



Per Sikora, PhD

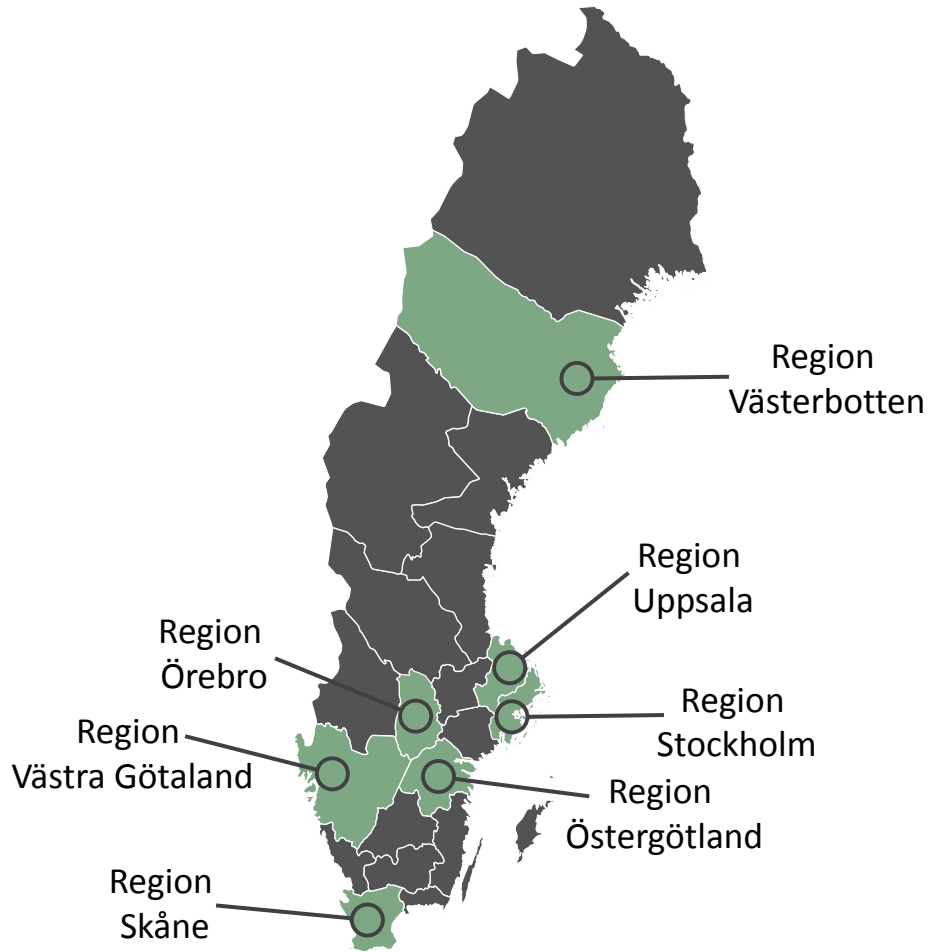
Chair of Informatics and Infrastructure, GMS

First Bioinformatician, Sahlgrenska University Hospital

# Sweden in Europe

5	 Sweden	450,295
7	 Germany	357,386 <sup>[5]</sup>
10	 Italy	301,338
11	 United Kingdom*	242,495

2	 Germany	82,887,000
5	 United Kingdom <sup>[Note 4]</sup>	66,040,229
6	 Italy	60,390,560
17	 Sweden	10,215,250



- Healthcare budget set by region
- Issues in sharing data between regions
- Heterogeneous diagnostic standards
- Plethora of EHR-Systems
- Small patient populations
- Little collaboration

# **How do we achieve precision health?**

## **(In Sweden)**

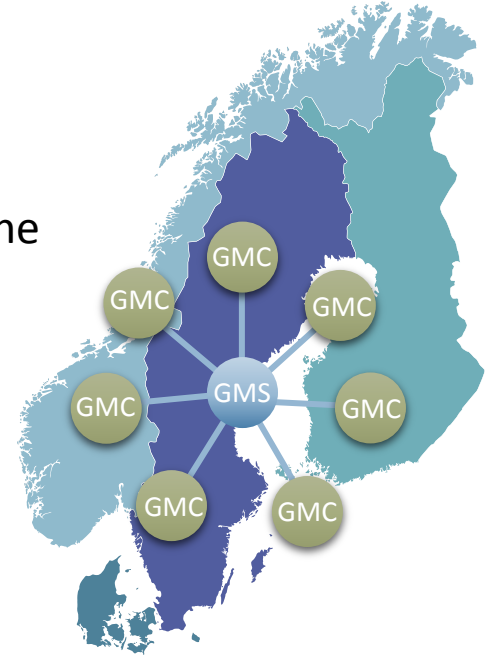
# Genomic Medicine Sweden



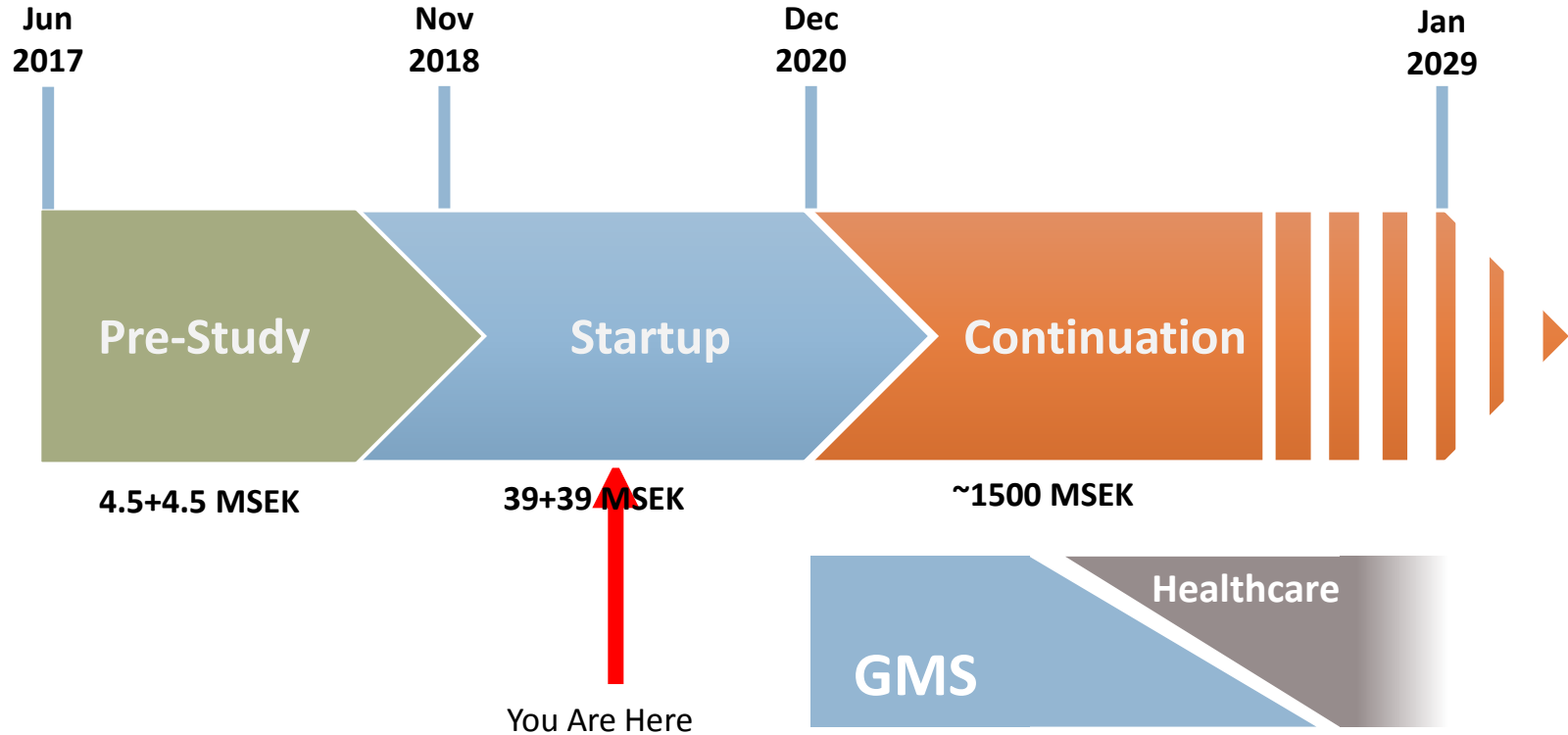


# GMS Mission Statement

- Modern diagnostics using NGS
  - Precision medicine – the correct treatment for everyone
  - Foster national collaboration within healthcare
  - Harmonize care across regions
  - Create unique national research opportunities
  - Further industry innovation and collaboration
- 
- Genomic Medicine Centers
  - New national structures and infrastructure in swedish healthcare

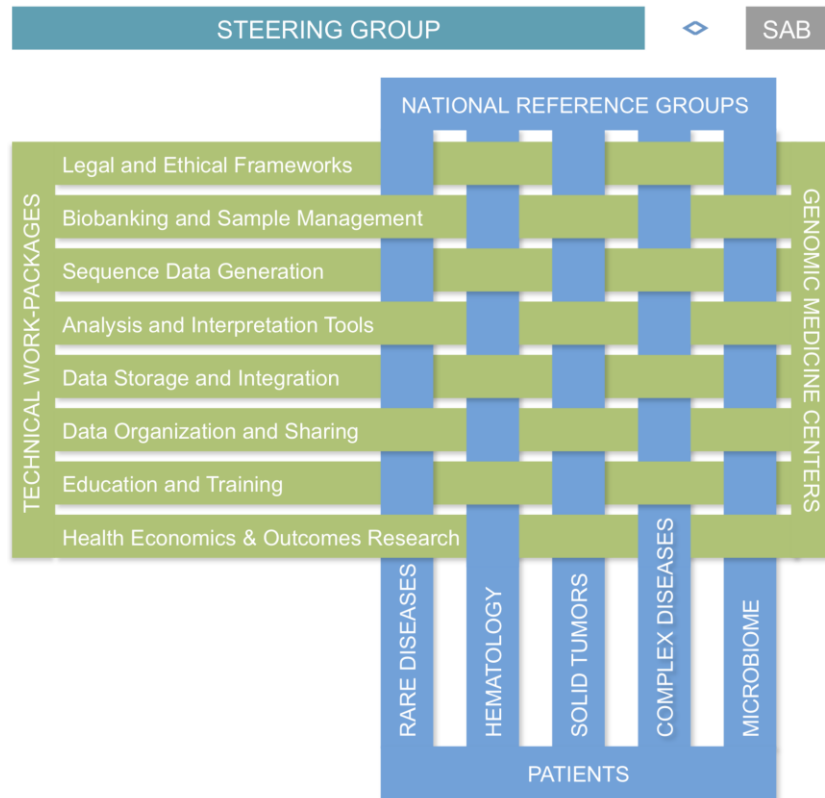


# GMS Project timeline

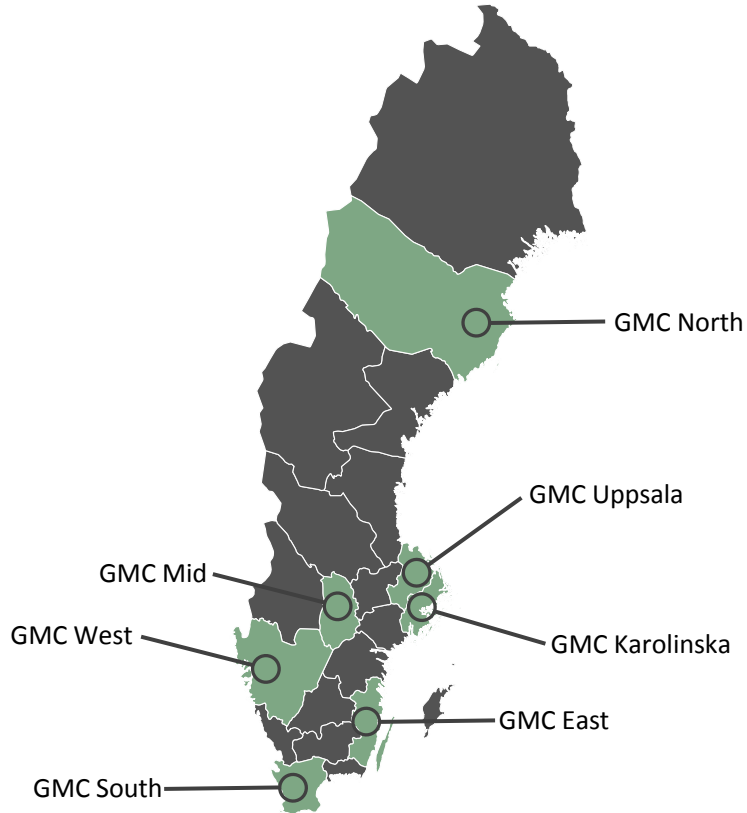




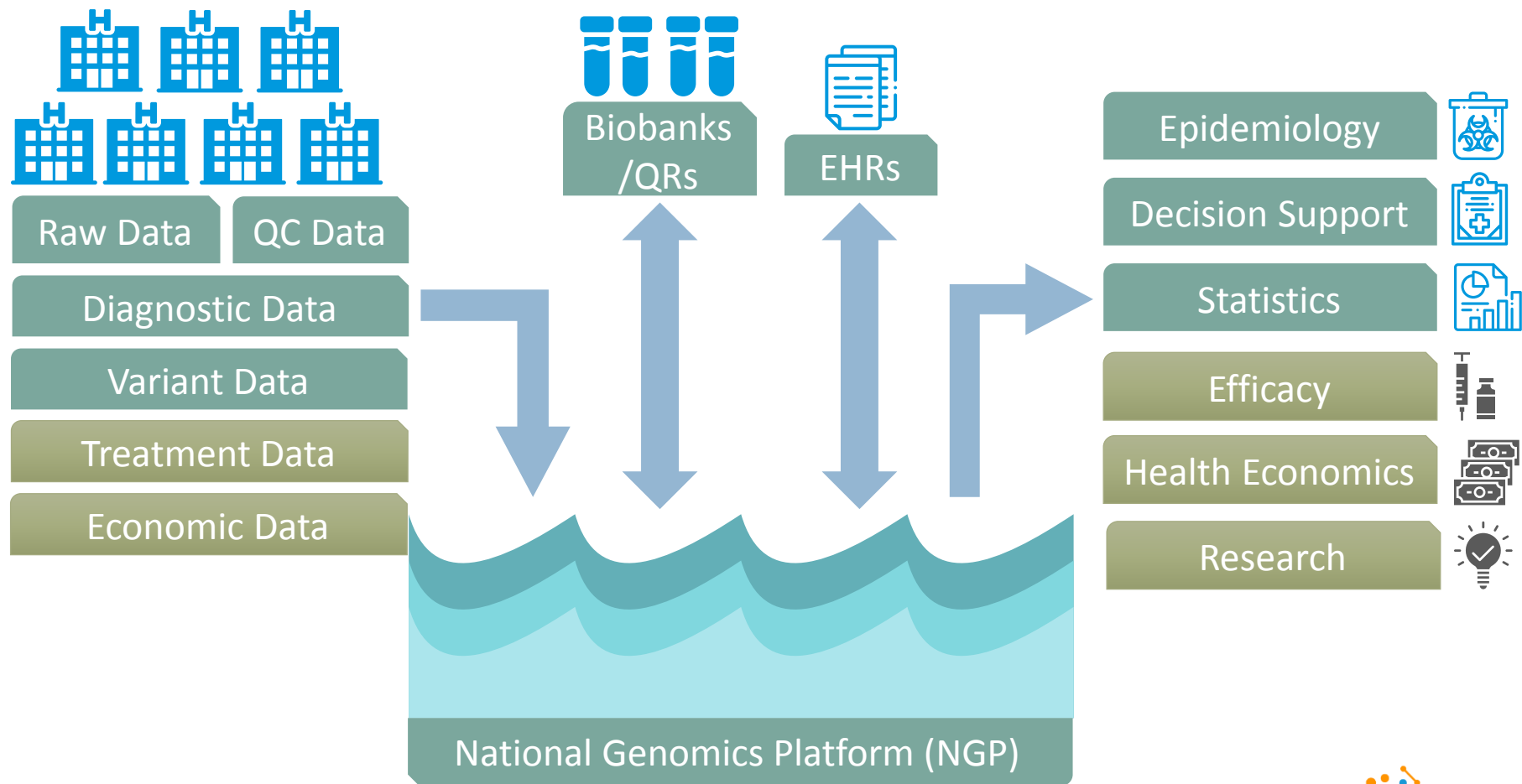
# GMS Organisation

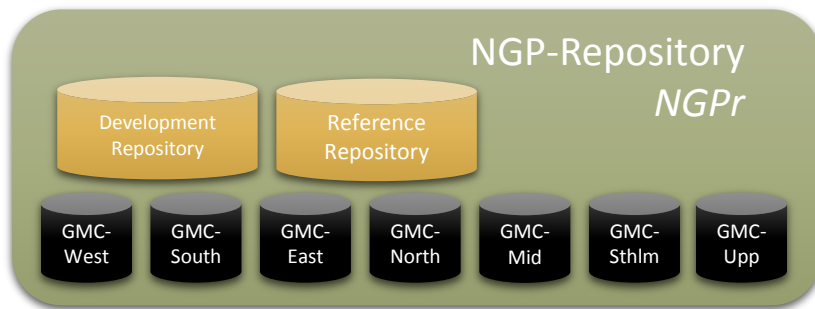
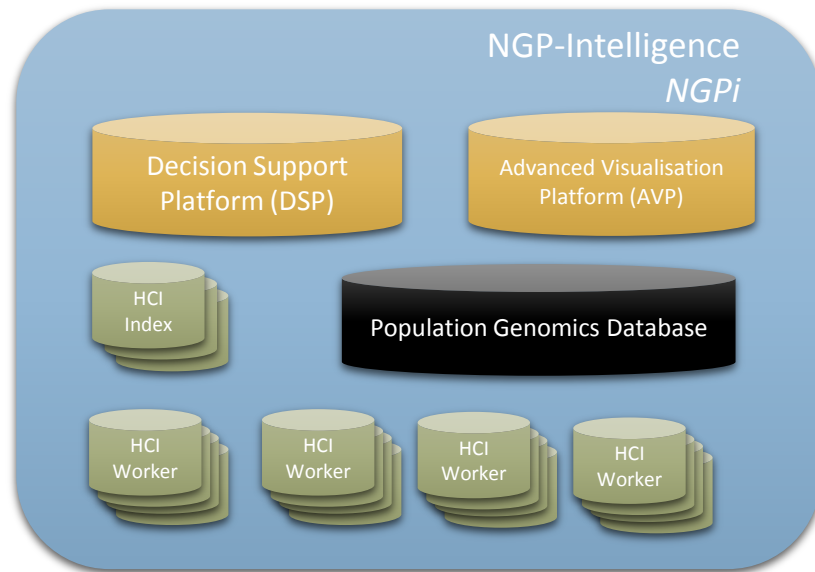
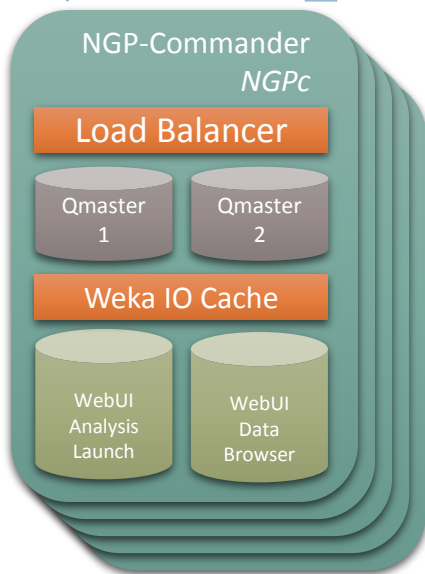
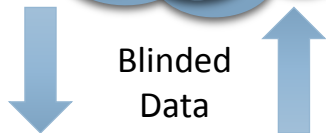
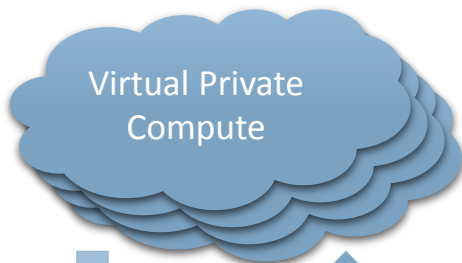


# Genomic Medicine Centers



- Organised by host university and university hospital
- Represents each region
- Supports specialised care
- Wide range of molecular diagnostic tools
- Part of national coordinated effort





# GMC-West/Sahlgrenska University Hospital

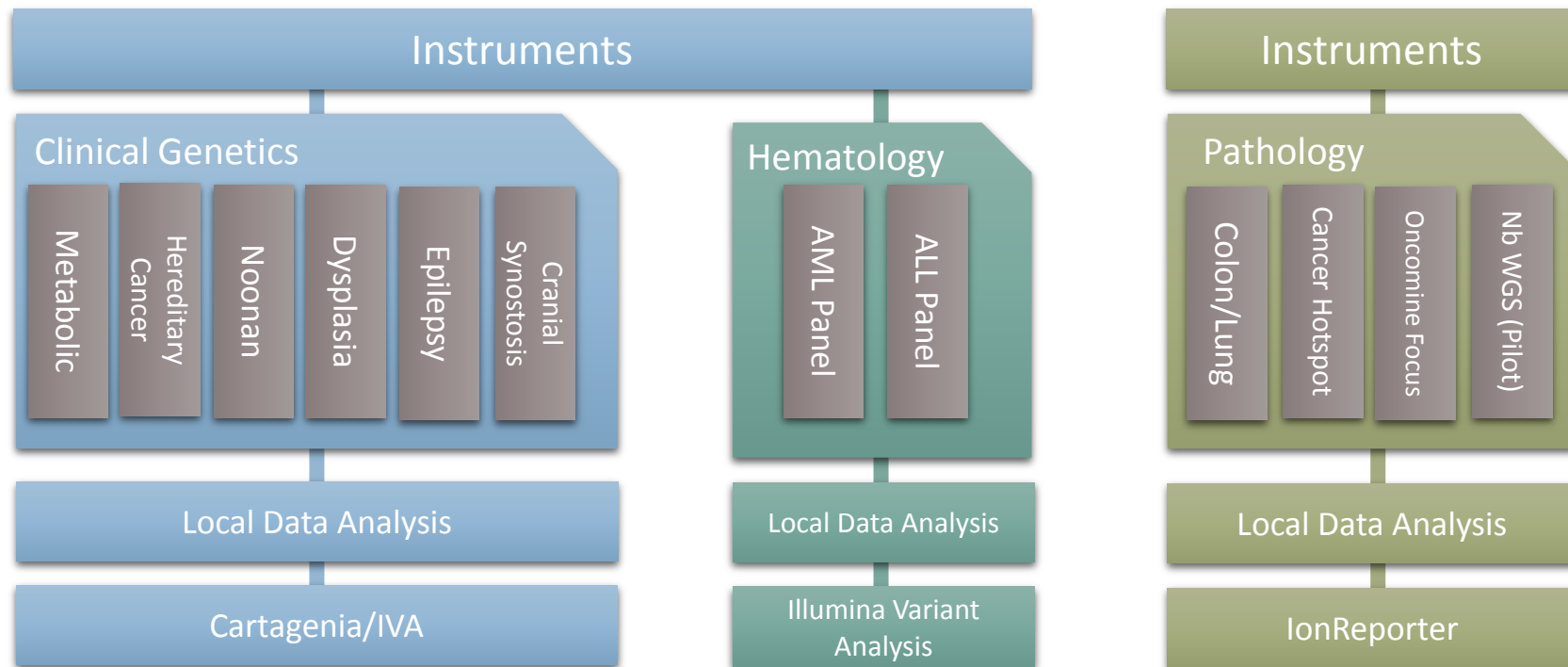


- Founded in 1772, Nicolaus Sahlgren
- 24 beds, 14 for paying customers
- Pehr Dubb first "Admiralty medicus"
- 2300 beds, 17000 employees
- Serves 1.7 million people

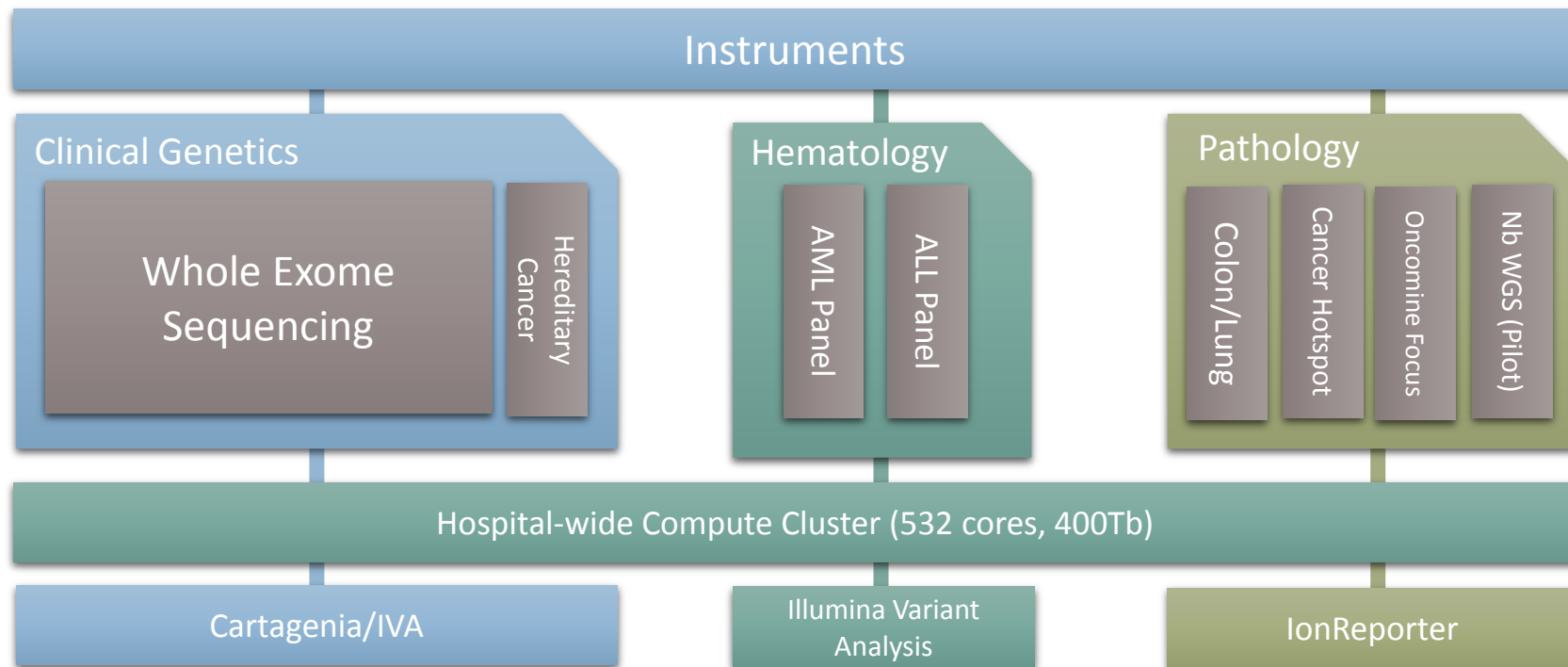
# NGS at Sahlgrenska



# Sahlgrenska - 2016

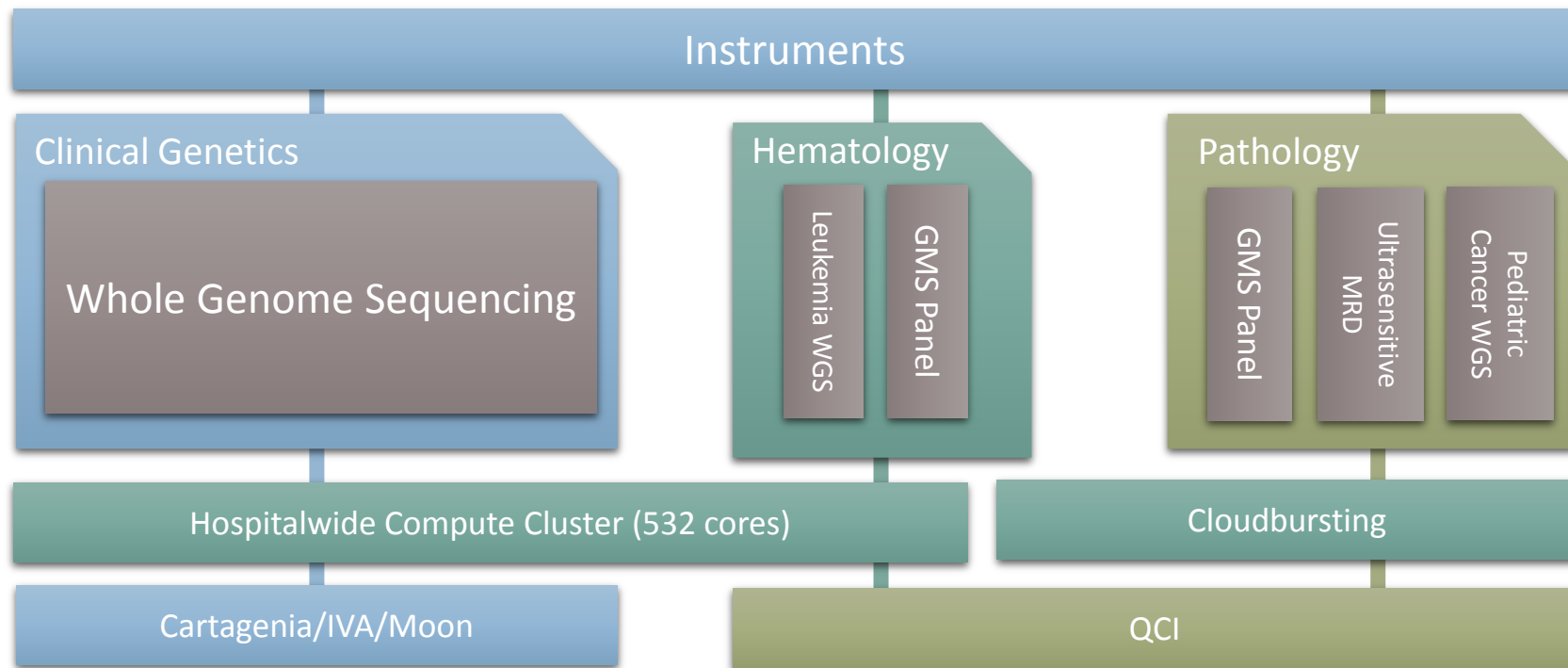


# Sahlgrenska - 2018





# Sahlgrenska - 2019

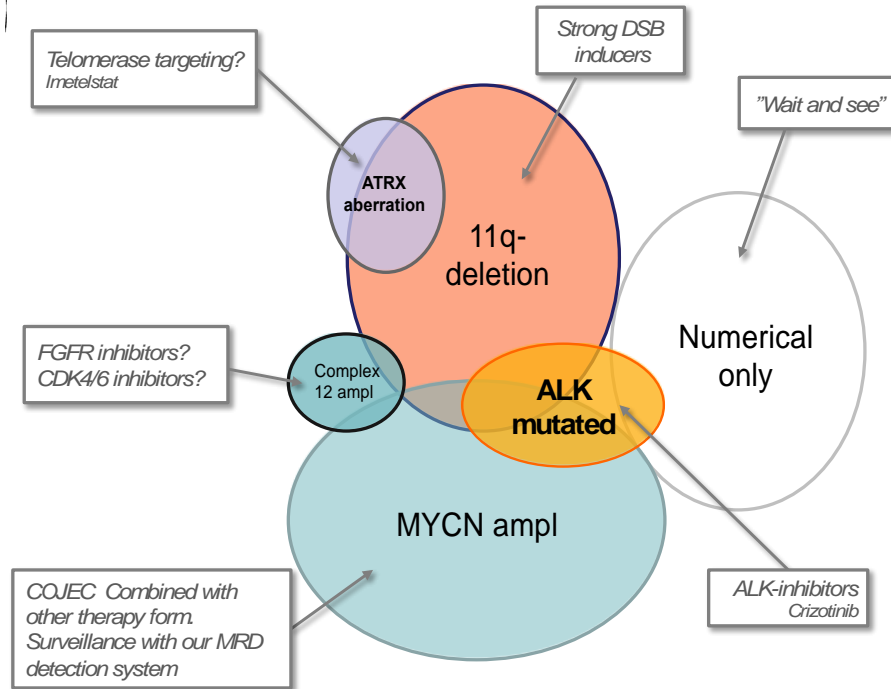


# Neuroblastoma –Pedaitric Cancer

Tumor in sympathetic nerve system in childhood

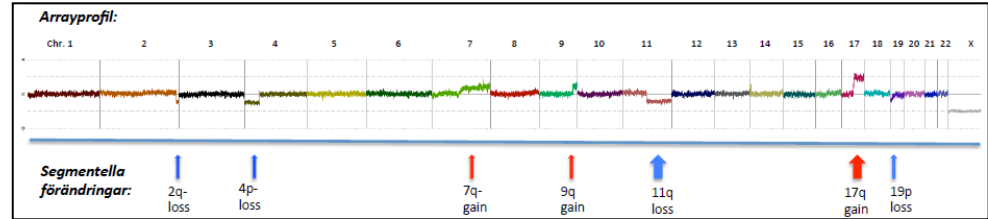


Biology directs treatment



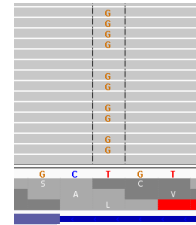
# Case Study

- Young male patient 16m
- Metastatic NB, 11q-
- DSB Inducers
- Treatment very toxic, anemic

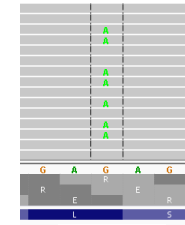


- WGS performed
  - Fanconi mutations
  - ALK-mut 1171T
- Change to treatment with 2nd generation ALK-inhib LDK378 (Ceritinib, Novartis)
- Child back in preschool

**Loss of function**  
**Maternal**      **Paternal**

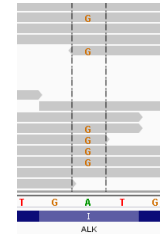


**FANCA**  
**Leu910Phe**



**FANCA**  
**Ss mut**

**Gain of function**  
**in tumor**



**ALK**  
**I1171T**



## Genomic Medicine Sweden

Genomic Medicine Sweden, GMS, paving the way for improved diagnostics, care and treatment.

[Read more](#)



### Sweden's ambitious national collaboration for genomic medicine

Our strategic mission is to foster collaboration between healthcare providers, academia and industry and realize precision medicine by effectively utilizing the rapidly advancing sequencing technologies to provide the highest standards of genomic testing in routine clinical care throughout Sweden.

**<http://genomicmedicine.se>**