

Genetics and Mosaicism

Residents Course | 04-06 October 2024 | Barcelona, Spain

FACULTY

Chair: Neil Rajan

Speakers: Pierre Vabres, Antonio Torrelo, Anna Dubois, Veronica Kinsler

COURSE DESCRIPTION

Genetic testing for genodermatoses is rapidly progressing, and in some countries is now in the hands of dermatologists. Recognising genetic conditions as well as mosaicism is especially important, as this can have implications for patient care and genetic counselling that have not been previously recognized.

A small faculty of European experts, with clinical and research experience in genetics and mosaicism, will lead this residential course to introduce genetic concepts relevant to germline and mosaic conditions clinically relevant to the care of patients with genetic skin diseases, and update on common clinical presentations and management.

LEARNING OBJECTIVES

The proposed training programme will outline basic concepts in genetics.

This will be followed by state-of-the-art clinical lectures on different types of germline and mosaic skin conditions.

Attendees will be invited to bring clinical cases (consented photographs and anonymised clinical information), which will be used for case-based discussions in workshops, and to develop diagnostic skills, specifically in relation to:

- When to consider a genetic diagnosis in the clinic, and the clinical signs that should prompt this.
- When to consider genetic testing, its advantages and limitations, and genetic counselling.
- What are the clinical diagnostic challenges in mosaicism and how to overcome them.
- How to manage patients with mosaicism, and when to think of involvement of other tissues beyond the skin.

PROGRAMME

The course will run over 3 days – Friday afternoon 4th October 2024, all day on Saturday 5th October and Sunday morning 6th October. The programme will include a pre-course online quiz on genetics and mosaicism. Participants will be required to submit a case presentation before the course for discussion at the meeting.

Friday, 04 October 2024 - Day 1

13:30-14:00 Arrival and participants check-in

14:00-14:20 Welcome and introductions

14:20-15:00 An introduction to genetics - Neil Rajan

15:00-15:30 Interactive session - Review of pre-course quiz responses - Anna Dubois

15:30-16:00 **Coffee Break**

16:00-17:30 Participant cases - short presentations - 10+5 mins questions x 6 - All faculty

17:30-18:00 Q&A session

18:00-18:15 Wrap-up and close of Day 1

Saturday, 05 October 2024 - Day 2

- 09:00-09:45 An introduction to mosaicism: Mosaic disorders affecting pigmentation - Veronica Kinsler
- 09:45-10:00 Relevant participant case x1
- 10:00-10:45 Genetic transmission of germline and mosaic conditions and family planning - Antonio Torrelo
- 10:45-11:00 Relevant participant case x1
- 11:00-12:00 Blaschkolinear pigmentation disorders - Pierre Vabres
- 12:00-13:00 **Lunch break**
- 13:00-13:30 Diagnostic testing of germline and mosaic diseases - Pierre Vabres
- 13:30-14:00 Faculty cases x 2 Exemplifying diagnostic strategies
- 14:00-14:30 GENotes - Resources for the clinician - Anna Dubois
- 14:30-15:00 **Coffee Break**
- 15:00-15:45 Clinical trials for mosaic conditions - Pierre Vabres
- 15:45-16:15 Q&A session
- 16:15-16:30 Wrap-up and close of Day 2

20:00-22:00 **Networking Dinner with faculty and participants**

Sunday, 06 October 2024 - Day 3

- 09:00-09:45 Genetic autoinflammatory diseases- Antonio Torrelo
- 09:45-10:00 Relevant participant case x1
- 10:00-10:45 Inherited skin tumour syndromes - Neil Rajan
- 10:45-11:00 Relevant participant case x1
- 11:00-11:30 **Coffee Break**
- 11:30-12:15 Mosaicism with neurological and endocrine phenotypes - Veronica Kinsler
- 12:15-12:30 Relevant participant case x1
- 12:30-13:00 Interactive quiz - Anna Dubois + All faculty
- 13:00-13:30 Course closing comments - "What we still don't know about genetics and mosaicism" - All faculty
- 13:30 Farewell and Depart

The programme might be subject to changes

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