



Rare Disease Day

Celebrating Royal Holloway's 10th annual Rare Disease Day event

Wednesday 26 February 2020



RARE DISEASE DAY.ORG



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Introduction

Thank you for joining us for our 10th annual celebration of Rare Disease Day.

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 2020 is 'we are the 300 million', emphasising the number of people affected by rare diseases worldwide.

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 2020 it conveys the message that we need to strive towards more equitable access to diagnosis, treatment, care and social opportunity.

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 the Treasurer and President-Elect of The British Society for Gene and Cell Therapy, and Chair of the Genetic Alliance UK. Dr Linda Popplewell is a lecturer in Biomedical Sciences 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.

It achieved 94% overall satisfaction from its students in the National Student Survey in 2019.



Programme

Time	Group A	Group B
09:30 - 10:00	Arrival and registration	
10:00 - 11:00	Lectures Professor Katie Normington Dr Linda Popplewell Professor Rafael Yáñez Mr Gennadiy Ilyashenko Queen's Lecture Theatre	
11:00 - 12:00	Lab activity Bourne 203	Exhibition zone Windsor foyers
12:00 - 13:00	Exhibition zone Windsor foyers	Lab activity Bourne 203
13:00 - 13:45	Lunch	
13:45 - 14:30	Disease Detective Bourne 203	Speed dating Bourne 203
14:30 - 15:15	Speed dating Bourne 203	Disease Detective Bourne 203
15:15 - 15:30	Evaluation and departure	

Our Speakers

Welcome

Professor Katie Normington, Deputy Principal,
Royal Holloway

Introduction to Rare Disease Day

Professor Rafael Yáñez, Advanced Gene and Cell
Therapy Lab, Department of Biological Sciences

Caring for a child with a rare disease

Mr Gennadiy Ilyashenko

Making sense with antisense

Dr Linda Popplewell, Lecturer in Biomedical
Sciences, Department of Biological Sciences



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 2020 exhibitors

We warmly welcome the following partners to our Rare Disease Day

- Action Duchenne
- Ashford and St Peter's Hospital
- AT Society
- Genetic Alliance UK
- Genetic Disorders UK
- Ichthyosis Support Group
- Rare Disease UK
- Royal Holloway Computer Science
- Royal Holloway History
- Spinal Muscular Atrophy UK
- SWAN UK
- The British Society for Gene and Cell Therapy
- The Haemochromatosis Society
- The Sickle Cell Society
- Treat SMA



CGCT

Centre of Gene
and Cell Therapy

royalholloway.ac.uk/cgct

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



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



Rare Disease Day:
royalholloway.ac.uk/rarediseaseday



treatsma.uk