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Table of Contents

List of Tables

1. Project Overview

MedGeNet is a project intended to expand the human expertise in clinical genetics and cancer genetics in our Mediterranean Partners Countries (MPCs), through the transfer of knowledge and technology between the Northern and Eastern/Southern rims of the Mediterranean which share a common burden of Genetics diseases.

Three main areas covered are:

- The creation of an Euro-Mediteranean network for Genetics Telecounselling and Telepathology together with the usage of the most advanced online database software and web-conferencing tools has permitted remote live interaction and collaboration between geneticists from European Countries and MPCs, this has reduced the need to hold face to face meetings, and reduce overall travel budget.

- Information and Communication Technology: the selection and installation of state of the art technologies in MPCs facilitates the cooperation in the field of Medical Genetics, the ongoing progress of collaborative research, the exchange in diagnostic best practices, information and protocol, the sharing of critical case histories facts and the immediate and permanent data accessibility through the build up of Internet based databases. Furthermore, an electronic microscope capable of scanning and storing thousands of tissue and DNA images technologically at the leading edge, provided to a Tunisian Partner Hopital Charles Nicolle (P13 HCN). It was the first step in creating the EuroMediterranean Telepathology network which is a big step forward in the overall diagnostic capability available at disposal to the medical staff.

- Improving the know-how of caregivers, using to the Remote Training Centres Network. Through the transfer of the needed technology and the establishment of didactic and logistic requirements set by the European experts, in MPCs the new centres have the chance to permit to their physicians and medical personnel to attend highly specialised courses in the field of Human Genetics. Completing the range of remote training items offered to partners, 5 E-learning Courses are available on the project portal. These course provide students in MPCs an increased awareness about the most relevant, innovative and updated findings achieved in the dynamic genetics domain. Thanks to MedGeNet professionals working in MPCs can update their scientific know-how at the same speed as their European colleagues, even though training centres are not physically located in their home countries.

In parallel to these 3 main areas of development, the project has also carried out an international research collaboration aimed at the scientific upgrading of an important Thalassemia diagnostic tool, the Thalassochip (based on the Arrayed Primer Extension technology), which thanks to the collection of DNA samples featuring relevant genetics variants collected in 6 different countries, the technical work performed by Asper Biotech (located in Tartu, Estonia) and the validation provided by CING (Cyprus) now supports 90 Talassemic mutations which is an enormous improvement over the previous 41, which means a screening potential raised by 120%. The overall activity results was disseminated through an international workshop totally organised by CING to transmit the results of the research and the renewed screening method to the scientific community.

Finally, the goal of dissemination was to develop a general awareness of the project achievements to Arabic speaking Mediterranean populations Genetics counselling free opportunities and prevention

basic rules within the familiar environment. To achieve these objectives dissemination products and events arranged by the Université Saint Joseph (Lebanon).

The dissemination activities were crowned whit a dedicated genetics conference held in Beirut, in a specific educatio nal DVD, in 2 different arabic pamphlets in Arabic addressing general issues touched by MedGeNet, in a promotional website, in a published survey within Arabic medical professionals outlining the professional routines referred to Genetics diseases.

2. Project Objectives and Results achieved

During the project several important results were achieved before the project was implemented. In particular:

A Telecounselling Service was set up. All the partners in the network have the possibility to request for a session, following an unanimously agreed model protocol. That was previously agreed upon It allows non European countries to take part in remote tele-counselling sessions with world renown Italian geneticists worldwide recognised as experts in Medical Genetics. A constant share of knowledge and competencies, is guaranteed together with a fruitful exchange of best diagnostic practices among countries sharing a common burden of genetic diseases. The Telecounselling system structure and interface are quite simples and user-friendly. The service works using a limited set of Internet based tools, which includes 2 online databases hosted by the MedGeNet web site, one dedicated to Cancer Genetics and another one for Dysmorphology, and a VOIP (Voice Over IP, in the specific case Skype) connection. The protocol, developed by EGF in collaboration with experts from the University of Bologna and agreed upon by partners testing the system, is a short document which provides a step by step set of rules to be applied by counselling applicants in their interaction with remote counsellors.

The Telepathology Service has also been set up. The Aperio Scanscope microscope is a powerful chromosome analysis microscope able to scan and store a large amount of images in its integrated database. It was bought by EGF and maintained for a period of testing in the Bologna University premises by the S.Orsola Hospital medical staff. Once Clinicians were sure of its full functionality and usability for MedGeNet purposes, the microscope was shipped to the Hopital Charles Nicolle (Tunisia). This partner currently hosts the service in its structure and makes it available to the entire network of members. The service can be used to scan and improve images enlarging them digitally by about 100 times. They are archived in the microscope database and are sent as digital slides and then proposed via e-mail to top level domain experts such as Prof. Eusebi (Bellaria hospital, Bologna, Italy), which provide to applicants at no cost, after image and other needed data analysis, their relevant opinion and advice regarding the examined case. When necessary, it is forecasted that live web conferences can be arranged between applicants and experts in order to address issues not definable by simple slides examinations, discuss relevant cases details and request eventual further clinical exams.

The Thalassochip has been upgraded and its overall diagnostic performance greatly improved. Based on the APEX technology (microarray-based assay for parallel detection of common mutations from beta-globin), the Thalassochip is able to perform a reliable Thalassemic mutation screening. The proactive research collaboration between CING, most of MPCs belonging to the MedGeNet network and the technical partner Asper Biotech, (an Estonian SME leader at continental level in the

biotechnology applied to genetics diseases industrial sector) has produced the integration onto the medical device of 49 new mutations in addition to the 41 already featured. The Chip now can detect 90 different types of common mutations from Beta-globulin and its diagnostic range has been increased by around the 120%, which permit the coverage of the largest majority of Beta-thalassemic mutation scientifically known at date.

An Educational DVD has been published**.** The University of Saint Joseph has collected several video-interviews of famous Arabic geneticists attending the 2nd Pan Arab Human Genetics Conference (November 20-22, 2007) in Dubai. These interviews have been then post-produced and put together by USJ in one single DVD, intended to be used as learning tool in Arabic speaking universities addressed to students of Human Genetics courses. The DVD has been printed in 50 copies, one each per target academic centre.

2 Booklets in Arabic have been drafted and printed in 10.000 copies each**.** They were distributed to public healthcare centres of the interested area (Middle East and North Africa countries). The aim was to distribute them around, free of charge, to the Arabic population addressing these healthcare centres. The 2 publications focus on common Mediterranean genetics diseases (i.e. Mediterranean fever) instructions and basic information to address to the families' prevention practices.

Knowledge in Genetics has been transferred to MPC partners**.** Undergraduate, postgraduate students, PhD and lecturers working in MPCs countries have been given the opportunity to receive an highly specialist formation in Human Genetics without moving from their locations, by attending the 5 free Internet EGF on demand courses available within the section called Remote Training of the MedGeNet web site (http://medgenet.tredueuno.it). The list of MedGeNet courses is the following: Molecular Cytogenetics and DNA Microarrays 2006; From Developmental Genes to Dysmorphology 2006; Genetic Counselling in Practice 2006; Bioinformatics for Molecular Biologists 2007, Medical Genetics 2007 In addition to these on demand courses, the 2008 edition of the course in Medical Genetics has been broadcasted live to MedGeNet partners belonging to the RTC International Network. All the remote training activities have been granted to the Consortium free of participations fees.

The extent of MPCs technology capacities and technical skills has been enlarged. MPCs have taken advantages of a transfer of technical equipment (needed to set up Remote Training Centres) and knowledge, in the frame of the Telepathology and Genetics Telecounselling services set up. They have been provided with extremely advanced devices such as the Virtual Microscope Aperio Scanscope and at the same time they acquired competencies in the usage of high-tech diagnostic techniques, the two factors combined now allow them to raise the level and quality of genetics services in the MPCs.

EGF training activities quality has been improved. MedGeNet Courses users have submitted online evaluation questionnaires which have been used to assess the overall quality of the Courses provided. Users' opinions were a key element used for the EGF Training staff to improve a work plan aimed at improving learning objects offered to courses participants.

3. Outlook

The MedGeNet project has to be considered a great success. It has created persistent links and relationships with the structures in this area. There were a number of occasions where the partners in far off eastern and southern Mediterranean countries reiterated their appeals for stronger integration into European Health initiatives. As an anecdote we put forward the case of the Universitè S. Joseph, in Beirut apologized for being a week late transmitting the program for the Conference which would have been held several months later (eventually held on 28 October 2008), as there was a war in their country and part of the hospital had been damaged. They wanted to be sure that as this was part of a deliverable we would be not be abandoning them and ensure our interest in their institute. Of course this only strengthened the commitment of the other consortium members as a whole. This anecdote describes the commitment of the partners in general and is an example of the group environment that was formed during the project, that up to today persists. This persistence of a "group" identity is our major asset and gives us high expectations for the future of our network.

The network will grow and continue to bring in new members thanks to interactions that are now common place. The geneticists in the eastern and southern Mediterranean countries commonly invite European Geneticists to speak and participate in their events while vice versa these geneticists and their students are commonly attending courses in Italy. In fact the Hôpital Charles Nicolle, Cairo University, and Chronic Care Center in Lebanon have all sent and students to the advanced medical genetics courses held in Ronzano and in Bertinoro Italy. This is a central component of our positive outlook for the future in that these students have returned to their home institutions and are acting as ambassadors for the genetics training curriculum in their countries. We have in fact registered several ongoing discussions regarding courses and curriculum which in fact were not part of the project itself. We assume this activity will expand, solidifying the relationships that have grown thanks to MedGeNet. Additionally the project foresaw and implemented Remote Training. It set up and equipped lecture halls so that participants in these countries could follow the courses held in Italy. It must be mentioned that this practice is continuing even now after the project and that centers including Centre Hospitalo – Université Mustapha and The Cyprus Institute of Neurology and Genetics have requested permanent relationships and the possibility to follow all of the courses in the future. The lead partner EGF provided five e-learning Courses (available on the MedGeNet web site) and many of the partners have requested access to additional material of the same nature. In fact EGF has been collecting multimedia material from Genetics courses for several years and has a wealth of materials on line. Partners such as the The Cyprus Institute of Neurology and Genetics and the National Center for Diabetes and Genetics Disorders in Jordan have asked for access to EGF's past courseware. It is expected that this cross-fertilization can only continue and grow now that the partners know of each other's existence and competencies.

Tele-counseling which was a focal part of the project phase has now become a reality. Although there is no official sanction from national or institutional management we have registered several cases where Geneticists in Universitè S. Joseph Lebanon, and the Hôpital Charles Nicolle have contacted Doctor Seri and Doctor Wischmeijer in the University of Bologna for advice on several occasions since the end of the project using the tele-counseling Database to exchange information. The outlook for the growing use of these systems is considered to be a given although legal and other regulations must be studied in further detail. In fact several projects in this area have already contacted or include partners in our project. The area of tele-counseling is of great interest and our

network has on the ground experience. Current projects are already referencing our work so we expect that future projects will also consider our activities. In particular within the consortium groups are forming for the promotion of some of the objectives in the projects. One example is the cooperation of between the Ministry of Health in Israel (and the Ono Accademic College, Kiryat Ono) and EGF and the FIRB - Fondo per gli Investimenti della Ricerca di Base in Italy. Other such synergies are imminent.

The Thalassemia diagnostic tool called Thalassochip (based on the Arrayed Primer Extension technology) has been a major coalescence factor during the project and is continuing today after the end of the project and future activity is already planned and underway between several of the partners in the project. Although from different countries and from cultures the partners commonly realise the efficacy of the chip developed in the project. It is already considered (in the restricted Sickel Cell and Thalassemia communities) to be the state of the art in the diagnosis of common mutations from Beta-globulin. The experience is also prompting the partners to jointly examine other possible areas where the same technologies can be employed. Asper Biotech, The Cyprus Institute of Neurology and Genetics, and the University of Bologna are already investigating replicating methodologies in the creation of similar chips for the diagnosis of Cystic Fibrosis. This area is subject for a number of research projects for which proposals are being written and is contemporaneously attracting the interest of private investors. The outlook is very favourable as the network has undoubtedly identified a major trend in genetics in an early phase. Having demonstrated competence and already organized training events and technology in this area the network is considered to be a stakeholder and expected to be referenced and included in future activities.

An interesting outlook for the project was perhaps overlooked in earlier phases of the project is its value of helping overcome some of the prejudices towards Genetics in some of the regions involved. In fact, it is clear that some of the areas are less endowed in terms of Genetics training and diagnostic units specifically because the administrations and the general public's awareness in these areas is not as developed as in other areas. The case of Lebanon is interesting in that the Universitè S. Joseph in Lebanon prepared simple brochures which describe common genetic disorders at a very entry level. These brochures highlight the act that genetic disorders are problems that affect families require a proper diagnosis and treatment and that Geneticists do exactly that and that many of the fears generated by the discussions in the media do not reflect the work their doctors are currently performing in the clinics. The success is hard to measure as only two brochures were actually produced and distributed in a rather low number but the doctors themselves perceived them as useful and described it to the other members of the consortium, who now also are planning similar activities, as in the end it is not a very expensive manner to reach the general public. The abovementioned center now currently plans to create a separate brochure for each of the disorders (which were summarily combined) in the first two brochures. Each of the brochures will then be planned to be distributed to family members in the clinics dealing with these disorders. This example is expected to be replicated by the other members of the network in their countries.

The two databases that were developed in the course of the project also represent a possible area to be used and exploited. Although legal issues regarding personal medical data and privacy issues may not allow all partners to use and leave medical information in this database data already exists and may be used for research purposes having clear regional data regarding genetic expression. Two possibilities exist. The first is that the databases can be used in future projects allowing the methodology and protocols to be used and the data to be enriched and expanded or a second

possibility that the data itself could be interesting for ongoing research projects in the fields of Dysmorphology and Thalassemia.

Generally speaking the outlook is good and concrete "persistent" collaborations are already happening. The consortium is confident that this collaborative network atmosphere will continue.

4. Implementation Overview

The MedGeNet project was made up of 3 implementation phases:

1) Infrastructure building up and user need analysis**.** During the first phase, the Coordinator focused on the setting up of the RTC, Telepathology and Telecounselling networks built on a common internet based technology. A questionnaire to determine the technical state of the art at partners sites was the basis for subsequent development. On the basis of this assessment EGF outlined the users requirements to be fulfilled by each of the centres to create the infrastructures. After survey result, the needed equipment has been detected and all partners were provided with any equipment need.

The provided equipment for Telecounselling and Telepathology, when needed, consisted of a laptop able to support web conferences and digital camera microphone and speakers to VOIP conversations. For RTC network, a set of particular didactic/technical requirements were sent to partners. In particular, to become RTC didactical needs and proper participate to Hybrid Courses partners were requested to use a classroom with a minimum capacity of 20 seats and to name a teacher responsible to organize and chair afternoon workshops. Being a fundamental tool to coordinate the project and provide useful information on the project itself to outside the Consortium, the MedGeNet portal was created and put online by EGF.

Thanks to MedGeNet the international RTC network managed by EGF was expanded by 6 new members from 6 different countries: Lebanon, Egypt, Jordan, Tunisia and Algeria. As of the Telecounselling informatics system, users needs were investigated trough an appropriate questionnaire prepared by the Egyptian National Research Centre, successively validated by EGF scientific staff and agreed upon by other partners. Since users needs analysis completed, the databases building up process has started and first Beta versions were ready to be tested at the end of the phase. By the end of this phase, which lasted 9 months (up till June 2007), all MPC partners were therefore technically able to attend genetics telecounselling sessions and 6 of them entered the RTC International network.

The work package leader CING has collected from other MCPs more than 600 DNA samples taken from Thalassemic genetics proneness carrier. These samples were sent to Asper Biotech in Estonia, which used them to detect the 49 new mutations which were then added to the ThalassoChip. In June 2007 the upgraded Chip version was proposed to CING for scientific validation.

During the same period the Université Saint Joseph (Lebanon) carried out a survey among caregivers of the Arabic area aimed at underlining the general awareness most common practices related genetics.

2) Tools Testing Phase. Started in July 2007, ended in June 2008. The networks created for Telecounelling and Telephatology purposes were implemented and tested EGF. In particular, the 2 databases were put online in the MedGeNet web site (http://medgenet.tredueuno.it). All partners were asked to try the data uploading system and to provide comments on it, in order to ameliorate

the overall performance of the two tools. The databases Beta version were upgraded according to user requirements. Afterwards, all partners were invited to begin to populate the database.

At the same time, EGF, on behalf of the hosting partner HCN the purchased an Aperio Scanscope microscope from Nikon Europe. The Aperio Scanscope is an advanced device able to work either as electronic microscope or as an highly performance scanner and electronic image warehouse. An in-depth analysis market analysis carried out by EGF showed its capabilities and confirmed it was the most adapt electronic tool to be used for Telephatology purposes. Thanks to its features it can be used for both Cytogenetics and Istopatholgical analysis.

All along this implementation phase the Aperio Scanscope was tested in the EGF premises. The dedicated software used to accumulate and archive virtual Chromosome images or specific tissues slides was installed and tested on the machine internal computer by the EGF staff. Simultaneously, microscope hosting partner prepared a model protocol to guide the Telepathology service. The protocol, which included the full explanation of service aims and a workflow explanation for potential users, was discussed by HCN with EGF and then proposed to the rest of the Consortium for approval.

In the meanwhile, the DNA Chip development, after having integrated the 49 new Thalassemic mutations, passed to validation of the extended diagnostic results. This part of the work was performed by CING, which applied for a trial period of 6 months the Chip on their own clinical cases, matching then the results obtained by the Chip with different diagnostic procedures already in use, to prove the scientific reliability of the instrument. The testing time ended with brilliant results so the new version of the Talassochip was approved as diagnostic tool and made ready to be officially proposed for a use to the international scientific community.

The statistical analysis arising from data extracted from Arabic caregivers questionnaires fulfilled was processed by the Université Saint Joseph. The final aim of this task was to outline the level of general awareness on genetic diseases among the medical personnel and their patients as well in the interested area (Middle East and Northern Africa), in order to better tailor and fine tune the dissemination products addressed to this type of audience, which were 2 booklets and the Educational DVD.

3) Services launch phase. In the last 6 months of the project, all the services provided by the project to the Consortium started to be fully operatives. EGF shipped the Aperio Scanscope Microscope to Tunis, which was then installed at the Hospital Charles Nicolle premises.

Prior to begin the workability of the Telepathology service, first contacts were taken with domain experts which were supposed to supply professional advices over scanned images stored in the microscope database based on tissues or Chromosome samples provided by partners applying for the telecounselling. After this step, a critical mass of samples were digitalized and first Telepathology session could take place.

The first Genetics Counselling session between the University of Bologna and the Charles Nicolle Hospital was video recorded and become a DVD which was then used by the EGF technical support to carry out a detailed analysis of the system functioning and detect eventual weak points of the live session development, in order to fix them. Same trial session record was sent to the entire partnership as a good practice sample, to show them how the Service really works.

Regarding the Thalassochip, after validation and its scientific assessment, an international demonstrative one-day demonstration workshop was organised by research leader institution CING and held in Nicosia (Cyprus), aimed at presenting to a relevant audience composed by scientist coming from Mediterranean countries the important findings obtained by developing the Chip for almost 2 years. Furthermore, during the workshops, in addition to the results showcase a session was organised which focused on practical clinical routines needed to exploit at best the Chip potential has been given.

Several young researchers attending the event were taught on how use in their laboratories this innovative application performing at the maximum potential its extended capacities, which up till today was not so common between Talassemic care centres. The Work Package leader CING, as well as the partner Bologna University, have long tradition and a expertise in the use of this tool, which has been initially created and then developed over the years thanks to the joint effort of these two institutions, Asper Biotech and other Italian universities. The main aim of this last step forward in upgrading the Chip carried out in the frame of the MedGeNet project, was not only to empower this already significant tool for keeping in line with the most recent genetics findings in the area of Talassemia mutations, but also to boost up the awareness on its existence and the knowledge on its high potential. The workshop held in Cyprus specifically addressed this objective and it can be said, after participants satisfaction questionnaires analysis, that the result was achieved. Nowadays these researchers are able to use the Thalassochip and to share their expertise with their colleagues throughout not only the Mediterranean are but all Europe, giving the chance to make this medical device a reference instrument for any health care professional carrying on diagnostic activities related to Thalassemia.

The Educational DVD produced and edited by USJ was released along with the 2 MedGeNet booklets. 50 Universities in Middle East and North Africa were sent free of charge with the video. They agreed to use it as an additional teaching resource in their Medicine Genetics courses both for undergraduate or postgraduate students. The DVD will be showed to courses attendees to give them an overview of the most advanced research carried out in genetics by some of the highest rewarded researchers of the Arabic world.

The booklets, printed in 10.000 copies each, have been distributed to around 250 Arabic health care institutions, in order to spread them for free to general public, in particular patients and their relatives. The aim was to sensitise population living in this specific geographic area on different issues related to wide spread genetics diseases, such as Mediterranean Fever or Thalassemia, in particular focusing on the best prevention practises that can be done in the everyday life to lower the impact of the incidence of these illnesses.

The project has been concluded with a public event which presented to attendees what has been done, for which reasons and the results achieved. The event has taken place on October 28th, in the frame of the 7th Annual Conference Lebanese Society of Family Medicine in front of an audience of around 250 people made by geneticists, hospital physicians and family doctors, health care institutions managers, policy makers and NGO's representatives. The project has been explained in all its details by Prof. Megarbané (USJ), who also presented the Educational DVD produced by the project and chaired the following debate with public centred on next years challenges for the Arabic countries regarding genetics diseases prevention. The discussion has also tackled the aspect of defining which policy or information campaign could help to change the wrong approach that many

people in Arabic countries, from intellectual elites up till any other citizen, have towards Human Genetics, finding the way to break the wall of prejudices that still persist in those countries.

5. Conclusion

After more than 2 years of cooperative work, the main goals set before project have been obtained. Beyond the specific objectives and results achieved during the project (as explained under paragraph 3 and 4 and fully detailed in the text of the submitted deliverables), it is relevant to stress how the MedGenNet Project has been able to contribute to the main INCO strategic objectives and priorities.

What we can conclude is that, thanks to the EC funded activities, MedGeNet has contributed to :

- strengthening research networks capacities and enabling participants to gain experience in international cooperation in research;
- supporting access to knowledge and expertise to an high extent with information, dissemination and training activities;
- network building and promoting the network (in a long term perspective) the participation of INCO targeted countries to the Framework Programme and other international cooperation activities.

Annex I - Plan for using and disseminating the knowledge

1. Exploitable knowledge and its Use

The exploitable results produced by the MedGeNet project at the end of the implementation phase are the following:

- 2 pamphlets in Arabic, printed in ten thousand copies, the first focused is on genetics disease prevention, the second one providing information and contact details for health care centres which provide free genetic counselling services in the Arabic speaking area. Both publications are addressed to the general public living in Middle East and North Africa countries. The dissemination mode of these publications is the free distribution to target readers through the by local public health care institutions.
- A 30 minutes Educational Video distributed free of charge to Universities located in the booklets diffusion area and meant to be used as a teaching resource for Medical Genetics graduate or undergraduate courses. The video consists in an edited video made up of several interviews with well-known Arabic geneticists taken in the occasion of the 2007 Pan-Arabic Genetics Conference (Dubai). It has been published in 50 copies.
- an upgraded version of the Thalassemia diagnostic tool called Thalassochip (APEX) featuring a total of 90 Thalassemia detectable mutations of which 49 non regarded in the previous APEX version, developed by the Cyprus Institute of Neurology and Genetics (Cyprus) in collaboration with Asper Biotech (Estonia). The release on the market of the upgraded version of the Chip represents only a first step in the development of a more performing technological diagnostic tool able to detect the complete pool of Thalassemia mutations known up-to-date. The Medicine Genetics is a fast evolving research field and the Thalassochip, even demonstrating a consistent step ahead toward an easy, sure and quick detection of Thalassemic factors in human blood samples, is surely not yet the definitive diagnostic answer to Thalassemia, the most concerning among existing genetics diseases in the Mediterraean area. This consideration, along with the wide spectrum of performance advances still to be implemented to the Thalassochip, opens a possible research pattern to be followed in the next years. The aim is to keep collecting DNA samples from Mediterranean countries other than those already considered during the project and to include the broadest range of uncovered thalassemic mutations to the Thalassochip. The development of this diagnostic device imply as well an high commercial exploitation prospective, which will be tackled in the immediate future by Asper Biotech, the SME which performed the technical part of the work and owns the product patent. The potential market of the tool is vast and embraces the whole Mediterranean area, especially the southern rim which includes country featuring an historical deficit in terms of medical technology update.
- the Genetic Telecounselling Network, which has been set up during the project, represents an important mix of knowledge and expertise developed by the network and is planned to be increased in its catchment area including new centre associated during the upcoming years. The technology transferred to MPCs will allow concerned partners to develop a series of genetics tests and diagnosis with an high degree of innovation for such a countries. At the same time the possibility to establish a direct contact top level Italian geneticists allows for

continues sharing of clinical data and medical expertise among the northern and the southern Mediterranean shores, this will result in a relevant gain of each other's knowledge in common genetics diseases and, secondarily, will improve professional respect and reputation on both sides. All above mentioned benefits have been included in a MedGenet Telecounselling (GT) information "package" which is going to be the main instrument to advertise the potential of this project finding that will permit to include new members to the network, likely other 15 GT points by the end of 2012. Actual members will play a basic role to achieve this quantitative goal: they will have the task to promote benefits package, distributing it at international conferences or meetings to potential new network members. By doing so, they would promote the GT service and describing their own positive direct experience as users, a decisive added value for attracting as much interest as possible toward it.

- the Telepathology Service introduced an innovative and technologically advanced system to share clinic history data, allowing a direct confrontation on difficult cases between domain experts and transferring top level technical and medical knowledge from European to MCPs. Likewise the Genetic Counselling Service the Consortium has planned to keep on exploiting the Telepathology results obtained within the EC funded period, broadening the numbers of health institutions applying to become users of the service. This goal will be reached expanding the service deployment area, which is currently limited to Arabic speaking countries, including European and African countries as well, on the ground that the scope of this service features a so various set of possible benefits that makes it potentially appealing for less and more developed health care systems equally. The virtual bank of DNA specimens stored in the microscope database is going to be doubled by the next two years under Hopital Charle Nicolle (Tunis, Tunisia) – institution hosting the microscope – coordination and leadership, trough the increased rate of tissues slides collection and digitalisation given by the affiliation of new networks members. The positive forecasts included in the sustainability plan associated to the future service development are based on the probable successful outcome of the call to apply for becoming service users addressed to all the numerous hospitals and universities (selected while project running) existing in Southern Mediterranean countries, which might found interest to receive free clinical advices from worldwide recognised experts. Further financing request to the European Commission may be envisaged to support the network growth.

- the Hybrid courses in Human Genetics and the associated setting up of new Remote Training Centres. Hybrid Courses are an innovative system for the transfer of knowledge at higher education level developed by EGF for the European School of Genetic Medicine courses, which is the genetics specialist school run by the Foundation itself. The basic scheme of such a type of courses is structured in 2 different session modes half-day long: on one hand the morning one which is made of remote attendance to live streaming webcasted lectures with the possibility for remote attendees to have direct interaction with lecturers addressing questions via Skype or e-mail; on the other hand the afternoon session which is made of students run workshop, based on the morning session talked topics. This blended remote learning system efficiently combine the passive teaching provided to attendees by morning lectures with a more proactive students involvement thanks to the after midday workshops. A set of technical and logistics requirements defined by EGF had to be met by applicant new

Remote Training Centres (RTCs) in order to be registered as network members. By transferring the needed technology equipment the MedGeNet project has permitted to increase the network size over any original expectations, from the starting level of 21 RTC to the final amount of 27, which means new 6 members coming from 6 different MCPs.
We, as MedGeNet consortium, plan to disseminate the advantages gained in becoming RTC network members through a series of awareness raising activities conducted by MCPs partners themselves within their national scientific communities and to empower the actual annual offer of Hybrid Courses, aiming at attracting potential new applicants. The target set by the Consortium is to double the score of 6 new RTCs by the end of 2010. It is an ambitious goal, but we assess it feasible as long as the scientific networks in which MCPs partners belong is being exploited at best, as these contacts represent a dissemination recipients area more than sufficient to attain the target. The commercial potential linked to the RTCs subscription to Hybrid Courses, which are provide under participation fees payment, will be exploited by the courses organiser entity, EGF.

Table 1 Exploitable knowledge and its Use: Overview table

Exploitable Knowledge (description)	**Exploitable product(s) or measure(s)**	**Sector(s)** of application	Timetable for commercial use	Patents or other IPR protection	**Owner** & Other Partner(s) involved
Improved diagnostic capacity related to Thalassemic diseases.	*Thalassochip*	*Medical*	*2009 & 2010*	*The partner who developed the tool already owns its property rights*	***P10 Asper Asper Biotech (owner)*** ***P03 CING*** *(responsible for scientific validation)*
Educational video	*DVD*	*Higher Education in Medicine Genetics*	*2009 & 2010*	*These products are IPR free. The video usage is given with no restrictions to targeted universities*	***P04 USJ*** *(author) and* ***P01 EGF*** *(editing and printing)*
Best practices in Mediterranean Genetics disease prevention disseminated	*2 pamphlets in Arabic*	*Health care policy*	*2009 & 2010*	*These products are IPR and provide no incomes because distributed at price 0*	***P04 USJ*** *(author) and* ***P01 EGF*** *(editing and printing)*
The Genetic Telecounselling Network	*Clinical information shares through online databases access.*	*Medical - research*	*2008 onward*	*The service is free for partners.*	***P01 EGF*** *(system administrator),* ***P06 Unibo*** *(counselling provider)*

Exploitable Knowledge (description)	**Exploitable product(s) or measure(s)**	**Sector(s)** of application	Timetable for commercial use	Patents or other IPR protection	**Owner** & Other Partner(s) involved
	Sessions of genetics counselling via web				
The Telepathology Service	*Human tissues specimens storage and worldwide retrieval system. Relevant clinical cases histories discussion among experts on remote.*	*Medical*	*2008 onward*	*The service is free for Partners.*	***P13 HCN** (service administrator)* ***P01 EGF** (technical assistance)* ***P06 Unibo** (counselling provider)*
The Hybrid courses in Human Genetics	*RTCs attendance to courses.*	*Higher Education in Medicine Genetics*	*2008 onward*	*No property license existing. Eventual incomes arising from subscriptions fees are enjoyed by EGF.*	***P01 EGF** (RTCs network administrator and Hybrid courses organiser)*

2. Dissemination of knowledge

Final Visibility Event: held in the frame of the 7th Lebanese Family Annual Conference on October 24th 2008. The educational DVD has been presented to the audience, about 250 people mainly composed by physicians, health policies makers, local NGOs and medical industries, gathered for the inauguration ceremony of the event. A debate chaired by organising partner USJ has followed the DVD presentation. A video of the event has been produced and delivered to the Commission.

Media release: the day after the Visibility Event (October 25th 2008) Prof. Adib (USJ) on behalf of the MedGenet network is interviewed in a popular talk show on air in a Lebanese national TV channel to speak about project aims and findings. Potential watchers were about 4M, no real data available.

Project website: http://medgenet.tredueuno.it. Since his put online on December 2006 it has been maintained and updated by EGF. It features both project members restricted access area and project dissemination/information sections free accessible. All relevant public documents were made downloadable from a webpage appended to the USJ Medical Genetic Unit portal. Around 50.000 site visitors and 300 documents downloads counted so far.

Informational Booklets: 2 booklets in Arabic released in December 2008 addressed to the general public and focused on Genetics diseases prevention practices and free counselling services have been printed in 20.000 copies (10.000 each) and freely distributed to patients and their relatives in 6 countries (Jordan, Lebanon, Syria, Egypt, Morocco, Tunisia), using as distribution means 200 public health care institutions.

Educational DVD: a 30 minutes long DVD has been produced in 50 copies by USJ (released on December 2008) on the base of several interview collected at the 2007 Pan-Arabic Genetics Conference (Morocco). It is made for higher education purposes, therefore it has been distributed to 50 universities in the Middle East – North Africa area, selected and contacted by local partners. It is meant to be used as teaching resource in undergraduate and graduate courses in Medical Genetics. Mention of the MedGeNet project scope, aims and contact details has been included in the DVD text. Surveying the involved Universities it has been forecasted that over 2500 students should be reached by this MedGeNet product by the 2010 academic year end.

Hybrid Courses: the 20th edition of the course in Medical Genetics organised by the European School of Medicine Genetics has become the first MedGeNet Hybrid Course has taken place (Bertinoro, 4-10 May 2008). 2 MedGeNet RTCs attended the course remotely, for a total of 40 students participating to morning live streaming lectures and related afternoon workshops. Attendance certificated and ECM credits are awarded to participants. After project end RTCs partners of MedGeNet will continue to enjoy for at least 5 years the possibility to apply free of charge to EGF Hybrid Courses, which on average are 7 per year.

Table 2 Dissemination of knowledge: Overview table

Actual Dates	Type	Type of audience	Countries addressed	Size of audience	Partner responsible /involved
October 25th 2008	*Media briefing*	*General public*	*Lebanon*	*Around 4 million*	***P04 USJ***
October 24th 2008	*Conference (MedGeNet final event)*	*Stakeholders*	*Middle East area*	*Around 250*	***P04 USJ***
December 2008	*Publications (2 booklets)*	*General Public*	*Arabic speaking countries*	*200 health care institutions – 20.000 readers foreseen*	***P04 USJ – P01 EGF** (editing and printing)*
December 2006	*Project web-site*	*General Public*	*The world*	*Any web user*	***P01 EGF** (site administrator) – **P04 USJ***
October 2008	*2 information flyers*	*General Public*	*Arabic speaking countries*	*Distributed along with the video*	***P04 USJ***
All project last	*Direct e-mailing*	*Stakeholders*	-	-	***P01 EGF – P04 USJ***
October	*Film/video*	*Higher education*	*Arabic*	*50*	***P04 USJ***

2008			*speaking countries*	*Universities – 2500 students*	

3. Publishable results

Educational DVD: published in 50 copies by USJ on October 2008, distributed free of charge to 50 Universities located in Arabic speaking countries. No IPRs protection is granted by the usage of the video, which is an open educational tool available for any interested person/institution. **(**For further information please contact: Prof. André Megarbane (USJ), megarbane@usj.edu.lb).

Booklets: published in 20.000 copies (10.000 each) by USJ. Distributed for free to 200 health care institutions in Arabic speaking countries, no IPRs are licensed. **(**For further information please contact*:* Prof. Salim Adib (USJ), salim.adib@usj.edu.lb).

Hybrid Courses: based on online streaming courses, it is a full structured learning platform provided solely by EGF (without external partners collaboration needed), which raises incomes from RTCs participation fees. Market field for such a service is higher education training in human genetics. Users target: universities, health care institutions and any public or private research centre all over the world, thanks to its location on the web. Its development is completed. MedGeNet partners have free access to the service granted for at least 5 years after project end. (For further information please contact: Giuseppe Curcio (EGF) giuseppe.curcio@eurogene.org– Tel. +39 (0)512088430).

Genetics Telecounselling Service: it is based on VoIP technology and top level human expertise in Genetics provided by relevant consortium members such as the Bologna University. The service consists in providing remote assistance to MCPs on determined clinical cases to the domain know-how owners. Major benefits are the easy worldwide accessibility to a large amount of clinical data on Cancer Genetics and Dismorphology coming from an international and genetically significant environment (Mediterranean area) for large spread diseases such as the Mediterranean fever, medical best practices exchange among important experts in the field and remote discussion of specific clinical cases. The service is implemented and fully operating since December 2008. It will be granted free of charge to project partners. No future commercial exploitation is foreseen. (For further information please contact: Alberto Goldoni (EGF) alberto.goldoni@eurogene.org – Tel. +39 (0)512070613).

Telepathology Service: based on scanned images of tissues specimens stored within the Aperio Scanscope microscope virtual archive, it allows a remote counselling activity through the analysis of virtual samples performed by world level experts voluntary associated to the project (i.e. Prof. Eusebi, Bellaria Hospital, Bologna Italy). Service administrator and technical equipment host is the Hospital Charle Nicolle (Tunis, Tunisia), technical support is provided by EGF. The service, fully operative since October 2008, is reserved to MedGeNet partners, which are granted of free membership with no time limits. No commercial exploitation is foreseen. (For further information please contact: Prof. Habiba Chaabouni (HCN) habiba.chaabouni@rns.tn).

Thalassochip (APEX**)**: upgraded version of the diagnostic tool for Thalassemic diseases. Developed by the CING (mutation recognitions scientific validation and dissemination within the scientific community) and Asper Biotech (technological upgrade). It features the traceability of 49 new Thalassemia mutations. Intellectual rights belong to Asper Biotech, which will enjoy the Thalassochip commercial exploitation. Under specific request project partners can be granted of a

free sample. (For further information please contact: Prof.Marina Kleantous (CING) marinakl@cing.ac.cy).