

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



### Our 2025 exhibitors

We warmly welcome the following partners to our Rare Disease Day

- Ataxia UK
- Beacon
- British Society for Gene and Cell Therapy
- Ehlers-Danlos Society
- Genetic Alliance UK
- Haemochromatosis UK
- Ichthyosis Support Group
- Orchard Therapeutics
- Royal Holloway AGCTlab
- Royal Holloway Augustin Lab
- Royal Holloway History Department
- Royal Holloway Wilkinson Lab
- The Sickle Cell Society
- UK Thalassaemia Society



## CGCT

Centre of Gene  
and Cell Therapy

[royalholloway.ac.uk/cgct](https://royalholloway.ac.uk/cgct)

Department of Biological Sciences:  
[royalholloway.ac.uk/biologicalsciences](https://royalholloway.ac.uk/biologicalsciences)



@RHULBioSci



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



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

# RARE DISEASE DAY



## Celebrating Royal Holloway's 15th annual Rare Disease Day event

### Friday 28 February 2025

# INTRODUCTION

## Thank you for joining us for our 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 theme of the day is 'More than you can imagine', in reference to the power of resilience and connection of those affected. 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, and 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 and Motor neuron 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 President of The British Society for Gene and Cell Therapy. Dr Alberto Malerba, Lecturer in Gene Therapy at Royal Holloway University of London, heads the Gene Medicine Laboratory for Rare Diseases.

### 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, nutritionally-enhanced or climate-resilient crops, 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. We also help our students to get their careers off to the best possible start, and rank in the top 20 in the UK for graduate prospects (The Guardian University Guide, 2025).

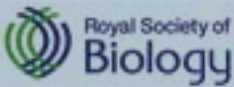


# PROGRAMME

Time	Group A	Group B
9.30-10am	Arrival and registration	
10-11am	<b>Lectures</b> Prof Mark Fellowes Prof Rafael J. Yáñez-Muñoz Dr Alberto Malerba Ms Rajeshri Badiani Location: Shilling Auditorium	
11am-12pm	<b>Lab activity</b> Location: Bourne 203	<b>Exhibition zone</b> Location: Windsor foyers
12-1pm	<b>Exhibition zone</b> Location: Windsor foyers	<b>Lab activity</b> Location: Bourne 203
1-1.45pm	Lunch (not provided)	
1.45-2.30pm	<b>Disease Detective</b> Location: Bourne 203	<b>Speed dating</b> Location: Bourne 202
2.30-3.15pm	<b>Speed dating</b> Location: Bourne 202	<b>Disease Detective</b> Location: Bourne 203
3.15-3.30pm	Evaluation and departure	

### Our Speakers

- **Welcome**, Prof Mark Fellowes (Pro-Vice-Chancellor of Academic Strategy, Planning and Resources)
- **Introduction to Rare Disease Day**, Prof Rafael J. Yáñez-Muñoz (Professor of Advanced Therapy at Royal Holloway University of London)
- **Gene therapy for Duchenne muscular dystrophy**, Dr Alberto Malerba (Lecturer in Gene Therapy at Royal Holloway University of London)
- **From a voluntary group to a formalized charity: the progress of FSHD UK**, Ms Rajeshri Badiani (Founder of FSHD UK)



All undergraduate degrees offered in the Department of Biological Sciences are accredited by the Royal Society of Biology.