SciLifeLab

AKADEMISKA SJUKHUSET



# Next Generation Sequencing in Clinical Diagnostics

4 April, 2014

Rudbeck Laboratory, Uppsala

Sweden

# Next-Generation Sequencing in Clinical Diagnostics

SciLifeLab and Uppsala University Hospital jointly organize the symposium Next-Generation Sequencing in Clinical Diagnostics 4 April, 2014. Internationally recognized clinicians and scientists will give state-of-the-art lectures and provide examples how next-generation sequencing can be applied in the diagnostic setting. The symposium will cover next-generation sequencing in diagnostics of cancer, hematological malignancies and inherited diseases. The diagnostic and bioinformatic challenges as well as ethical questions, such as reporting of incidental findings, will also be discussed. The newly established national SciLifeLab Clinical Sequencing Facility will also report on its current and planned activities.

The overall aim of this open symposium is to give important insights how next-generation sequencing can be transferred into the health-care system, but also to discuss future directions within this rapidly progressing and very promising field.

The symposium will be held at Rudbeck Laboratory in Uppsala.

Welcome!

For the Clinical Sequencing Facility,



Richard Rosenquist Brandell
Platform director



Johan Rung
Facility Manager

Application Deadline: March 20, 2014

For registration, updated information about the event, including practical information please visit: http://www.scilifelab.se/events/next-generation-sequencing-in-clinical-diagnostics/

The symposium is free of charge (on a first come basis).

# Program at a glance - April 4

| | | |
|---|---|---|
| 8.00-9.00 | Registration | |
| 9.00-9.10 | Welcome | |
| 9.10-9.30 | SciLifeLab & Uppsala University Hospital | Kerstin Lindblad-Toh<br>Sune Larsson, Uppsala |
| 9.35-9.55 | History of sequencing | Ulf Pettersson, Uppsala |
| 10.00-10.15 | Clinical Sequencing Facility | Richard Rosenquist, Uppsala |
| 10.15-10.45 | Coffee | |

## Session I: Solid tumors (chair: Johan Botling)

| | | |
|---|---|---|
| 10.45-11.00 | Facility activities in solid tumors | Johan Botling, Uppsala |
| 11.00-11.40 | Insights into the architecture of cancer genomes by massively parallel sequencing | Martin Peifer, Cologne |
| 11.45-12.05 | NGS in pheochromocytoma and other endocrine tumors | Joakim Crona, Uppsala |
| 12.10-12.30 | Targeted next-generation sequencing in solid tumors | Lotte Moens, Uppsala |
| 12.30-13.20 | Lunch | |

## Session II: Hematological malignancies (chair: Lucia Cavelier)

| | | |
|---|---|---|
| 13.20-13.35 | Facility activities in hematology | Lucia Cavelier, Uppsala |
| 13.35-14.15 | Amplicon next-generation deep-sequencing in a hematology diagnostics laboratory | Alexander Kohlmann, Munich |
| 14.20-14.50 | High-throughput studies of immune repertoires in health and disease | Kostas Stamatopoulos, Thessaloniki |
| 14.55-15.15 | RNA-sequencing in pediatric ALL | Jessica Nordlund, Uppsala |
| 15.15-15.40 | | Coffee |

## Session III: Inherited diseases (chair: Marie-Louise Bondeson)

| | | |
|---|---|---|
| 15.40-15.55 | Facility activities in inherited diseases | Marie-Louise Bondeson, Uppsala |
| 15.55-16.35 | Next-generation sequencing in genetic diagnostics: how clinical research labs are evolving | Marcel Nelen, Nijmegen |
| 16.40-17.00 | Exome sequencing for explorative diagnostics of Mendelian traits | Niklas Dahl, Uppsala |
| 17.05-17.35 | Incidental findings from NGS data: how to handle them? | Ulf Kristoffersson, Lund |
| 17.40-17.55 | Bioinformatic strategies in the new facility | Johan Rung, Uppsala |
| 17.55-18.00 | Concluding remarks | |
| 18.30 | Mingle at Navet BMC | |

---

**For more information please contact:**

Richard Rosenquist (richard.rosenquist@igp.uu.se)

Johan Rung (johan.rung@scilifelab.uu.se)