

ESHG Syndromology Course Programme 2024

Tuesday 22 nd October				
11:15	11:30	15	Course Introduction	
11:30	12:15	45	Clinical pattern recognition in Dysmorphology	Dian Donnai
12:15	13:00	45	Fundamentals of clinical examination and case presentation in dysmorphology	Jill Clayton-Smith
13:00	14:00	60	Lunch	
14:00	14:45	45	Craniosynostosis	Andrew Wilkie
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	Craniofacial and clefting syndromes	Sofia Douzgou Houge
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	
19:30			Evening Meal at	

Wednesday 23 rd October				
09:00	10:00	60	tbd	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	Sponsored talks
12:15	13:00	45	Chromatin disorders and epigenatures	Siddharth Manish Banka
13:00	14:00	60	Lunch	
14:00	14:45	45	Syndromes of the Ras MAPK and PI3K pathways	Emma Burkitt Wright
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	Community and patient engagement	The Whitworth group
17:15	18:00	45	Participants' cases	All Faculty
18:00			Back to the Hotel	
19:30			Course Dinner at	

Thursday 24 th October				
09:00	10:00	60	Skeletal syndromes (incl. Limb)	Mohnish Suri
10:00	10:45	45	Facial analysis technology in dysmorphology	Bert De Vries
10:45	11:15	30	Coffee	
11:15	12:15	60	Metabolic dysmorphic syndromes	Charulata Deshpande
12:15	13:00	45	Under-and overgrowth syndromes	Sofia Douzgou Houge
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	

ESHG course supported by - MRCC, MFT, MCGM, UoM, NIHR Manchester BRC, EpiGenRare, ERN-ITHACA, UNIQUE