

Nuclear Envelope-linked Rare Human Diseases: from Molecular Pathophysiology towards Clinical Applications – Euro-Laminopathies

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The EMAUG research group shall carry out the objectives of Unit n° 7 within the project “Nuclear Envelope-linked Rare Human Diseases: from Molecular Pathophysiology towards Clinical Applications – Euro-Laminopathies” (contract n°. LSHM-CT-2005-018/690). In particular, for EMA-UG, Prof Manfred Wehnert, (Medizinische Fakultät, Institut für Humangenetik) shall be in charge of the identification of new mutations that could cause laminopathies, of establishing cell cultures from biopsies derived from genetically characterised patients, and/or of collecting cell cultures and of performing genetic characterisation of the cells. Prof. Manfred Wehnert shall provide the Laboratorio di Biologia Cellulare e Microscopia Elettronica IOR with all the studied samples in order to allow further metabolic studies at IOR.

The scientific aim of the EMAUG contribution to the Euro-Laminopathy project is to use a functional candidate approach to associate further genes encoding components of the nuclear lamina and nuclear envelope to genetic human diseases including clinical phenotypes like Emery-Dreifuss muscular dystrophy, dilated cardiomyopathies and others. Moreover, the project is expected to add to the molecular elucidation of laminopathies by providing defined mutations in *LMNA*, *STA* and *ZMPSTE24* genes resulting from molecular genetic diagnostic cases.