

Professor Anna Lindstrand is a specialist in Clinical Genetics. Director of the Clinical Genetics diagnostic laboratory at Karolinska University Hospital and group leader for Rare Diseases research group at Karolinska Institutet. She is active in Genomic Medicine Sweden (GMS), both as co-chair of GMS - Rare Diseases and a member of the GMS management group.

Annas main areas of research are the study of rare genetic diseases, both clinically and at the molecular level to improve genetic diagnostics, increase knowledge about genotype-phenotype correlations and further understanding of disease biology. Especially interested structural genomic variation, how it forms and how it causes human genetic disease. Detailed characterization of chromosomal rearrangements is performed with a variety of methodologies and next generation sequencing platforms. Findings are functionally validated in induced pluripotent stem cells and zebrafish embryos.

Annas group has pioneered the implementation of WGS-karyotyping or cytogenomics in clinical diagnostics as well as research. They have developed several novel bioinformatic tools for the analysis of structural variants from whole genome sequencing data. The pipelines have been implemented in clinical diagnostic at the Karolinska University Laboratory, increasing the diagnostic yield of WGS based gene panels and trios. Most recently she has evaluated different sequencing technologies, including long read sequencing, to detect and characterize structural variants in rare disease patients.