

Agenda – Genetics and Precision Medicine in Rare Diseases

Lisbon, 18-19 May 2023

Day 1

Session name/Topic	Speaker	Time
Introduction		2.30 pm
Precision medicine: the model of monogenic diabetes	Carolina Neves	3.00 pm
From clinical suspicion to genetic testing	Carolina Neves	3.20 pm
Genetic variants and clinical impact	João Filipe Raposo	3.40 pm
Break		4.10 pm
Current Methodologies for Genetic Diagnosis for rare diseases	Mafalda Bourbon	4.30 pm
Classification of genetic variants according to current guidelines	Paulo Dario	5.15- 6.00 pm
Dinner		19.30 pm

Day 2.

Session name/Topic	Speaker	Time
Recruitment and Regulatory Issues	Dulce do Ó	9.00 am
Data interpretation in clinical research	Rogério Ribeiro	9.45 am
Break		10.30 am
Precise medicine studies in evaluating therapeutic response – does it exist?	Philippe Froguel	11.00 am
Working Groups: research project design – precise medicine in a rare disease	Rogério Ribeiro	11.45 am
Lunch		1.00 pm
Working Groups presentations	Carolina Neves	2.00 pm
Closing remarks	Carolina Neves Rogério Ribeiro	3.30 pm

