



Introducing diagnostic applications of '3Gb-testing' in human genetics

EU –FP7 project grant agreement no 602269

Regional 3Gb-TEST meeting on Whole Genome sequencing: (WGS) NGS as tool in diagnostics.

March 19, 2015
University Hospital Central Building
C-block, Lecture room 3
Lund , Sweden

DAY 1 Thursday March 19th

Chair: Bert Bakker, Leiden , Netherlands

11.00 Welcome to Lund, & Short Introduction
(Ulf Kristofferson, Lund, Sweden)

11.05 The sequencing methods, NGS terminology, types of variants detected.
(Bert Bakker, Leiden, The Netherlands)

11.45 Validation and clinical utility NGS The critical steps in the process;
EuroGentest guidelines for diagnostics NGS,
(Chris Mattocks, Salisbury, UK)

12.25 Lunch break

13.30 Tools to interpret sequence data, strength and weaknesses of databases.
(Tina Smets, Leuven, Belgium)

14.10 External Quality Assessment for NGS-based diagnostics
(Nicola Wolstenholme, Manchester, UK)

14.50 Functional validation of variants;
RNA, protein, expression, functional assays
(Bert Bakker, Leiden, the Netherlands)

15.30 Tea break

16.00 Future of health care, WGS personalized Medicine
(T.b.c., Wessex, UK)

16.45 ELSI aspects; ethical, legal and social issues
(*T.b.c.*)

17.30 Group discussions on a practical work flow
WGS, Exome or Targetted? When, what, how.
For each method used, discuss shortcomings or needs
Methods to decide on pathogenicity?
Reporting, When, what, how

~ 18.30 Closing of the symposium

Attendance is free, registration is needed:

Registration https://forms.lumc.nl/lumc2/meeting_Lund

Hotel: Lund has a number of hotels at different prices in the city centre, please consult www.bookings.com or www.hotels.com for more information and booking.

Venue: University Hospital Central Building, C-block, Lecture room 3
(Föreläsningssal 3)

Website: www.3Gb-TEST.eu