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 November 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 *David Adams, Wellcome Sanger Institute, UK*
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

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 *Diana Blaydon, Queen Mary University of London, UK*
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: AI 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
17:05	**18:05**	**Session 7: Keynote speaker**
		Chair: Joanna Jackow-Malinowska, Kings College London, UK
17:05	18:05	Human skin in health and disease: lessons from genomics *Muzlifah Haniffa, Wellcome Sanger Institute, UK*
18:05	**19:05**	**Poster session with drinks reception**
		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	Gene therapy for epidermolysis bullosa *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	Unravelling sporadic vascular malformations due to mosaic RAS/MAPK variants: new therapeutic perspectives *Maanasa Polubothu, University College London, UK*
11:40	12:20	Therapeutic Gene Editing Technologies in Dermatology *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	Grab bag lunch
13:10		Coach departures for Cambridge train station and city centre