Rare Disease Day
Bridging health and social care
Tuesday 26 February 2019
Introduction

This special day aims to draw attention to rare diseases and the millions of people who are affected by them, highlighting the need for more research and funding to help sufferers and their families. The theme for 2019 is ‘Bridging health and social care’, emphasising the importance of communication between different services to meet patients’ best interests.

In Europe, a rare disease is defined as one with an incidence of less than 1 in 2,000 people. While each of them is rare, there are 6–8,000 rare diseases, which together affect 3.5 million people in the UK, taking 20% of all health care costs. Rare diseases are a serious public health concern and an international priority, but unfortunately often very little is known about them by health professionals and the public alike.

People who are affected by a rare disease will mostly receive symptomatic and palliative care because there are hardly any curative treatments. Often, relatives will have to stop working and become full-time carers.

Rare Disease Day encourages us to continue finding ways to work together. In 2019 it conveys the message that it is critical to bridge the gaps between medical, social and support services to improve the lives of people living with a rare disease and their families.

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, Ataxia telangiectasia, Duchenne muscular dystrophy, Motor neuron disease and Severe Combined Immunodeficiencies (referred to as ‘child in a bubble’ disease). Our scientists are at the forefront in developing innovative techniques in gene based therapy.

Today’s Organisers

Prof Rafael J. Yáñez-Muñoz, Professor of Advanced Therapy and Director of the Centre of Gene and Cell Therapy, is the Editor-in-Chief of Gene Therapy, the Treasurer of The British Society for Gene and Cell Therapy and Chair of the Genetic Alliance UK. Professor George Dickson (co-organiser) is Chair of Molecular Cell Biology in the School of Biological Sciences and a founder member of The British Society for Gene and Cell Therapy.

School of Biological Sciences

The School of Biological Sciences at Royal Holloway has earned an international reputation for the world-class quality of our research which is at the forefront of scientific discovery. We offer an exciting range of undergraduate degree programmes and research opportunities ranging from gene therapy, tropical diseases, GM technology, vaccine technology and neuroscience, to animal behaviour, conservation, ecology and biodiversity.

The School places a strong emphasis on practical teaching, with state-of-the-art equipment and an exceptionally supportive environment for all students.

It achieved 88% overall satisfaction from its students in the National Student Survey in 2018.
# Programme

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<td>09:00 - 09:30</td>
<td>Arrival and registration</td>
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<td>09:30 - 10:30</td>
<td>Lectures&lt;br&gt;Professor Paul Layzell&lt;br&gt;Professor Rafael Yáñez</td>
<td>Lectures&lt;br&gt;Professor George Dickson&lt;br&gt;Dr Jayne Spink</td>
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<td>Boilerhouse Auditorium</td>
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<td>10:30 - 11:30</td>
<td>Lab activity&lt;br&gt;Bourne 203</td>
<td>Exhibition zone&lt;br&gt;Windsor foyers</td>
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<tr>
<td>11:30 - 12:30</td>
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<td>Lab activity&lt;br&gt;Bourne 203</td>
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<td>12:30 - 13:15</td>
<td>Lunch</td>
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<td>13:15 - 14:00</td>
<td>Disease Detective&lt;br&gt;Bourne 203</td>
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<tr>
<td>14:00 - 14:45</td>
<td>Speed dating&lt;br&gt;Bourne 203</td>
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## Our Speakers

**Welcome**  
Professor Paul Layzell, Principal, Royal Holloway

**Introduction to Rare Disease Day**  
Professor Rafael Yáñez, Advanced Gene and Cell Therapy Lab, School of Biological Sciences

**Illuminating the Rare Reality**  
Dr Jayne Spink, Chief Executive, Genetic Alliance UK; Chair, Rare Disease UK

**Delivering gene therapy for muscular dystrophy**  
Professor George Dickson, Professor of Molecular Cell Biology
Spotlight on Science

Lesson starters

Why should we care about rare diseases?
Prof Rafael Yáñez

Learning objectives:
By the end of this activity day you should be able to:
• Define what a rare disease is, and name some examples of genetic diseases
• Describe how genomic mutations can cause rare diseases
• Explain why rare diseases are important
• Understand why gene and cell therapies are considered so promising
• Consider some ethical issues of relevance to rare diseases

Points to discuss:
• Are all rare diseases genetic, and are all genetic diseases rare?
• How do genetic diseases arise and why are there so many?
• What organs and tissues can be affected by rare diseases?
• Why do rare diseases have disproportionate health and social costs?
• How can rare diseases be treated? How can engineered viruses be used as medicines?
• Why don’t we screen newborns for all genetic diseases?
• Why are the marketed treatments so expensive?

For other Spotlight on Science A level topics, visit royalholloway.ac.uk/spotlightonscience

Our 2019 exhibitors
We warmly welcome the following partners to our Rare Disease Day
• Ashford and St. Peter’s Hospitals
• Action Duchenne
• Action for A-T
• Genetic Alliance UK
• Muscular Dystrophy UK
• Rare Disease UK
• Shine
• Spinal Muscular Atrophy UK
• SWAN UK
• The British Society for Gene and Cell Therapy
• The Ichthyosis Support Group
• Treat SMA
• Tuberous Sclerosis Association

CGCT
Centre of Gene and Cell Therapy
royalholloway.ac.uk/cgct

School of Biological Sciences:
royalholloway.ac.uk/biologicalsciences

@RHULBioSci

Advanced Gene and Cell Therapy Laboratory (AGCTlab):
agctlab.org

Rare Disease Day:
royalholloway.ac.uk/rarediseaseday