



*Telethon Institute of
Genetics and Medicine
Naples*

Student Handbook 2017-2018



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Dean's Message



Dear Students,

We are delighted to get to know you and help you on your way to pursuing a career in medical sciences. TIGEM and its researchers are devoted to training scientists in human genetics. The Institute provides a unique environment for students with a deep commitment to their PhD studies and offers a multidisciplinary curriculum offering theoretical and practical training in cell biology of genetic diseases, systems biology and functional genomics, and molecular therapy.

We encourage you to take full advantage of the resources and opportunities at TIGEM, as our philosophy of education is built on the belief that our students should recognize the intrinsic rewards of an education in science.. We provide a research environment which fosters creative thinking and encourages intellectual curiosity. Our perspective on learning is original and based off of collaboration and personal wellbeing; instead of emphasizing memorization, grades or class rank, we stress participation in hypothesis-driven research, interdisciplinarity, by which our scientists, post-docs and trainees, work together to solve problems focused around understanding the mechanisms of human genetic diseases, development of strong research skills and dedication to responsible, professional behavior.

TIGEM's PhD programme includes training courses, seminars, and data clubs, allowing PhD students to interact with international scientists and practice presenting their thesis data, stimulating the exchange of ideas and improving students' language and presentation skills and critical perspective, all essential components to a successful research or teaching career in the medical sciences.

Over the years our PhD students have pursued successful scientific careers in Italy, Europe and elsewhere, have published in leading scientific journals and obtained prestigious fellowships and awards, which are all indicators of the success of our training programmes.

Be sure to enjoy your experiences as a developing scientist and to always seize the opportunities that will help you in reaching your goals. We look forward to meeting you here at TIGEM and welcome you to experience the adventure of making a scientific discovery first hand.

Sincerely yours,

Graciana Diez-Roux

TIGEM and surroundings



What is TIGEM?

The Telethon Institute of Genetics and Medicine (TIGEM) is an international research centre dedicated to the study of rare genetic disease.

It was founded in 1994 by the Telethon Foundation, one of Italy's major non-profit organizations, to promote the advancement of research aimed at the diagnosis, prevention and cure of human genetic diseases.

TIGEM's mission is to understand the mechanisms of genetic diseases and to develop therapeutic and preventive strategies.

TIGEM provides state-of-the-art laboratory instruments, workplaces resources and a welcoming environment to its staff, students, and visiting scientists, allowing them to use their talents and fully contribute to the productivity of this research entity. Since its creation the Institute's research goals have progressively evolved from identifying the genes responsible for genetic diseases to examining gene function and delving into the cellular mechanisms behind these diseases. Significant investments have been made in the field of therapeutic applications; in particular, gene delivery and transfer approaches

has helped to develop promising solutions for the treatment of inherited eye diseases and inborn errors of metabolism, and systems biology and functional genomics have been key elements for the development of novel approaches to identifying regulatory gene networks.

Research at TIGEM is multidisciplinary and incorporates three main research tactics:

- Cell Biology of Genetic Diseases that aims to understand disease mechanisms through the in depth study of the molecular basis of how cells are organized, how they accomplish their individual tasks and cooperate to form tissues and systems, and how mutations in disease genes alter these functions.
- Genomics Medicine that focuses on the development and application of integrated experimental and computational tools in mammalian systems to study genetic diseases and either develop or monitor therapeutic approaches.
- Molecular Therapy that aims to develop novel strategies for the therapy and prevention of genetic diseases.



TIGEM Location

Since its establishment, the Institute has grown considerably- over 220 members, including 18 independent research groups, graduate students, post-doc fellows, technicians and administration, work together towards a greater cause.

TIGEM has moved to Pozzuoli, a town located near Naples, in spring 2014. The Institute is located in the “Comprensorio Olivetti”, the structure of a former factory built in 1954. The “Comprensorio Olivetti” is a unique example of intelligent insertion of an industrial structure into an environment of great natural beauty. The factory was built with the desire to combine the needs of an industrial society with the values of a traditional society, in line with the ideas of Adriano Olivetti.

5000 square meters were converted into 4 open space laboratories, 4 meeting rooms, over 28 offices for researchers and administrative staff, spaces dedicated to microscopy, cell culture and bioinformatics, and an auditorium capable of seating 180 people for business, training and scientific dissemination.

Pozzuoli is just few kilometres away from Naples, which hosts the largest concentration of academic and research institutes in Southern Italy. The region of which Naples is the capital, Campania, hosts nine universities (Università degli Studi di Napoli Federico II, Università degli Studi della Campania Luigi Vanvitelli, Università degli Studi di Napoli Parthenope, Università degli Studi di Napoli L'Orientale, Università degli Studi Suor Orsola Benincasa, Università degli Studi di Salerno, Università degli Studi del Sannio, Università Telematica Giustino Fortunato, Università Telematica Pegaso) and a

number of important research and medical centers of scientific excellence.

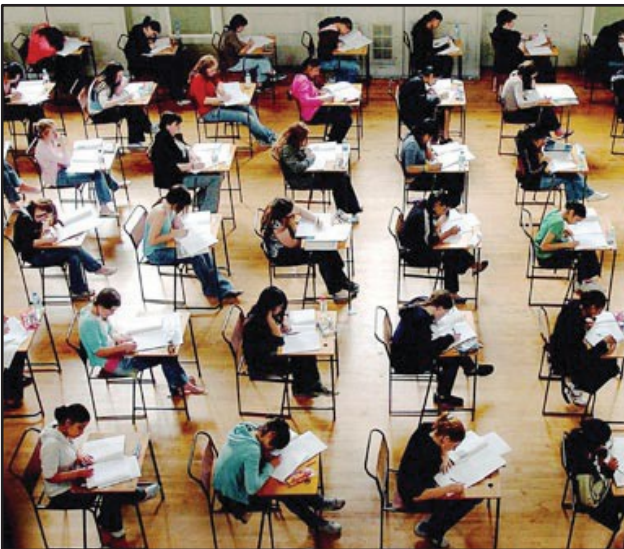
These institutes often work as a consortium, and their collaboration makes it possible to share scientific events and organize research programs with public and private research institutions, as they do with TIGEM.

Funding at TIGEM

TIGEM is a daughter research centre of the Telethon Foundation. Telethon's funding covers structural and research costs and is renewed every five years through a rigorous revision process, comprising the individual evaluation of research projects and a site visit to the Institute conducted by an international group of scientists. Investigators at the Institute are funded by prestigious international funding agencies such as the European Union (ERC), the National Institute of Health, the European Molecular Biology Foundation (EMBO) and the Wellcome Trust, among others. Furthermore TIGEM has sealed an alliance with Shire and BioMarin, two leader pharmaceutical companies, yet remains fully independent in its self-governance and research programs. Through the years, the Institute has played a leading role in EU-funded research and has been part of many EU-funded consortia.

TIGEM PhD Programmes

Students are a vital resource at TIGEM, as they are the main vehicle driving the quality of research and by contributing to the formation of an inspiring scientific environment.

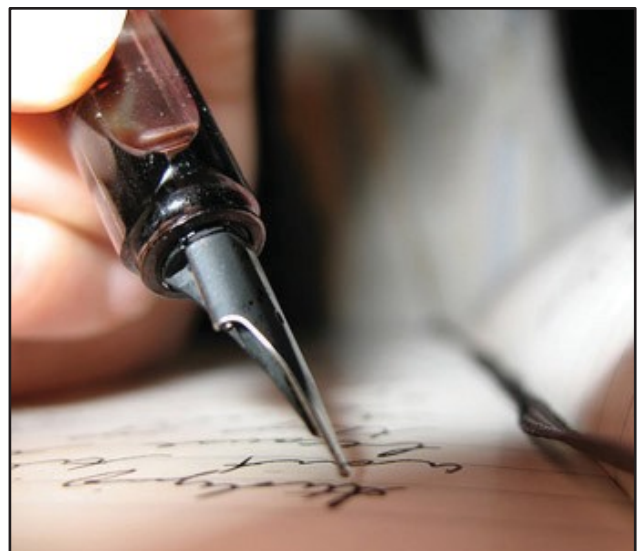


Graduate students may join TIGEM by way of two different PhD programmes designed for self-motivated science graduates wishing to become scholars in the medical sciences. The Open University (OU) programme offers preparation in those planning a career in human genetics, and that of The European School of Molecular Medicine (SEMM) in Systems Medicine.

For all graduate programmes, TIGEM functions as a sponsoring establishment, providing enrolled students with laboratory spaces, facilities, resources, and educational activities. The university of enrollment, instead, represents the Research School and awarding body, conferring the student with the final (UK (OU) or Italian (SEMM)) PhD title. There are currently 20 PhD students enrolled at TIGEM, and 137 students have successfully completed their

graduate studies at TIGEM since the programmes were created.

The establishment of international programmes at TIGEM is of great value because they enhance the prestige of the Institute abroad and allows us to recruit international talented students. This offers TIGEM researchers and students the opportunity to interact and collaborate with foreign entities. Notwithstanding, TIGEM also collaborates with local regional universities such as the University of Salerno (UNISA) to promote additional graduate training opportunities for Italian students.



PhD Programme in Human Genetics



The Open University

Goal

The goal of the PhD programme is to prepare doctorate students for research careers in all aspects of human genetics.

Length:

3 (+1) years

Research School/ Awarding Body

The Open University (OU), Milton Keynes, UK

Affiliated Research Centre:

The Telethon Institute of Genetics and Medicine (TIGEM), Naples, IT

PhD Programme Coordinator

Sandro Banfi

Training

TIGEM's PhD curriculum provides coursework, high-tier scientific seminars, and an annual data club programme. Moreover, PhD students also participate in the two-day TIGEM retreat, which is organized every 18 months, and includes scientific presentations, poster sessions and intense dialogue about new discoveries and research strategies.

Supervision and Mentoring:

Graduate students are constantly monitored throughout the graduate curriculum. Each student is assigned a supervisor (or tutor), a co-tutor (optional) and an external supervisor with whom the student meets periodically to discuss his/her research progress. A written report, as well as an oral presentation, summarizing the student's progress in thesis work, is due at the end of each year, according to OU and TIGEM requirements. Moreover, a meeting with an ad hoc on-site Thesis Committee composed of staff members and with a Third Party Monitoring member, who are both appointed to help students in their path towards obtaining PhD, are also compulsory and scheduled on an annual basis.

Working language:

English

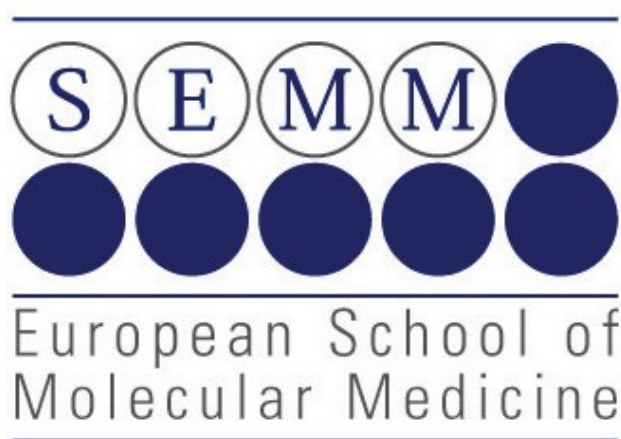
Fellowship

Financial support is provided to all the students participating in the programme, either by TIGEM fellowships or external grants for the perspective three (or four)-year period.

Key dates

- Application: May/June
- Examination: September/October
- Starting date: October/November

PhD Programme in Systems Medicine

**Goal:**

The aim of the PhD programme is to promote the training of young scientists in the field of post-genomics and molecular medicine, with a particular focus on genetic diseases.

Length:

4 years

Research School/ Awarding body:

Università degli Studi di Napoli Federico II, Naples, IT

Affiliated Research Centre:

The Telethon Institute of Genetics and Medicine (TIGEM), Naples, IT

Collaborating Establishment/s:

The Centre for Genetic Engineering (CEINGE), Naples, IT
The Stazione Zoologica Anton Dohrn of Naples (SZN), Naples, IT

PhD programme Coordinator:

Diego di Bernardo

Training:

The sponsoring establishment provides coursework, a high-tier programme of scientific seminars, and an annual data club programme throughout the graduate curriculum. Moreover, graduate students also participate in the two-day TIGEM retreat, which is organized every 18 months, and includes scientific presentations, poster sessions and intense dialogue about new discoveries and research strategies.

Supervision and Mentoring:

PhD students are constantly monitored throughout the graduate curriculum. Each student is assigned a supervisor (or tutor), an internal tutor and an external supervisor with whom the student meets periodically to discuss his/her research progress. A written report, as well as an oral presentation, summarizing the student's progress in thesis work, is due at the end of each year, according to SEMM and TIGEM requirements. Moreover, a meeting with an ad hoc on-site Thesis Committee of staff members, who are appointed to help students in their path towards the obtainment of their PhD, is also compulsory and scheduled on an annual basis.

Working language:

English

Fellowship:

All the students participating in the programme are supported either by SEMM fellowships or external grants for the perspective four-year period.

Key dates:

- Application: June-August
- Examination: September
- Starting date: October (1st day of the month every year)

PhD Programme in Human Genetics - Study Plan

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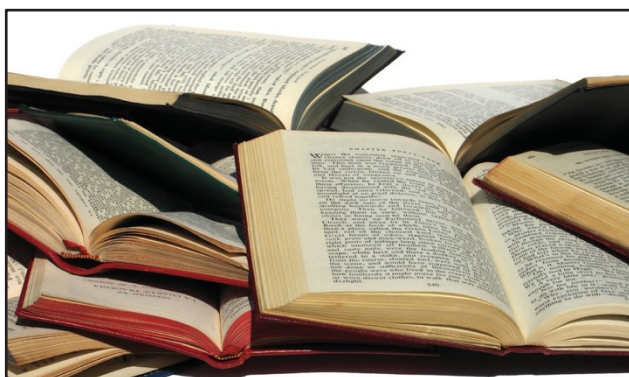
Study Plan at a Glance

Study Plan at a Glance

Admission procedure

All PhD programmes are open to both national and foreign students with no restrictions on nationality, gender, culture, race or religion. Suitable applicants will be evaluated based on their academic achievement, scientific potential and motivation to excel in the doctoral programme, and letters of recommendation. All the graduate students admitted to either of the two PhD programmes are supported by a fellowship for the perspective three- or four- year period.

Which graduate programme to choose

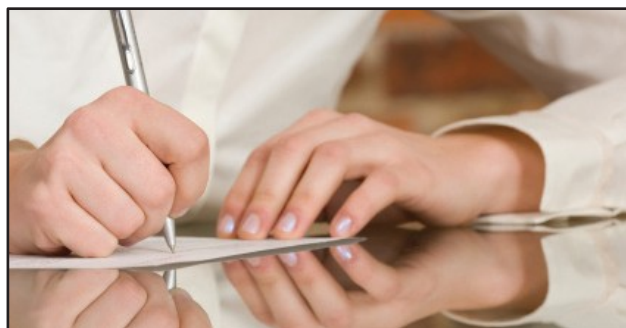


Although TIGEM's PhD programmes are addressed to both national and foreign students, the OU PhD programme awards the student with a UK PhD title and is designed for foreign students wanting to spend their PhD study period in Italy or for Italian students with the goal of obtaining a PhD title that is recognized internationally and thus favours mobility to a foreign country. Supervision and mentoring throughout the student's graduate studies, as well as doctoral thesis writing and oral defence are necessary according to UK PhD standards.

The SEMM PhD programme may include both Italian and foreign students since the selection process, and supervision and mentoring of the student throughout the graduate studies, follow international standards.

After the course of the programme, students are conferred with an Italian PhD title.

Eligibility



All applicants must hold a second level academic degree, the Italian Laurea Specialistica, Master of Science, or equivalent, from an accredited university or college before matriculation.

Applicants expecting to be awarded their university degree can also apply, provided that they receive the award before the examination date on to avoid exclusion from the selection.

The following list of scientific academic degrees are considered valid for application:

- Bioinformatics
- Biological sciences
- Biomedical engineering
- Biotechnology
- Chemistry
- Dentistry
- Electronics engineering
- Human nutrition sciences
- Medicine
- Natural sciences
- Pharmacy and industrial pharmacy
- Physics
- Veterinary medicine



How to apply

The admission procedure requires the submission of an application, regardless of which PhD programme the student ends up choosing. The application period is launched every year through the TIGEM web site (www.tigem.it) where interested students can find all the details relevant to the application process (application forms, templates of letters of recommendation, important notes, etc.), as well as other information of interest.

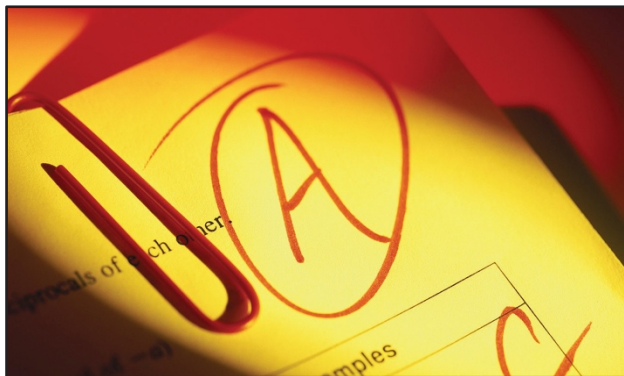
Letters of recommendation are an essential part of the application for most graduate programmes. TIGEM prefers recommendations from instructors who are familiar with the applicant's work and with the field in which the candidate expects to study.

The specific details on how to apply are described in the following pages and are given separately for each graduate programme.



OU PhD programme

Candidates applying to the OU PhD programme should submit the application form and two reference letters, by mail or by e-mail (see contact details below; for the application form and reference letter template please consult the TIGEM web site).



Applicants should inform their referees of their application to the OU PhD programme and provide them with the appropriate template, which can be downloaded directly from the TIGEM web site. Reference letters sent by mail should always be placed in sealed envelopes and signed by the referee on the back of the envelope. These may be sent together with the application form (by the student), or separately (by the referee). Reference letters may also be sent by e-mail, but only by the referees, as indicated below.

Valeria Rotoli
TIGEM
Via Campi Flegrei, 34
80078,
Pozzuoli,
Naples
Italy
Fax: +39-081-19230651
e-mail: phd.program@tigem.it

It is important to remember that:

1. Applications not accompanied by reference letters will not be taken into consideration;
2. It is the applicant's responsibility to ensure that the referees fill in the appropriate template and send the letters within the deadline for application;
3. Letters of recommendation may also be sent directly by the student, together with the application form, provided that the student receives the reference letters from the referees in a pre-sealed envelope, signed on the back by each referee.

SEMM PhD programme

To apply at this PhD Program you must have at least a B2-level English proficiency certification. Applications are exclusively on-line, as indicated on the SEMM web site (www.semm.it). The application consists of the application form (which can only be submitted online during the application process) and two reference letters (which can only be submitted through the online system; during the application process, the applicant is required to provide the email address of the referees, who will then receive an automatic email with the instructions for the letter submission. After each letter submission, the applicant receives a confirmation email. It is the applicant's responsibility to ensure that the referees send the letters within the application deadline). The application runs through June to (the exact date is only available at the time of application, please consult the SEMM web site).

It is important to note that:

1. Applications not accompanied by letters of recommendation will be discarded;
2. The applicant should make sure the referees send the letters within the deadline for application.

Selection



Admission to the programme is granted on a competitive basis and on the student's distinguished ability to excel in the doctoral programme. A pre-selection generally takes place soon after the deadline. An ad hoc examination board screens the applications received and short lists candidates on the basis of the information provided in the application form and in the reference letters. The examination board attributes a score to each candidate; a minimum score is required to be admitted to the graduate programme.

Only pre-selected candidates are invited to TIGEM or to the Research School and admitted to the examination process. Candidates must be in

possession of a legally valid ID card or equivalent to take the exams. The examination differs slightly among graduate programmes.

Details are given separately for each graduate programme.

OU PhD programme

The exam is held in English and consists of:

- an oral seminar on the experimental work carried out by the candidate during his/her undergraduate studies, or the results of the student's past experience in a research laboratory;
- individual interviews with TIGEM group leaders as an assessment of the applicant's scientific potential and motivation.

Knowledge of English is assessed during the interviews and seminar presentation.

Please note that selected candidates will be put forward to The Open University for registration. Furthermore the registration for the OU PhD in our Affiliated Research Centre is subject to Open University approval.

SEMM PhD programme

The exam is held in English and consists of:

- a written exam (multiple choice test) on general aspects of biochemistry, molecular and cell biology, and genetics;
- an oral exam aimed at evaluating the scientific background of the candidate, as well as his/her motivation to succeed in a demanding programme.

Knowledge of English is assessed during the written and oral exams.

Individual interviews with TIGEM group leaders or faculty members are held after the oral examination as an additional assessment of the applicant's scientific potential and motivation.

Fellowship

All graduate students admitted to either of the two PhD programmes are supported by a fellowship provided by the Institute or the Research Schools for the perspective three- or four year period. The current annual, gross salary for doctoral students studying at TIGEM is, on average, between 18.000,00-20.000,000 euro/year.

All PhD students enrolled at TIGEM, irrespective of the programme they join, are expected to participate in the following aspects of the PhD course.

Training

- Attendance at thematic courses during the first three years of their study;
- Participation in high-tier scientific seminars on an annual basis;
- Participation in an annual programme of data clubs.
- The three-day course for third-year students covers CV preparation, presentation skills and science communication.

Research

- Perform full-time research activity in a TIGEM research group.

Review

- Submission of a PhD thesis proposal at the end of the first year of study;
- Submission of a 6-monthly written report;
- Presentation of their data and research progress in a lab data club once a year;
- Meet with the on-site Thesis Committee on a yearly basis.
-

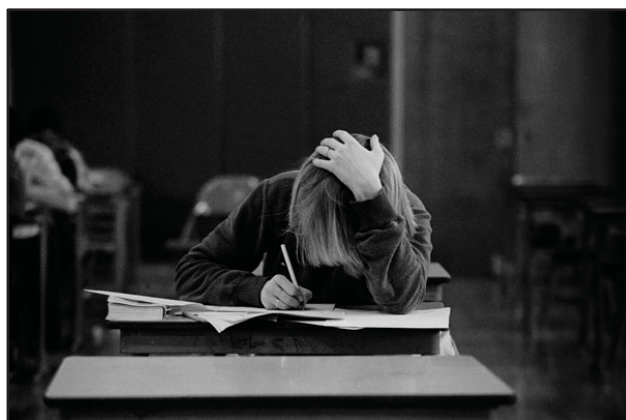
Courses

The courses, held during spring/summer term, are taught by TIGEM faculty members.

- The two-day induction course (only for first-year students) provides information regarding experimental design and ethics, covering the basics for students approaching research. The first year is crucial for graduate students as during this period they outline their working research hypothesis and develop the methodology and critics that will help them in conducting a doctoral research project;
- The four-week course (course A and B) rotates

every two years. The four-week course covers four different topics, one topic administered per week. Three of these course topics are theoretical, and the other, practical. At the end of each week the students take a test on the topic of the week, to check is own progress..

The courses are compulsory, and students are required to pass each test, with the penalty of repeating the course in the case of unsatisfactory results.



Seminar series

TIGEM organizes scientific seminars, often in collaboration with other scientific and medical institutions, from September to July. The presentations are given by the research staff within TIGEM and invited speakers, which are internationally renowned in various fields of biological sciences, including human genetics.

Data clubs

The data club programme also runs from September to July and occurs once a week. Data club is designed to improve the students' language and presentation skills and encourage exchange of information, discussion and collaboration between lab groups. Students are expected to attend data clubs weekly and to present their data annually. They receive feedback from the Thesis Committee after their presentations. Each data club session is chaired by the speakers of the previous week. In this way, students can also improve their discussion leading skills.



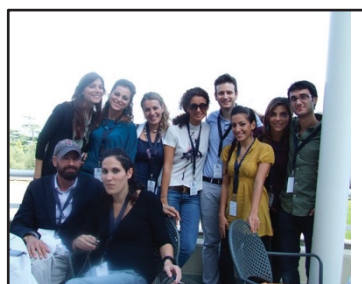
Web resources

A general description of the graduate programmes offered at TIGEM, including the admission procedure, is available on the TIGEM web site (www.tigem.it)

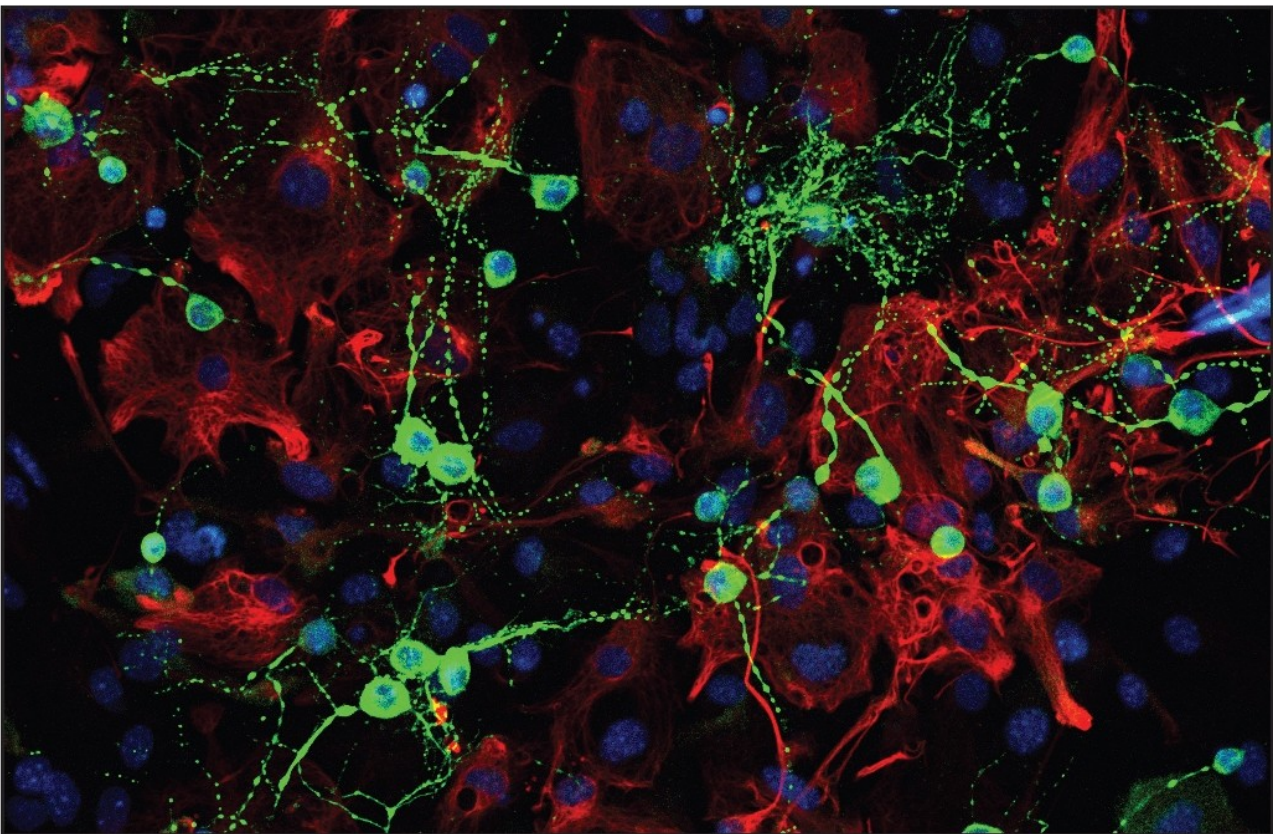
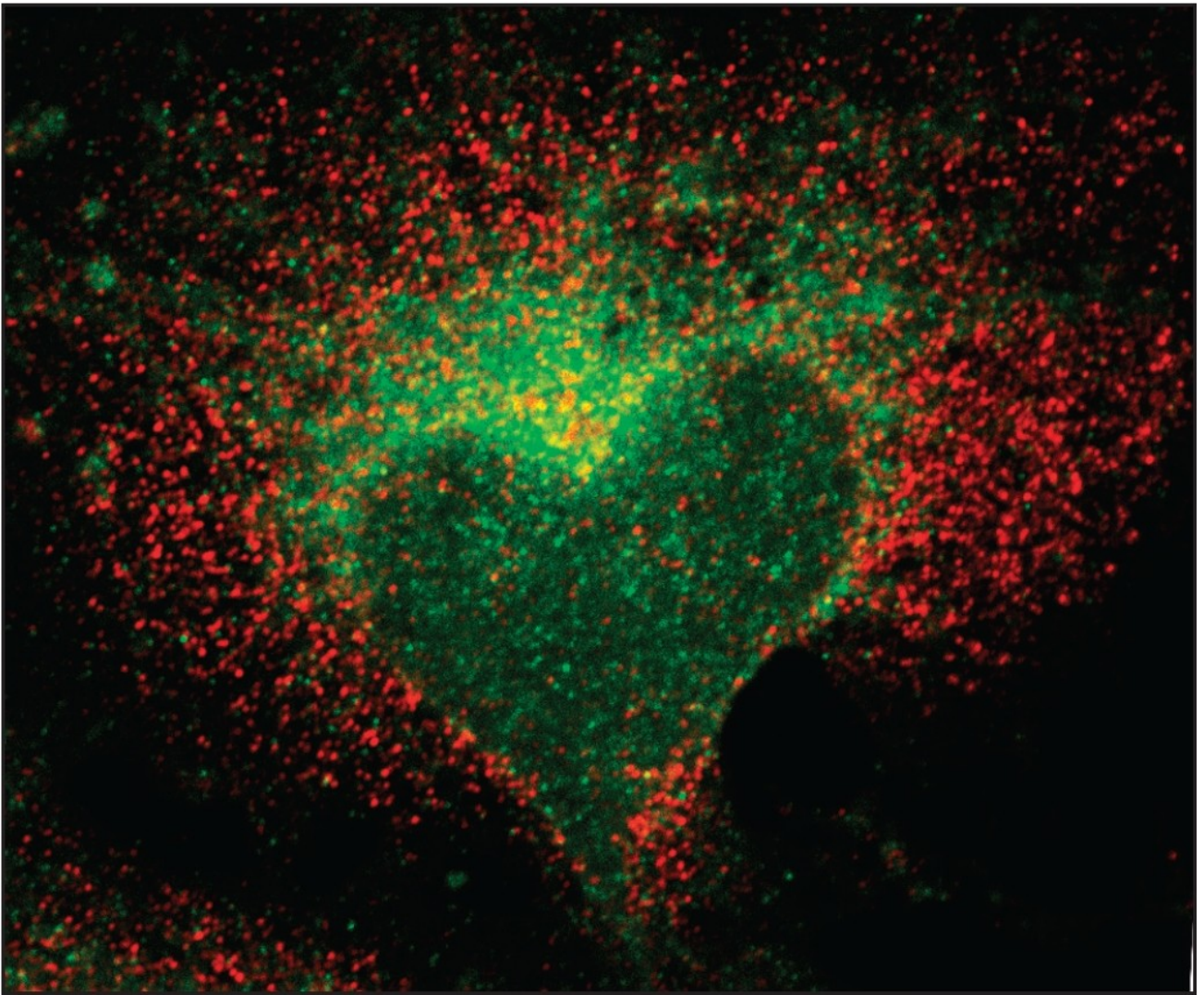


Retreat

PhD students also participate in the TIGEM retreat, which is organized approximately every 18 months in an informal setting to encourage scientific exchange, communication and bonding among members of the research staff. The retreat lasts two to three days and includes scientific presentations, poster sessions and special activities. The last TIGEM retreat, was held in Rome.



Induction course (only 1 st year)		
Introduction to scientific methodology and Experimental design		
Ethics In Science		
Presentation skills		
4-weeks courses (1 st and 2 nd year)		
A	B	
1. Cell Biology of Genetic Diseases		
Membrane trafficking	Cell signalling	
Title	Title	
The drug discovery process - the special case of cystic fibrosis	Sorting out membrane traffic to the lysosome: Ubiquitin, ESCRTs and protein quality control Systems	
Endomembrane signaling in metabolism	Cellular Dynamics imaged in real time and in 3D: the mechanism of intraluminal vesicle formation mediated by ESCRT-III and Vps4	
Membrane trafficking and cell polarity	Lysosome Function, Dysfunction and Neurodegenerative Disease	
Cellular environment: implication for brain development and maldevelopment	The RNA world	
Mass spectrometry based proteomics	Expanding the functions of autophagy regulators to the nucleus	
2. Systems Biology and Functional Genomics		
Statistics and bioinformatics	High-Throughput Genomics and quantitative biology	
Statistical Analysis of data	Introduction to Next Generation Sequencing	
1) Network Biology I e II 2) Functional Genomics	Quantitative microscopy	
SINEUPs: a new functional class of antisense long non coding RNA s that activate translation	Polymer models of chromatin organization	
Shaping the blood: Lessons from chromatin and single cell RNA dynamics	Transcriptomics for the investigation of cellular mechanisms and drug discovery	
Understanding Cellular Heterogeneity” an abstract is attached		
Investigate cell fate decisions through integrative genomics		
3. Molecular Therapy		
Gene therapy	Therapy for genetics diseases	
Adeno-associated virus (AAV) vectors-based Gene Therapy	Gene therapy for inborn errors of metabolism	
Retrovirus/Lentivirus vector-based vectors Gene Therapy	Towards Clinical Translation of Safe and Effective Hematopoietic Stem Cell Gene Editing for the Correction of SCID-X1	
Adenovirus-based Ad) vectors-based Gene Therapy	Therapies for lysosomal storage diseases	
Clinical trial1: GMP-vector production & Regulatory Aspect	Gene therapy for retinal degeneration: rods, cones and rod-like cones	
Clinical trial2: Clinical Protocol; exam		
4. Practical Course		
Medaka fish as model system for biomedical research	Vector production	
Advanced light microscopy in modern biomedical research.	Electron microscopy in modern biomedical research	
Development and validation of cell- based high content imaging assays	Introduction to the behavioral genetics	
NGS technology and application	NGS Analysis of WES in families	
3-days courses (only third year)		
CV writing		
Presentation skills		
Fellowship/Grant Application		



During the first year of the curriculum, it is crucial for the students to rapidly adjust to the intensive educational experience that lies behind obtaining a PhD degree.



Soon after matriculation students are matched with a supervisor and assigned to a TIGEM research group in which they conduct their research project throughout their PhD studies.

The month of matriculation depends on the graduate programme the student joins, and is generally October for SEMM PhD programme (PhD programme in systems medicine) or January for OU PhD programme (PhD programme in human genetics).

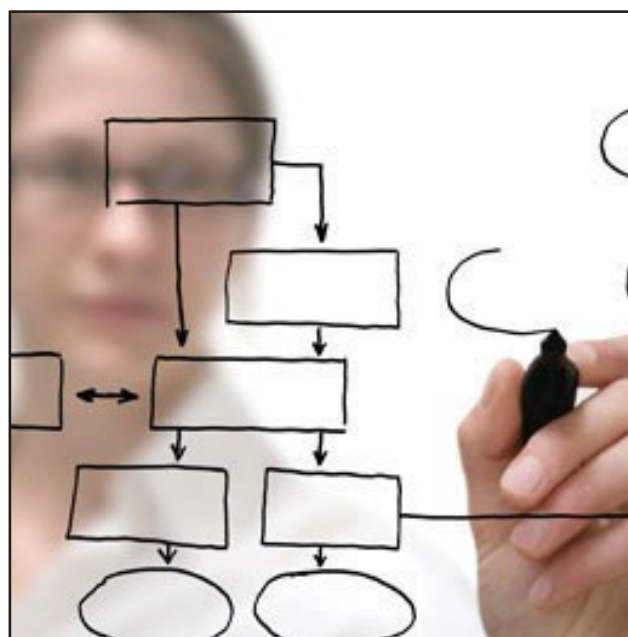
The first months are spent settling into the research group and laboratory and interacting and making friends with other colleagues and research members. Doctoral research and educational activities are carried out from the very beginning. During the subsequent spring-summer term students take the induction two-day and four-week courses.

By the end of the first year, students draft their PhD thesis proposal in which they introduce their topic and working hypothesis and outline the experiments to be executed during the subsequent three or four years. The thesis proposal is composed of the following sections: title, abstract of research, specific

aims, background, preliminary data (if available, not a requirement), methods, and references. The total length of the proposal, including references, should not exceed 10 pages. The experiments proposed should be rational and feasible to be performed in a suitable time frame. For each proposed experiment, students should discuss difficulties, possible outcomes and interpretations of the results.

Students are also expected to orally present their research proposals, which is evaluated by an ad hoc Thesis Committee, composed of the student's supervisor and two other participants (TIGEM group leaders or mature post docs).

The oral defence consists of a short presentation followed by a discussion during which the student is asked to respond to questions raised by the commission to establish the student's depth of knowledge and analytical ability on the thesis topic. Usually, this presentation coincides with a TIGEM data club, and therefore consists of a first session open to all TIGEM researchers and of a second session



whose participation is restricted to the student and the thesis commission.

If the thesis commission considers the proposed experiment inadequate the study will be asked to rewrite the proposal, or in extreme cases, resign. It is important to underline that the proposed experiments, at the time of thesis proposal submission, and even the project per se, may change during the following years of the student's curriculum depending on the results obtained.

Students are also assigned an external supervisor from the OU, with whom the student meets at the end of the first year of study to discuss their progress and future strategies and individual problems with the on-site Thesis Committee.



Only for OU students

- Probationary Period Report. Only OU students are expected to submit a Probationary Period Report at the end of the first year (11 months after matriculation) that substitutes and is equivalent to the PhD Thesis Proposal discussed above. According to OU requirements, the "Probationary Period Report should involve a major review of the student's progress to determine which degree s/he is to be registered for, i.e. PhD or MPhil". Students and supervisors will be informed by the Research School and provided with the appropriate forms for the Probationary Period Report submission. Moreover, before the submission of the Probationary Period Report, OU students must also conduct an oral discussion (mini-viva) in which they defend their research project and progress, to the members of the Thesis Committee (excluding the student's director of studies) or to two other TIGEM investigators. The mini-viva can be considered as an "exercise" for the student's final defence at the end of his/her graduate studies.
- The skills audit/portfolio. The skills audit is only compulsory for OU students and should be completed soon after the PhD project commences. It consists of a series of questions, each relating to particular skills or sets of skills that the student may already have acquired in past experiences, skills with which the student may need additional training before using that skill, or even skills irrelevant to the student's particular research project. The Research School provides the student with a date by which the audit should be completed, upon the supervisor's agreement, and then the audit is submitted to the student's ePortfolio. The student's ePortfolio is a disk storage that keeps a record of skill achievements. It may contain items such as literature reviews, copies of presentation slides and certificates for laboratory safety courses. After consulting with the supervisor, the student should complete a benchmark assessment of his/her research skills in the categories specified. The student and the supervisor can then decide what training is required to gain the skills necessary for the PhD project. As the student acquires skills, s/he should upload records of those achievements into the ePortfolio. At regular periods (every 6 months), the student's supervisor will view and verify the content of this ePortfolio. Having verified the evidence is appropriate; a statement outlining the generic transferable skills that the student has acquired throughout the PhD will be provided. This may aid the student in preparing CVs for job interviews and helping with the identification of the next career step.
- Third Party Monitoring. The Open University has created a system of support called the Third Party Monitoring, which is a research staff member that meets with the student once a year to 1) provide pastoral support to the student outside of the supervisory environment, 2) identify and resolve potential problems, including resource management problems (staff time, equipment, technical issues, etc), and, more importantly, 4) provide an impartial view if difficulties arise.

After fulfilling the requirements of the PhD Thesis Proposal, concentrate their efforts on their research project. Nonetheless, course work continues throughout the course of the programme. During the spring/summer terms the students will follow the four-week courses and attend seminars and data clubs.



Students' research progress continues to be reviewed annually by the Thesis Committee and by the students' external supervisors.

Students present a written report every six months, in which they describe results accomplished, problems encountered and changes envisioned to remedy those problems, and finally future research strategies and possible outcomes of the doctoral research project.

The Thesis Committee always remains a point of reference for the student throughout his / her

studies. As in the first year, the student is expected to discuss the data and developments of his/her doctoral research project in a public session (generally a TIGEM data club) once a year and in the presence of the committee members, followed by the feedback session restricted to the student and the thesis committee.

Meetings with the student's external supervisor and Third Party Monitoring staff member (OU students only) are also scheduled.

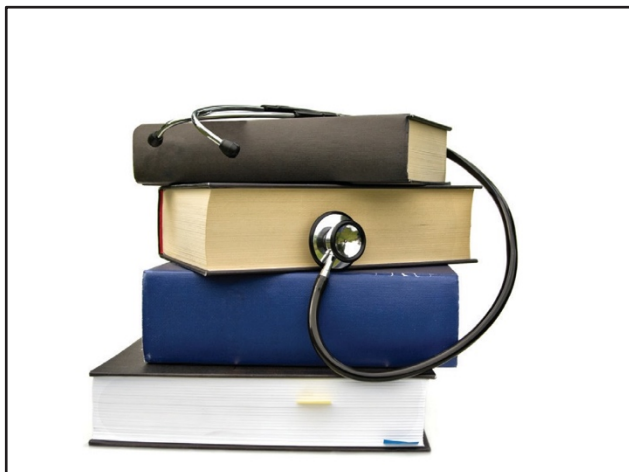


Only for OU students

OU students are expected to present an annual written report, submitted in February. Students and supervisors will be informed by the Research School in due time of the deadline and provided the appropriate forms.

Third and Fourth Year

The third- and fourth-year PhD student is expected to follow seminars and data clubs from July to September. During the spring/summer terms the students will follow the three-day “Complementary Skills” course. During the third year, if the student fulfills the requirements as evaluated by the commission, s/he will receive the



official authorization to write his/her doctoral thesis. Third year SEMM students must also present their doctoral research progress as a written report to an *ad hoc* commission composed of their Supervisor, Internal Supervisor and External Supervisor. OU students continue to refer to the Third Party staff for support.

During the last year, students submit a written doctoral thesis, which is evaluated and then presented and discussed in a public lecture attended by the supervisors and an External Examination Committee. Students are awarded the PhD degree after successful thesis defence. A high quality publication of the experimental work carried out during the research period, or at least submission of a manuscript concerning such work, is required prior to the discussion of the doctoral thesis.

Only for OU students

The oral defence of the doctoral thesis is restricted to the student, the student’s supervisor (optional, s/he may only act as observer), and the Examination Panel. The latter is nominated by the student’s supervisor following the appointment criteria regulated by the Research School, and must be approved by the OU Research Degrees Committee before the student submits his/her doctoral thesis.

A satisfactory thesis defence and fulfillment of the necessary credit workload for educational activities results in the award of the PhD degree.



Extracurricular activities

For a PhD student, life at TIGEM is much more than attending courses, seminars and data clubs and performing full-time research. The Institute involves students in many extracurricular activities, such as the “TIGEM Open Doors” event, which is organized to increase public awareness on research in genetic diseases. Students often get together outside of work to spend an enjoyable evening at Naples’ San Carlo theatre, the cinema, or at a favorite local restaurant or pizzeria to enjoy the exquisite Neapolitan cuisine.

“TIGEM Open Doors”

As the name of the project implies, the primary objective of the event is to open the Institute to others, welcoming local primary, secondary and

high school students to promote Telethon’s mission by participating in science dissemination of research and by aiding researchers in defining their research activities.



The event “TIGEM Open Doors” is a unique opportunity for PhD students and PostDocs to talk to a younger crowd about biology and medicine. Basic scientific knowledge is provided to students with a brief intro, generally presented by a TIGEM senior PostDoc. The presentation is then followed by a visit to the laboratories, such as the microscopy, DNA and cellular lab centers, where, under the supervision of PhD students, primary, secondary and high school students can explore the equipment and techniques generally used in a research laboratory.

From this experience, PhD students learn how to explain their research in more simple terms, and in general, how to communicate to a broader audience, which is crucial for public speaking, a necessary prerequisite for a future research or teaching career. TIGEM dedicated ten days in 2017/2018 to this important and popular event. The event saw the participation of roughly 2000 students coming from schools of Naples, the Campania region and the whole Italy.

Fund raising

Throughout their studies, PhD students may also be involved in fund-raising for research in genetic diseases through the solidarity marathon, which has been broadcasted on national Italian TV annually since Telethon's inception in 1990; the Telethon Foundation has taken its name from this event, formed from a portmanteau of television and marathon.

Telethon: the Television Marathon

Fund-raising for research in genetic diseases, by way of the solidarity marathon broadcasted annually on Italian RAI TV channels for 7 days in December has been behind Telethon's success ever since its foundation. In fact, the Italian Telethon was the first solidarity marathon to have ever been presented on Italian television, increasing public awareness of human genetic diseases, and in particular on rare genetic diseases, the most neglected by pharmaceutical companies and the local government. Although largely inspired by former initiatives (Telethon USA, Telethon France, more details given in the official Telethon web site: www.telethon.it), the activity, organization and management of the Italian Telethon Foundation, and of its four intramural

research institutes, is completely independent from the other non-profit organizations.

Since 1991, Telethon Italy has invested over 300 million euros in research and funded 2,261 research projects on more than 400 human genetic diseases, which range from basic research to clinical trials.

The research portfolio includes intramural research (performed in four institutes, including TIGEM) and extramural research through grants to universities, public and non-profit research institutes in Italy.

In Telethon's 7-day TV marathon research staff and students from TIGEM participate in TV programmes and talk shows. Researchers and students have the chance to talk about their research activities, illustrating the current progress and future potential in this research field in a setting completely different from the laboratory, and to a much broader audience. The heart of the TV marathon is the collection of Telethon's "short stories", produced by professional movie directors, which describe the life of courageous patients living with genetic diseases. These stories, as well as the interviews with Telethon researchers and students, help to increase public awareness of the social burden and struggle associated with living with genetic diseases.

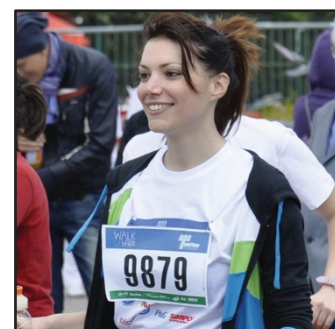


Walk of Life

Since 2012, Telethon has held its Walk of Life yearly as another initiative in battling rare genetic disease. Walk of Life offers another opportunity to gather funds for the precious research and resources needed in order to provide a possible cure for these diseases and spread awareness to support those who are affected. This event is organized to attract participants of all types. There is a 10-kilometer race for those who want to participate in a competitive manner, and there are the non-competitive 3- and 5-kilometer walks for those who simply want to enjoy the day and reflect calmly on the sentimental meaning of the event. Walk of Life spans over a period of a few weeks, divided into different phases that attract other organizations with the same interests of the Telethon Foundation. Telethon is thus able to collaborate with these organizations with a common goal: to fight against rare genetic disease.

While Telethon works year round to promote its mission in helping those with genetic diseases, this event offers yet another opportunity to put these disorders into the spotlight. Between 6-8 million of these rare diseases exist, but unfortunately, they often overlooked by private and public pharmaceutical companies and research organizations. Walk of Life thus represents a race against time; the quicker we can spread awareness, the faster we can delivery cures to those that suffer from rare, often lethal, genetic diseases. Telethon holds this event all over Italy and makes it possible for people in different cities to join this effort. Telethon dedicates each Walk of Life in participating cities to specific patients who have bravely shared their stories. It allows those participating in the walk to feel connected to those whom they are helping and to feel personally responsible for the wellbeing of those around them. Participating in the Walk of Life is simple. To join

one needs to make a small donation of 10 euro or 5 euro for children up to 12 years of age at a sign-up center near them. One small thought, when put together with many others, becomes a large force against genetic diseases, and allows our researchers to continue examining and solving the mechanisms behind these disorders. Thus Telethon not only relies on its researchers in reaching its objectives, but the generosity and empathy of those that participate in these events.



Students Thoughts



Alessia Romano
SEMM PhD student
Year of enrollment: 2012

When I started working at Tigem as a research fellow, I had just come back from an experience abroad. Quickly I came to appreciate TIGEM's international environment of this institute its efficient organization. So I decided that TIGEM was the institute where I wanted to do my PhD and I applied for one of its PhD programs. Since starting my PhD studies here I am convinced that I made the right choice. At Tigem I have the possibility to work on a very stimulating research project and the opportunity to learn something new everyday. All the researchers are very happy to share their knowledge with younger students and are always available to discuss your results and to give you precious advice. Furthermore everyone at Tigem is really friendly and welcoming, encouraging newcomers to join its big family.



Sara Barbato
OU PhD Students
Year of enrollment: 2012

The PhD program at TIGEM is a great opportunity for young aspiring students to discover what it means "to do research" and start a scientific carrier. The Telethon Institute gives students the opportunity to work in a friendly, young environment, which is educational and challenging and at the same time. Thanks to the Institute's international links and collaborations, students also have the opportunity to spend part of their PhD programs in a foreign country, allowing them to experience new cultures and work environments. Studying as a PhD student at TIGEM has made me realize that a PhD degree acts as a springboard not only for a career in research, but also for life in general.



Simona Ventre

SEMM PhD student

Year of enrollment: 2011

I am a PhD student of Systems Medicine working at Telethon Institute of Genetics and Medicine (TIGEM). My childhood interest in science prompted me to pursue a career in research. My interest in research stemmed in part from my love for science magazines, specifically from how they envisioned future possibilities in genetic engineering, and from my curiosity about the possibility of helping others people with my work.

I have had my best work experience at TIGEM, as it gave me the incredible opportunity to work on a very interesting internationally-funded project and offered me a multicultural and stimulating environment and a sense of fulfillment in life. At TIGEM research is held at very high standards, and while it is challenging, it is not just a job. Here the work environment is very informal and friendly. Our lab group leaders are very young and offer a helping hand in giving suggestions for PhD students' projects. The institute offers its students many science-related activities and training, such as lab meetings and data clubs. Our meetings and data clubs are designed to help us practice sharing our data; it is very important for us to strengthen our self-confidence and presentation skills, so these presentations give us the possibility to receive precious feedback and suggestions to overcome technical and conceptual issues related to our scientific projects. Every week at TIGEM we welcome guests renowned internationally in the sciences to give scientific seminars. These weekly occasions allow us the opportunity to meet scientific leaders from all around the world and help us to determine which fields we are interested in. Moreover they inform us of the current leading research and shed light on the research institutes involved in the similar projects, a fundamental step

toward a good scientific networking and a promising career.

TIGEM offers advanced scientific instruments and equipment, making it possible to perform high quality experiments and take advantage of great computer and scientific assistance and many specialized core facilities that provide scientific and technical support for our experiments.

Since we are fortunate to have a family-like work environment, we often arrange outings outside of work so that we can spend some of our free time together. Our institute's location, Naples is the crown jewel of Southern Italy, and here the people are friendly, welcoming, and always willing to help. The unique and picturesque city in which TIGEM is located offers us the possibility to a cultural experience as well, from the beauty of the sea to the amazing typical food of this area.



Francesca Zappa

OU PhD student

Year of enrollment: 2012

The PhD program at TIGEM is a great opportunity for young aspiring students to discover what it means "to do research" and start a scientific carrier. The Telethon Institute gives students the opportunity to work in a friendly, young environment, which is educational and challenging and at the same time. Thanks to the Institute's international links and collaborations, students also have the opportunity to spend part of their PhD programs in a foreign country, allowing them to experience new cultures and work environments.

Studying as a PhD student at TIGEM has made me realize that a PhD degree acts as a springboard not only for a career in research, but also for life in general.

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Explore the web page: www.telethon.it

