

Genomics for diagnostics of Rare Diseases TRAINMALTA meeting

Meeting Room St Barbara (5th)/Anna de Paepe (6th); Convent van Chievres 1st Floor, Begijnhof
Parking code 3542

Monday 5th November

Coffee: 08.30-09.00

Session 1: Introduction of genomics for diagnostics

9.00-10.30 Discussion leader Kathleen Freson

- Kathleen Freson: Welcome
- Chris Van Geet: Why do we need a genetic diagnosis in the clinic?
- Willem H Ouwehand: A decade of genomics
- Kate Downes: Gene panel testing for diagnostics of bleeding & thrombosis: what can you expect?
- Anniek Corveleyn: Overview of genomics in molecular diagnostics @UZLeuven
- *30min discussion*

10.30-11.00 Coffee

Session 2 : Analysis of Whole Genomes and Mendeliomes

11-12.30 Discussion leader Anniek Corveleyn

- Karyn Megy: Pertinent findings in Rare Diseases pilot phase of the 100,000 Genomes Project
- Ernest Turro: Application of new analysis methods to the 100,000 Genomes Project
- Erika Souche: Pipelines for germline mutation detection in clinical exomes and whole genomes
- Francesca Borg Carbott: Highlights from the Malta NGS Project
- *30min discussion*

Lunch 12.30-13:30 (Infirmierie, Faculty Club)

Session 3: Bioinformatic tools for analyzing genomes and non-coding elements

13.30-15.00 Discussion leader Willem H Ouwehand

- David Carbonez: Overview of the RNA-seq services provided by Genomics Core Leuven
- Eberer JP: VAR.mt
- Zeynep Kalender Atak: Gene regulation bioinformatics
- Sebastiaan Vanuytven: Variant detection in single cell RNAseq data
- *30min discussion*

15.00-15.30 Coffee

Session 4: Some special examples of rare diseases unraveled with genomics

15.30-16.30 Discussion leader: Rosienne Farrugia

- Evelien Van Hoof and Liesbeth Keldermans: Next-generation sequencing in molecular diagnostics: beyond single nucleotide variants.
- Manisha Padmakumar: Precision functional genetics for autism patients with abnormal platelets
- Veerle Labarque: KDSR a gene for skin or bone marrow disease
- *20min discussion*

16.45 Departure with bus for Beer tasting with Horses and a “Beer-and-Food-paring” menu

Tuesday 6th November

Coffee : 08.30-09.00

Session 1 (Room Anna De Paepe)

9.00-10.30 Using functional genetics to understand novel genes for rare diseases (part 1)

- Jessica Heremans: Visualizing granule biogenesis in megakaryocytes
- Matt Sims: Grey Platelet Syndrome: more than a simple bleeding disorder
- Analisse Cassar: Investigating the Effects of a Natural Extract on the Terminal Differentiation of Leukaemia
- Nick Gleadall: From genotype to actionable data for blood matching
- *30min discussion*

10.30-10.45 Coffee

Session 2

11-12.30 Using functional genetics to understand novel genes for rare diseases (part 2)

- Oriana Mazzitelli: Immunology & Tcells
- Erika Van Nieuwenhove: Unexpected relation between IKZF1 and autoimmunity
- Kathleen Freson: Unexpected relation between IKZF5 and thrombocytopenia
- Lore De Kock: DAP3 and congenital dyserythro/megakaryo-poiesis
- *30min discussion*

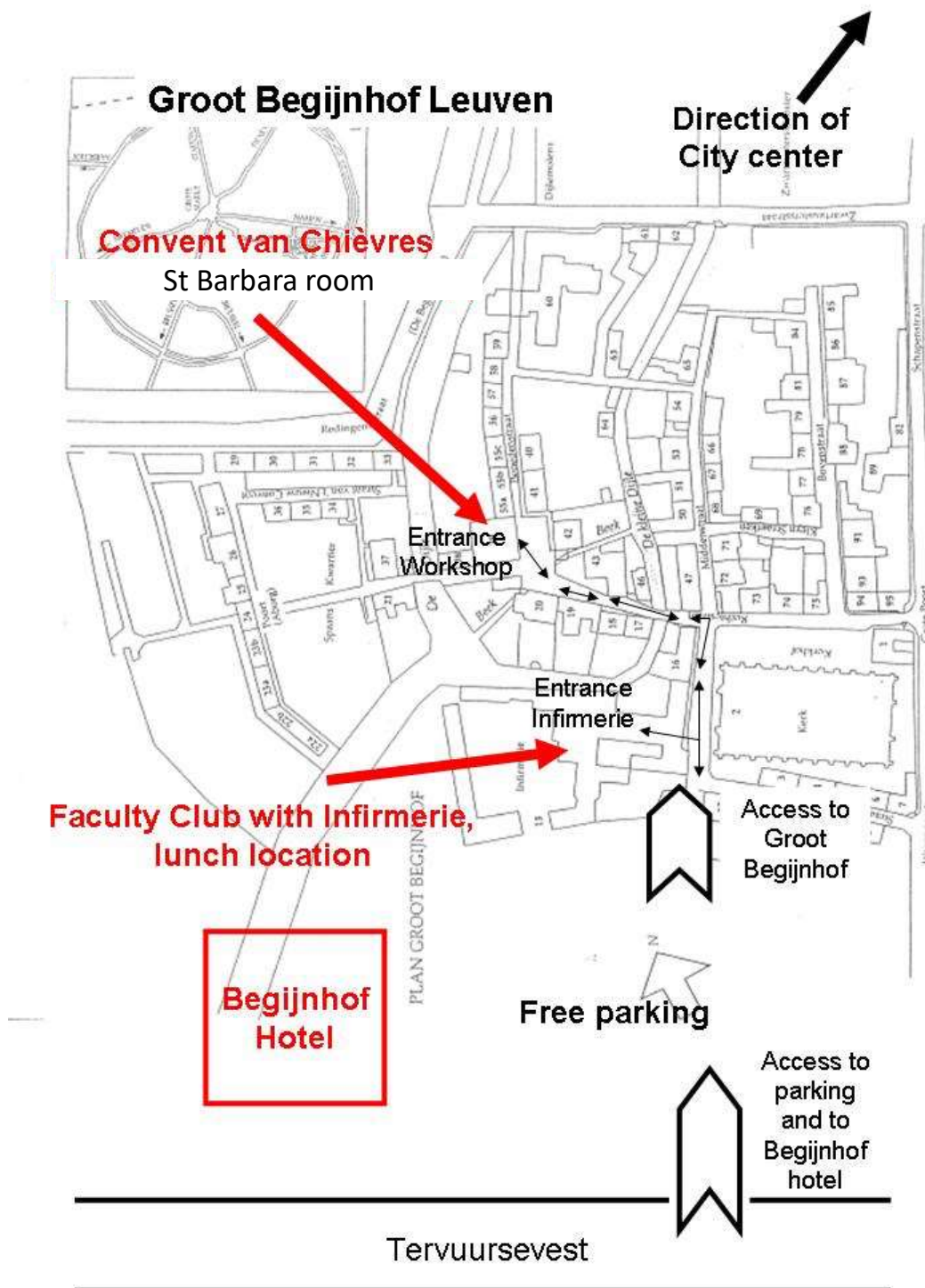
Lunch 12.30-14 (Infirmierie)

Session 3

13.30-15.00

- Closure of TRAINMALTA: Overview by Rosienne
- Discussions

15.00-16 Coffee



Information about the Begijnhof hotel for our visitors from abroad

<http://www.bchotel.be/en/>

Researchers attending from University of Malta

Chanelle Cilia
Jessica Debattista
Rosienne Farrugia
Francesca Borg Carbott
Analissee Cassar
Jean Paul Ebejer
Oriana Mazzitelli

Researchers attending from University of Cambridge, Department of Haematology

Kate Downes
Karyn Megy
Matt Sims
Ernest Turro
Willem H Ouwehand
Nick Gleadall

Researchers attending from University of Leuven, Center for Molecular and Vascular Biology

Kathleen Freson
Chantal Thys
Jessica Heremans
Manisha Padmakumar
Lore De Kock
Mara de Reys
Chris Van Geet
Kathelijne Peerlinck
Veerle Labarque

Researchers attending from University of Leuven, Centre of Human Genetics

Kris Van den Bogaert
Liesbeth Keldermans
Evelien Van Hoof
Karen Willekens
Amelie Van Eesbeeck
Marijke Bauters
Valerie Race
Erika Souche
Anniek Corveleyn
David Carbonez
Sebastiaan Vanuytven
Zeynep Kalender Atak

Researchers attending from University of Leuven, Laboratory of Genetics of Autoimmunity

Erika Van Nieuwenhove