

New and Developing Technologies for Genetics Diagnostics



National Genetics Reference Laboratory (Wessex)

5th and 6th July 2010
Salisbury District Hospital, UK

Monday 5th July 2010

12:00 Lunch

13:00 Welcome

Nick Cross (NGRL W)

00:35 Next generation sequencing of multiple genes in parallel in genetically heterogeneous disorders after enrichment

Hans Scheffer (Nijmegen)

13:35 **Session 1: Next Generation Sequencing in diagnostics** *Chair: Chris Mattocks*

00:20 TBA

Ann Curtis (Newcastle)

00:20 Developing targeted enrichment re-sequencing on the SOLiD platform

Howard Martin (Cambridge)

00:20 Software and IT

Graham Taylor (Leeds)

00:25 Target enrichment using selector probes

Mats Nilsson (Uppsala)

15:15 Tea

15:45 **Session 2: Chips and arrays**
Chair: John Crolla

00:35 Molecular karyotyping: From postnatal to pre-implantation genetic diagnosis

Joris Vermeesch (Leuven)

00:20 Rapid Diagnosis of Familial Hypercholesterolemia in British Patients

Lourdes Palacios (Progenika)

00:25 Bac on Beads

Susan Gross (New York)

00:20 Comparison of array platforms

Shuwen Huang (NGRL W)

17:40 Finish

Tuesday 6th July 2010

9:00 **Session 3: Acquired abnormalities**

Chair: Nick Cross

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|-------|---|----------------------------------|
| 00:35 | Interrogating whole cancer genomes with next generation sequencing | Peter Campbell (Sanger) |
| 00:25 | Targeted enrichment and resequencing of whole exomes for the identification of somatically acquired variants in breast cancer | Patrick Tarpey (Sanger) |
| 00:15 | Cold PCR | Phil Eastlake
(Transgenomics) |

10:30 **Coffee**

11:00 **Session 4: Epigenetics**

Chair: Deborah Mackay

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| 00:20 | AutoMeDIP-seq: a cost effective approach toward whole genome methylation analyses | Lee Butcher (London) |
| 00:20 | Methylation profiling using the Infinium platform in Follicular Lymphoma | Jude Fitzgibbon (London) |
| 00:20 | Methylation Analysis with EpiTYPER™ | Henning Gohlke (Sequenom) |

12:00 **Lunch**

13:00 **Session 5: New sequencing technologies – The future**

Chair: Graham Taylor

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| 00:30 | Towards sequencing single DNA molecules with a protein nanopore | Andrew Heron (Oxford) |
| 00:30 | Developments and applications of next-generation sequencing in an academic medical hospital | Johan den Dunnen (Leiden) |
| 00:25 | Single Molecule Real-Time Nucleic Acid Sequencing-by-Synthesis Using Quantum-dot (Qdot™) Nanocrystal-Labeled DNA Polymerases with FRET-Based Detection | Peter Vander Horn (Life Technologies) |

14.30 **Keynote**

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| 00:40 | Deciphering Developmental Disorders | Nigel Carter (Sanger) |
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Closing remarks and discussion

Graham Taylor (Leeds)

15.30 **Refreshments and Finish**

CPD credits applied for

For more details see www.ngri.org.uk/Wessex