

**11. RARE DISEASES<sup>i</sup>**

Level II

**Department of Experimental and Clinical Medicine (DMSC)**

<b>Course coordinators</b>	Francesco Annunziato Coordinator Cristina Scaletti Scientific Coordinator
<b>Executive Committee</b>	Francesco Annunziato Cristina Scaletti Fabio Marra Cecilia Berni Domenica Taruscio
<b>Contact person for information regarding teaching organization, class schedule, course content</b>	Cristina Scaletti <a href="mailto:cristina.scaletti@unifi.it">cristina.scaletti@unifi.it</a> Rosina De Cario <a href="mailto:rosina.decario@unifi.it">rosina.decario@unifi.it</a>
<b>Practical-professional profile of the course and industry sector of reference</b>	<p>The Master on Rare Diseases will enable the acquisition of skills to address the overall management of the rare disease patient from clinical to care, social, research, ethical and legislative aspects through an integrated and multidisciplinary approach toward diseases that are a public health problem and are often burdened by diagnostic and therapeutic difficulties, clinical severity, chronic course, disabling outcomes and heavy repercussions on the rare disease patient's family.</p> <p>Rare diseases constitute a large and heterogeneous group of human diseases (about 8-9,000) and are defined as such because of their low incidence in the population (they affect no more than 1 in 2,000 inhabitants in the European Union).</p> <p>Precisely because of these characteristics, specifically in terms of low incidence per disease but high numbers of diseases known to date, they constitute a significant health problem and affect millions of people and families worldwide. Their numerosity, low incidence, and heterogeneity often mean that it is difficult for the patient to obtain an early and appropriate diagnosis and a timely treatment where it exists. Add to this the often chronic-invalidating disease course and the relevant individual, family, and social burden.</p> <p>Also of paramount importance are the necessary research inputs in terms of early diagnosis, pathogenetic mechanisms, and new therapies.</p> <p>Approximately 80% of cases are genetic in origin, and the remaining 20% are multifactorial diseases derived from individual susceptibility and other factors (e.g., some environmental and dietary factors) or from the interaction between genetic and environmental causes. There is a significant difference in the age at which they appear; some may occur prenatally, some at birth or during childhood, and some in adulthood.</p> <p>These characteristics necessitate a comprehensive cognitive approach that can provide the tools for professionals to take charge of the rare disease patient and to develop and consolidate new research areas in an integrated vision between care and research.</p> <p>These objectives constitute the educational purpose of the Master Course, knowing that the Region of Tuscany has been involved for years in the field of Rare Diseases and, in particular, in the implementation of the new Rare Disease Network, including the organization of specialized facilities of high competence for rare diseases or groups of diseases, in the definition of diagnostic, therapeutic and assistance pathways (DTAP), in the implementation and promotion of neonatal screening, in support of participation in research calls dedicated to Rare Diseases and in international networks (ERNs), of which many</p>

	<p>of the Tuscan centers are members.</p> <p>The reference market consists of:</p> <ul style="list-style-type: none"> <li>• Rare Disease Referral Centers or Specialist Groups at university or territorial hospital health care companies also with reference to expertise in the construction of Diagnostic-Therapeutic-Assistance Pathways for Rare Diseases</li> <li>• multidisciplinary group activities for specific rare diseases, research laboratories, centers for the development of new technologies for the diagnosis and treatment of diseases or groups of rare diseases</li> <li>• pharmaceutical and biotechnological industries</li> <li>• public and private National and European entities involved in the care and organization of rare diseases (e.g., European Reference Networks (ERNs), patient associations).</li> </ul> <p>The Master course is aimed at individuals interested in developing concrete professionalism in the field of Rare Diseases recognition in order to integrate the training acquired during university studies and, in particular:</p> <ul style="list-style-type: none"> <li>• Know about Rare Diseases and their complexity by framing them within the ERN groupings, national and regional regulatory context, and collaborate in the management and implementation of the Rare Disease Network</li> <li>• Know the groups of Rare Diseases and their specificities</li> <li>• Create and manage patient pathways (DTAP) for patients with rare diseases within the health care and social welfare system</li> <li>• Taking charge of rare disease patients and their families within regional pathways</li> <li>• Deepen and promote research on rare diseases, including the means of accessing specific calls for proposals</li> <li>• Stimulate knowledge about rare disease therapies, genetics and gene therapies, orphan drugs, and research in this area</li> <li>• Learn about new technologies to implement the cross-cutting approach toward Rare Diseases.</li> </ul>
<p><b>Access prerequisites</b></p>	<p>Master's degree obtained in accordance with the system under Ministerial Decree No. 270/2004 (or specialist degree under Ministerial Decree No. 509/1999 equated under I.D. July 9, 2009) in one of the following classes</p> <ul style="list-style-type: none"> <li>• LM-1 Cultural Anthropology and Ethnology</li> <li>• LM-6 Biology;</li> <li>• LM-8 Industrial Biotechnology</li> <li>• LM-9 Medical, Veterinary and Pharmaceutical Biotechnology;</li> <li>• LM-14 Modern Philology</li> <li>• LM-15 Philosophy, Literature and History of Antiquity</li> <li>• LM-16 Finance</li> <li>• LM-17 Physics;</li> <li>• LM-18 Computer Science;</li> <li>• LM-21 Biomedical Engineering;</li> <li>• LM-22 Chemical Engineering</li> <li>• LM-25 Automation Engineering;</li> <li>• LM-27 Telecommunications Engineering;</li> <li>• LM-29 Electronic Engineering;</li> <li>• LM-31 Management Engineering</li> </ul>

	<ul style="list-style-type: none"> <li>• LM-32 Computer Engineering</li> <li>• LM-33 Mechanical Engineering</li> <li>• LM-38 Modern Languages for Communication and International Cooperation</li> <li>• LM-39 Linguistics</li> <li>• LM-40 Mathematics</li> <li>• LM-51 Psychology</li> <li>• LM-52 International Relations</li> <li>• LM-54 Chemical Sciences</li> <li>• LM-55 Cognitive Sciences</li> <li>• LM-56 Economics</li> <li>• LM-60 Nature Sciences</li> <li>• LM-61 Human Nutrition Sciences</li> <li>• LM-62 Political Science</li> <li>• LM-63 Public Administration Science</li> <li>• LM-77 Economic and Business Sciences</li> <li>• LM-78 Philosophical Sciences</li> <li>• LM-82 Statistical Sciences</li> <li>• LM-83 Actuarial and Financial Statistics</li> <li>• LM-85 Pedagogical Sciences</li> <li>• LM-87 Social Service and Social Policy</li> <li>• LM-88 Sociology and Social Research</li> <li>• LM-90 European Studies</li> <li>• LM-91 Information Society Techniques and Methods</li> <li>• LM-92 Theories of Communication</li> <li>• LM-93 Theories and Methodologies of E-Learning and Media Education</li> <li>• LM/SNT1 Nursing and midwifery</li> <li>• LM/SNT2 Rehabilitation health professions</li> <li>• LM/SNT3 Technical health professions</li> <li>• LM/SNT4 Prevention health professions</li> <li>• LM/SC-GIU Legal Science</li> </ul> <p>Single-cycle degree obtained according to the system under Ministerial Decree No. 270/2004 (or specialist degree under Ministerial Decree No. 509/1999 equated under I.D. July 9, 2009) in one of the following classes:</p> <ul style="list-style-type: none"> <li>• LM-13 Pharmacy and Industrial Pharmacy</li> <li>• LM-41 Medicine and Surgery</li> <li>• LM-46 Dentistry and Dental Prosthetics</li> <li>• LMG/01 Law</li> </ul> <p>Degree awarded according to a system prior to Ministerial Decree No. 509/1999 of closely related content, deemed suitable by the Executive Committee or a Commission specifically appointed by it.</p>
<b>How the admission procedure takes place</b>	Selection by academic qualifications
<b>Duration</b>	12 months
<b>Teaching methods</b>	Distance Learning - Synchronous/A-synchronous - Moodle Platform
<b>Language of instruction</b>	Italian
<b>Attendance requirements</b>	minimum 70%
<b>Location of the course</b>	Department of Experimental and Clinical Medicine

	Region of Tuscany - Via Taddeo Alderotti n. 26N - 50139 Florence
<b>Foreseen lecture schedule</b>	Classes are held on Fridays
<b>Examinations procedures and schedule</b>	There will be mid-term verifications in ways that will be identified later.
<b>Final examination</b>	The final examination consists of the presentation of a paper on the skills acquired.

<b>Available places and enrolment fees</b>	
<b>Full-fee students</b>	
Minimum number	5
Maximum Number	20
Enrolment fee	€2500
<b>SINGLE MODULES</b>	
None planned	

<b>Description of the activities and training objectives of the internship</b>	Students also carry out an internship period aimed at completing and integrating their knowledge in the field of rare diseases as well as applying the concepts learned in face-to-face lectures and declined in general aspects and specific aspects for disease groups. In particular, the care of patients with rare diseases and their families, ways to manage the complexity of issues related to the need for early diagnosis, research, innovative therapies, new technologies, and social and regulatory aspects are explored. Observational activity. 150 total hours of internship.
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<sup>i</sup> This document is a translation of the form A.1 relating to the characteristics of the course attached to the Decree of the Deputy number 873 (record 158006) of 25th of July 2022, drafted in Italian and issued on the Master | Didattica | Università degli Studi di Firenze | UniFI and which therefore constitutes the only official document. This English translation cannot be used for legal purposes and has the sole purpose of supplying information in English on the content of the public notice.