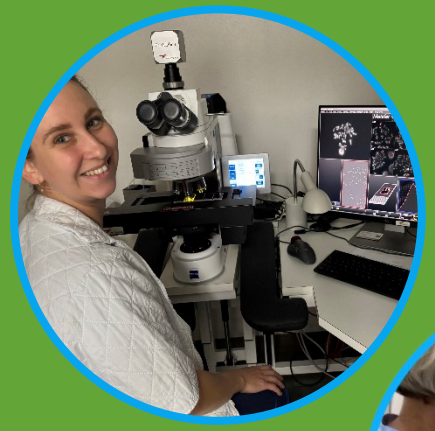
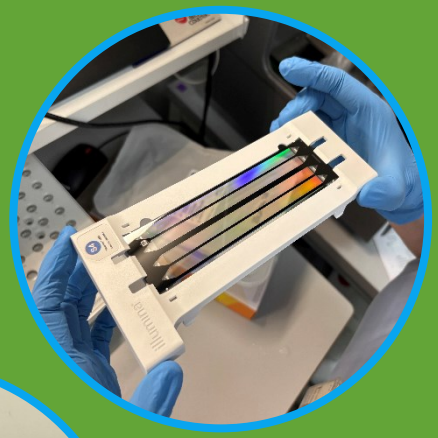
Research unit of Clinical Genetics, University of Southern Denmark
Department of Clinical Genetics, Odense University Hospital
Department of Clinical Research, University of Southern Denmark
Clinical Genome Center (CGC)








ANNUAL REPORT 2025

OUH
Odense
University Hospital

cgc
Clinical Genome Center

SDU

CONTENTS

WELCOME

The past year has been characterised by strong engagement and a meaningful progress across the research unit at the Department of Clinical Genetics. Our activities have focused on hereditary cancer and rare genetic disorders, supported by the application and development of advanced genetic and bioinformatic methods in close collaboration with the Clinical Genome Center (CGC) core facility.

Several PhD projects have been successfully completed and defended, and new doctoral and student-led projects have been initiated, reflecting an active and engaged research environment.

In February 2025, the kick-off reception for the new flagship center CIMP – Center for Integrated Multi-Omics in Precision Medicine – was held together with the University of Southern Denmark. The new center will work to reduce overtreatment in prevalent diseases, initially targeting breast cancer, osteoporosis, and multiple sclerosis. CIMP joins research capacity from the University of Southern Denmark (SDU) and Odense University Hospital (OUH) and is headed by Professor Vijay Tiwari and Professor Mads Thomassen.

The Research Committee of the Department of Clinical Genetics has met regularly throughout the year and has led the development of the department's forthcoming research strategy. This strategy, which will be presented shortly, reflects our shared ambition to define the department's primary research areas, strengthen the development of novel genetic methodologies, and enhance our focus on research education and capacity building.

I would like to express my sincere gratitude to everyone who contributes to research within the department. I extend my warm thanks to our national and international collaborators for their valued partnerships, to the funds whose support makes our work possible, and to our patient representatives, whose engagement and perspectives are essential in ensuring that our research remains relevant and of benefit for patients.

Together, these contributions form a strong foundation for the continued development of research at the department.

With this, it is my pleasure to present the department's research activities in the following pages.

Kind regards,

Anja

Head of Research Anja Lisbeth Frederiksen
MD, PhD, Professor

Ongoing PhD studies (1)

Caroline Hey Bækgaard, Project title: *"NIPT 2.0: Non-Invasive Fetal Genomic Screening".*
Supervisors: **Associate Professor Martin Larsen, MD, PhD Pernille Tørring** and **MSc, PhD Qin Hao,** Department of Clinical Genetics, OUH, and **Associate Professor Lene Sperling**, Department of Gynaecology and Obstetrics, OUH.

Katrine Saldern Aagaard, Project title: *"PreGene – tracing fetal footprints".*
Supervisor: **Lene Sperling, Associate Professor Karina Hjort-Pedersen**, Department of Gynaecology and Obstetrics, OUH, **Pernille Tørring** and **Martin Larsen**, Department of Clinical Genetics, OUH.

Maria Lissel Isaksson, Project title: "*Genetic causes of short stature in children".*
Supervisor: **Professor Lilian Bomme Ousager** and **Associate Professor Martin Larsen,** Department of Clinical Genetics, **Associate Professor Dorte Hansen,** H.C. Andersen Childrens' Hospital, OUH.

Astrid Skov Midtiby, 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.

Ongoing PhD studies (2)

Louise Adel Jensen, 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, OUH. Co-Supervisor: **Professor Thomas van Overeem Hansen**, Department of Clinical Genetics, Rigshospitalet.

Emilie Boye Lester, 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.

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

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

Completed PhD studies (1)

Project title: "***Integrated genomic analysis of breast tumors. A possible tool towards reduction of overtreatment".***
PhD defence: 8 October 2025

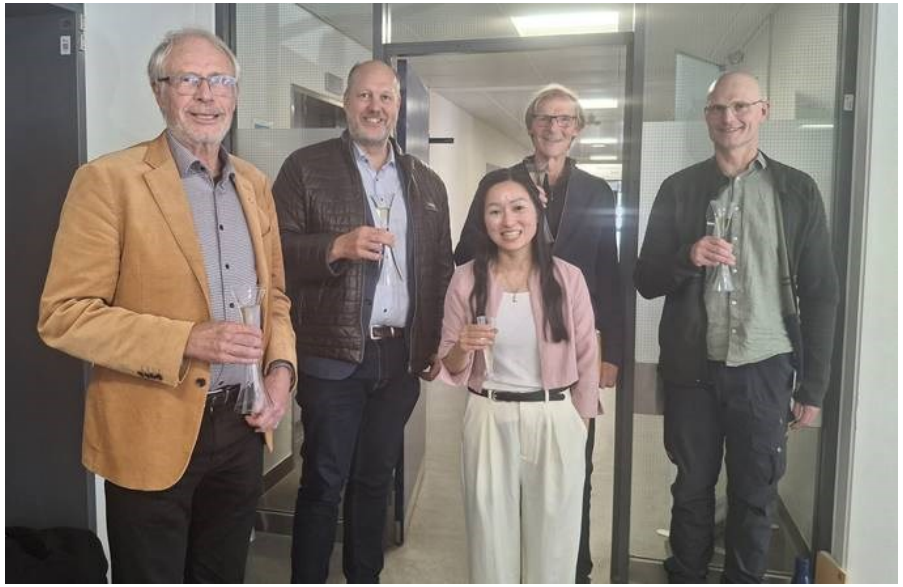
The PhD project comprised three studies focused on predicting clinical outcomes in systemically untreated breast cancer (BC) patients. The main aim was to contribute to reducing overtreatment, as more than 90% of current BC patients receive adjuvant systemic therapy, although up to 40% may not actually benefit from it.

Study I evaluated the relative ability of mRNA- and long non-coding RNA (lncRNA)-based signatures to predict recurrence among low-risk BC patients. The prognostic utility of these RNA molecules was also compared using two distinct classification strategies.

In Study II, it was hypothesized that integrating mRNA, lncRNA, and miRNA gene expression data could improve patient classification performance. The resulting multi-omics classifier significantly outperformed the individual datasets in the most clinically relevant setting of ≥99% sensitivity.

Study III involved whole exome sequencing of the patient cohort. Somatic variant calling for single nucleotide variants (SNVs) and insertion-deletion mutations (indels) was performed, followed by in-depth analyses to gain deeper biological and functional insights into the somatic genetic landscape of low-risk BC.

Nhu Do, PhD



Main supervisor: **Professor Torben Kruse.** Co-supervisor: **Professor Mads Thomassen,** Dep. of Clinical Genetics, OUH. Evaluation Committee: **Professor Mårten Fernö,** Medical Oncology, Lund University, **Professor Claus Lindbjerg Andersen**, Dep. of Clinical Medicine and Molecular Medicine, Aarhus University, and (not in photo) **Professor Hans Christian Beck** (chair), Dep. of Clinical Research, SDU.

Completed PhD studies (2)

Zainab Hikmat, PhD

Project title: "***Predicting response to medical treatment of inflammatory bowel disease using transcriptomic methods on intestinal biopsies: a prospective cohort study of personalised medicine".***
PhD defence: 1 December 2025.

Zainabs project focuses on patients with inflammatory bowel disease (IBD) which encompasses ulcerative colitis (UC) and Crohn's disease (CD). Patients with IBD are treated with biological therapy in the form of Tumour Necrosis factor inhibitors (anti-TNF), however, approximately 50% do not achieve remission following treatment. Therefore, the aim of this project was to use multi-omics methods, transcriptomics, proteomics and metabolomics to identify predictive biomarkers for treatment response.

Main supervisor: **Professor Mads Thomassen**, the Department of Clinical Genetics, OUH.
Co-supervisors: **Professor Vibeke Andersen**, Molecular Diagnostics and Clinical Research at the University Hospital of Southern Denmark, Aabenraa; **Dr. med. Tue Bjerge Bennike,** Department of Health Science and Technology, Aalborg University, **Molecular biologist, PhD Maja Dembic,** Department of Clinical Genetics, OUH.

PhD studies, in collaboration with other departments

Ongoing:

Amalie Elton Baisgaard, PhD student
Project title: *"Single cell sequencing spinal cord"*
Supervisors: **Associate Professor Jacek Lichota**, Department of Health Science and Technology, Aalborg University. Co-supervisor: **Associate Professor Mark Burton**, Department of Clinical Genetics, OUH.

Maria Line Foged, PhD student
Project title: *"The role of de novo somatic mutations in the pathogenesis and prediction of progressive multiple sclerosis"*
Supervisors: **Professor Zsolt Illes**, Department of Neurology. Co-supervisors: **Professor Mads Thomassen**, Department of Clinical Genetics, OUH, **Professor Vijay Tiwari**, Department of Molecular Medicine, SDU.

Completed:

Christine Fribert Thusgaard, PhD
PhD defence: 22 August 2025

Title: "***Circulating tumor DNA - A potential clinical marker in Ovarian High-Grade Serous Carcinomas***".

Main supervisor: **Associate Professor Kirsten Marie Jochumsen**, Department of Clinical Research, SDU. Co-supervisors: **Professor Mads Thomassen** and **Professor Torben A. Kruse,** Department of Clinical Genetics, OUH.

Ongoing Master studies

Dalal Ali Alyas, Medical student
Project title: *"Clinical variability in carriers of 16p13.11 duplications: a retrospective cohort study evaluating the need for cardiac surveillance"*
Supervisors: **Professor Lilian Bomme Ousager** and **PhD Qin Hao,** Department of Clinical Genetics, OUH.

Oliver Christoffer Knudsen, student of Bioinformatics
Project title: "*Investigating the impact of gene expression changes in hormone-treated transgender patients"*
Supervisor: **Associate Professor Mark Burton, MD, PhD Louise Lehmann Christensen**, Department of Clinical Genetics, OUH.

Emma Linnea Thonning Jørgensen, Medical student, undergraduate research year
Project title: "*RNA expression changes of cardio-metabolic pathways after initiation of sex hormone intervention in transgender individuals"*
Supervisors: **Louise Lehmann Christensen, Mark Burton**, Department of Clinical Genetics, **Professor Dorte Glintborg**, Department of Medical Endocrinology, OUH.

Kathrine Ladefoged Vestkær, student of Biomedicine
Project title: "*Investigation of the human endometrial transcriptome in RIF patients"*
Supervisor: **Associate Professor Mark Burton** , Department of Clinical Genetics, OUH.

Sofia Tziraki, Student of Computational biomedicine
Project title: *"Mapping the methylation landscape: Comprehensive genomic mapping of differential methylation and epigenetic architecture based on nanopore sequencing data"* Supervisors: **Associate Professor Martin Larsen. PhD student Caroline Hey Bækgaard**, Department of Clinical Genetics, OUH, **PhD Jesper Grud Skat Madsen,** Department of Biochemistry and Molecular Biology.

Completed 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**, Department of Clinical Genetics, OUH.

Emilie Kofoed Hansen, Medical student.
Project title: *"NGC hereditary heart disease: Diagnostic yield and genotype-phenotype correlation".* Supervisors: **Professor Lilian Bomme Ousager, PhD Qin Hao**, Department of Clinical Genetics, OUH, and **PhD Thomas Thomas Morris Hey.**

Frederik Møller Larsen, Student of Biomedicine. Project title: *"Mutational profiles of gastroesophageal cancers during treatment and identification of emerging treatment-resistant mutations".*
Supervisors: **Professor Mads Thomassen**, **PhD student Mikkel Møller Larsen** and **PhD student Alexander Venzel Rudbeck**, Department of Clinical Genetics, OUH.

Jeppe Hannibal Niemann, Health IT student. Project title: *"Language Model for Genetic Diagnostics".*
Oliver Rønholt Grimm, Health IT student. Project title: *"Language Models for Generating Summaries of Clinically Important EMR-notes in the Genetic Analysis Process".*
Both projects involved 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 **MD PhD Pernille Tørring**, Department of Clinical Genetics, OUH**.**

Solvej Engtoft Johansen, Medical student.
Project title: *"Characterization of the Birt-Hogg-Dubé phenotype".* Supervisors: **Professor Lilian Bomme Ousager** and **Associate Professor Henriette Roed Nielsen**, Department of Clinical Genetics, OUH.

Completed ISA Projects

Sofia Tziraki, Student in Computational biomedicine
ISA Project title: ”*A Novel Method for detecting Uniparental Disomy from Nanopore Methylation Sequencing Data”*
Supervisors: **Associate Professor Martin Larsen. PhD student Emilie Boye Lester**, Department of Clinical Genetics, OUH.

Matilde Skovbjerg Slot, Student in Biomedicine
ISA Project title: “*Comparison of long-read and short-read sequencing of breast cancer patients to verify mutational signatures”*
Supervisors: **Professor Mads Thomassen, PhD student Louise Adel Jensen**, Department of Clinical Genetics, OUH.

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
- **Bjørk Ditlev Marcher Larsen,** MSc, PhD
- **Britta Schlott Kristiansen,** MD
- **Charlotte Brasch Andersen**, MSc, PhD, Associate Professor
- **Henriette Roed Nielsen,** MD, PhD, Associate Professor
- **Jens Michael Hertz**, MD, DMSci, 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, Associate Professor
- **Mark Burton**, MSc, PhD, Associate Professor
- **Martin J. Larsen**, MSc, PhD, Associate Professor
- **Pernille M. Tørring**, MD, PhD
- **Qin Hao**, PhD
- **Sepideh Sadegh**, MSc, PhD
- **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 at the 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 research fields.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (1)

Pernille M. Tørring, MD, PhD

Pernille is a senior consultant in the Department of Clinical Genetics and is involved in several national and international research initiatives. Her primary research interests include prenatal/fetal genetics and vascular disorders, including Hereditary Hemorrhagic Telangiectasia (HHT).

In the field of prenatal and fetal genetics, Pernille is currently co-supervising two PhD projects addressing key questions in prenatal genetic diagnostics: *NIPT 2.0 – Non-Invasive Fetal Genomic Screening*, and *PreGene – tracing fetal footprints*. She is also a core member of the Center for Fetal Genetics.

With over 15 years of experience, Pernille has been part of the national HHT center, contributing to numerous clinical and research projects. She recently was senior author of a significant study providing strong evidence for the Knudsonian two-hit mutation mechanism in the formation of arteriovenous malformations (AVMs) in HHT. In addition, she collaborates closely with VASCERN – the European Reference Network for rare vascular diseases – further contributing to both clinical care and translational research in vascular genetics.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (2)

Mark Burton, MSc, PhD, Associate Professor. Bioinformatician at the Clinical Genome Center (CGC)

Mark is involved in data analysis for a wide range of CGC users, but also in advising and mentoring students in data analysis and statistical design. His primary focus (and speciality) is associated with RNA-related data, such as bulk RNA sequencing, single cell RNA sequencing and microarray gene expression data analysis.

Over the past year, CGC has experienced a growing demand for scRNAseq analysis from research groups at OUH and SDU, as well as from groups outside the Region of Southern Denmark and abroad.

Most scRNAseq analyses have targeted neuropsychiatric or neurodegenerative disorders. One such project involved sequencing brain organoids derived from children with Attention Deficit/Hyperactivity Disorder (ADHD), conducted in collaboration with the Department of Psychiatry at OUH and a Swiss research group. The results revealed imbalances in the ratio between excitatory and inhibitory neurons, as well as cell type specific transcriptional and pathway alterations related to neuronal development and synaptic signaling. Additional analyses have been performed on organoids derived from patients diagnosed with Bipolar Disorder, Autism, or adults with ADHD. Three other brain related projects focused on transcriptional alterations and changes in cellular composition in mice experiencing a stroke, effects of feeding with high- or low-fat diets, and motor neuron impairment due to a pathogenic variant mutation in the *SOD1* gene, respectively.

A key methodological achievement this year was the development of custom probes capable of detecting a specific somatic *JAK2* variant at the single-cell level—a variant mutation associated with myeloproliferative neoplasms and known to increase the risk of other inflammatory diseases. This innovation enables precise mapping of variant bearing cells within heterogeneous tissues and provides a powerful tool for studying clonal architecture and disease evolution.

Across all projects, the integration of scRNAseq, microarray data, and targeted variant expression measurements has strengthened our ability to interpret complex biological systems. This year's work generated high-resolution datasets, expanded our network of collaborations, and advanced the Center's mission to support precision medicine through cutting-edge transcriptomic data analysis.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (3)

Katrine Saldern Aagaard, PhD student

Project title: "*PreGene – tracing fetal footprints*"

Katrines project aims to improve prenatal genetic diagnostics by clarifying how ultrasound findings relate to underlying genetic disease. Rapid advances in sequencing technologies have significantly shaped the field of fetal genetics, yet a key challenge remains the limited understanding of how specific prenatal findings correlate with genetic conditions. This gap affects both variant interpretation and the strategic use of extended genetic testing.

The project consists of three complementary studies.

- Study 1 evaluates the diagnostic yield of current prenatal genetic testing strategies and how they relate to different categories of ultrasound findings and pregnancy outcomes.
- Study 2 characterises prenatal ultrasound features in children with confirmed genetic diseases to identify patterns that may support earlier recognition of these conditions.
- Study 3 investigates aborted fetuses with unexplained malformations using deep exome sequencing to detect somatic variants that could explain the phenotype and inform recurrence counselling.

The project is carried out in collaboration between Clinical Genetics and Fetal Medicine.

Supervisor: Associate Professor Lene Sperling, MD, PhD, Department of Gynaecology and Obstetrics, OUH.
Co-supervisors: Associate Professor Karina Hjort-Pedersen, Department of Gynaecology and Obstetrics, Pernille Tørring, and Martin Larsen, Department of Clinical Genetics, OUH.

CURRENT RESEARCH FIELD

Researchers and their research fields, example (4)

Caroline Hey Bækgaard, PhD student

Project title: "*desNIPT 2.0: Non-Invasive Fetal Genomic Screening*"

The primary goal of Caroline's project is to further develop and clinically validate the next-generation NIPT 2.0 assay to establish a comprehensive, non-invasive prenatal screening platform.

The project is derived from Center of Fetal Genetics and builds on a close collaboration between Department of Clinical Genetics and Department of Obstetrics and Gynecology, Odense University Hospital.
Non-invasive prenatal testing (NIPT), based on analysis of cell-free fetal DNA in maternal blood, has transformed prenatal screening by reducing the need for invasive procedures. However, first-generation NIPT mainly detects common chromosomal aneuploidies such as trisomy 21 and does not capture most rare monogenic disorders.

The project aims to develop the NIPT test, which has the potential to complement current prenatal screening by enabling the identification of a much broader range of genetic disorders – including those that cannot be detected by ultrasound. By integrating trio exome sequencing, the ability to identify both de novo and inherited genetic variants is enhanced.

Supervisors: Martin Larsen, Pernille Tørring, Department of Clinical Genetics, OUH, and Lene Sperling, Department of Gynaecology and Obstetrics, OUH.

Selected grants (1)

PhD student Louise Adel Jensen received DKK 48,000 from Fonden til Lægevidenskabens Fremme (A.P. Møller Fonden). The funds will be used in Louise's project on hereditary breast cancer, specifically for laboratory work: "In this project, we are performing whole-genome sequencing of tumour and blood samples from patients with hereditary breast cancer or suspected hereditary breast cancer. The aim is to identify mutation signatures that can be used to divide patients into molecular subgroups. This work builds on findings from Lars Andersen's PhD project and aims to improve risk stratification and genetic counselling".

PhD student Malthe Genét Lindenskov received DKK 650,000 in support of his PhD project: "Bone regulation of glucose-homeostasis" (GlucoBone).

PhD student Caroline Hey Bækgaard received PhD salary for 1 year from SDU and PhD salary for 1 year from the PhD funds of the Region of Southern Denmark.

MD Stine Bjørn Gram, PhD, received DKK 25,000 from Dansk Dermatologisk Selskab. The funds are to be used for a project on national whole genome sequencing for rare hereditary skin diseases.

GRANTS

Selected grants (2)

Professor Mads Thomassen and his research team received DKK 1.5 million from the Region of Southern Denmark's fund for personalised medicine. The funds have been allocated to the project: *"Circulating tumor DNA as a prognostic marker in primary breast cancer. A potential tool to reduce overtreatment"*.

Associate Professor Martin Larsen received DKK 785,000 from the Region of Southern Denmark's fund for Personalised Medicine 2025. The funds are allocated for the FetalScreen Study: *"Advancing Non-Invasive Prenatal Screening — Comprehensive Detection of Severe Fetal Disorders from a Single Maternal Blood Draw".*

GRANTS

FLAGSHIP CENTER

Center for Integrated Multi-Omics in Precision Medicine (CIMP), 1/2

The Center for Integrated Multi-Omics in Precision Medicine (CIMP) is a flagship research center between the University of Southern Denmark (SDU) and Odense University Hospital (OUH) focusing on reducing overtreatment of patients through precision medicine. CIMP functions as an interdisciplinary platform where research, clinical insight and advanced multi-omics and bioinformatic analyses are brought closer together to improve the basis for decision-making in patient treatment. The center is jointly led by Professor Vijay Tiwari and Professor Mads Thomassen and also includes Professor Thomas Levin Andersen, Professor Richard Röttger, Professor Zsolt Illes and project coordinator Sara Gregson. In addition, a number of other local, national and international partners are affiliated with the center.

Although modern medicine has made great strides, many patients receive standardised treatments that do not sufficiently take into account individual differences in disease and treatment response. This may mean that some patients receive treatment with limited effect while being exposed to unnecessary side effects, which also puts unnecessary pressure on healthcare resources. CIMP has been established to address this challenge by integrating multi-omics research, clinical expertise and advanced bioinformatic analysis into the development of more individualised and patient-centred treatment decisions.

FLAGSHIP CENTER

Center for Integrated Multi-Omics in Precision Medicine (CIMP), 2/2

CIMP is currently working on projects in three disease areas: breast cancer, multiple sclerosis and osteoporosis. In breast cancer, CIMP is working to improve decisions about post-operative treatment so that patients who do not benefit from further treatment can be spared unnecessary treatment and side effects. In multiple sclerosis, research is being conducted to identify which patients continue to benefit from treatment and which patients can safely reduce or stop treatment in order to minimise complications from long-term immunosuppressive therapy. In the field of osteoporosis, CIMP is working to improve treatment strategies when discontinuing denosumab to prevent rapid bone loss and reduce the risk of serious fractures.

Overall, CIMP is an ambitious and interdisciplinary center that aims to contribute to making future treatment more tailored to the individual patient, thereby improving patients' quality of life and supporting a more sustainable healthcare system.

CIMP was approved as new research flagship in December 2024 by SDU's and OUH's research councils and deans and directors from SDU and OUH.

INVOLVEMENT IN EUROPEAN REFERENCE NETWORKS (ERN)

European Reference Networks share knowledge on rare diseases between health care providers across Europe. The purpose is to develop clinical expertise within specialties, to improve treatment of rare diseases and to increase networks and ensure equal access to rare disease treatment. At present, there are 24 networks across Europe. Odense University Hospital is a member of 9 of these networks.

On the basis of our expertise in genetics, the Department of Clinical Genetics participates in the three ERNs ITHACA, VASCERN and SKIN and cooperates with further two networks: ERN BOND (rare bone diseases) and Endo-ERN (rare endocrine diseases).

ERN ITHACA

ITHACA is the European Reference Network for rare malformative conditions, intellectual disabilities and neurodevelopmental disorders. The network today consists in 71 expert clinical centres and 45 European Patient Advocacy Groups (EPAGs). The network promotes improved patient care, research, and access to information in patients with rare congenital malformations and intellectual disability, through the exchange of expertise, experience, and resources.

Professor Lilian Bomme Ousager, the Department of Clinical Genetics, is lead of ERN ITHACA at OUH in close collaboration with H. C. Andersens Childrens' Hospital.

ERN ITHACA meets twice a year, recently in Bergen, Norway, where Professor Lilian Bomme Ousager and MD, PhD, Louise Lehmann Christensen participated from OUH.
The network meetings include one day of research presentations, and the network has the opportunity to share case studies on an ongoing basis – an initiative which was started in 2024 in order to enable the network to utilise the broad collective expertise.

In 2025, the first Clinical Guideline on Kleefstra Syndrome was elaborated (published in January 2026).

INVOLVEMENT IN EUROPEAN REFERENCE NETWORKS (ERN)

VASCERN HHT – Danish Contribution
VASCERN is the European Reference Network for Rare Multisystemic Vascular Diseases, established to improve care for patients with complex and rare vascular disorders across Europe. Within VASCERN, VASCERN HHT focuses specifically on Hereditary Haemorrhagic Telangiectasia (HHT).

Denmark is represented by Professor Anette Kjeldsen, the Department of Otorhinology, and MD, PhD Pernille M. Tørring, the Department of Clinical Genetics. The VASCERN HHT group participates in monthly virtual meetings, supporting structured European collaboration and cross-border case discussions. Over 50 complex clinical cases have been reviewed within the network.
Key achievements include the development of a European patient pathway, publication of multiple Dos and Don'ts for General Care, three position statements, more than 10 peer-reviewed publications, webinars and the completion of the European HHT Registry project. Furthermore, two 'Based on Evidence' European (BEE) Meetings (2023 and 2025) further strengthened collaboration among experts, clinicians, and patient representatives.

Given that HHT is a rare disorder, European networking through VASCERN continues to be invaluable in improving patient care and outcomes.

ERN SKIN
ERN SKIN is the European Reference Network for rare and complex skin disorders. At OUH, the Danish ERN SKIN activities are coordinated through the Department of Dermatology, which provides highly specialized diagnosis, treatment, and follow-up of patients with rare skin disorders.

Through ERN SKIN, OUH collaborates on clinical expertise, knowledge sharing, and case discussions. The network also facilitates joint research initiatives, development of clinical guidelines, and education aimed at improving diagnosis and care for patients with rare skin diseases across Europe.

OUH is represented by MD, PhD Annette Schuster, the Department of Dermatology, and MD, PhD Stine Bjørn Gram, the Department of Clinical Genetics. Stine Bjørn Gram participated in the ERN-Skin Board Meeting in Paris in 2025.

CENTER FOR RARE AND COMPLEX DISEASES (CAKS)

The Center for Hereditary and Complex Diseases (CAKS) is a virtual center working with all relevant departments at OUH to establish and update patient pathway descriptions for selected rare, hereditary and complex diseases.

In 2025, CAKS worked with the database unit in the Region of Southern Denmark to implement the treatment database, CAKSsyd. CAKSsyd provides an overview of patients associated with the various disease groups in CAKS, including the status of the care programme for each individual patient. In addition, CAKSsyd makes it possible to optimise workflows around the multidisciplinary conferences in CAKS, freeing up time for clinical discussions and research collaboration.

New diseases added in 2025:

- *BAP1* – tumor predisposition syndrome
- m.3243A>G mitochondrial diabetes – mitochondrial disease

In October 2025, the third CAKS Annual meeting was held, organised by CAKS and the ERN Secretariat (Secretariat for the European Reference Networks at OUH). This Annual Meeting focused on two themes: 1. Rare bone diseases, and 2. The many kinds of support services available for people with rare diseases.

A big ‘Thank you’ to the speakers: Chair of Rare Diagnoses **Liselotte Wesley Andersen**, Senior Consultant **Lars Folkestad**, the Department of Endocrinology, OUH, and Senior Consultant **Anders Jørgen Schou**, H.C. Andersen Children's and Youth Hospital, OUH.

We look forward to seeing you all at the next Annual meeting in 2026.

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 material 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 61 publications/articles written by 34 different authors from our research unit, 21 of these are authors of two or more publications.

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The research committee supports research in the department (for patients and healthcare) and ensures interaction between research, clinical practice and other functions.

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