

11. RARE DISEASESⁱ	
Second Level	
Department of Experimental and Clinical Medicine (DMSC)	
Course coordinator	Francesco Annunziato Coordinator Cristina Scaletti Scientific Coordinator
Organizing committee	Francesco Annunziato Cristina Scaletti Fabio Marra Cecilia Berni Domenica Taruscio
Contact person for information regarding course organization, the schedule of classes and course content	Cristina Scaletti cristina.scaletti@unifi.it Linda Di Pietro linda.dipietro@unifi.it
Practical-professional profile of the course and reference job market	<p>The Master in Rare Diseases provides students with the skills to address the overall management of the rare disease patient, from clinical to care, social, research, ethical and legislative aspects, through an integrated 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 patient's family. 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>These characteristics, and in particular the large number of diseases with low incidence known to date, make rare diseases a significant health problem affecting millions of people and families worldwide.</p> <p>Their numerosity, low incidence and heterogeneity often mean that it is difficult for the patient to obtain an early correct diagnosis and timely treatment, where available. In addition, the course of the disease is often chronic-invalidating and represents a significant burden for patients, their families, and society.</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 stemming 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 others in adulthood.</p> <p>These characteristics call for a comprehensive exploratory approach that can provide the tools for professionals to take charge of the rare disease patient and to develop and consolidate new research areas, integrating care and research. These are the educational objectives 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 organisation of specialised facilities of high competence for rare pathologies or groups of pathologies, in the definition of diagnostic, therapeutic and assistance paths (PDTA), in the implementation and promotion of neonatal screening, in promoting participation in research calls dedicated to Rare Diseases and in international networks (ERN), of which many Tuscany centres are members.</p>

	<p>Possible employers include:</p> <ul style="list-style-type: none"> • Rare Disease Referral Centres or Specialist Groups at university or community hospitals, also with reference to expertise in the development of Diagnostic-Therapeutic-Assistance Pathways for Rare Diseases; • multidisciplinary group activities for specific rare diseases, research laboratories, centres for the development of new technologies for the diagnosis and treatment of diseases or groups of rare diseases; • pharmaceutical and biotechnological industries; • public and private national and European institutions involved in the management and organization of rare diseases (e.g., European Reference Networks (ERNs), patient associations). <p>The Master course is for individuals interested in developing concrete professionalism in recognizing Rare Diseases so as to fill gaps in knowledge acquired during university studies and, in particular:</p> <ul style="list-style-type: none"> • Know about Rare Diseases and their complexity, placing them within ERN groupings, national and regional regulatory contexts, and collaborating in the management and implementation of the Rare Disease Network • Know the groups of Rare Diseases and their specificities • Create and manage patient pathways (DTAP) for patients with rare diseases within the healthcare and social welfare system • Take charge of rare disease patients and their families within regional pathways • Deepen and promote research on rare diseases, including ways of accessing specific calls for proposals • Stimulate knowledge about rare disease therapies, genetics and gene therapies, orphan drugs, and research in this area • Learn about new technologies to implement a transversal approach to Rare Diseases.
<p>Admission requirements</p>	<p>Master's degree awarded in accordance with the system under Ministerial Decree No. 270/2004 (or Master's degree under Ministerial Decree No. 509/1999, equivalent under I.D. of July 9, 2009) in one of the following classesⁱⁱ</p> <ul style="list-style-type: none"> • LM-1 Cultural Anthropology and Ethnology • LM-6 Biology • LM-8 Industrial Biotechnology • LM-9 Medical, Veterinary and Pharmaceutical Biotechnology • LM-14 Modern Philology • LM-15 Philosophy, Literature and History of Antiquity • LM-16 Finance • LM-17 Physics • LM-18 Computer Science • LM-21 Biomedical Engineering • LM-22 Chemical Engineering • LM-25 Automation Engineering

	<ul style="list-style-type: none"> • LM-27 Telecommunications Engineering; • LM-29 Electronic Engineering • LM-31 Management Engineering • LM-32 Computer Engineering • LM-33 Mechanical Engineering • LM-38 Modern Languages for Communication and International Cooperation • M-39 Linguistics • LM-40 Mathematics • LM-51 Psychology • LM-52 International Relations • LM-54 Chemistry • LM-55 Cognitive Sciences • LM-56 Economics • LM-60 Nature Sciences • LM-61 Human Nutrition • LM-62 Political Science • LM-63 Public Administration • LM-67 Sciences and Techniques of Preventive and Adapted Motor Activities • LM-77 Economics and Business • LM-78 Philosophy • LM-82 Statistics • LM-83 Actuarial and Financial Statistics • LM-85 Pedagogical Sciences • LM-87 Social Service and Social Policy • LM-88 Sociology and Social Research • LM-90 European Studies • LM-91 Information Society Techniques and Methods • LM-92 Theories of Communication • LM-93 Theories and Methodologies of E-Learning and Media Education • LM/SNT1 Nursing and midwifery • LM/SNT2 Rehabilitation health professions • LM/SNT3 Technical health professions • LM/SNT4 Health professions for prevention • LM/SC-GIU Legal Science <p>Single-cycle Master's degree awarded in accordance with the system under Ministerial Decree No. 270/2004 (or a Master's degree under Ministerial Decree No. 509/1999, equivalent under I.D. of 9 July 2009) in one of the following classes:</p> <ul style="list-style-type: none"> • LM-13 Pharmacy and Industrial Pharmacy • LM 41 Medicine and Surgery • LM 46 Dentistry and Dental Prosthetics • LMG/01 Law <p>Degree awarded under a system prior to Ministerial Decree No. 509/1999 of closely related content, deemed suitable by the Organizing Committee or by a Committee specifically appointed by it.</p>
Admission procedure	Selection based on qualifications
Duration	12 months
Teaching methods	Distance – synchronous/asynchronous – Moodle Platform

Language the course will be delivered in	Italian
Attendance requirement	at least 70%
Course location	Department of Experimental and Clinical Medicine, Largo Brambilla 3, Florence Regione Toscana, Via Taddeo Alderotti 26, Florence
Foreseen lecture days	Classes are held on Fridays
Exam procedures and schedule	There will be mid-term exams. The methods for delivering these exams will be decided later.
Final exam	The final exam consists in the presentation of a paper on the knowledge acquired.

Number of places available and enrolment fees	
Full-fee students	
Minimum no. of places	5
Maximum no. of places	25
Enrolment fee	2,500 Euros
Single modules	
None	

Description of traineeship activities and training objectives	<p>Students also carry out a traineeship period aimed at completing and integrating their knowledge in the field of rare diseases as well as applying the concepts learned in in-person lectures and declined in general and specific aspects for each disease group. 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.</p> <p>Observational activity. 150 total hours of traineeship.</p>
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ⁱ 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 652 (record 154925) of 13th of July 2023, 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.

ⁱⁱ As modified with the Decree of the Deputy number (record) of