Rare Disease Day

Celebrating Royal Holloway’s 12th annual Rare Disease Day event

Monday 28 February 2022
10am-3.30pm
Introduction

Thank you for joining us for our 12th annual celebration of Rare Disease Day, our second virtual event.

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 people affected and their families. The message for 2022 is ‘Share your colours’, raising awareness and generating change for the 300 million people worldwide living with a rare disease, their families and carers.

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 more than 9,600 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 very few 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 2022 the Day is focused on the importance of improving diagnosis and treatment for all of those who are living with a rare disease.

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 Neurofibromatosis. Our scientists are at the forefront in developing innovative techniques in gene-based therapy.

Today’s Organiser
Prof Rafael J. Yáñez-Muñoz, Professor of Advanced Therapy and Director of the Centre of Gene and Cell Therapy, is President of The British Society for Gene and Cell Therapy.

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

In the National Student Survey 2021 we achieved 80% overall satisfaction from our students, and we rank 3rd in the UK for Graduate Career after 15 months (The Guardian University Guide, 2022).
## Programme

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<th>Time</th>
<th>Activities</th>
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<tr>
<td>10-11am</td>
<td><strong>Lectures</strong></td>
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<tr>
<td></td>
<td><strong>Introduction to Rare Disease Day</strong>, Prof Rafael J. Yáñez-Muñoz, Prof of Advanced Therapy, Royal Holloway, University of London</td>
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<td><strong>The British Society for Gene and Cell Therapy</strong>, Robyn Bell, British Society for Gene and Cell Therapy</td>
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<td><strong>Gene Therapy for Neuromuscular Diseases</strong>, Prof Linda Popplewell, Prof in Molecular Therapies, Royal Holloway, University of London</td>
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<td>11.15am-12pm</td>
<td><strong>Exhibition</strong></td>
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<td><strong>From operating theatre to country houses</strong> – a creative approach to rare disease awareness, Ceridwen Hughes, Same but Different</td>
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<td><strong>The Student Voice Prize</strong>: Grow with the rare community, Blayne Baker, Beacon</td>
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<td>12.15-1pm</td>
<td><strong>Speed dating</strong></td>
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<td><strong>Meet Royal Holloway undergraduate students and representatives from several stakeholder charities</strong> (British Society for Gene and Cell Therapy, Genetic Alliance UK, Spinal muscular atrophy UK, Cystic Fibrosis Trust, Unique and RareQoL) to talk about uni life and rare disease</td>
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<td>1pm-2pm</td>
<td><strong>Lunch break</strong></td>
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<td>2-3pm</td>
<td><strong>‘Lab’ activity</strong></td>
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<td><strong>X-linked recessive inheritance</strong>, Dr Katie Lloyd-Jones, Royal Holloway, University of London</td>
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<td>3-3.30pm</td>
<td><strong>Evaluation and close</strong></td>
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### Our Speakers
- **Prof Rafael J. Yáñez Muñoz**  
  Professor of Advanced Therapy at Royal Holloway University of London
- **Robyn Bell**  
  from the British Society for Gene and Cell Therapy
- **Prof Linda Popplewell**  
  Professor in Molecular Therapies at Royal Holloway University of London

### Our Virtual Format during the COVID-19 pandemic
- The MS Team links to join the event will have been sent to the lead teachers.
- If your school is participating in the optional hands-on activity (2-3pm), your teachers will have prepared the required resources.
- Additional resources are available at [royalholloway.ac.uk/rdd](http://royalholloway.ac.uk/rdd)

All undergraduate degrees offered in the Department of Biological Sciences are accredited by the Royal Society of 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/studying-here/schools-and-colleges/

Our 2022 exhibitors
We warmly welcome the following partners to our Rare Disease Day
• Beacon
• British Society for Gene and Cell Therapy
• Cystic Fibrosis Trust
• Genetic Alliance UK
• RareQoL
• Same but Different
• Sickle Cell Society
• Spinal Muscular Atrophy UK
• Unique

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

@RHULBioSci
Department of Biological Sciences:
royalholloway.ac.uk/biologicalsciences

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

Rare Disease Day at Royal Holloway:
royalholloway.ac.uk/rarediseaseday