



Genomic Medicine Sweden – building a national infrastructure for precision medicine within a regionalized healthcare system

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Precision diagnostics



Healthcare



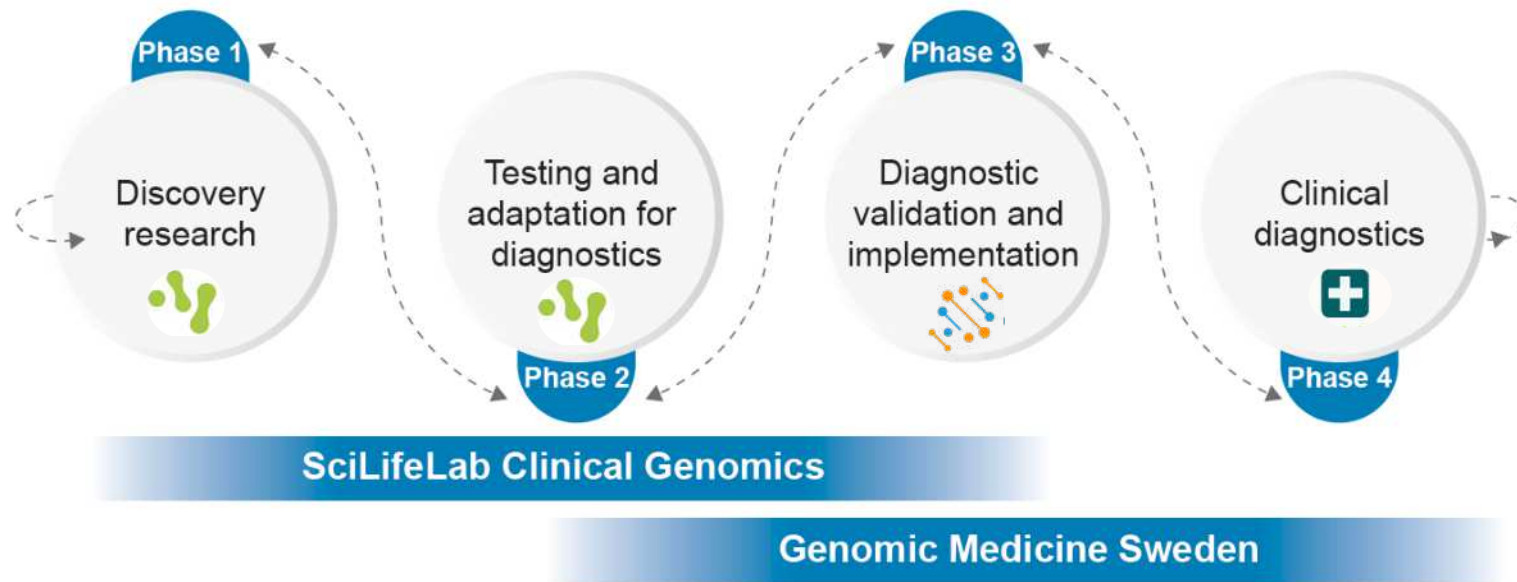
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Clinical Genomics Platform
(optimize and adapt for diagnostics)

Genomic Medicine Sweden
(broad national implementation)

Implementing precision medicine in a regionally organized healthcare system in Sweden





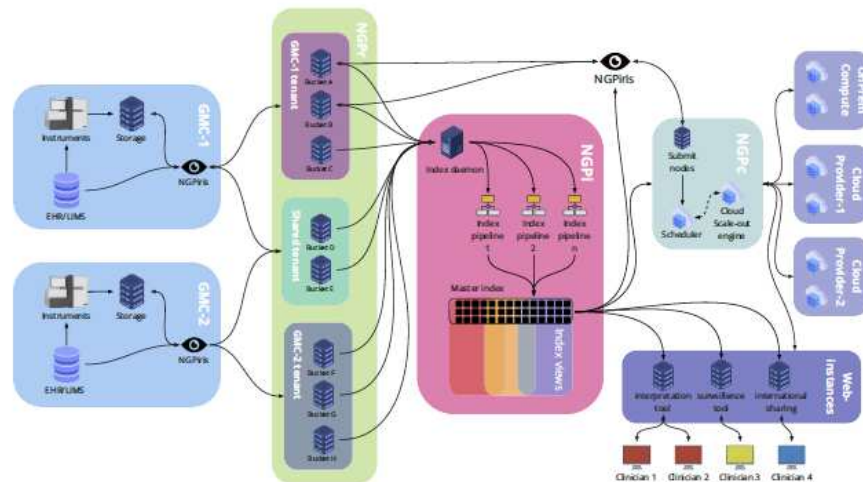
GMS coordinates implementation of precision medicine across Sweden

- **Clinically focused** – bottom-up initiative
- **National** - 7 Genomic Medicine Centers
- **Harmonised** - national standards, guidelines and methods
- **Cost-effective** – coordinated implementation
- **Common IT infrastructure** – National Genomics Platform

Offer patients in all healthcare regions equal access to precision medicine through a nation-wide collaborative effort

National Genomics Platform

- National resource for healthcare, research and innovation (4,5 PB)
- Link to electronic health records and quality registries
- Genomics data can be shared across the country within research projects with ethical approval
- Ongoing governmental inquiry on secondary use of health data



Per Sikora



Tobias Strid



Valtteri Wirta



Katarina Nyström



Focus areas - diagnosis and treatment

**Rare
diseases**



Hematology



Cancer
Solid tumors



Pediatric
cancer



**Microbiology/
Infectious
diseases**



**Pharmaco-
genomics**



**Complex
diseases**



Whole-genome sequencing in rare diseases

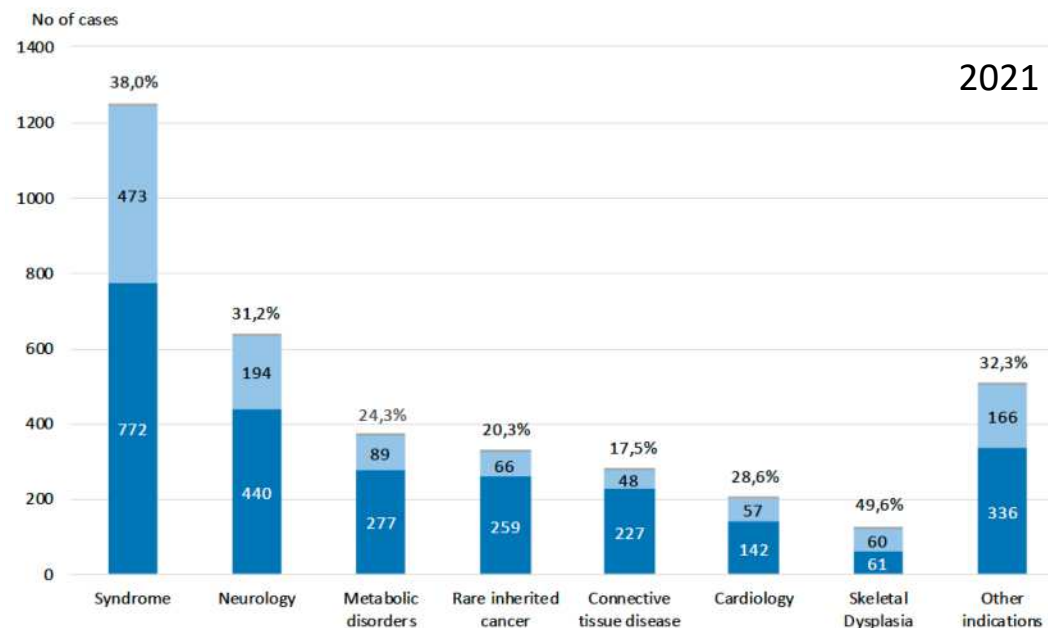
5 800 patients were analyzed with WGS within healthcare in 2022

WGS is available at 4 GMCs

Introducing RNA-sequencing



Digital consent project



Anna Lindstrand



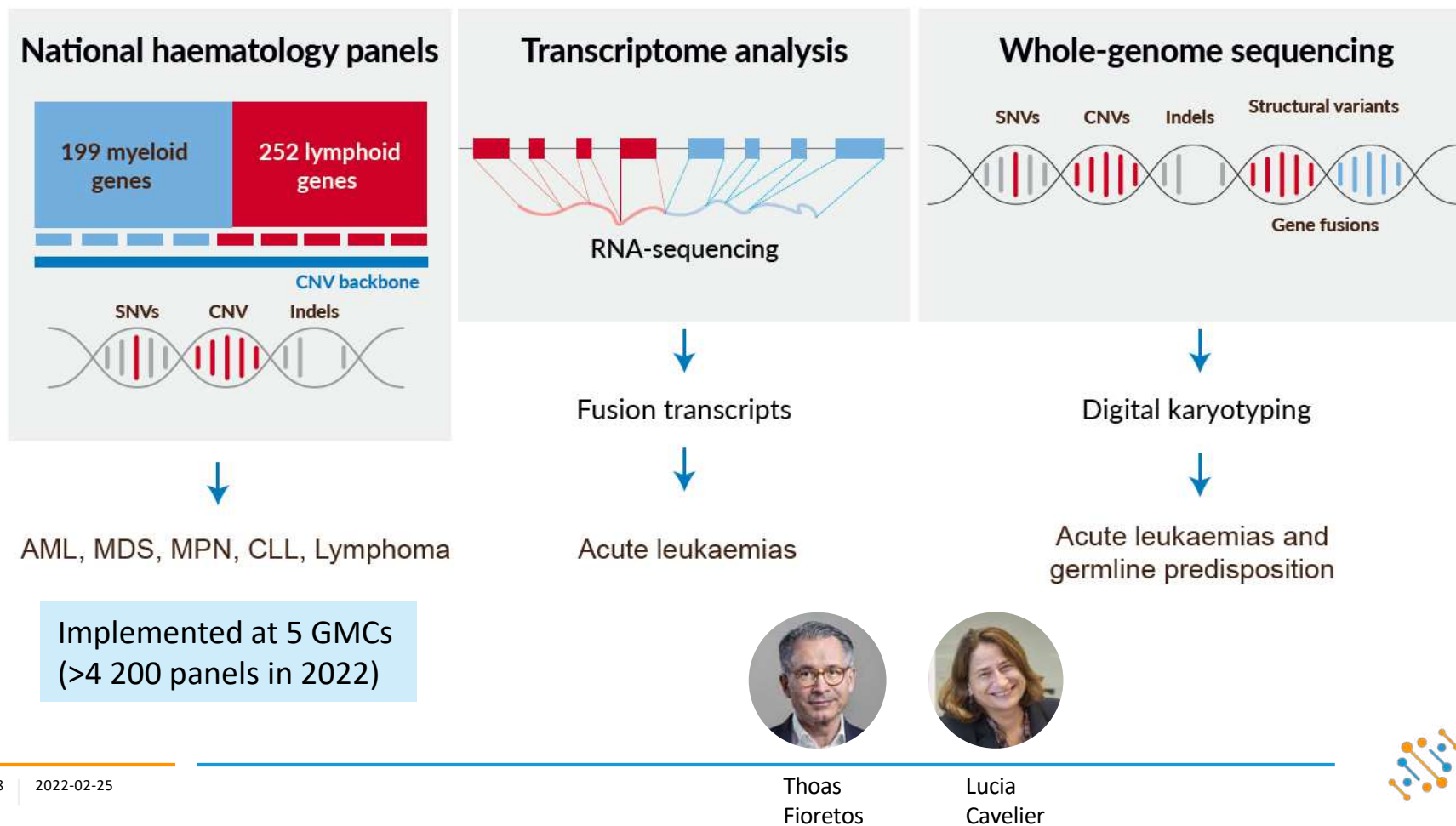
Hans Ehrencrona



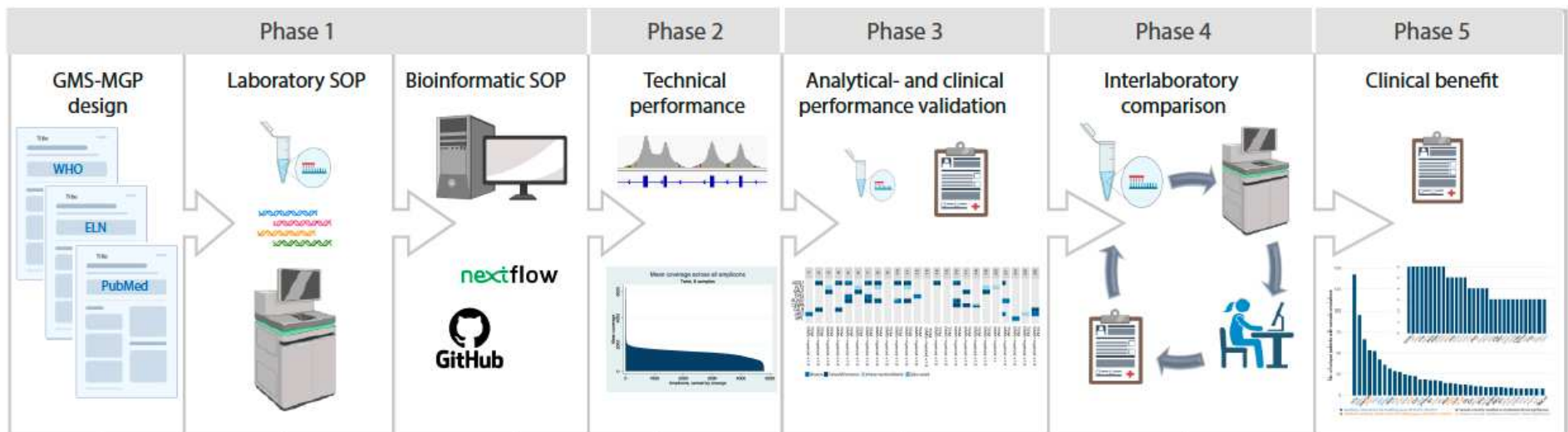
Lovisa Lovmar



Strategy genomic profiling in hematological malignancies

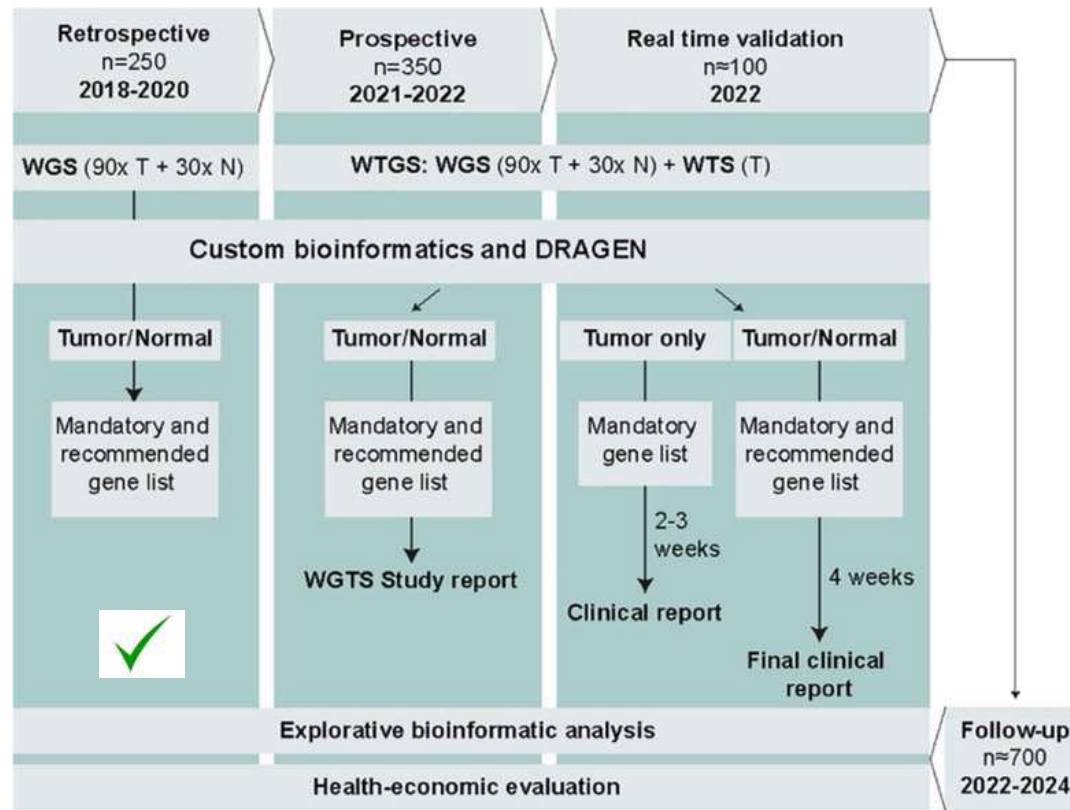


National validation of the GMS Myeloid Gene Panel



Orsmark-Pietras et al, submitted

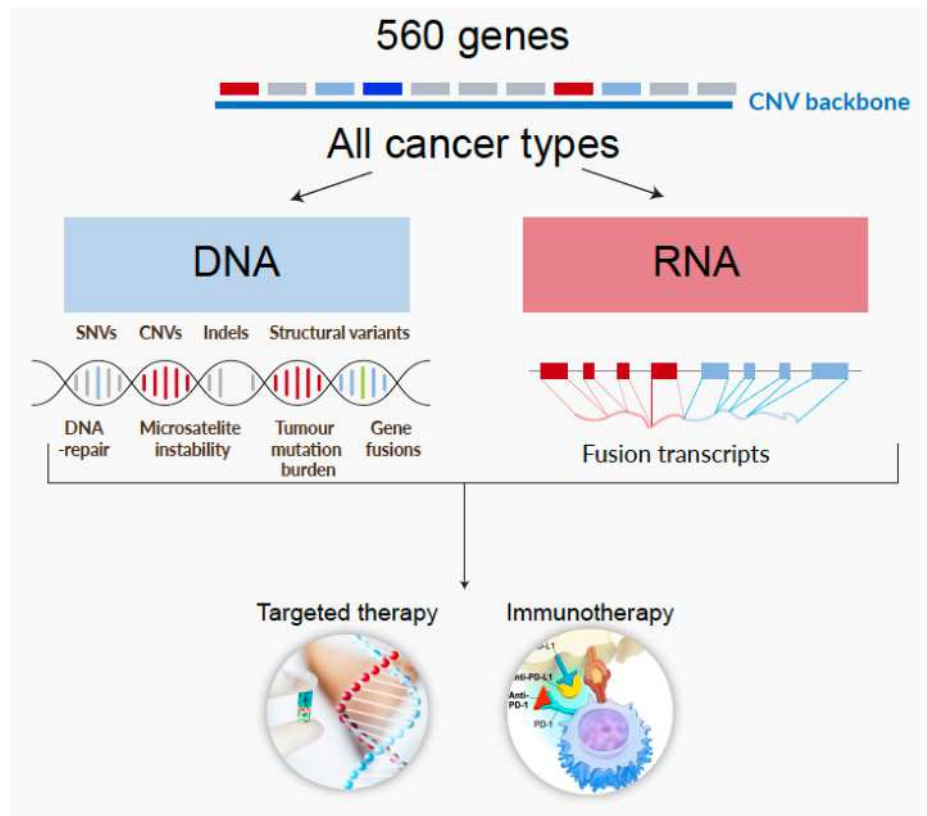
Validation and implementation of WGTs in acute leukemias



Prospective phase

- 352 patients consented
- 325 sequenced
- 191 patients compared to SoC
- Very high concordance to SoC

Comprehensive genomic profiling in solid tumors



Ongoing implementation of
GMS560 at all 7 GMCs

National pilot projects in breast,
ovarian and lung cancer

Work is ongoing with liquid biopsies



Anders Edsjö



Johan Botling

Test Bed Sweden for Precision Health in Cancer



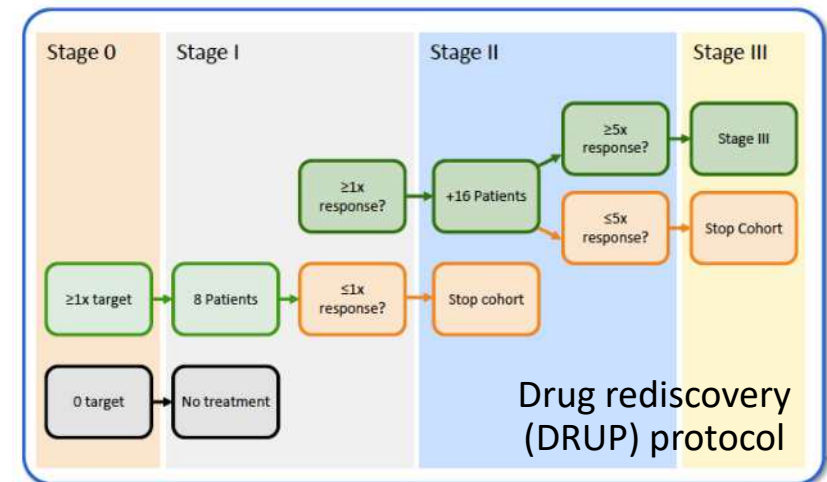
- Increased availability of clinical trials
- Improved precision diagnostics and prevention
- More effective treatments



Ebba Hallersjö Hult



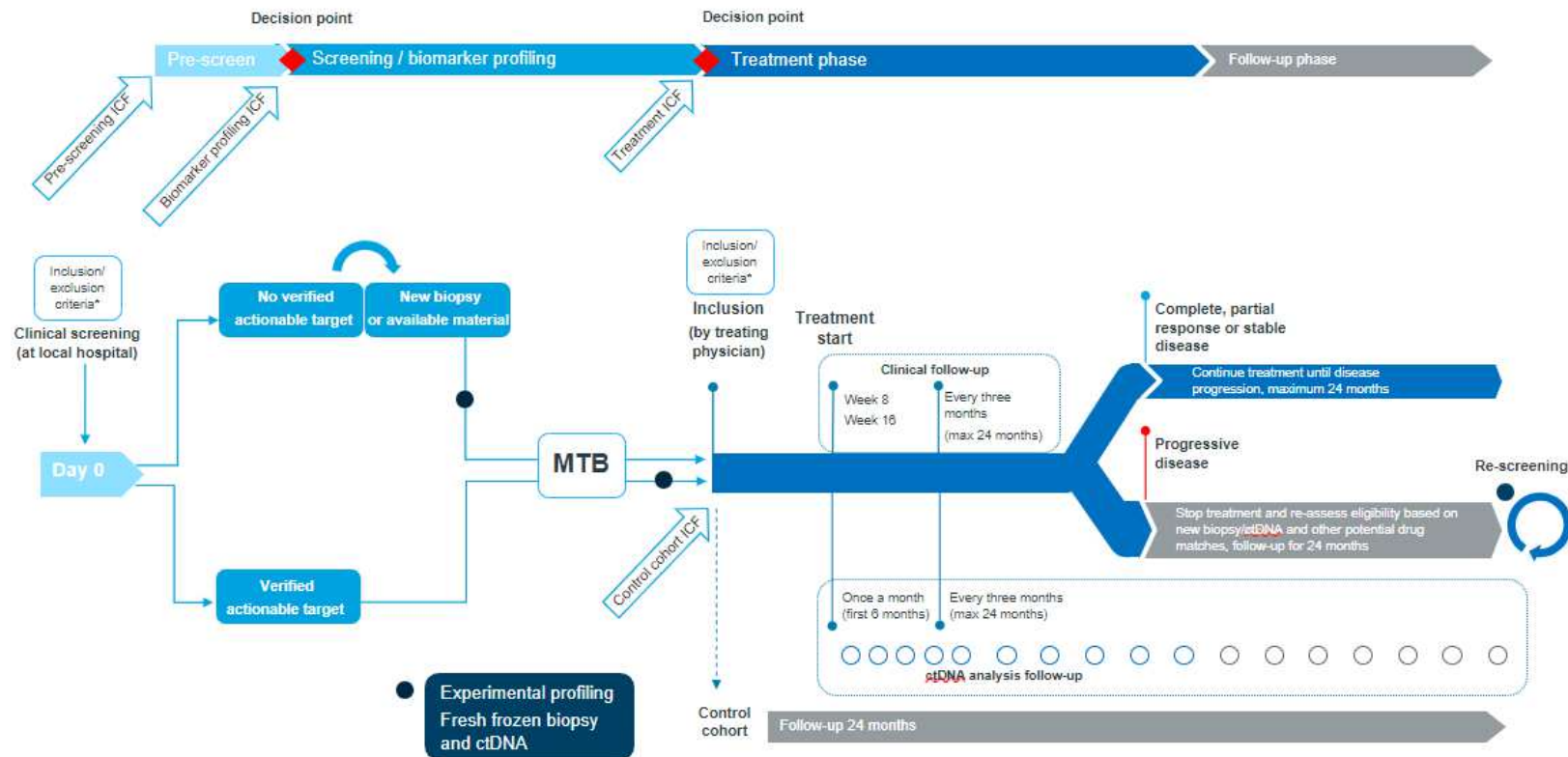
Anders Edsjö



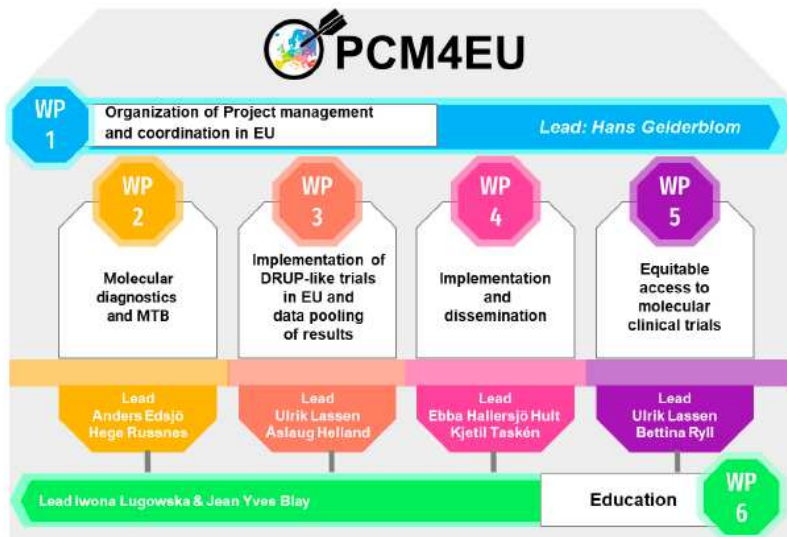
Goal Q1 2024: start up national precision cancer medicine (DRUP) trial



National PCM Study Sweden

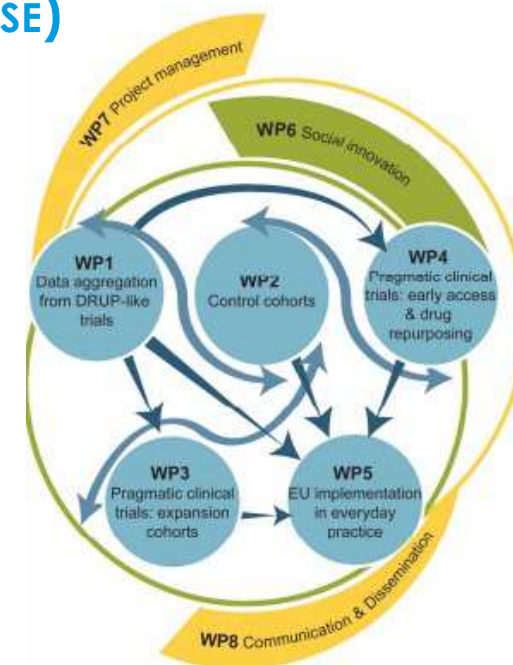


Personalised Cancer Medicine for all EU citizens



15 countries, based on the DRUP trial concept
Coordinated by Hans Gelderblom, the Netherlands

PRECISION CANCER MEDICINE REPURPOSING SYSTEM USING PRAGMATIC CLINICAL TRIALS (PRIME-ROSE)

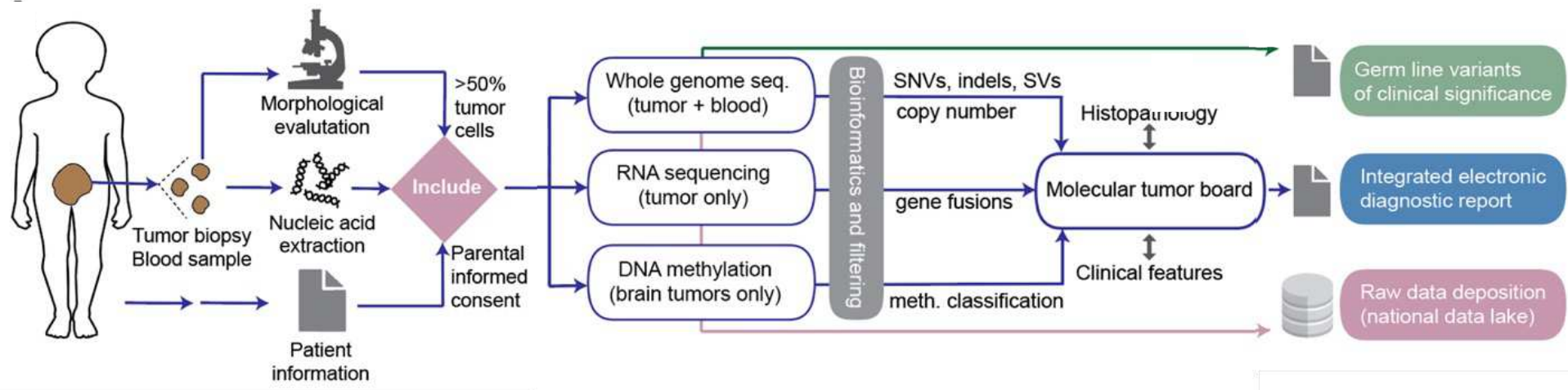


Coordinated by Kjetil Taskén, Norway

GMS Childhood cancer



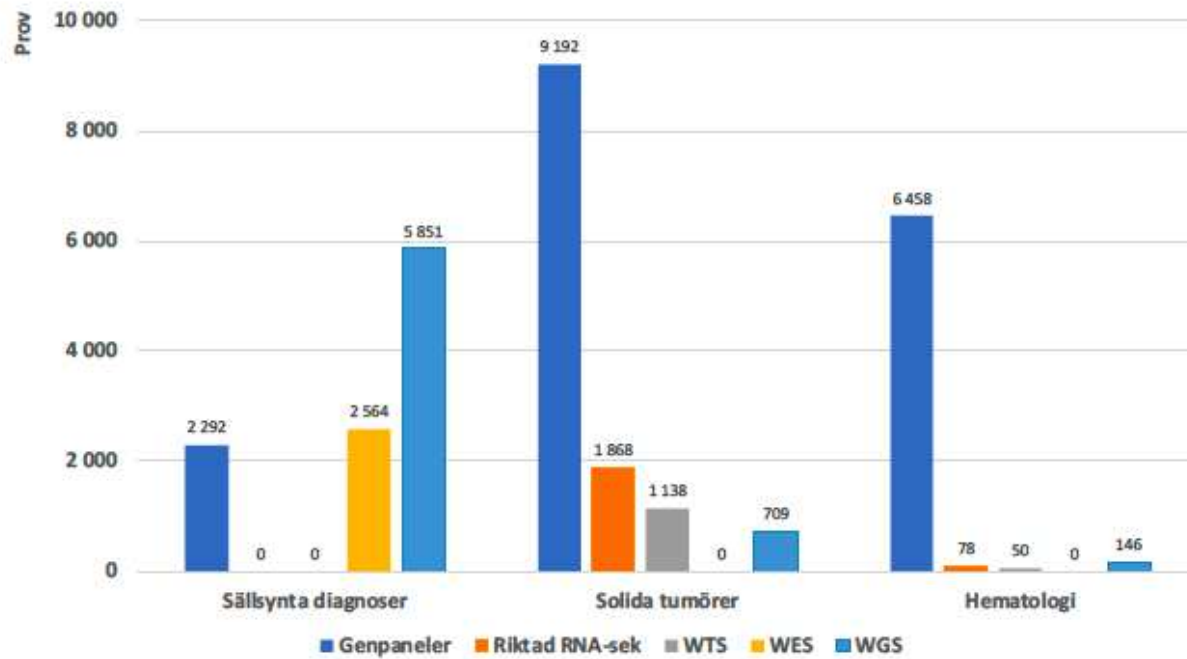
David Gisselsson-Nord



Clinically relevant genetic alterations detected in 91%
Potential targeted therapy in 24% (Wadensten et al, JCO Precision Oncology 2023)



Clinical sequencing performed in 2022



2018-2022

WGS/WES in rare diseases
25 000

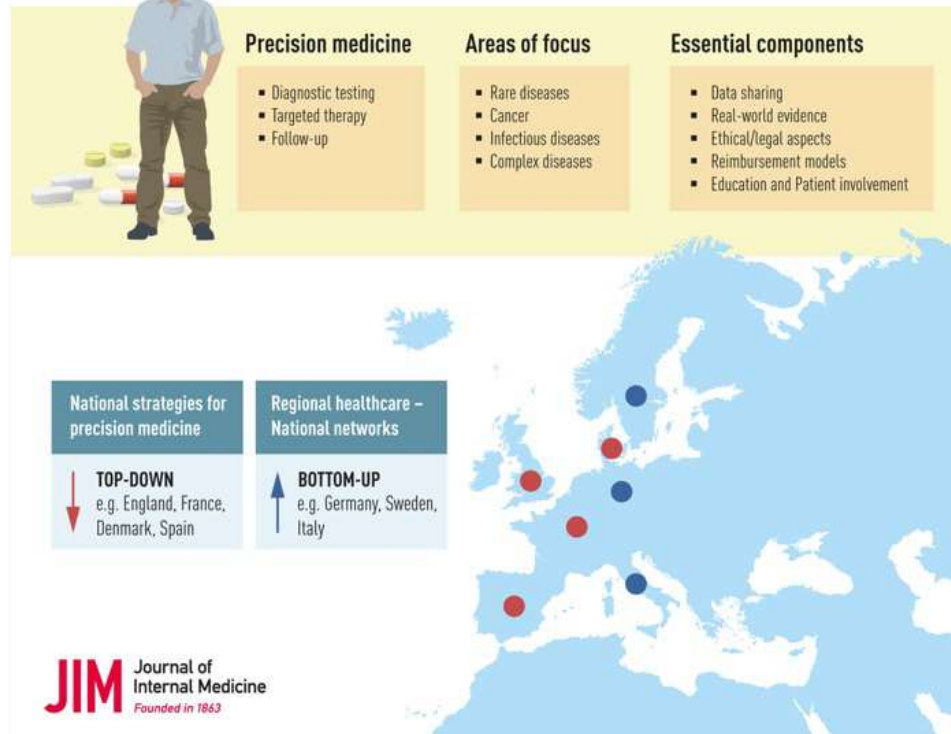
WGS in cancer
1 200

Gene panels in cancer
55 500

International collaboration



Implementation of precision medicine in healthcare – a European perspective



5 dedicated reviews:

- European perspective on PM
- Rare Diseases
- Hematological malignancies
- Solid tumors
- Complex diseases

To be successful we need to collaborate

National



International



Local

Karolinska University Hospital and Karolinska Institutet establish joint precision medicine centre

NEWS 2021-02-26

Karolinska University Hospital, Karolinska Institutet and Region Stockholm have been working together for just under a year to increase the pace of introduction of precision medicine, which briefly means diagnostics, treatment and follow-up at the right time and adapted to the patient's individual circumstances.



Precision Medicine Center Karolinska

Important lessons - the way forward

- Cooperation is required at all levels: national, regional and international
 - Strategic partnership between the state and healthcare regions
 - Permanent national infrastructure for precision medicine including all stakeholders
- Data sharing and secondary use of data not possible today
 - Governmental inquiry on secondary use ongoing
- Continue implementing technologies together with SciLifeLab
 - Multi-modal diagnostics
 - Clinical decision support tools/AI
- Investments in national PM trials are essential
- Need to educate the profession, patients and society



Nobel Symposium - 20-22 September 2023

Precision Medicine Transforms Healthcare: A New Trajectory for Research and Innovation



Venue: Nobel Forum, Karolinska Institutet, Stockholm



Thank you for listening!

www.genomicmedicine.se

