

Manchester European Society of Human Genetics Syndromology and Dysmorphology Course 2024



Location: Postgraduate Centre, The Blvd, Manchester, M13 9NQ

Tuesday 22 nd October				
11:15	11:30	15	Course Introduction	Sofia Douzgou
11:30	12:15	45	Fundamentals of Dysmorphology	Dian Donnai
12:15	13:00	45	Syndromes of the Ras MAPK and PI3K pathways	Emma Burkitt Wright
13:00	14:00	60	Lunch	
14:00	14:45	45	Craniofacial and clefting syndromes	Sofia Douzgou Houge
14:45	15:30	45	The Brain as a clue to syndrome diagnosis	Abhijit Dixit
15:30	16:00	30	Coffee break	
16:00	16:45	45	Chromatin disorders	Siddharth Banka
16:45	17:15	30	Quiz: The eye as a clue to syndrome diagnosis	Kay Metcalfe
17:15	18:00	45	Participants' cases	All Faculty
18:00			Back to the Hotel	

Wednesday 23 rd October				
09:00	10:00	60	TADs and related syndromes	Gunnar Douzgos Houge
10:00	10:45	45	Sequencing and data re-analysis	Adam Jackson
10:45	11:15	30	Coffee	
11:15	12:15	60	Beyond DNA sequencing and disorders of the non-coding genome	Stefan Barakat
12:15	13:00	45	Episignatures and syndromes	Siddharth Banka
13:00	14:00	60	Lunch	
14:00	14:45	45	Craniosynostosis	Andrew Wilkie
14:45	15:30	45	Quiz: Hair, nails, skin and teeth as a clue to syndrome diagnosis	Sofia Douzgou Houge
15:30	16:00	30	Coffee break	
16:00	16:45	45	Syndromes of the Connective tissue and TGF-β signalling pathway	Bert Callewaert
16:45	17:15	30	Extending the conversation about what it means to be rare	The Whitworth Group
17:15	18:00	45	Participants' cases	All Faculty
18:00			Back to the Hotel	
19:30			Course Dinner	

Thursday 24 th October					
09:00	10:00	60	Skeletal and limb syndromes	Mohnish Suri	
10:00	10:45	45	Under-and overgrowth syndromes	Sofia Douzgou Houge	
10:45	11:15	30	Coffee		
11:15	12:15	60	Metabolic dysmorphic syndromes	Charulata Deshpande	
12:15	13:00	45	Facial analysis technology in dysmorphology	Tzung-Chien Hsieh	
13:00	14:00	60	Lunch		
14:00	14:30	30	Clinical trials in rare diseases	Simon Jones	
14:30	15:00	30	Resources for patients and families	Sarah Wynn	
15:00	16:00	60	Participants' cases	All Faculty	
16:00			End of course		

The ESHG Course is supported by:







