



# Rare Disease Day

Monday 28 February 2011

**Programme of events**



*Rare Disease Day*

Royal Holloway  
University of London



# Rare Disease Day



Rare Disease Day

In Europe a rare disease is defined as one with an incidence of less than 1 in 2000 people. While each of them is rare, there are more than 7,000 rare diseases, which together affect 24 million or six percent of people in Europe. 80 percent of rare diseases have identified genetic origins whilst others are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative. 50 percent of rare diseases affect children. Collectively, rare diseases are a serious public health concern, taking up 20 percent of all health care costs, but they are often little known to health professionals and the public alike.

Rare Disease Day on 28 February has been established as an international event to raise awareness of these diseases, with a particular campaign focus on 'Rare but Equal - rare diseases and health inequalities.'

Rare Disease Day is open to everyone. Patient groups, health professionals, researchers and their collaborative partners – coordinated by national alliances – are working together to organise a multitude of public events to draw attention to rare diseases and the millions of people who are affected by them. The campaign aims to promote the inclusion of rare diseases in the Third EU Public Health Programme, which will decide the policy and funding priorities in the years ahead.



## Rare disease research at Royal Holloway

Royal Holloway is a leading institution in the development of novel therapies for rare diseases, including Spinal muscular atrophy, Duchenne muscular dystrophy and Severe Combined Immunodeficiency (referred to as 'child in a bubble' disease). Our scientists are at the forefront in developing innovative techniques in gene based therapy. They have developed safer methods to deliver therapeutic genes and are at the forefront of 'Genome Surgery', the technology that allows actual repair of genetic mutations in affected cells.

Visit our Rare Disease Day website at [www.rhul.ac.uk/rarediseaseday/](http://www.rhul.ac.uk/rarediseaseday/)

## Organisers

Dr Rafael Yáñez and Professor George Dickson, Centre for Biomedical Sciences, School of Biological Sciences, Royal Holloway, University of London

## Sponsors



## Exhibitors



## Afternoon events (3-6 pm)

### 3 pm Registration, Picture Gallery (refreshments available 3-6 pm)

- Introduction, Dr Rafael Yáñez, Organiser
- Welcome, Professor Philip Beesley, Dean of Science
- Rare Disease and Rare Disease Day, Julian Walker, Genetic Alliance UK

### 4 pm Picture Gallery Exhibition and Satellite venues:

- **Exhibitor stands**  
Patient Societies, CREST awards, College Support Services... see our exhibitor logos on page 2
- **Posters from our young scientists**  
Ask them everything you ever wanted to know about their work and life in the lab, and make sure they speak your language!
- **DNA Origami**  
Visit the British Society for Gene Therapy stand and Lynda, their Young Scientist Board member.
- **Lab Visits**  
(Require booking at our registration desk on arrival)  
Scientists spend half their lives applying for grants... come and see why.
- **Career Networking Event** (4-5 pm, Poster area)  
Wondering about the future? Meet young and more experienced scientists and ask them what a career in science is like.
- **Debate: Living with a Rare Disease**  
(5-5:45 pm, Large Board Room, limited space available; screen viewing from larger venue tba), led by Joseph Irwin, father of a baby with Spinal muscular atrophy. Listen to people with rare diseases, carers and relatives sharing their experience, or join them on the panel if you are one of them.
- **PC Lab Activity: Researching Rare Diseases Online**  
(Requires booking at our registration desk on arrival)  
Let our Final Year students guide you to rare disease information on the internet, and complete their questionnaire
- **Screening of BBC Horizon: 'A Decade of the Human Genome'** (Small Board Room)  
Watch some of the UK's best gene therapists, plus mice that cannot help having a drink... or two!

Louise and David have Fanconi Anaemia. Both will require bone marrow transplants, and for David there is currently no match. For children like David we look to gene therapy to provide life saving treatments in the not-too-distant future.

### 4.10 pm Windsor 0-02.

Screening of 'Extraordinary Measures', with Brendan Fraser, Harrison Ford and Keri Russell, about the quest for a cure for Pompe Disease.

### Early evening events (5.45-8.15 pm)

#### 5.45 pm Windsor Building Foyer. Light refreshments 6.15 pm Windsor Auditorium. Main Session

- Introduction, Dr Rafael Yáñez and Professor George Dickson, Organisers
- Welcome by Professor Geoff Ward, Vice-Principal, Royal Holloway
- Gene Hunting in the Amish Community, Mike Patton, Professor of Medical Genetics & Head of Medical Genetics Section, St George's, University of London
- What is Gene Therapy? Dr Rafael Yáñez, Senior Lecturer in Genetics, School of Biological Sciences, Royal Holloway
- The Personal Perspective, Mr Srinivas Madipalli, a young person affected by Spinal muscular atrophy
- Clinical Trials for Cystic Fibrosis, Eric Alton, Professor of Gene Therapy and Respiratory Medicine & Chair in Gene Therapy, Imperial College
- Treating Duchenne Muscular Dystrophy: are we there yet?, Professor George Dickson, Director, Institute of Biomedical & Life Sciences, Royal Holloway
- Round Table with all participants and questions from the audience

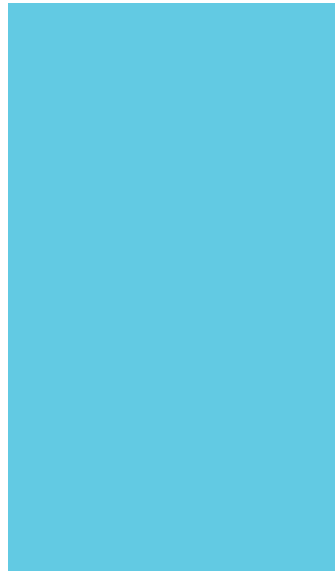
### 8.15 pm Official close, Professor Philip Beesley, Dean of Science

### Late evening (8.30 pm till late)

#### 8.30 pm Café Scientifique at Tommys Bar.

An opportunity for our student volunteers to talk about science with the experts. Other students and people welcome, but name must be supplied in advance to [RDD@rhul.ac.uk](mailto:RDD@rhul.ac.uk)





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