

NIHR Rare Diseases Translational Research Collaboration

EURORDIS Meeting, Madrid
30th May 2015

Dr. Marijcke Veltman
Operational Director

RD-TRC

Co-Chairs: Dr. John Bradley (Cambridge BRC)
 Prof. Patrick Chinnery (Newcastle BRC)

Build a Translational Research Collaboration of NIHR Biomedical Research Centres, Biomedical Research Units and Clinical Research Facilities by:

- ▶ Harnessing and developing NHS research infrastructure to support patient-centred research into rare diseases in England.
- ▶ Facilitate collaboration between NIHR research infrastructure, clinical researchers, NHS organisations, research funders and life sciences.
- ▶ Increase in-depth phenotyping and link this to data on genomic abnormalities.
- ▶ Promote the nation's reputation as a world-class centre for translational research.
- ▶ Establishing a national database of patients with rare diseases.
- ▶ Investing in training and capacity building.
- ▶ NIHR Funding: £20M, 2012 – 2017

Themes

THEME

- Cancer
- Cardiovascular
- Dementia & Neurodegen. Disease
- Eye disease
- Eye disease (Co-Lead)
- Gastrointestinal
- Immunological disorders
- Metabolic & Endocrine
- Musculoskeletal
- Neuromuscular disorders
- Respiratory disease
- Skin
- Renal disease
- Non-malignant haematology
- Paediatrics (cross cutting)

LEAD

TBC

Hugh Watkins
Nick Wood
Graeme Black
Andrew Webster
David Jones
Adrian Thrasher
Steve O'Rahilly
Paul Wordsworth
Mike Hanna
Eric Alton
John McGrath
Fiona Karet
Irene Roberts
Tim Barrett

Lead NIHR CENTRE

Royal Marsden BRC
Oxford BRC
UCL BRC
(Manchester CRF)
Moorfields BRC
Newcastle BRC
GOSH BRC
Cambridge BRC
Oxford BRU
UCL BRC
Brompton BRU
Guy's/Kings BRC
Cambridge BRC
Oxford BRC
Birmingham CRF

Deputy Theme Leads

THEME

- Cancer
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DEPUTY LEAD

Naureen Starling

Houman Ashrafian

Not nominated

N/A – Eye (Co-Lead)

Gideon Hirschfield

Lucy Wedderburn

Robert Semple

Kassim Javaid

Not nominated

Robert Stockley

Jemima Mellerio

Detlef Bockenhauer

Ami Nathwani, Willem Ouwehand, Anna Schuh

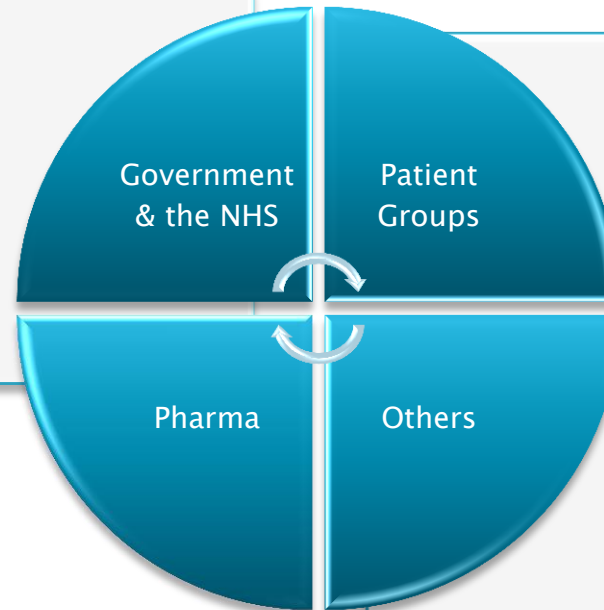
Not nominated

Strategic Oversight Group

▶ RD-TRC Co-Chair	Dr. John Bradley	Cambridge BRC
▶ RD-TRC Co-Chair	Prof. Patrick Chinnery	Newcastle BRC
▶ Operational Director	Dr. Marijcke Veltman	Cambridge BRC
▶ Informatics Director	Dr. Roger James	Cambridge BRC
▶ NOCRI Representative	Dr. Oliver Rausch	NOCRI
▶ NIHR Training Lead	Prof. David Jones	Newcastle BRC
▶ NIHR BioResource RD	Prof. Willem Ouwehand	Cambridge BRC
▶ Rare Disease UK	Alastair Kent	Genetic Alliance UK
▶ Assoc. of Med. Res. Charities	Dr. Liz Philpots	AMRC
▶ Partner Organisations	Prof. Dame Janet Thornton	EMBL-EBI
▶ Industry	Dr. Kevin Lee	Pfizer
▶ International Representative	Prof. Kate Bushby	Newcastle BRC
▶ Lay Representatives (n=2)	TBA	

Collaborators

- Department of Health
- National Institute for Health Research (NIHR)
- National Office for Clinical Research Infrastructure (NOCRI)
- NIHR Biomedical Research Centres
- NIHR Clinical Research Facilities
- NIHR Biomedical Research Units
- NIHR BioResource – Rare Diseases
- NIHR Clinical Research Network
- Genomics England Ltd.
- *NHS England & Public Health England*



- Genetic Alliance
- Rare Diseases UK
- INVOLVE
- Cohort specific patient groups via studies
- NHS Trust and NIHR Infrastructure

- Other research funders
- European Bioinformatics Institute
- Orphanet
- The International Rare Diseases Research Consortium (IRDiRC)

In Depth Phenotyping Projects

Round one (2013–2015 funding; **closed**).

- 48 Applications across 12 topic specific and the cross cutting theme
- 22 Projects funded
- 16 Projects awarded additional funding for biomarker profiling

Round two (2015–2017 funding; **closed**).

- 111 Expressions of Interest; 56 invited to submit full applications
- 14 Projects funded (5 continuations from Round 1)

Industry Call (2015–2017 **joint-funding**; **open**). To date

- 17 Expressions of Interest received
 - 9 Full applications invited
 - 1 Study co-funded
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National Database

- ▶ OpenClinica as the ‘hub’ system for the national federated database
 - Aligned to other work from cancer, the BioResource – Rare Diseases and the work of Genomics England
- ▶ The IT infrastructure and facilities for a secure OpenClinica facility integrated with the main NHS [N3] network and systems was designed and commissioned
- ▶ Training support and documentation made available to the RD–TRC projects
 - Training delivered to over 50 RD–TRC researchers in 6 England-wide locations

Fellowships

Fellowships

- ▶ 40 applications across 14 topic specific and the cross cutting themes
- ▶ 12 fellowships funded; 7 PhD, 5 post doctoral

Masters

- ▶ MPhil in Rare Diseases (Cambridge)
 - Taught component, including scientific methodology
 - Research project
 - First students enrolled October 2014

Patient & Public Governance

- ▶ Each Study has Patient & Public Involvement
- ▶ RD-TRC
 - Patient & Public Oversight Group
 - Each Theme represented
 - RD-TRC Executive
 - Twice per year
 - Strategic Oversight Group
 - Theme Leads Meetings

Rare Disease Day 2015

Partner-sites

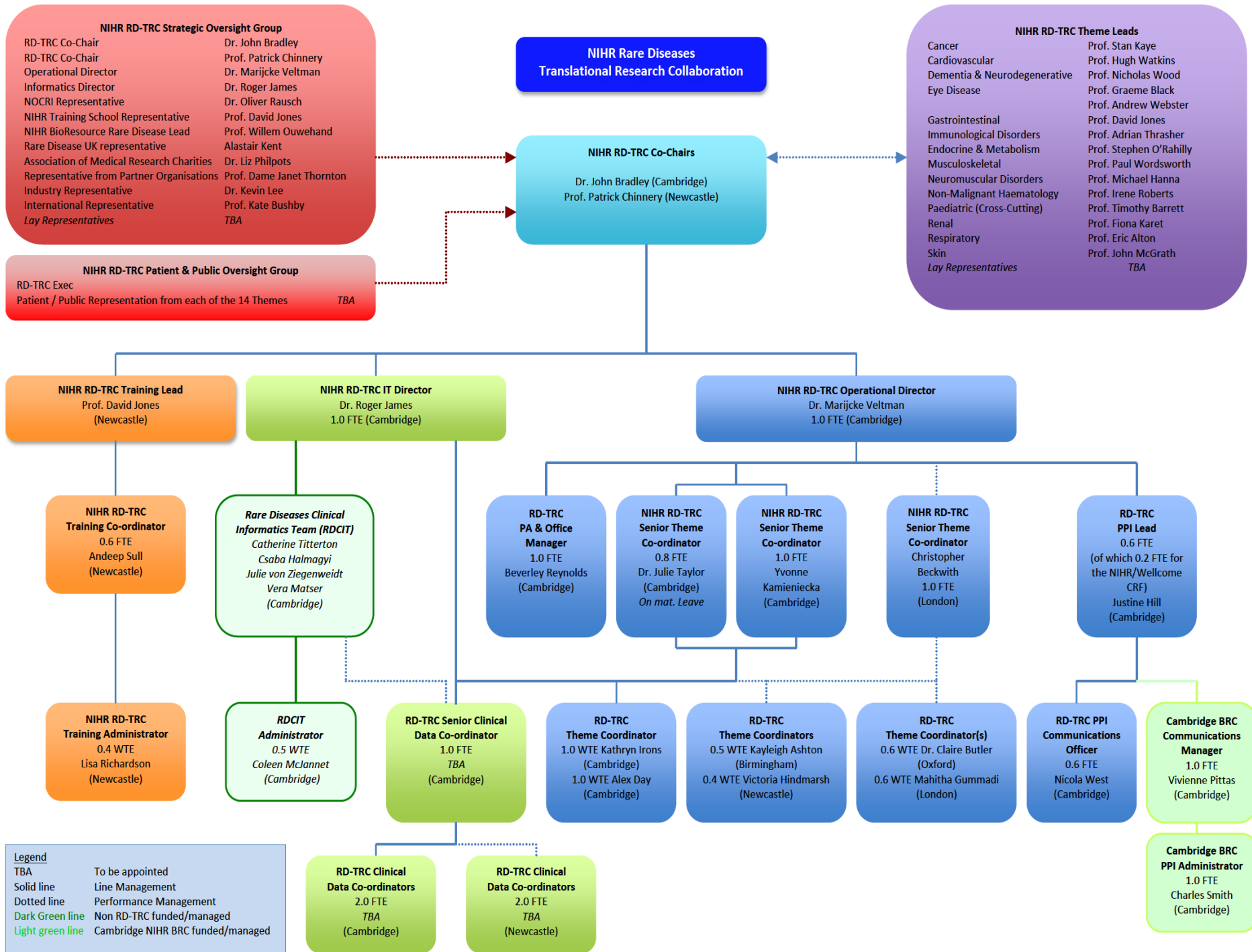
- Birmingham CRF
- Birmingham Children's Hospital
- Cambridge BRC
- Great Ormond Street BRC
- Guy's & St. Thomas' BRC
- Moorfields Hospital BRC
- Newcastle BRC
- Royal Brompton BRU
- Southampton Respiratory BRU
- UCL BRC



Rare Diseases Translational Research Collaboration

Rare Disease Day 2015





Contact Details

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Cambridge, UK

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