



# Rare Disease Day

Celebrating Royal Holloway's 11th  
annual Rare Disease Day event

Friday 26 February 2021



ROYAL  
HOLLOWAY  
UNIVERSITY  
OF LONDON

# Introduction

**Thank you for joining us for our 11th annual celebration of Rare Disease Day, our first 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 2021 is 'Rare is many, Rare is strong, Rare is proud', emphasising the number of people affected by rare diseases, their community strength and pride.

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 2021 the Day aims to improve knowledge of rare diseases amongst the general public while encouraging researchers and decision makers to address the needs of those living with rare diseases.

## **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 Organisers**

Prof Rafael J. Yáñez-Muñoz, Professor of Advanced Therapy and Director of the Centre of Gene and Cell Therapy, is President-Elect of The British Society for Gene and Cell Therapy. Dr Linda Popplewell is a Reader in Molecular Therapies in the Department of Biological Sciences and she is also on the board 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 2020 it achieved 100% overall satisfaction for our Biology and Zoology degrees, and 95% for our Biomedical and Biochemistry degrees.



# Programme

Time	Activities
10:00 - 11:00	<b>Lectures</b> <b>Introduction to Rare Disease Day</b> , Prof Rafael J. Yáñez-Muñoz <b>Genomics, health innovation and rare disease</b> , Baroness Nicola Blackwood <b>How to train your virus</b> , Prof Alan L. Parker
11:00 - 12:00	<b>Lab activity</b> <b>X-linked recessive inheritance</b> , Dr Katie Lloyd-Jones <i>Use the lab activity pack from the resource box</i>
12:00 - 13:00	<b>Exhibition</b> <b>RAREolutions, a Rare Disease Competition</b> , Jo Balfour, Cambridge Rare Disease Network <b>DNA bracelets and tattoos</b> , Robyn Bell and Lisa McArthur, British Society for Gene and Cell Therapy. <i>Use the activity pack from the resource box</i> <b>From operating theatre to country houses – a creative approach to rare disease awareness</b> , Ceridwen Hughes, Same but Different
13:00 - 13:45	<b>Lunch</b>
13:45 - 14:25	<b>Nurse-led innovation</b> <b>An augmented reality game for young people with lifelong conditions</b> , Roald Dahl's Marvellous Children's Charity, StoryFutures and Play Well For Life
14:25 - 15:15	<b>Speed dating</b> Meet Royal Holloway undergraduate students and representatives from several stakeholder charities (Findacure, Haemochromatosis UK, Shine, Sickle Cell Society and TreatSMA) to talk about uni life and rare disease
15:15 - 15:30	<b>Evaluation and close</b>

## Our Speakers

### Prof Rafael J. Yáñez Muñoz

Professor of Advanced Therapy at Royal Holloway University of London

### Baroness Nicola Blackwood

of North Oxford,  
Chair of Genomics England,  
Member of the House of Lords Science & Technology Select Committee

### Prof Alan L. Parker

Professor of Translational Virotherapies at Cardiff University

## Our Virtual Format during the COVID-19 pandemic

We have prepared and shipped boxes of resources for each participating student, to combine hands-on activities at home, information, virtual presentations and meetings. Additional resources are available at [royalholloway.ac.uk/rdd](http://royalholloway.ac.uk/rdd). We acknowledge our partners British Society for Gene and Cell Therapy, Gain, Haemochromatosis UK, Roald Dahl's Marvellous Children's Charity, Shine, Spinal Muscular Atrophy UK and StoryFutures for providing materials.



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/](http://royalholloway.ac.uk/studying-here/schools-and-colleges/)

## Our 2021 exhibitors

We warmly welcome the following partners  
to our Rare Disease Day

- British Society for Gene and Cell Therapy
- Cambridge Rare Disease Network
- Findacure
- Gain
- Haemochromatosis UK
- Play Well For Life
- Roald Dahl's Marvellous Children's Charity
- Same but Different
- Shine
- Sickle Cell Society
- Spinal Muscular Atrophy UK
- StoryFutures
- TreatSMA



Centre of Gene  
and Cell Therapy  
[royalholloway.ac.uk/cgct](http://royalholloway.ac.uk/cgct)

## CGCT



@RHULBioSci  
Department of Biological Sciences:  
[royalholloway.ac.uk/biologicalsciences](http://royalholloway.ac.uk/biologicalsciences)



Advanced Gene and Cell  
Therapy Laboratory (AGCTlab):  
[agctlab.org](http://agctlab.org)



Rare Disease Day at Royal Holloway:  
[royalholloway.ac.uk/rarediseaseday](http://royalholloway.ac.uk/rarediseaseday)



British Society for Gene and Cell Therapy  
[www.bsgct.org](http://www.bsgct.org)