Jeanesse Scerri

Cardiogenetics: Bridging the Gap

Duration	4 weeks
Short Bio	Dr. Jeanesse Scerri is a biomedical scientist with over 10 years' experience in molecular biology and genetics. She is the scientific team leader at the Molecular Biology & Genetics laboratories, Pathology Department, Mater Dei Hospital, physically situated at the University of Malta, where she has introduced and is spearheading various clinical services including next generation sequencing (NGS) for cardiogenetic conditions and tumour molecular profiling. She is experienced in research with international collaborations, having obtained her PhD in breast cancer research in 2019. She is the lead of the 1+MG/Genome of Europe Maltese National Mirror Group 4 (Good sequencing practice/Development of standards for clinical interpretation) and supporting lead of the National Mirror Group 12 (Genome of Europe) in the National Genomics Infrastructure, and a member of the EU4Health Action Grant: CAN.HEAL: Cancer Diagnostics & Treatment for All; Genomics for Public Health.
Home Institution	Mater Dei Hospital (Ministry for Health)
Host Institution	Amsterdam University Medical Centre (UMC)
Project Description	The project consisted of two main arms: • The diagnostic arm, whereby knowledge about the cardiogenetic diagnostic service in a state-of-the-art reference European laboratory was obtained by observing and discussing technical aspects with the laboratory teams and attendance to weekly multidisciplinary team meetings (MDTs) at the UMC, which included scientific staff, cardiologists, geneticists, and other professionals, allowing for the observation of interdisciplinary collaboration and decision-making processes. A successful online meeting between the two institutions was also held, where the Maltese cardiogenomic diagnostic service was presented, and current issues, such as the low genetic diagnostic yield in Maltese long QT syndrome patients, were discussed. • The research arm, whereby advanced technologies and functional assays for the investigation of novel variants and variants of unknown significance (VUS) in hereditary cardiac pathologies were observed. These included patch clamp, optical mapping, and cardiac tissue slicing. Weekly informal research group updates and interdisciplinary research meetings were also attended to gain familiarity with the research and collaborative environment at the UMC.

Personal Statement	The outcomes of this exchange will lead to improvements in routine laboratory operations at Molecular Pathology & Genetics. The project's deliverables, including a gap analysis and report on the diagnostic area and a presentation on observed functional research techniques, were shared with colleagues at various levels within the Ministry for Health. The main achievement of this exchange was the bridging of the two Institutions for future collaborative opportunities. Whilst the Host Institution has all the infrastructure for advanced functional research, the Home Institution has a population with a unique genetic pool and the continuous discovery of novel variants that are associated with hereditary cardiac conditions but are of as yet uncertain clinical significance. This creates the perfect scenario for fruitful collaboration. The network created with researchers at the Host Institution will be important in future collaboration, including research projects that can be carried out between the two Institutions to answer scientific questions arising from diagnostic cases. Similar initiatives at various levels, with the support of the Home Institution and ERNs, are recommended. Supporting healthcare professionals in participating in ERN fellowship exchanges is an ideal investment in their professional development and that of the institution. Collaboration with highly specialized teams in specific networks can strengthen the understanding and management of rare diseases in a small country like Malta.