

Nationally coordinated Whole Genome Sequencing of individuals with a suspected rare genetic disease

The swedish GMS-RD consortium ¹

BACKGROUND Genomic Medicine Sweden Rare Diseases (GMS-RD) is one of seven national reference groups within the Swedish GMS initiative. The nationwide network consists of six regional genomic medicine centers (GMCs) representing all Swedish University Hospitals and their corresponding universities.

OVERALL MISSION of GMS-RD:

- To ensure that all patients in Sweden with a suspected rare genetic disease will have equal access to the most appropriate genomic investigations based on the patient's symptoms.
- To coordinate the competence and resources in Sweden for performing and interpreting genomic data to ensure high quality as well as cost-effectiveness.
- To develop informatics resources to facilitate data sharing, variant interpretation and to enable genetic research.



PILOT STUDY First, we will conduct a nation wide pilot study where individuals with intellectual disability and a suspected rare disease are offered whole genome sequencing (WGS) based trio analysis. By introducing clinical WGS in all GMCs, our study will pave the way for clinical WGS as a routine diagnostic tool in many different rare diseases.

The primary aim of the WGS pilot study is related to the question of added diagnostic value of WGS in Swedish healthcare, but will also serve as a starting point for building a Swedish genotype-phenotype database to support the clinical

interpretation of a WGS result in a corresponding population context, and to facilitate national and international data sharing.

The data that is generated from this study will be used for method development to further increase the sensitivity of WGS and thus minimize the need for (and the cost of) additional genetic analyses. The project will also serve as a catalyst for a national coordination of the analysis pipeline, all the way from patient sample to analysis report. Furthermore, we will use the data to expand our knowledge about the genetic basis of rare diseases.

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