Rigshospitalet

CURRENT RESEARCH FIELD

Researchers and their research fields, example (3)

Maria Lissel Isaksson, PhD student

Title of project: ***Genetic causes of short stature in children***

Marias project examines the genetic causes of low height in children and will contribute knowledge to improve, develop and target the assessment of children with low height in the future.

The project is a collaboration between Department of Clinical Genetics and the H. C. Andersen Children's Hospital and involves whole genome sequencing of a group of children with low height as well as investigation of RNA expression, global DNA methylation (episignature) and possible identification of specific imprinting defects on blood samples from the participants.

The project also includes method development and testing of long-read sequencing with nanopore technology to determine copy number variants (CNVs), single nucleotide variants (SNVs) and methylation patterns in the patient group. The study will also establish a cohort of children with low height and create a biobank that can form the basis for future follow-up studies.

Supervisor: **Professor Lilian Bomme Ousager**, Department of Clinical Genetics, OUH
Co-supervisors: **Associate Professor Martin Larsen** and **PhD Stine Bjørn Gram**, Department of Clinical Genetics, OUH, and **Associate Professor Dorte Hansen**, H. C. Andersen Children’s Hospital.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (4)

Sepideh Sadegh, MSc in Computational Biology, PhD

Sepideh is a bioinformatician who contributes to data analysis for the users of the Clinical Genome Center (CGC) and provides support to PhD students with their data analysis.

She is primarily involved in cancer genomics projects at the CGC. Her current work focuses on circulating tumor DNA (ctDNA), a promising biomarker that can potentially improve cancer survival rates by enabling monitoring of treatment responses and the early detection of relapses. Additionally, ctDNA holds potential as a tool for prognosis. Liquid biopsies utilizing ctDNA offer several advantages over traditional methods like invasive tissue biopsies or PET/CT imaging, by being less invasive and providing quicker, more accessible results.

Sepideh has been developing computational methods for tumor-informed single-nucleotide variant detection of ctDNA in ovarian cancer using whole-genome sequencing data. Detecting ctDNA is particularly challenging due to its low abundance in the bloodstream. To address this, various noise-reduction strategies are implemented to optimize detection accuracy and improve the reliability of the method.

The utility of the newly developed ctDNA detection methods is also being explored through testing and evaluation in other cancer types, including breast and kidney cancers.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (5)

Steffen Møller Bøttger, MSc in Molecular Biology, PhD

Steffen works as a bioinformatician at the Clinical Genome Center (CGC), where he am involved in several projects spanning different research fields with cancer genetics being the largest.
Some of these projects are:

MESTAR: A subproject focused on identifying the clonal evolution of metastatic breast cancer in three patients who have not received treatment. The biological material from these patients consists of biopsies from primary tumors, metastasis, and plasma for circulating tumor DNA. The project has provided valuable insights into breast cancer development in the rare context of untreated patients.

ATEROSOMMUT: A still very active and data-heavy project focused on the etiology of atherosclerotic plaques.

ENIGMA: A multicenter, collaborative study aimed at finding the best approach for investigating the complexity of splice variants caused by mutations in the genes, BRCA1 and BRCA2. The project is based on long-read sequencing, and, among others, Steffen has been working on a tool to isolate splice isoforms.

Nanopore long-read genome sequencing: This project is specifically focused on harnessing the increased information found in long-reads compared to classical NGS data. Steffen is particularly involved in the use and development of tools for the identification and analysis of structural chromosomal rearrangements.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (6)

Stine Bjørn Gram, MD, PhD

Stine completed her PhD in April 2024. Since then, she has been working 60% of her time in the clinic, where she is training to become a specialist in clinical genetics, and spending 40% of her time on clinical research.

Stine's PhD focused on the clinical and genetic characterization of patients with palmoplantar keratoderma, a rare hereditary skin disease. She is now continuing her research on this condition, working on several different projects, including:
- Discovering new genes linked to the disease in previously undiagnosed patients.
- Characterizing specific subgroups of patients with pathogenic variants in specific genes.
- Investigating quality of life in patients with palmoplantar keratoderma.
- Publishing novel and rare variants identified in daily clinical practice.

Stine has been co-supervisor of Master student Mette Lykke Jørgensen and is co-supervisor of PhD student Maria Lissel Isakson.

In May 2024, she spent a week at the Department of Clinical Genetics at University Medical Center Groningen, who are experts in hereditary skin diseases. Afterwards, Peter van der Akker, MD, PhD, agreed to be international mentor of Stine guiding her through the next steps of her career, and they are now working to arrange future collaborative projects.

At the end of 2024, Stine received the OUH Career Grant, which ensures that she can continue her clinical research on palmoplantar keratoderma and other rare genetic skin diseases.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (7)

Qin Hao, Molecular Biologist and Bioinformatician, PhD

Current project:
Qin is working on a project that investigates the effects of genetic background on the differential manifestation of Neurofibromatosis type 1 (NF1). In this work, two manifestations have been chosen: plexiform neurofibromas (PNFs) and optic pathway glioma (OPGs). These two features are relatively common in NF1 children and are both associated with severe prognoses. The goal is to identify genes associated with the risk of developing PNFs and OPGs in patients with NF1.

Qin is part of several other exciting projects:
DesNIPT project: the aim is to optimise the NIPT method to be able to detect genetic changes in the fetus at an earlier stage of pregnancy with greater sensitivity and more precision. This would enable patients to make better choices.

SWEA project: data was collected from around 6,000 patients tested with the SWEA panel over the last 10 years. The variant classification was re-investigated in order to identify methods that can improve variant classification and thereby improve the diagnostic yield.

HHT project: Qin and colleagues contribute to the genetics part in a collaboration with the HHT center, where a lot of exciting projects are underway. One ongoing project is to study the two-hit hypothesis in AVM formation. So far, different AVM tissues from the same patient have been selected and somatic variants have been investigated.

PhD grants

Katrine Saldern Aagaard was granted DKK 1,665,843 from the Karen Elise Jensen Foundation for her upcoming PhD project:
'Clinical and genetic counselling in fetal life and childhood - overview and new perspectives'.

The project will generate new knowledge about the relationship between abnormal findings in prenatal screening and genetic diseases. The project will be based at the Centre for Fetal Genetics.
Supervisor: **Chief physician Lene Sperling,** Department of Gynaecology and Obstetrics. Co-supervisors: **PhD Pernille Tørring; Associate professor Martin Larsen,** Department of Clinical Genetics, **PhD Karina Hjort-Pedersen,** Department of Gynaecology and Obstetrics.

Malthe Folmer Genét received a PhD grant of DKK 628,000 from Steno Diabetes Center Odense for his upcoming project:
'*Bone Regulation of Glucose Homeostasis'.*

The study is a cross-sectional case-control study with the aim, through clinical glucose studies and genetic analyses of patients with the hereditary bone condition osteopetrosis (CLCN7), to:

1) clarify whether hormone secretion from bone contributes to the regulation of glucose balance
2) determine whether epigenetic biases contribute to variable phenotype.

Supervisor: **Professor Anja L Frederiksen**. Co-supervisors: **Professor Mads Thomassen,** Department of Clinical Genetics; **Professor Kurt Højlund**, Steno Diabetes Center Odense.

PHD GRANTS

Selected grants

PhD student Louise Adel Jensen received DKK 48,000 from Brødrene Hartmanns fond for medical purposes. The grant will be used to continue the work that Carl and Ellen Hertz's grant was given to, that is to analyse VUS in breast cancer-predisposing genes using somatic (tumour node) gene profiles.

Stine Bjørn Gram, MD, PhD, received DKK 260,000 for 20% research time from the OUH Research Career Fund 2024.

The grant will be used for the project *'Advancing the Understanding of Palmoplantar Keratoderma and Rare Genetic Skin Diseases'.*

Martin J Larsen, Associate Professor, MSc in Bioinformatics, PhD, received DKK 3.4 million from Novo Nordisk Foundation.
The grant will be used for the continuation of the DES-NIPT project. The project is in collaboration with Department D. The aim of the project is to develop and test a non-invasive screening test for pregnant women to identify monogenic diseases in the foetus. The project title is:
'The Future is Non-Invasive: Revealing Severe Fetal Disorders from a Single Draw of Maternal Blood - Pioneering a Comprehensive Genetic Screening Test'.

GRANTS

Selected grants

Professor Mads Thomassen and his research group received DKK 3.9 million to analyse circulating tumour DNA (ctDNA).The funding is part of a large EU grant entitled PREMIO COLLAB:
‘*Personalised response monitoring in oncology: co-creating clinical trials in advanced breast cancer.’*

Moreover, Mads and his research group received a DKK 4 million grant from Independent Research Fund Denmark for research on the treatment of breast cancer patients. The project will investigate and develop better methods to identify breast cancer patients who do not need chemotherapy or other medical treatment to fight their disease. Project title:
‘*Precision Medicine through Integrated Multi-Omics to Prevent Overtreatment of breast cancer (PreMO-BC)’.*

In addition, together with Professor Thomas van Overeem Hansen from Department of Genetics, Rigshospitalet, Mads received the RH/OUH joint research pool for the research project:
‘*Classification of MSH6 variants of uncertain significance (VUS) as benign or pathogenic using high-throughput functional analysis and tumor sequencing for improved precision genomic medicine’.*

GRANTS

CENTER FOR RARE AND COMPLEX DISEASES (CAKS) AND THE ERN SECRETARIAT

Center for Rare and Complex Diseases (CAKS) collaborates with all relevant specialties at OUH, establishes and updates patient pathways and provides genetic counselling for selected rare, hereditary and complex diseases.

In 2024, CAKS has worked to improve the administration of the diseases associated with CAKS. The work has included optimisation of registration, implementation of ORPHA codes specific to rare diseases and development of the treatment database CAKSsyd in collaboration with the Database Unit in the Region of Southern Denmark. CAKSsyd is expected to be operational at the beginning of 2025 and will provide an overview of patients affiliated with CAKS as well as the opportunity to optimise workflows around the multidisciplinary conferences in CAKS, freeing up time for clinical discussions of courses.

Number of patients connected to CAKS 31/12 2024: 811
Addition of new diseases in 2024: Phelan-McDermid syndrome, Cowden syndrome.

The OUH Secretariat for participation in the European Reference Networks (ERN) provides administrative support to the 9 ERN networks at OUH. The Department of Clinical Genetics participates in 3 of these networks.

In October 2024, CAKS and the ERN Secretariat organised its second annual meeting with a focus on rare diseases, transition from child to adult and the function of CAKS:

- Many thanks to the speakers: Nurse Julie Bech Olesen, Consultant Anders Jørgen Schou, Professor Henrik Christesen, Department of Paediatrics, and Consultant Dorte Glintborg, Department of Endocrinology.
- We look forward to seeing you all at the next annual meeting in 2025!

CAKS Annual meeting 2024

Patient representatives

Anna S. Andersen and Anne Vestergaard Youssufi are patient representatives at the Department of Clinical Genetics. Anna and Anne help us with a range of tasks including:

- Help identify relevant research questions that address the needs and challenges experienced by patients
- Improve research quality to ensure it aligns with real-world patient needs
- Advise on methods to recruit participants in a patient-friendly manner
- Assist in preparing information materials to ensure patients comprehend the study's purpose, their role and potential implications
- Provide input on ethical issues in research
- Contribute to data interpretation from a patient's perspective
- Act as a bridge between research and patient organisations
- Advise on how to share research findings clearly and relevantly
- Help increase funding opportunities through patient involvement

…and many other tasks.

Anna and Anne also participate in research seminars and research meetings whenever relevant. Their involvement contributes with a value to our department for which we are grateful.

For more information on patient representatives at OUH, visit Center for Forskning Sammen med Patienter og Pårørende (ForSa-P)

Anna S. Andersen

Anne Vestergaard Youssufi

PATIENT REPRESENTATIVES

PUBLICATIONS

The below list comprises 79 publications/articles written by 33 different authors from our research unit, 24 of these are authors of two or more publications.

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1. **Founder Variants in KRT5 and POGLUT1 Are Implicated in Dowling-Degos Disease.** Sheetal Kumar, Oleg Borisov, Carlo Maj, Damian J. Ralser, Aytaj Humbatova, Sandra Hanneken, Astrid Schmieder, Janina Groß, Laura Maintz, Andre Heineke, Jana Knuever, Christina Fagerberg, Laurent Parmentier, Waltraud Anemüller, Vinzenz Oji, Iliana Tantcheva-Poór, Regina Fölster-Holst, Joerg Wenzel, Peter M. Krawitz, Jorge Frank, Regina C. Betz. *Journal of Investigative Dermatology*.2024jan.;144(1):181-184. doi: 10.1016/j.jid.2023.04.03

2. **Severe lympho-depletion, abrogated thymopoiesis and systemic EBV positive T-cell lymphomaof childhood, a case**. Anders Asmussen, Leticia Quintanilla-Martinez, Martin Larsen, Christina Fagerberg, Marie Bækvad-Hansen, Maja Bech Juul, Kate Rewers, Klas Raaschou-Jensen, Mike Bogetofte Barnkob, Michael Boe Møller, Kristian Assing. *Leukemia and Lymphoma*. 2024jan.;65(1):118-122. Epub 2023okt. 23. doi:10.1080/10428194.2023.2264425

3. **Mapping the daily rhythmic transcriptome in the diabetic retina.** Silk RP, Winter HR, Dkhissi-Benyahya O, Evans-Molina C, Stitt AW, Tiwari VK, Simpson DA, Beli E. *Vision Research*. 2024jan.;214:108339. doi:10.1016/j.visres.2023.108339

4. **The association between frailty and perceived physical and mental fatigability: The Long Life Family Study.** Patel, Ruhee; Cosentino, Stephanie; Zheng, Esther Zhiwei; Schupf, Nicole; Barral, Sandra; Feitosa, Mary; Andersen, Stacy L.; Sebastiani, Paola; Ukraintseva, Svetlana; Christensen, Kaare; Zmuda, Joseph; Thyagarajan, Bharat; Gu, Yian; Long Life Family Study (LLFS). *Journal of the American Geriatrics Society*. 2024jan.;72(1):219-225. doi: 10.1111/jgs.18624

5. **Genetic associations with longevity are on average stronger in females than in males.** Zeng, Yi; Chen, Huashuai; Liu, Xiaomin; Song, Zijun; Lei, Xiaoyan; Lv, Xiaozhen; Cheng, Lingguo; Chen, Zhihua; Bai, Chen; Yin, Zhaoxue; Lv, Yuebin; Lu, Jiehua; Li, Jianxin; Land, Kenneth C; Yashin, Anatoli I; O'Rand, Angela M; Sun, Liang; Yang, Ze; Tao, Wei; Gu, Jun; Gottschalk, William; Tan, Qihua; Christensen, Kaare; Hesketh, Therese; Tian, Xiao-Li; Yang, Huanming; Egidi, Viviana; Caselli, Graziella; Robine, Jean-Marie; Wang, Hualin; Shi, Xiaoming; Vaupel, James W.; Lutz, Michael W; Nie, Chao; Min, Junxia. *Heliyon*. 2024jan. 15;10(1):e23691. doi:10.1016/j.heliyon.2023.e23691

6. **Male with an apparently normal phenotype carrying a BRCA1 exon 20 duplication in trans to a BRCA1 frameshift variant.** Block, Ines; Mateu-Regué, Àngels; Do, Thi Tuyet Nhu; Miceikaite, Ieva; Sdogati, Daniel; Larsen, Martin J; Hao, Qin; Nielsen, Henriette Roed; Boonen, Susanne E; Skytte, Anne-Bine; Jensen, Uffe Birk; Høffding, Louise K; De Nicolo, Arcangela; Viel, Alessandra; Tudini, Emma; Parsons, Michael T; Hansen, Thomas V O; Rossing, Maria; Kruse, Torben A; Spurdle, Amanda B; Thomassen, Mads. *Breast Cancer Research*. 2024jan. 9;26:6. doi: 10.1186/s13058-023-01755-9

PUBLICATIONS

7. **Frail inner limiting membrane maculopathy suggested to describe a new retinal Alport-like condition with two variants in three generations of females**. Petersen SD, Belmouhand M, Hertz JM, Fagerberg C, Brasch-Andersen C, Grauslund J, Munier FL, Larsen M. *Ophthalmic Genetics*.2024;45(3):281-288. Epub 2024jan. 10. doi: 10.1080/13816810.2023.2294844

8. **The Protective Effect of Familial Longevity Persists After Age 100: Findings From the Danish National Registers.** Galvin A, Pedersen JK, Wojczynski MK, Ukraintseva S, Arbeev K, Feitosa M, Province MA, Christensen K. *The Journals of Gerontology Series A*. 2024jan. 1;79(1). doi: 10.1093/gerona/glad164

9. **Variants in the WDR44WD40-repeat domain cause a spectrum of ciliopathy by impairing ciliogenesis initiation.** Accogli A, Shakya S, Yang T, Insinna C, Kim SY, Bell D, Fagerberg C, Larsen MJ et al. *Nature Communications*. 2024jan. 8;15(1):365. doi: 10.1038/s41467-023-44611-2

10. **Transcription Factor Regulation of Gene Expression Network by ZNF385D and HAND2 in Carotid Atherosclerosis**. Tan M, Andersen LJ, Bruun NE, Lindholm MG, Tan Q, Snoer M. *Genes*. 2024feb.;15(2):213. doi: 10.3390/genes15020213

11. **A Mass Spectrometry-Based Proteome Study of Twin Pairs Discordant for Incident Acute Myocardial Infarction within Three Years after Blood Sampling Suggests Novel Biomarkers**. Beck HC, Skovgaard AC, Mohammadnejad A, Palstrøm NB, Nielsen PF, Mengel-From J, Hjelmborg J, Rasmussen LM, Sørensen M. *International Journal of Molecular Sciences*. 2024feb. 24;25(5):2638. doi: 10.3390/ijms25052638

12. **Pericardial delta like non-canonical NOTCH ligand 1(Dlk1) augments fibrosis in the heart through epithelial to mesenchymal transition**. Jensen, Charlotte Harken; Johnsen, Rikke Helin; Eskildsen, Tilde; Baun, Christina; Ellman, Ditte Gry; Fang, Shu; Bak, Sara Thornby; Hvidsten, Svend; Larsen, Lars Allan; Rosager, Ann Mari; Riber, Lars Peter; Schneider, Mikael; De Mey, Jo; Thomassen, Mads; Burton, Mark; Uchida, Shizuka; Laborda, Jorge; Andersen, Ditte Caroline. *Clinical and Translational Medicine*. 2024feb.;14(2):e1565. doi:10.1002/ctm2.1565

13. **High risk of ischaemic stroke amongst patients with hereditary haemorrhagic telangiectasia.** Kofoed MS, Tørring PM, Christensen AA, Lange B, Kjeldsen AD, Nielsen TH. *Eur J Neurol*. 2024 Feb;31(2):e16128. Doi: 10.1111/ene.16128. Epub 2023 Nov 13.PMID: 37955551

14. **Decoding gene regulatory circuitry underlying TNBC chemoresistance reveals biomarkers for therapy response and therapeutic targets.** Lusby R, Zhang Z, Mahesh A, Tiwari VK. *NPJ Precision Oncology*. 2024mar. 12;8(1):64. doi: 10.1038/s41698-024-00529-6

15. **The Depressiveness, Quality of Life and NEO-FFI Scale in Patients with Selected Genodermatoses**. Bartlomiej Wawrzycki, Magdalena Fryze, Radosław Mlak, Alicja Pelc, Katarzyna Wertheim-Tysarowska, Anette Bygum, Aleksandra Wiktoria Kulbaka, Dariusz Matosiuk, Aldona Pietrzak. *Journal of Clinical Medicine*. 2024mar.;13(6):1624. doi: 10.3390/jcm13061624

PUBLICATIONS

16. **Large-scale genome-wide association study of 398,238 women unveils seven novel loci associated with high-grade serous epithelial ovarian cancer risk.** Barnes, Daniel R.; Tyrer, Jonathan P.; Dennis, Joe; Leslie, Goska; Bolla, Manjeet K.; Lush, Michael; Aeilts, Amber M.; Aittomäki, Kristiina; Andrieu, Nadine; Andrulis, Irene L.; AntonCulver, Hoda; Arason, Adalgeir; Arun, Banu K.; Balmaña, Judith; Bandera, Elisa V.; Barkardottir, Rosa B.; Berger, Lieke P.V.; Berrington de Gonzalez, Amy; Berthet, Pascaline; Białkowska, Katarzyna; Bjørge, Line; Blanco, Amie M.; Blok, Marinus J.; Bobolis, Kristie A.; Bogdanova, Natalia V.; Brenton, James D.; Butz, Henriett; Buys, Saundra S.; Caligo, Maria A.; Campbell, Ian; Castillo, Carmen; Claes, Kathleen B.M.; Colonna, Sarah V.; Cook, Linda S.; Daly, Mary B.; Dansonka-Mieszkowska, Agnieszka; de la Hoya, Miguel; DeFazio, Anna; DePersia, Allison; Ding, Yuan Chun; Domchek, Susan M.; Dörk, Thilo; Einbeigi, Zakaria; Engel, Christoph; Evans, D. Gareth; Foretova, Lenka; Fortner, Renée T.; Fostira, Florentia; Foti, Maria Cristina; Friedman, Eitan; Frone, Megan N.; Ganz, Patricia A.; Gentry-Maharaj, Aleksandra; Glendon, Gord; Godwin, Andrew K.; González-Neira, Anna; Greene, Mark H.; Gronwald, Jacek; Guerrieri-Gonzaga, Aliana; Hamann, Ute; Hansen, Thomas v.O.; Harris, Holly R.; Hauke, Jan; Heitz, Florian; Hogervorst, Frans B.L.; Hooning, Maartje J.; Hopper, John L.; Huff, Chad D; Huntsman, David G.; Imyanitov, Evgeny N.; Izatt, Louise; Jakubowska, Anna; James, Paul A.; Janavicius, Ramunas; John, Esther M.; Kar, Siddhartha; Karlan, Beth Y.; Kennedy, Catherine J.; Kiemeney, Lambertus A.L.M.; Konstantopoulou, Irene; Kupryjanczyk, Jolanta; Laitman, Yael; Lavie, Ofer; Lawrenson, Kate; Lester, Jenny; Lesueur, Fabienne; Lopez-Pleguezuelos, Carlos; Mai, Phuong L.; Manoukian, Siranoush; May, Taymaa; McNeish, Iain A.; Menon, Usha; Milne, Roger L.; Modugno, Francesmary; Mongiovi, Jennifer M.; Montagna, Marco; Moysich, Kirsten B.; Neuhausen, Susan L.; Nielsen, Finn C.; Noguès, Catherine; Oláh, Edit; Olopade, Olufunmilayo I.; Osorio, Ana; Papi, Laura; Pathak, Harsh; Pearce, Celeste L.; Pedersen, Inge S.; Peixoto, Ana; Pejovic, Tanja; Peng, Pei-Chen; Peshkin, Beth N.; Peterlongo, Paolo; Powell, C. Bethan; Prokofyeva, Darya; Pujana, Miquel Angel; Radice, Paolo; Rashid, Muhammad U.; Rennert, Gad; Richenberg, George; Sandler, Dale P.; Sasamoto, Naoko; Setiawan, Veronica W.; Sharma, Priyanka; Sieh, Weiva; Singer, Christian F.; Snape, Katie; Sokolenko, Anna P.; Soucy, Penny; Southey, Melissa C.; StoppaLyonnet, Dominique; Sutphen, Rebecca; Sutter, Christian; Teixeira, Manuel R.; Terry, Kathryn L.; Thomsen, Liv Cecilie V.; Tischkowitz, Marc; Toland, Amanda E.; Van Gorp, Toon; Vega, Ana; Velez Edwards, Digna R.; Webb, Penelope M.; Weitzel, Jeffrey N.; Wentzensen, Nicolas; Whittemore, Alice S.; Winham, Stacey J.; Wu, Anna H.; Yadav, Siddhartha; Yu, Yao; Ziogas, Argyrios; Berchuck, Andrew; Couch, Fergus J.; Goode, Ellen L.; Goodman, Marc T.; Monteiro, Alvaro N.; Offit, Kenneth; Ramus, Susan J.; Risch, Harvey A.; Schildkraut, Joellen M.; Thomassen, Mads; Simard, Jacques; Easton, Douglas F.; Jones, Michelle R.; ChenevixTrench, Georgia; Gayther, Simon A.; Antoniou, Antonis C.; Pharoah, Paul D.P.; GEMO Study Collaborators; EMBRACE Collaborators; kConFab Investigators; Ovarian Cancer Association Consortium; Consortium of Investigators of Modifiers of BRCA1 and BRCA2. *MedRxiv*, 2024 Mar 4:202402.29.24303243. DOI: 10.1101/2024.02.29.24303243

17. **Basal-epithelial subpopulations underlie and predict chemotherapy resistance in triple-negative breast cancer.** Inayatullah M, Mahesh A, Turnbull AK, Dixon JM, Natrajan R, Tiwari VK. *EMBO Molecular Medicine*. 2024apr. 15;16(4):823-853. Epub 2024mar. 13. doi: 10.1038/s44321-024-00050-0

18. **Prevalence and Patient Characteristics of Ectodermal Dysplasias in Denmark.** Laura Krogh Herlin, Sigrun A J Schmidt, Xenia Buus Hermann, Kirsten Rønholt, Anette Bygum, Annette Schuster, Ulrikke Lei, Mette Mogensen, Gabrielle R Vinding, Malene Djursby, Hanne Hove, Jenny Blechingberg, Lise Graversen, Trine H Mogensen, Hans Gjørup, Sinéad M Langan, Mette Sommerlund. *JAMA Dermatology*. 2024maj15;160(5):502-510. Epub 2024mar. 14. doi: 10.1001/jamadermatol.2024.0036

PUBLICATIONS

19. **Regulating PCCA gene expression by modulation of pseudo exon splicing patterns to rescue enzyme activity in propionic acidemia.** Spangsberg Petersen, Ulrika Simone; Dembic, Maja; Martínez-Pizarro, Ainhoa; Richard, Eva; Holm, Lise Lolle; Havelund, Jesper Foged; Doktor, Thomas Koed; Larsen, Martin Røssel; Færgeman, Nils J.; Desviat, Lourdes Ruiz; Andresen, Brage Storstein. *Molecular Therapy Nucleic Acids*. 2024mar.12;35(1):102101. doi: 10.1016/j.omtn.2023.102101

20. **Worldwide trends in underweight and obesity from 1990 to 2022: a pooled analysis of 3663 population-representative studies with 222 million children, adolescents, and adults**. NCD Risk Factor Collaboration (NCD-RisC), Nowell H. Phelps, Rosie K. Singleton, Bin Zhou, Rachel A. Heap, Anu Mishra, James E. Bennett, Christopher J. Paciorek, Victor PF Lhoste, Rodrigo M. Carrillo-Larco, Gretchen A. Stevens, Andrea Rodriguez-Martinez, Honor Bixby, Jennifer L. Baker, Julie Aarestrup, Shoaib Afzal, Kristine Allin, Lars Bo Andersen, Lars Ängquist, Peter Bjerregaard, Stig E. Bojesen, Kaare Christensen, Camilla T. Damsgaard, Thomas M. Dantoft, Louise Eriksen, Aleksander Giwercman, Jytte Halkjær, Torben Jørgensen, Peter Lund Kristensen, Lars Lind, Allan Linneberg, Jing Liu, Liping Liu, Anja L. Madsen, Kim F. Michaelsen, Line T. Møllehave, Erik Lykke Mortensen, Børge G. Nordestgaard, Merete Osler, Kim Overvad, Ida Maria Schmidt, Peter Schnohr, Stine Schramm, Eugène Sobngwi, Morten Sodemann, Thorkild IA Sørensen, Anne Tjonneland, Janne S. Tolstrup, Qian Wang, Ying Wei Wang, Yang Yang. *The Lancet*. 2024mar. 16;403(10431):1027-1050. doi: 10.1016/S0140-6736(23)02750-2

21. **Clinical and genetic characterisation of palmoplantar keratoderma.** Gram SB. *Syddansk Universitet. Det Sundhedsvidenskabelige Fakultet,* 2024. 186s. doi: 10.21996/zfct-mv75

22. **Splice-Switching Antisense Oligonucleotides Correct Phenylalanine Hydroxylase Exon 11 Skipping Defectsx and Rescue Enzyme Activity in Phenylketonuria.** Martinéz-Pizarro A, Alvarez M, Dembic M, Lindegaard CA, Castro M, Richard E, Andresen BS, Desviat LR. *Nucleid Acid Ther*. 2024;34(3): 134-142

23. **Generation of two patient specific GABRD variants and their isogenic controls for modeling epilepsy**. Morad Kamand, Reema Taleb, Methi Wathikthinnakon, Fadumo Abdullahi Mohamed, Said Pasalar Ghazanfari, Denis Konstantinov, Jonas Laugård Hald, Bjørn Holst, Charlotte Brasch-Andersen, Rikke S. Møller, Johannes R. Lemke, Ilona Krey, Kristine Freude, Abinaya
Chandrasekaran. *Stem Cell Research*. 2024apr.;76:103372. doi: 10.1016/j.scr.2024.10337

24. **In vivo effect of vaginal seminal plasma application on the human endometrial transcriptome: a randomized controlled trial.** Catalini L, Burton M, Egeberg DL, Eskildsen TV, Thomassen M, Fedder J. *Molecular Human Reproduction*. 2024apr. 1;30(5):gaae017. doi:10.1093/molehr/gaae017

25. **Plasticity in cell migration modes across development, physiology, and disease.** Pourjafar M, Tiwari VK. *Frontiers in Cell and Developmental Biology*. Apr2024;12:1363361. doi:10.3389/fcell.2024.1363361

26. **Golfspillerens purpura.** Drivenes JL, Bygum A. *Tidsskrift for Den norske legeforening*. 2024apr.23;144(5). doi: 10.4045/tidsskr.23.0604

PUBLICATIONS

27. **Meta-analysis of ACE inhibitor–induced angioedema identifies novel risk locus.** Mathey, Carina M.; Maj, Carlo; Eriksson, Niclas; Krebs, Kristi; Westmeier, Julia; David, Friederike S.; Koromina, Maria; Scheer, Annika B.; Szabo, Nora; Wedi, Bettina; Wieczorek, Dorothea; Amann, Philipp M.; Löffler, Harald; Koch, Lukas; Schöffl, Clemens; Dickel, Heinrich; Ganjuur, Nomun; Hornung, Thorsten; Buhl, Timo; Greve, Jens; Wurpts, Gerda; Aygören-Pürsün, Emel; Steffens, Michael; Herms, Stefan; Heilmann-Heimbach, Stefanie; Hoffmann, Per; Schmidt, Börge; Mavarani, Laven; Andresen, Trine; Sørensen, Signe Bek; Andersen, Vibeke; Vogel, Ulla; Landén, Mikael; Bulik, Cynthia M.; Bygum, Anette; Magnusson, Patrik K.E.; von Buchwald, Christian; Hallberg, Pär; Rye Ostrowski, Sisse; Sørensen, Erik; Pedersen, Ole B.; Ullum, Henrik; Erikstrup, Christian; Bundgaard, Henning; Milani, Lili; Rasmussen, Eva Rye; Wadelius, Mia; Ghouse, Jonas; Sachs, Bernhardt; Nöthen, Markus M.; Forstner, Andreas J.; Estonian Biobank Research Team; DBDS Genomic Consortium. *Journal of Allergy and Clinical Immunology*. 2024apr.;153(4):1073-1082. Epub 2024. doi: 10.1016/j.jaci.2023.11.921

28. **Identification of a founder variant AAGAB c.370C>T, p.Arg124Ter in patients with punctate palmoplantar keratoderma in Southern Denmark.** Gram SB, Jørgensen ASF, Bygum A, Brusgaard K, Ousager LB**.** *Clinical Genetics*. 2024maj;105(5):561-566. Epub 2024feb. 4. doi: 10.1111/cge.1448
29. **Systemic inflammation in relation to exceptional memory in the Lond Life Family Study (LLFS)**. Ruhee Patel, Kaare Christensen**,** et al; Long Life Family Study (LLFS). I: Brain, Behavior, and Immunity – Health, Bind 37, 100746, 05.2024

30. **Enhanced resolution profiling in twins reveals differential methylation signatures of type 2 diabetes with links to its complications**. Christiansen, Colette; Potier, Louis; Martin, Tiphaine C.; Villicaña, Sergio; Castillo-Fernandez, Juan E.; Mangino, Massimo; Menni, Cristina; Tsai, Pei Chien; Campbell, Purdey J.; Mullin, Shelby; Ordoñana, Juan R.; Monteagudo, Olga; Sachdev, Perminder S.; Mather, Karen A.; Trollor, Julian N.; Pietilainen, Kirsi H.; Ollikainen, Miina; Dalgård, Christine; Kyvik, Kirsten; Christensen, Kaare; van Dongen, Jenny; Willemsen, Gonneke; Boomsma, Dorret I.; Magnusson, Patrik K.E.; Pedersen, Nancy L.; Wilson, Scott G.; Grundberg, Elin; Spector, Tim D.; Bell, Jordana T.. *EBioMedicine*. 2024maj;103:105096. doi:10.1016/j.ebiom.2024.10509

31. **Comprehensive Noninvasive Fetal Screening by Deep Trio-Exome Sequencing.** Miceikaitė I, Hao Q, Brasch Andersen C, Fagerberg CR, Torring PM, Kristiansen BS, Ousager LB, Sperling L, Ibsen MH, Löser K, Larsen MJ. *Obstetrical and Gynecological Survey*. 2024maj 1;79(5):261-263. doi: 0.1097/01.ogx.0001023596.15300.92

32. **Ensemble-based classification using microRNA expression identifies a breast cancer patient subgroup with an ultralow long-term risk of metastases.** Block I, Burton M, Sørensen KP, Larsen MJ, Do TTN, Bak M, Cold S, Thomassen M, Tan Q, Kruse TA. *Cancer Medicine*. 2024maj;13(9):e7089. doi: 10.1002/cam4.7089

33. **Plantar keratoderma and curly hair as a diagnostic clue of cardiomyopathy risk.** Gram SB, Brusgaard K, Bygum A, Christensen AH, Ousager LB**.** *Journal of Dermatology*. 2024maj;51(5):e143-e144. Epub 2024mar. 25. doi: 10.1111/1346-8138.17192

34. **Genome-Wide Epistatic Network Analyses of Semantic Fluency in Older Adults.** Qihua Tan, Weilong Li, Marianne Nygaard, Ping An, Mary Feitosa, Mary K. Wojczynski, Joseph Zmuda, Konstantin Arbeev, Svetlana Ukraintseva, Anatoliy Yashin, Kaare Christensen, Jonas Mengel-From. *International Journal of Molecular Sciences*. 2024maj;25(10):5257. doi: 10.3390/ijms25105257

PUBLICATIONS

35. **Single-Cell Multi-Omics Map of Cell Type-Specific Mechanistic Drivers of Multiple Sclerosis Lesions.** Elkjaer ML, Hartebrodt A, Oubounyt M, Weber A, Vitved L, Reynolds R, Thomassen M, Rottger R, Baumbach J, Illes Z. *Neurology(R)neuroimmunology & neuroinflammation*. 2024maj 1;11(3):e200213. doi:10.1212/NXI.0000000000200213

36. **Well-differentiated G1 and G2 pancreatic neuroendocrine tumors: a meta-analysis of published expanded DNA sequencing data**. Andersen KØ, Detlefsen S, Brusgaard K, Christesen HT. *Frontiers in Endocrinology*. 2024;15:1351624. doi: 10.3389/fendo.2024.1351624

37. **IDENTIFYING SOMATIC MUTATIONS IN AGGRESSIVE NON-HODGKIN B-CELL LYMPHOMA WITH ENHANCED SPECIFICITY USING LIQUID BIOPSY: A COMPARATIVE STUDY OF CELL-FREE DNA AND FFPE TISSUE**. Vimalathas G, Hansen MH, Thomassen M, Møller MB, Nyvold CG, Larsen TS. *HemaSphere*. 2024jun.;8(S1):4000-4001.

38. **Screening of dermatology drugs for aberrant use-patterns: An application of epidemiologic estimates and measures of inequality in drug use.** Delvin T, Bygum A, Lund LC, Andersen JH, Hallas J. *British Journal of Clinical Pharmacology*. 2024jun.;90(6):1450-1462. Epub 2024mar. 12. doi:10.1111/bcp.16037

39. **Epigenome-wide analysis of frailty: Results from two European twin cohorts.** Jonathan KL Mak, Asmus Cosmos Skovgaard, Marianne Nygaard, Laura Kananen, Chandra A Reynolds, Yunzhang Wang, Ralf Kuja-Halkola, Ida K. Karlsson, Nancy L Pedersen, Sara Hägg, Mette Sørensen, Juulia Jylhävä. *Aging Cell*. 2024jun.;23(6):e14135.doi: 10.1111/acel.14135

40. **RNA expression profiling of peritoneal metastasis from pancreatic cancer treated with Pressurized Intraperitoneal Aerosol Chemotherapy (PIPAC).** Detlefsen S, Burton M, Ainsworth AP, Fristrup C, Graversen M, Pfeiffer P, Tarpgaard LS, Mortensen MB. *Pleura and Peritoneum*. 2024 jun.;9(2):79-91.Epub 2024. doi: 10.1515/pp-2024-0001

41. **Klinisk genetik. Medicinsk Kompendium.** Ousager LB, Kibæk Nielsen I, Sunde L. I Hauge EM, Ainsworth MA, Poulsen SD, red. 20. udg. Bind 2. Munksgaard. 2024. s. 1947-1957

42. **Cognitively high-performing oldest old individuals are physically active and have strong motor skills - a study of the Danish 1905 and 1915 birth cohorts.** Hermansen M, Nygaard M, Tan Q, Jeune B, Semkovska M, Christensen K, Thinggaard M, Mengel-From J. *Archives of Gerontology and Geriatrics*. 2024jul.;122:105398. doi: 10.1016/j.archger.2024.10539

43. **Drugst.One - a plug-and-play solution for online systems medicine and network-based drug repurposing**. Andreas Maier, Michael Hartung, Mark Abovsky, Klaudia Adamowicz, Gary D Bader, Sylvie Baier, David B Blumenthal, Jing Chen, Maria L Elkjaer, Carlos Garcia-Hernandez, Mohamed Helmy, Markus Hoffmann, Igor Jurisica, Max Kotlyar, Olga Lazareva, Hagai Levi, Markus List, Sebastian Lobentanzer, Joseph Loscalzo, Noel Malod-Dognin, Quirin Manz, Julian Matschinske, Miles Mee, Mhaned Oubounyt, Chiara Pastrello, Alexander R Pico, Rudolf T Pillich, Julian M Poschenrieder, Dexter Pratt, Nataša Pržulj, Sepideh Sadegh, Julio Saez-Rodriguez, Suryadipto Sarkar, Gideon Shaked, Ron Shamir, Nico Trummer, Ugur Turhan, Rui-Sheng Wang, Olga Zolotareva, Jan Baumbach. *Nucleic Acids Research*. 2024jul. 5;52(W1):W481-W488. doi: 10.1093/nar/gkae388

PUBLICATIONS

44. **GDNF/GFRA1signaling contributes to chemo- and radioresistance in glioblastoma.** Avenel ICN, Ewald JD, Ariey-Bonnet J, Kristensen IH, Petterson SA, Thesbjerg MN, Burton M, Thomassen M, Wennerberg K, Michaelsen SR, Kristensen BW. *Scientific Reports*. 2024jul. 31;14:17639. doi: 10.1038/s41598-024-68626-x

45. **Genotype-histotype-phenotype correlations in hyperinsulinemic hypoglycemia.** Larsen AR, Brusgaard K, Christesen HT, Detlefsen S. *Histology and Histopathology*. 2024jul.;39(7):817-844. Epub2024jan. 12. doi: 10.14670/HH-18-709

46. **Markers of Mitochondrial Function and DNA Repair Associated with Physical Function in Centenarians**. Sanchez-Roman, Ines; Ferrando, Beatriz; Holst, Camilla Myrup; Mengel-From, Jonas; Rasmussen, Signe Høi; Thinggaard, Mikael; Bohr, Vilhelm A; Christensen, Kaare; Stevnsner, Tinna V. *Biomolecules*, 2024 Jul 26;14(8):909

47. **Late diagnosis of partial 3β-hydroxysteroid dehydrogenase type 2deficiency - characterization of a new genetic variant.** Cagla Margit Øzdemir, Mette Mølby Nielsen, Jani Liimatta, Clarissa D. Voegel, Rawda Naamneh Elzenaty, Victor S. Wasehuus, Marie Lind-Holst, Marie Juul Ornstrup, Stine Bjørn Gram, Lilian Bomme Ousager, Christa E. Flück, Claus H. Gravholt. *Endocrinology, Diabetes & Metabolism Case Reports*. 2024jul.1;2024(3):e230090. doi: 10.1530/EDM-23-0090

48. **Exploring the prognostic value of circular RNAs in pancreatic ductal adenocarcinoma using genome-wide expression profiling**. Ørbeck, Siri Vreim; Jakobsen, Theresa; García-Rodríguez, Juan Luis; Burton, Mark; Rasmussen, Lukas Gammelgaard; Ewald, Jesper Dupont; Fristrup, Claus Wilki; Pfeiffer, Per; Mortensen, Michael Bau; Kristensen, Lasse Sommer; Detlefsen, Sönke. *Pancreatology*. 2024aug.;24(5):706-718. Epub 2024apr. 24. doi:10.1016/j.pan.2024.04.004.

49. **Spontaneous Mucocutaneous Ulcerations: A Quiz**. Drivenes JL, Bygum A. *Acta Dermato-Venereologica*. 2024aug. 5;104:adv41006. doi: 10.2340/actadv.v104.41006

50. **Definition, acronyms, nomenclature, and classification of angioedema (DANCE): AAAAI, ACAAI, ACARE,and APAAACI DANCE consensus.** Avner Reshef, Thomas Buttgereit, Stephen D Betschel, Teresa Caballero, Henriette Farkas, Anete S Grumach, Michihiro Hide, Ankur K Jindal, Hilary Longhurst, Jonathan Peter, Marc A Riedl, Yuxiang Zhi, Werner Aberer, Mohamed Abuzakouk, Tariq Al Farsi, Nashat Al Sukaiti, Mona Al-Ahmad, Sabine Altrichter, Emel Aygören-Pürsün, Maria Luisa BaezaNoemi Anna Bara, Andrea Bauer, Jonathan A Bernstein, Isabelle Boccon-Gibod, Hanna Bonnekoh, Laurence Bouillet, Zenon Brzoza, Anette Bygum, Oscar Calderon, Regis de Albuquerque Campos, Freya Helena Campos Romero, Mauro Cancian, Herberto Jose Chong-Neto, George Christoff, Stefan Cimbollek, Danny M Cohn, Timothy Craig, Inna Danilycheva, Razvigor Darlenski, Aurélie Du-Thanh, Luis Felipe Ensina, Daria Fomina, Luz Fonacier, Atsushi Fukunaga, Asli Gelincik, Pedro Giavina-Bianchi, Kiran Godse, Mark Gompels, Margarida Goncalo, Maia Gotua, Guillermo Guidos-Fogelbach, Mar Guilarte, Alicja Kasperska-Zajac, Constance H Katelaris, Tamar Kinaciyan, Pavel Kolkhir, Kanokvalai Kulthanan, Marcin Kurowski, Elena Latysheva, Antti Lauerma, David Launay, Ramon Lleonart, William Lumry, Alejandro Malbran, Ramzy Mohammed Ali, Iman Nasr, Sandra Nieto-Martinez, Claudio Parisi, Ruby Pawankar, Macarena Piñero-Saavedra, Todor A Popov, Grzegorz Porebski, Alicia Prieto Garcia, Polina Pyatilova, Michael Rudenko, Bulent Enis Sekerel, Faradiba Sarquis Serpa, Farrukh Sheikh, Frank Siebenhaar, Angèle Soria, Maria Staevska, Petra Staubach, Marcin Stobiecki, Simon Francis Thomsen, Massimo Triggiani, Anna Valerieva, Solange Valle, Nguyen Van Dinh, Carolina Elisa Vera Ayala, Anna Zalewska-Janowska, Andrea Zanichelli, Markus Magerl, Marcus Maurer. *Journal of Allergy and Clinical Immunology*. 2024aug.;154(2):398-411.e1. Epub 2024apr. 25. doi: 10.1016/j.jaci.2024.03.024

PUBLICATIONS

51. **Deleterious ZNRF3 germline variants cause neurodevelopmental disorders with mirror brain phenotypes via domain-specific effects on Wnt/β-catenin signaling**. Boonsawat, Paranchai; Asadollahi, Reza; Niedrist, Dunja; Steindl, Katharina; Begemann, Anaïs; Joset, Pascal; Bhoj, Elizabeth J.; Li, Dong; Zackai, Elaine; Vetro, Annalisa; Barba, Carmen; Guerrini, Renzo; Whalen, Sandra; Keren, Boris; Khan, Amjad; Jing, Duan; Palomares Bralo, María; Rikeros Orozco, Emi; Hao, Qin; Schlott Kristiansen, Britta; Zheng, Bixia; Donnelly, Deirdre; Clowes, Virginia; Zweier, Markus; Papik, Michael; Siegel, Gabriele; Sabatino, Valeria; Mocera, Martina; Horn, Anselm H.C.; Sticht, Heinrich; Rauch, Anita. *American Journal of Human Genetics.* 2024sep.;111(9):1994-2011. doi: 10.1016/j.ajhg.2024.07.016

52. **Effects of smoking on clinical treatment outcomes amongst patients with chronic inflammatory diseases initiating biologics: secondary analyses of the prospective BELIEVE cohort study.** Maja Graves Rosenkilde Larsen*, Silja Hvid Overgaard, Sofie Ronja Petersen, Karen Mai Møllegaard, Heidi Lausten Munk, Anders Bathum Nexøe, Henning Glerup, Tanja Guldmann, Natalia Pedersen, Sanaz Saboori, Jens Frederik Dahlerup, Christian Lodberg Hvas, Karina Winther Andersen, Mohamad Jawhara, Ole Haagen Nielsen, Fredrik Olof Bergenheim, Jacob Broder Brodersen, Anette Bygum, Torkell Ellingsen, Jens Kjeldsen, Robin Christensen, Vibeke Andersen. *Scand J Immunol,* 2024 Sept,100(3):e13395.

53. **Phenotypic variability related todominant UCHL1mutations: about three families with optic atrophy and ataxia.** Marelli C, Ramond F, Vignal C, Blanchet C, Frost S, Hao Q, Bocquet B, Nadjar Y, Leboucq N, Taieb G, Benkirane M, Hersent C, Koenig M, Meunier I. *Journal of Neurology*. 2024sep.;271(9):6038-6044. Epub 2024. doi: 10.1007/s00415-024-12574-z

54. **Nano Imprint: A DNA methylation tool for clinical interpretation and diagnosis of common imprinting disorders using nanopore long-read sequencing.** Bækgaard CH, Lester EB, Møller-Larsen S, Lauridsen MF, Larsen MJ. *Annals of Human Genetics*. 2024sep.;88(5):392-398. Epub 2024maj 1. doi: 10.1111/ahg.12556

55. **Expansion of the neurodevelopmental phenotype of individuals with EEF1A2 variants and genotype-phenotype study.** Alix Paulet, Cavan Bennett-Ness, Faustine Ageorges, Detlef Trost, Andrew Green, David Goudie, Rosalyn Jewell, Minna Kraatari-Tiri, Juliette PIARD, Christine Coubes, Wayne Lam, Sally Ann Lynch, Groeschel Samuel, Francis Ramond, Joël Fluss, Christina Fagerberg, Charlotte Brasch-Andersen, Konstantinos Varvagiannis, Tjitske Kleefstra, Bénédicte GérardMélanie Fradin, Antonio Vitobello, Romano Tenconi, Anne Sophie Denommé-Pichon, Aline Vincent-Devulder, Tobias Haack, Joseph A. Marsh, Lone Walentin Laulund, Mona Grimmel, Angelika Riess, Elke de Boer, Sergio Padilla-Lopez, Somayeh Bakhtiari, Michael C. Kruer, Jonathan Levy, Alain Verloes, Catherine M. Abbott, Lyse Ruaud. *European Journal of Human Genetics*. 2024sep.;32(9):1144-1149. Epub 2024feb. 15. doi:10.1038/s41431-024-01560-8

56. **Novel epigenetic biomarkers for hematopoietic cancer found in twins**. Clemmensen SB, Frederiksen H, Mengel-From J, Heikkinen A, Kaprio J, Hjelmborg JVB. *Acta Oncologica* (Stockholm, Sweden). 2024sep. 18;63:710-717. doi: 10.2340/1651-226X.2024.40700

57. **Achenbachs syndrom: Ugens billede.** Drivenes JL, Bygum A. *Ugeskrift for Læger*. 2024okt.14;186(20):V72038. Epub 2024sep. 2. doi: 10.61409/V72038

PUBLICATIONS

58. **An Elderly Man with Facial Oedema and a Photosensitive Rash: A Quiz.** *Acta Dermato-Venereologica*. Drivenes JL, Bygum A. 2024okt. 30;104:adv42012. doi: 10.2340/actadv.v104.42012

59. **Deep Genome Sequencing Uncovers Extensive Genetic Heterogeneity in Early Human Placentas**. Miceikaite I, Fagerberg C, Brasch-Andersen C, Torring P, Kristiansen BS, Hao Q, Sperling L, Larsen MJ. 2024. doi: 10.21203/rs.3.rs-5140201/v1

60. **Germline pathogenic variants in RNF43 in patients with and without serrated polyposis syndrome**, Heidi Hesselø Brinch, Anna Byrjalsen, Zuzana Lohse, Andreas Ørslev Rasmussen, John Gásdal Karstensen, Britta Schlott Kristiansen & Anne Marie Jelsig. TIdsskrift: *Fam Cancer*. 2024 Nov 15;24(1):3. doi: 10.1007/s10689-024-00428-6. https://rdcu.be/d08TL

61. **Genomic profiling and expanded use of targeted anticancer drugs in solid cancers with exhausted evidence-based treatment options (PRECODE): study protocol of a prospective, non-randomized, cohort study**. Hansen, Karin Holmskov; Lyng, Maria Bibi; Kodahl, Annette Raskov; Asmussen, Jon Thor; Arshad, Arman; Petersen, Henrik; Krogh, Lotte; Ehmsen, Sidse; Kristensen, Thomas Kielsgaard; Ditzel, Henrik J. *BMC Medical Genomics*. 2024nov. 21;17:274. doi: 10.1186/s12920-024-02033-z

62. **Age-Dependent DNA Methylation Variability on the X-Chromosome in Male and Female Twins**. Tan Q, Alo H, Nygaard M, Sørensen M, Saleh A, Mengel-From J, Christensen K. *Epigenomics.* 2024nov.;8(4):43. doi: 10.3390/epigenomes8040043

63. **Association Study of DNA Methylation Age and Bone Health Phenotypes in Older Twins**. 2024. Sørensen M, Christensen K, Kassem M. Poster session præsenteret ved *The Gerontological Society of America 2024 Annual Scientific Meeting*, Seattle, USA. November 2024

64. **Molecular Composition and Kinetics of B Cells During Ibrutinib Treatment in Patients with Chronic Lymphocytic Leukemia.** Veyhe, Sólja Remisdóttir; Cédile, Oriane; Dahlmann, Sara Kamuk; Krejcik, Jakub; Abildgaard, Niels; Høyer, Thor; Møller, Michael Boe; Thomassen, Mads; Juul-Jensen, Karen; Frederiksen, Henrik; Dybkær, Karen; Hansen, Marcus Høy; Nyvold, Charlotte Guldborg. *International Journal of Molecular Sciences*. 2024nov. 22;25(23):12569. doi:10.3390/ijms252312569

65. **Reclassification of an FBN1 variant emphasizes the importance of segregation analysis, information sharing, and multidisciplinary teamwork in understanding genetic variants in health and disease**. Lildballe DL, Markholt S, Lyngholm CD, Hao Q, Fagerberg C, Nielsen DG, Svensmark JH, Diness BR, Gregersen PA. *American Journal of Medical Genetics*, Part A. 2024nov.;194(11):e63795. Epub 2024. doi:10.1002/ajmg.a.63795

66. **Reclassification of Two MLH1 Variants of Uncertain Significance Utilizing Clinical and Functional Data.** Frederiksen, Jane Hübertz; Birkedal, Ulf; Bachmann, Sarah; Eliesen, Elisabeth Victoria; Rasmussen, Lene Juel; Pedersen, Katja Venborg; Al-Zehhawi, Lana; Boonen, Susanne E.; Krogh, Lotte; Rønlund, Karina; Graversen, Lise; Assenholt, Jannie; Schmiegelow, Kjeld; Wadt, Karin; Gerdes, Anne Marie; Hansen, Thomas v.O. *Molecular Genetics and Genomic Medicine*. 2024nov.;12(11):e70026. doi:10.1002/mgg3.70026

PUBLICATIONS

67. **The Moderating Influence of School Achievement on Intelligence: A Cross-National Comparison**. Emilie R. Hegelund, Erik Lykke Mortensen, Trine Flensborg-Madsen, Jesper Dammeyer, Kaare Christensen, Matt McGue, Christoph H. Klatzka, Frank M. Spinath, Wendy Johnson. *Behavior Genetics*. 2024. doi: 10.1007/s10519-024-10203-z

68. **A Sensitive and Transparent Method for Tumor-Informed Detection of Circulating Tumor DNA in Ovarian Cancer Using Whole-Genome Sequencing**. Thusgaard CF, Sadegh S, Jochumsen KM, Kruse TA, Thomassen M. *International Journal of Molecular Sciences* . 2024dec.;25(24):13349. doi: 10.3390/ijms25241334

69. **Clinical and Genetic Findings in Patients With Palmoplantar Keratoderma.** Gram SB**,** Brusgaard K**,** Lei U, Sommerlund M, Vinding GR, Sleire SOK, Bygum A, Ousager LB, Christensen AH, Fast SP, Bach R. *JAMA Dermatology* 2024dec. 4. Epub2024dec. 4. doi: 10.1001/jamadermatol.2024.4824

70. **Expanding the phenotype and genotype spectrum of TAOK1 neurodevelopmental disorder and delineating TAOK2 neurodevelopmental disorder**. Nour Elkhateeb, Renarta Crookes, Michael Spiller, Lisa Pavinato, Flavia Palermo, Alfredo Brusco, Michael Parker, Soo-Mi Park, Ariana Costa Mendes, Jorge M Saraiva, Trine Bjørg Hammer, Lusine Nazaryan-Petersen, Tahsin Stefan Barakat, Martina Wilke, Elizabeth Bhoj, Rebecca C Ahrens-Nicklas, Dong Li, Tomoki Nomakuchi, Eva H Brilstra, David Hunt, Diana Johnson, Sahar Mansour, Kathryn Oprych, Sarju G Mehta, Konrad Platzer, Franziska Schnabel, Henriette Kiep, Helene Faust, Gillian Prinzing, Kimberly Wiltrout, Jessica A Radley, Alvaro H Serrano Russi, Isis Atallah, Belinda Campos-Xavier, David J Amor, Angela T Morgan, Christina Fagerberg, Ulla A Andersen, Charlotte B Andersen, Emilia K Bijlsma, Lynne M Bird, Sureni V Mullegama, Andrew Green, Bertrand Isidor, Benjamin Cogné, Janna Kenny, Sally A Lynch, Shauna Quin, Karen Low, Theresia Herget, Fanny Kortüm, Rebecca J Levy, Jennifer L Morrison, Patricia G Wheeler, TaraChandra Narumanch, Kristina Peron, Nicole Matthews, Jillian Uhlman, Lauren Bell, Lewis Pang, Ingrid Scurr, Rebecca S Belles, Bonnie Anne Salbert, Gerald Bradley Schaefer, Sarah Green, Andrea Ros, Agustí Rodríguez-Palmero, Tanja Višnjar, Karin Writzl, Pradeep C Vasudevan, Meena Balasubramanian. *Genet Med*. 2024 Dec 27;27(3):101348 doi: 10.1016/j.gim.2024.101348

71. **FOXO-regulated OSER1 reduces oxidative stress and extends lifespan in multiple species**. Jiangbo Song. Zhiguan Li, Lei Zhou, Xin Chen, Wei Qi Guinevere Sew, Héctor Herranz, Zilu Ye, Jesper Velgaard Olsen, Yuan Li, Marianne Nygaard, Kaare Christensen, Xiaoling Tong, Vilhelm A Bohr, Lene Juel Rasmussen, Fangyin Dai. *Nature Communications*. 2024dec.;15:7144. doi: 10.1038/s41467-024-51542-z

72. **Genome-wide methylation profiling differentiates benign from aggressive and metastatic pituitary neuroendocrine tumors.** Jotanovic, Jelena; Boldt, Henning Bünsow; Burton, Mark; Andersen, Marianne Skovsager; Bengtsson, Daniel; Bontell, Thomas Olsson; Ekman, Bertil; Engström, Britt Edén; Feldt Rasmussen, Ulla; Heck, Ansgar; Jakovcevic, Antonia; Jørgensen, Jens Otto L.; Kraljevic, Ivana; Kunicki, Jacek; Lindsay, John R.; Losa, Marco; Loughrey, Paul Benjamin; Maiter, Dominique; Maksymowicz, Maria; Manojlovic-Gacic, Emilija; Pahnke, Jens; Petersenn, Stephan; Petersson, Maria; Popovic, Vera; Ragnarsson, Oskar; Rasmussen, Åse Krogh; Reisz, Zita; Saeger, Wolfgang; Schalin-Jäntti, Camilla; Scheie, David; Terreni, Maria Rosa; Tynninen, Olli; Whitelaw, Ben; Burman, Pia; Casar-Borota, Olivera. *Acta Neuropathologica*. 2024dec.;148(1):68. doi: 10.1007/s00401-024-02836-5

PUBLICATIONS

73. **Impact of gluten intake on clinical outcomes in patients with chronic inflammatory diseases initiating biologics: Secondary analysis of the prospective multicentre BELIEVE cohort study**. Gregersen, Laura; Jessen, Pernille Dyhre; Lund, Helene Wiencke; Overgaard, Silja Hvid; Hikmat, Zainab; Ellingsen, Torkell; Kjeldsen, Jens; Pedersen, Andreas Kristian; Petersen, Sofie Ronja; Jawhara, Mohamad; Nexøe, Anders Bathum; Bygum, Anette; Hvas, Christian Lodberg; Dahlerup, Jens Frederik; Bergenheim, Frederik Olof; Glerup, Henning; Henriksen, Rikke Holm; Guldmann, Tanja; Hvid, Lone; Brodersen, Jacob; Munk, Heidi Lausten; Pedersen, Natalia; Saboori, Sanaz; Nielsen, Ole Haagen; Heitmann, Berit Lillenthal; Haldorsson, Thorhallur Ingi; Christensen, Robin; Andersen, Vibeke. *Scandinavian Journal of Immunology*. 2024dec.;100(6):e13409. Epub 2024. doi: 10.1111/sji.13409

74. **Longitudinal associations between fruit and vegetable intakes and depressive symptoms inmiddle-aged and older adults from four international twin cohorts.** Annabel P. Matison, Anbupalam Thalamuthu, Victoria M. Flood, Vibeke S. Catts, Kaare Christensen, Marianne Nygaard, Nancy L. Pedersen, Perminder S. Sachdev, Simone Reppermund, Karen A. Mather, The Interplay of Genes and Environment across Multiple Studies (IGEMS) consortium. *Scientific Reports*. 2024dec.;14(1):29711. doi: 10.1038/s41598-024-79963-2

75. **Microfibrillar-associated protein 4 as a predictive biomarker of treatment response in patients with chronic inflammatory diseases initiating biologics: secondary analyses based on the prospective BELIEVE cohort study**. Bjørk K Sofíudóttir, Heidi L Munk, Robin Christensen, Sören Möller, Silja H Overgaard, Grith L Sorensen, Karen M Møllegaard, Jessica Pingel, Anders B Nexøe, Henning Glerup, Tanja Guldmann, Natalia Pedersen, Jens Frederik Dahlerup, Christian L Hvas, Karina W Andersen, Mohamad Jawhara, Ole Haagen Nielsen, Fredrik Olof Bergenheim, Anette Bygum, Jesper R Davidsen, Signe Bek Sørensen, Jacob B Brodersen, Jens Kjeldsen, Vibeke Andersen, Torkell Ellingsen. *Rheumatology International*. 2024dec.;44(12):2935-2947.Epub 2024okt. 28. doi: 10.1007/s00296-024-05744-9

76. **Microglia induce an interferon-stimulated gene expression profile in glioblastoma and increase glioblastomaresistance to temozolomide.** Sørensen MD, Olsen RFS, Burton M, Kavan S, Petterson SA, Thomassen M, Kruse TA, Meyer M, Kristensen BW. *Neuropathology and Applied Neurobiology*. 2024dec.;50(6):e13016.doi: 10.1111/nan.13016

77. **Mosaic loss of Y chromosome and the association to mortality in Danish men aged 56-100 years**. Hozakowska-Roszkowska, Dominika Marzena; Mengel-From, Jonas; Hristozova, Teodora K; Pedersen, Jacob Krabbe; Jeune, Bernard; Andersen-Ranberg, Karen; Hjelmborg, Jacob V B; Christensen, Kaare; Röttger, Richard; Nygaard, Marianne. *Mechanisms of Ageing and Development*. 2024dec.;222:111979. Epub 2024.doi: 10.1016/j.mad.2024.111979

78. **Multi-omics association study of DNA methylation and gene expression levels and diagnoses of cardiovascular diseases in Danish Twins**. Skovgaard AC, Mohammadnejad A, Beck HC, Tan Q, Soerensen M. *Clinical Epigenetics*. 2024dec.;16:117. doi: 10.1186/s13148-024-01727-6

79. **Ruthenium Drug BOLD-100 Regulates BRAFMT Colorectal Cancer Cell Apoptosis through AhR/ROS/ATR Signaling Axis Modulation**. Griffin, Daryl; Carson, Robbie; Moss, Debbie; Sessler, Tamas; Lavin, Deborah; Tiwari, Vijay K.; Karelia, Shivaali; Kennedy, Richard; Savage, Kienan I.; McDade, Simon; Carie, Adam; Pankovich, Jim; Bazett, Mark; Van Schaeybroeck, Sandra. *Molecular Cancer Research*. 2024dec. 1;22(12):1088-1101. doi: 10.1158/1541-7786.MCR-24-0151

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