

## Course Programme

Pre-course		Introduction to genomics - genetics variation	
		Aim of session: Understand the fundamental concepts of genetic variation, gene expression, and regulation.	
Video	20 mins	Introduction to the human genome Andrew Read, University of Manchester, UK	
Video	15 mins	Gene expression and regulation - Analysis of the transcriptome Hans Hennies, University of Huddersfield, UK	
Video	13 mins	Variation in the human genome - extend and source of variation Sara Brown, University of Edinburgh, UK	
Video	17 mins	Variation in the human genome, part II - How can we make sense of this?  Sara Brown, University of Edinburgh, UK	
Start (GMT)	Finish (GMT)	Presenter details	
Monday	/ 11 Nove	ember 2024	
12:00	13:00	Registration, lunch and networking	
13:00	13:10	Welcome	
		Wellcome Connecting Science: Nagehan Ramazanoglu Bahadir - Wellcome Connecting Sceince, UK	
		Scientific Programme Committee:	
		Ajoy Bardhan, University of Birmingham, UK	
		Joanna Jackow-Malinowska, Kings College London, UK	
		David Kelsell, Queen Mary University of London, UK	
		Edel O'Toole, Queen Mary University of London, UK	
13:10	14:00	Introductions (ice-breaker activity)	
14:00	14:30	Quiz on pre-course material	
		Chair: Joanna Jackow-Malinowska, Kings College London, UK Ajoy Bardhan, University of Birmingham, UK	
		Comfort break	
14:40	16:20	Session 1: Repair mechanisms and cancer	
		Aim of session: Explore the mechanisms of DNA repair in skin cells and their implications for cancer	
		Chair: David Kelsell, Queen Mary University of London, UK	
14:40	15:25	Dermatlas: The Genomic Atlas of Dermatopathology <u>David Adams, Wellcome Sanger Institute, UK</u>	
15:25	16:10	Title TBC Sophie Momen, St John's Institute of Dermatology, UK	
16:10	16:20	Session discussion	
16:20	16:50	Refreshment break and networking	



## 11-13 November 2024

16:50	18:30	Session 2: Epigenetics
		Aim of session: Identify epigenetic mechanisms and non-coding genomic elements and their functions in skin biology.
		Chair: Edel O'Toole, Queen Mary University of London, UK
16:50	17:10	Role of histone modifications in genome regulation  Pradeep Madapura, Queen Mary University of London, UK
17:10	17:30	RNA-based mechanisms orchestrating the cell cycle and genome stability in cancer
		Lovorka Stojic, Barts Cancer Institute, UK
17:30	18:30	Workshop
18:30	19:30	Poster session
		Aim of session: Showcase and discuss your recent research findings and advancements in skin biology
		Bar open (card payments only)
19:30		Dinner
19:30		Bar open (card payments only)



Tuesday 12 November 2024					
09:30	11:00	Session 3: Genomic literacy (workshop)			
		Aim of session: Learn to perform basic variant interpretation using clinical case histories and genetic reports			
		Chair: David Kelsell, Queen Mary University of London, UK			
09:30	11:00	Genetic variation in clinical care - tools, tips and pitfalls?  Elizabeth Jones, Saint Mary's Hospital Manchester and University of Manchester, UK			
		Thomas Cullup, Great Ormond St Hospital, UK			
11:00	11:45	Refreshment break and networking			
11:45	13:25	Session 4: Genomic technologies in skin disease			
		Aim of session: Explore advanced techniques and applications of long read sequencing and transcriptomic studies in skin research Chair: Joanna Jackow-Malinowska, Kings College London, UK			
11:45	12:30	Long read sequencing for skin research			
		John Common, A *STAR Skin Research Labs, Singapore			
12:30	13:15	Transcriptomic studies of mouse paw skin in health and disease <u>Diana Blaydon, Queen Mary University of London, UK</u>			
13:15	13:25	Session discussion			
13:25	14:25	Lunch and networking			
14:25	15:45	Session 5: Rare disease patient voice			
		Aim of session: Understand and integrate patients' expectations for new treatments Chair: Edel O'Toole, Queen Mary University of London, UK			
14:25	14:50	Ichthyosis, the scale of the problem  Mandy Aldwin- Easton. Ichthyosis Support Group (ISG). UK			
14:50	15:15	Pain, isolation, and the life-changing work of PC Project  Janice Schwartz, Pachyonychia Congenita Project, USA- VIRTUAL			
15:15	15:45	Panel discussion			
15:45	16:15	Refreshment break and networking			
16:15	17:05	Session 6: Al and genomics			
		Aim of session: Explore the potential of AI in skin research Chair: Ajoy Bardhan, University of Birmingham, UK			
16:15	17:00	Generative machine learning to model cellular perturbations  Mo Lotfollahi, Wellcome Sanger Institute, UK			
17:00	17:05	Comfort Break			
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17:05	18:05	Session 7: Keynote speaker Chair: Joanna Jackow-Malinowska, Kings College London, UK			
17:05	18:05	The human skin: development and disease  Muzlifah Haniffa, Wellcome Sanger Institute, UK			
18:05	19:05	Poster session with drinks reception			
10.03	13.03	Aim of session: Showcase and discuss your recent research findings and advancements in skin biology			
19:05		Dinner			
19:05		Bar open (card payments only)			



Wednesday 13 November 2024				
09:30	10:30	Session 8: Therapies (keynote speaker)		
		Aim of session: Understand the significance of genetic diagnosis in skin disease and its role in developing effective therapies		
		Chair: Joanna Jackow-Malinowska, Kings College London, UK		
09:30	10:30	Title TBC		
		Peter Marinkovich, Stanford University, USA		
10:30	11:00	Refreshment break and networking		
11:00	12:00	Session 9: Therapies (continued)		
		Aim of session: Understand the significance of genetic diagnosis in skin disease and its role in developing effective therapies		
		Chair: Edel O'Toole, Queen Mary University of London, UK		
11:00	11:40	Title TBC		
		Maanasa Polubothu, University College London, UK		
11:40	12:20	Title TBC		
		Joanna Jackow-Malinowska, Kings College London, UK		
12:20	12:30	Session discussion		
12:30	12:50	Consolidation and Closing remarks		
		Scientific Programme Committee:		
		Ajoy Bardhan, University of Birmingham, UK		
		Joanna Jackow-Malinowska, Kings College London, UK		
		David Kelsell, Queen Mary University of London, UK  Edel O'Toole, Queen Mary University of London, UK		
12:50	13:00	O Grab bag lunch		
13:10		Coach departures for Cambridge train station and city centre		
13.10		Coach departures for Cambridge train station and dity centre		