

PRACTICAL INFORMATION

Venue

Sir Bob Burgess Building University of Leicester
11 Putney Road Leicester LE2 7TF
Located within the Freeman's Common redevelopment,
near Welford Road.

By Car & Parking

Freemen's Common Multi-Storey Car Park
90 Aylestone Road, Leicester LE2 7LB
Parking charge: Approximately £12 per day.
Contactless payment only.

By Train

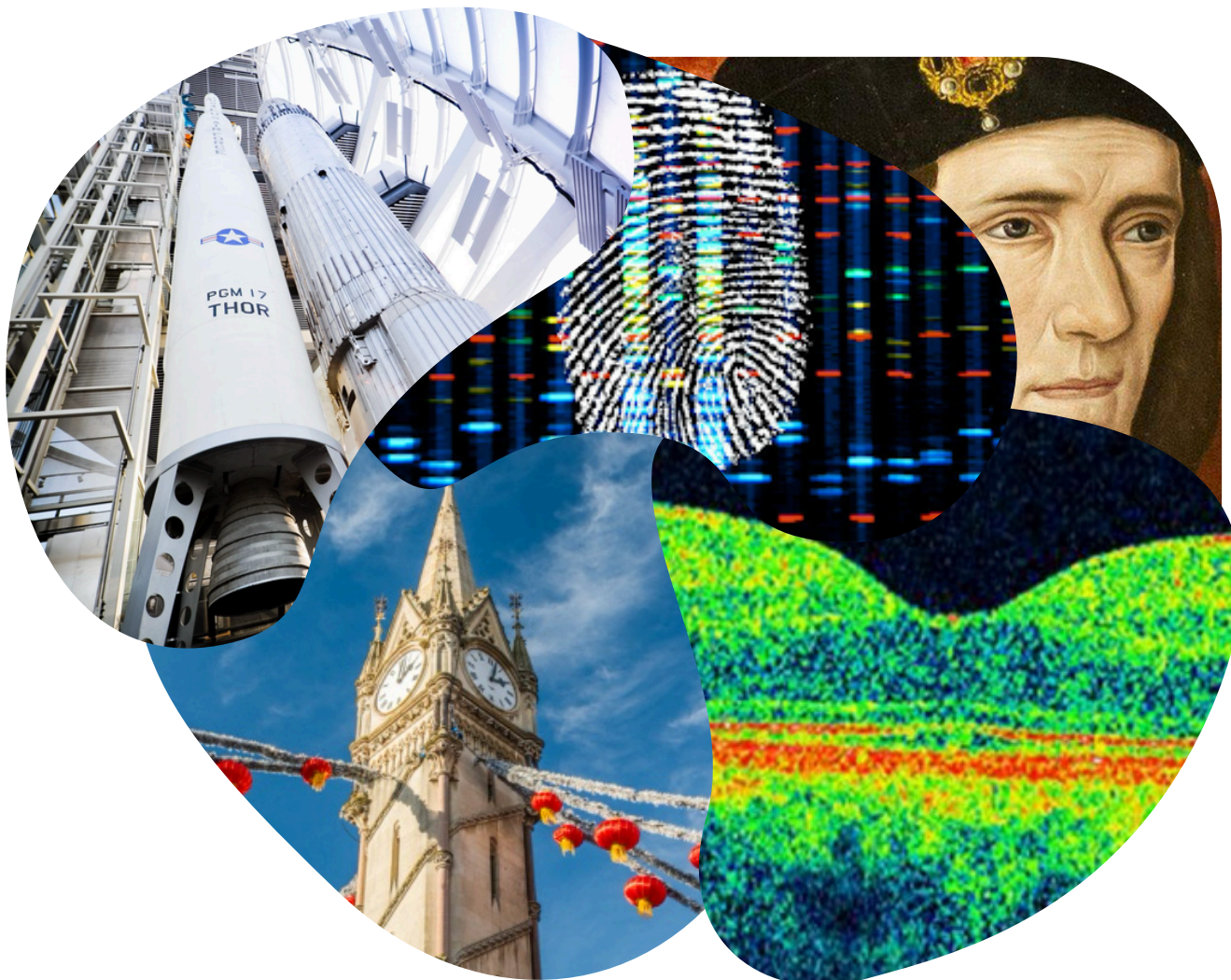
Leicester Railway Station is approximately a **20-minute walk** from the venue.

On leaving the station, head south along London Road,
then continue via Welford Road to Putney Road.

Taxis available outside the station entrance
can take you to campus in 5 minutes.

On the Day

- Registration opens at **08:30**
- Please wear your name badge
- Lunch and refreshments are provided
- Poster viewing during all break periods



PROGRAMME

08:30 *Registration & arrival*

09:15 **Welcome** *Mervyn Thomas*

SESSION 1 · 09:20 – 10:45 · Emerging Spectrum of Ocular Genetic Disease · K. Stephenson / C. Stanton

09:20 Refining diagnostic pathways for infantile nystagmus *Gail Maconachie*

09:40 Novel approaches to paediatric imaging *Zhanhan Tu*

10:00 Gene to guttae: genetic mechanisms to clinical consequences of Fuchs' endothelial corneal dystrophy *Siyin Liu*

10:20 Beyond single genes: polygenic and Mendelian pathways converge on the fovea *Callum Hunt*

10:40 *Session discussion*

10:45 – 11:05 · Refreshments & poster viewing

SESSION 2 · 11:05 – 11:55 · Early Career Researchers · Mariya Moosajee

11:05 **ECR presentations**

11:05 Cone synapse disruption drives visual loss in murine model of RPGR-mediated retinal degeneration *Chloe Brotherton*

11:15 Mitochondrial stress and autophagy disruption precede photoreceptor death in mouse models of IRD *Fay Newton*

11:25 Mutational spectrum of RHO-associated autosomal dominant retinitis pigmentosa in a large UK cohort and clinical features of the three most prevalent variants *Mark Hughes*

11:35 Reinventing the panel: Solving additional families with developmental glaucoma in the 100,000 Genomes Project *Nicky Cronbach*

11:45 Advancing towards an mRNA therapy for RLBP1-retinopathy *Sara Romero-Vázquez*

KEYNOTE LECTURE · 11:55 – 12:40

"Molecular genetics of inherited retinal diseases: past, present, and future"

Professor Carlo Rivolta

University of Basel & University of Leicester

12:40 – 13:40 · Lunch · Poster viewing · Annual General Meeting: All members welcome!

SESSION 3 · 13:40 – 15:00 · Genetic Counselling, Patient Perspective & Industry Collaborations · Julian Barwell

13:40 Genetic counselling: incidental findings and ethical dilemmas *Maria Echevarria / Julian Barwell*

13:55 Patient Perspective *Ramadan Younes*

14:10 Media portrayal of genetic eye disease *Vanessa Rodwell*

14:20 Patient reported exposure to and uptake of bogus ophthalmic treatments *Mariya Moosajee*

14:30 Updates on trials, regulatory process and communication *Aniz Girach*

14:50 *Session discussion*

15:00 – 15:15 · Break & meet the sponsors

15:15	Seedcorn Award	
15:15	Seedcorn UKEGG Grant Award 2025	<i>Dr Danyun Zhao</i>
15:20	Seedcorn UKEGG Grant Award Winner Announcement 2026	

SESSION 4 · 15:30 – 16:50

Clinical Cases & Modelling Disease · P. Vasudevan / O. Mahroo

15:30	Systemic disease and the eye: collaborative model of care between clinical genetics and ophthalmology	<i>Pradeep Vasudevan</i>
15:45	Clinical Cases	
15:45	SLC24A1 mutations are a cause of autosomal recessive retinitis pigmentosa	<i>Imran H. Yusuf</i>
15:55	Expanding the genotypic landscape of congenital stationary night blindness in an ethnically diverse Canadian population	<i>Kevin Gregory-Evans</i>
15:05	Hypomorphic IDUA genotypes with residual alpha-L-iduronidase activity are associated with retinitis pigmentosa in patients without syndromic MPS I disease	<i>Siyang Lin</i>
16:15	Preliminary Findings from the LUCE-1 Phase 1/2 Study: Single Sub-Retinal Administration of Dual AAV Gene Therapy, AAVB-081, for Usher Syndrome Type 1B (USH1B) Retinitis Pigmentosa (RP).	<i>Jayashree Sahni</i>
16:25	Stickler Syndrome: Insights from an Irish Cohort and the Need for A Dedicated Care Pathway	<i>Bridget Moran</i>
16:40	AI in IRD diagnostics and prognostication	<i>Nikolas Pontikos</i>
16:50	Insights from IRD disease models in a dish	<i>Cecile Méjécase</i>
17:10	<i>Session discussion</i>	
17:20	Prizes & close	

17:30 – 18:00 · *Networking reception*