

# SHINING A LIGHT ON GENETIC DISORDERS

Tuesday 18 June 2019  
18:00 - 20:00



## PROGRAMME

18:00 – 19:00

### Co-Presentations on the genetics of disease

**IGMM Host: Professor Wendy Bickmore**, MRC Human Genetics Unit (HGU) Director

**Event Comperer: Hazel Lambert**, Public Engagement with Research Manager, College of Medicine, University of Edinburgh

18:00 - 18:30

### The genetics of Colorectal Cancer

**Farhat Din**, Clinical Consultant and Research Scientist and one of her patients, **Debbie Webster** and **Susan Farrington**, Senior Scientist will share their personal perspectives about Bowel Cancer.

**John Scott, MSP** will talk about Lynch Syndrome, a rare a genetic bowel condition.

18:30 - 18:50

### The genetics of Aicardi-Goutières syndrome (AGS)

**Professor Yanick Crow**, clinical geneticist and **Robert Paterson**, the father of one of Yanick's patients will share their personal perspectives of Aicardi-Goutières syndrome.

18:50 - 19:15

### Discussion Session

#### Panel Members

- Farhat Din, Clinical Consultant and Research Scientist on Colorectal Cancer, Cancer Research UK Edinburgh Centre, MRC IGMM
- Colorectal Cancer patient
- Susan Farrington, Senior Scientist on Colorectal Cancer, Cancer Research UK Edinburgh Centre, MRC IGMM
- John Scott, MSP
- Professor Yanick Crow, clinical geneticist, CGEM, MRC IGMM
- Aicardi-Goutières syndrome (AGS) patients: The Paterson Family
- Natalie Frankish, Genetic Alliance UK
- Carol Porteous, Patient and Public Involvement Advisor, UoE Clinical Research Facility

19:15 - 20:00

### Networking Reception with Drinks & Canapés

## IGMM Nucleus, First Floor

- Talk to our researchers about their posters on genetic conditions – answering the key questions that people receiving a diagnosis ask
- Information stalls from support groups and relevant health agencies
- Chat to our researchers as you have fun with hands on activities

## LAY POSTERS

## DROP IN ACTIVITIES

1	<p><b>The SCOTTY Study</b> Genetic Disorder: Colorectal Cancer Genetics Poster Presenter: <b>Donna Markie</b> Malcolm Dunlop Colorectal Cancer Genetics Research Group MRC HGU &amp; CRUK Edinburgh Centre - MRC IGMM</p>	10	<p><b>Aniridia - Aberrant gene control affects vision</b> Genetic Disorder: Aniridia (an eye malformation) Poster Presenter: <b>Shipra Bhatia</b> Wendy Bickmore Research Group MRC HGU, MRC IGMM</p>
2	<p><b>Colorectal cancer: Living Well Beyond Diagnosis</b> NHS Lothian Colorectal Cancer Nursing Team <b>Susan McKeen &amp; Linda Sherwood</b> NHS Lothian</p>	11	<p><b>Genetic Eye Malformations</b> Genetic Disorders: Microphthalmia &amp; Aniridia (eye malformations) Poster Presenter: <b>Lusaidh Owen</b> David FitzPatrick Research Group MRC HGU, MRC IGMM</p>
3	<p><b>Colorectal Cancer Genetics- Polyposis Syndromes</b> Genetic Disorder: Colorectal Cancer Poster Presenter: <b>Paz Freile</b> Farhat Din, Susan Farrington &amp; Malcolm Dunlop Research Groups CRUK Edinburgh Centre, MRC IGMM</p>	12	<p><b>Genetic causes of Late-Onset Retinal Degeneration (L-ORD)</b> Genetic Disorder: L-ORD (an eye condition) Poster Presenter: <b>Chloe Stanton</b> Molecular Genetics of Vision-related Traits &amp; Quantitative Trait Loci Research Groups MRC HGU &amp; CGEM - MRC IGMM</p>
4	<p><b>Aicardi-Goutieres syndrome &amp; the interteronopathies</b> Genetic Disorder: AGS Poster Presenter: <b>Yanick Crow</b> Yanick Crow Research Group MRC HGU &amp; CGEM - MRC IGMM</p>	13	<p><b>Keratoconus</b> Genetic Disorder: Keratoconus (an Eye Condition) Poster Presenter: <b>Zuhair Mustafa</b> Veronique Vitar &amp; Quantitative Trait Loci Research Groups MRC HGU, MRC IGMM</p>
5	<p><b>Moving Toward Novel Therapeutics for Ovarian Cancer</b> Genetic Disorder: Ovarian Cancer Poster Presenter: <b>Zahra Massoud</b> Liz Patton Research Group MRC HGU, MRC IGMM</p>	14	<p><b>Improving Diagnosis of Myalgic encephalomyelitis /Chronic fatigue syndrome</b> Condition: ME / CFS Poster Presenter: <b>Joshua Dibble</b> Chris Ponting Research Group MRC HGU, MRC IGMM</p>
6	<p><b>Fishing for melanoma treatments</b> Genetic Disorder: Skin Cancer Poster Presenter: <b>Allesandro Brombin</b> Liz Patton Research Group MRC HGU, MRC IGMM</p>	15	<p><b>Understanding the causes of Depression</b> Condition: Genetic susceptibility to mental illness Poster Presenter: <b>Matthew Thorpe</b> Pippa Thomson CGEM, MRC IGMM</p>
7	<p><b>Primary ciliary dyskinesia (PCD)</b> Genetic Disorder: PCD Poster Presenter: <b>Fraser McPhie</b> Eleanor Mills Research Group MRC HGU, MRC IGMM</p>	16	<p><b>People Power - Scottish Population Biobanks</b> Generation Scotland, Viking &amp; Orcades Poster Presenter: <b>Caroline Hayward</b> QTL in Health and Disease research programme MRC HGU, MRC IGMM</p>
8	<p><b>Understanding Hutchinson-Gilford Progeria Syndrome</b> Genetic Disorder: HGPS Poster Presenter: <b>Nattaphong Rattanavirotkul</b> Tamir Chandra Research Group MRC HGU, MRC IGMM</p>	17	<p><b>Discovering a new medicine - not just a 'Eureka' moment!</b> The medicinal chemistry lab designs potential novel drugs &amp; performs pre-clinical testing Poster Presenter: <b>Cecilia Ayala</b> Asier Unciti-Broceta Group CRUK Edinburgh Centre</p>
9	<p><b>Investigating the basis of Cornelia de Lange Syndrome</b> Genetic Disorder: CdLS Poster Presenter: <b>Rob Foster</b> David FitzPatrick Research Group, MRC HGU, MRC IGMM</p>		

Check Your Cheek Cells (Microscopy)  
Strawberry DNA extraction  
Connecting Codes to Cures  
Mighty Mitochondria  
Power of Partnerships  
Puzzle & Fluorescent Cell Staining Game

## INFORMATION STALLS

**Genetic Alliance UK** -  
Natalie Frankish

**University of Edinburgh Clinical Research Facility** -  
Carol Porteous, Patient and Public Involvement Advisor

The **SHARE** Register

Primary Ciliary Dyskinesia (**PCD**) Support UK