

## Contact

### Clinical Genetics

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**For more information about the department, please visit our webpage**

[www.akademiska.se/sv/Verksamheter/Akademiska-laboratoriet/Klinisk-genetik/](http://www.akademiska.se/sv/Verksamheter/Akademiska-laboratoriet/Klinisk-genetik/)

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**Welcome to Clinical Genetics  
at Uppsala University Hospital in Uppsala**



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UPPSALA UNIVERSITY HOSPITAL

## Department of Clinical Genetics

Clinical Genetics is a medical discipline that serves all specialties where genetic inquiries are present. We offer expertise in prevention, diagnostics and follow-up of various genetic diseases for the entire healthcare region of Uppsala-Örebro.

Collaboration takes place at a regional, national and international level. We participate in expert groups and multidisciplinary rounds involving pediatrics, prenatal diagnostics, hematology and oncology. Training and teaching is a part of the daily work.

## Patient Reception

Patient reception is divided into three sections: Oncogenetics, pediatric genetics and general genetics.

We receive patients and families with various suspected or clinically verified congenital genetic diseases and syndromes as well as individuals/families with suspicion of familial cancer. We also offer genetic counselling and guidance within both hospital and wider healthcare region.

## Laboratory

Our laboratory consists of cytogenetics and molecular genetics and conducts accredited activities. We perform various types of analyses, regarding both congenital and acquired genetic conditions, by using the latest and most advanced laboratory techniques like Next-Generation Sequencing (NGS).

## Research

Our department carries development and research concerning syndromes, monogenic diseases and hematological malignancies, in collaboration with other laboratories like *Science for Life* laboratory. Biobank-based laws are applied to all samples stored in our facilities.

## Centre for Rare Diseases

Our Centre for Rare Diseases helps people with rare diseases to have the same opportunity for diagnosis, adequate treatment and community service.

## The Department of Clinical Genetics employees

Clinical geneticists/specialists in pediatrics, hematology and oncology

Residents in clinical genetics

Geneticists

Nurses

Genetic counselors

Biomedical analysts

Laboratory assistants

Medical secretaries

Administrators



Next Generation Sequencing (NGS)