S. Passamonti, S. Gustincich, T. Lah Turnšek, B. Peterlin, R. Pišot, P. Storici (Eds.)
CONFERENCE PROCEEDINGS with an analysis of innovation management and knowledge transfer potential for a smart specialization strategy
ISBN 978-88-8303-572-2 / e-ISBN 978-88-8303-573-9. EUT, 2014.

SIGN PROJECT TELEGENETICS - PROVIDING REMOTE GENETIC SERVICES IN THE CROSS-BORDER REGION OF ITALY AND SLOVENIA

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Abstract — This paper describes the telegenetics component of the SIGN project. Telegenetics is a subfield of telemedicine applied to clinical genetics. In the SIGN project, telegenetics aims to provide remote genetic services in the cross-border region of Italy and Slovenia. More specifically, the goal is to develop remote genetic services in areas without them and to provide best expertise in clinical genetics without the need to travel large distances (patients or professionals). The major telegenetics activities within the SIGN project are the development of remote genetic services: for genetic counseling, for expert to expert communication, and for communication between the partners of the project. To accomplish the activities, these major tasks had to be done: testing and selection of video-conferencing equipment and software for secure data sharing and exchange; development of work protocols; end-user education; installation, configuration and testing of necessary hardware, software and developed protocols; and evaluation of user satisfaction (both patients and genetic service providers). The preliminary evaluation results show promising user-satisfaction. They also highlight areas where further improvement of the remote services is possible.

Index Terms — SIGN, telemedicine, telegenetics, genetic counseling, video-conferencing, cross-border collaboration

1 INTRODUCTION

Telemedicine utilizes information and telecommunications technology to transfer medical information for diagnosis, therapy and education. Telegenetics is subfield of telemedicine applied to clinical genetics.

In the SIGN project, telegenetics has these major goals:

- To develop remote genetic services in regions without them.
- To provide best expertise in clinical genetics without the need to travel large distances (patients or professionals).
- To use innovative health information technology (videoconferencing).
- As more specific goals, which are at the same time expected results, the telegenetic part of the SIGN project aims to achieve:
- Better access to genetic services for those patients which do not have genetic services available in their region of residence without the need to travel to a remote center.
- Access to better or more advanced genetic services for the patients who have a local genetic center, but who need more specialized genetic services only available at bigger genetic centers.
- Improved communication between the Slovenian and Italian genetic service providers.
- Reduced travel costs.

2 TELEGENETICS ACTIVITIES IN THE SIGN PROJECT

The major telegenetics activities within the SIGN project are:

- Development of remote genetic services for genetic counseling
- Development of remote genetic services for expert to expert communication
- Remote communication between the partners of the project

To accomplish the above mentioned activities, these major tasks had to be done:

- Testing and selection of video-conferencing equipment
- Testing and selection of software for secure data sharing and exchange
- Development of work protocols (workflow)
- End-user education
- Installation, configuration and testing of necessary hardware, software and developed protocols
- Evaluation of user satisfaction (both patients and genetic service providers)

In what follows, these tasks are described in more detail.

3 TESTING AND SELECTION OF VIDEO-CONFERENCING EQUIPMENT

We did extensive market research and testing of available video-conferencing software and equipment. Finally, we decided to use standard Windows or Mac iOS computers with additional software tools and video-conferencing hardware. We decided to use the WebEx video-conferencing tool when there are more than two participants in the meeting, and Skype for point-to-point (two participants) meetings, including remote genetic counseling. We selected suitable full HD (high definition) video-cameras, microphones, loudspeakers, printers and scanners.

4 TESTING AND SELECTION OF SOFTWARE FOR SECURE DATA SHARING AND EXCHANGE

During some of the telegenetic activities a lot of medical information is exchanged. Some of this information is sensitive. This kind of information is not suitable to be sent by ordinary email because of security concerns and because of email attachment limitations. Therefore, we investigated tools for data sharing and exchange such as Wuala, SkyDrive, Google Drive, iCloud, Dropbox, SugarSync and similar. Finally, we decided to use Wuala because it encrypts the data before it leaves the user computer and before it is stored in the cloud.

5 PROTOCOLS FOR THE MAJOR TELEGENETIC ACTIVITIES

5.1 Telegenetic counseling and estimate of genetic risk predisposition

The genetic counseling room is equipped with a full HD video-camera, a microphone, desktop computer with large screen, printer and scanner. The equipment is operated by a genetic nurse who also introduces the patient to the whole procedure. Skype is used for video-conferencing because it is simple, efficient, supports HD video, and all of the SIGN partners were already familiar with it (Figure 1). The genetic counselor uses similar equipment. The counselor can be: in another room in the same building (used for testing purposes), in another genetic centre in the same country, or in another genetic centre in another country.
The patients usually provide relevant medical documentation a few days before the counseling. This documentation is scanned and made available to the genetic counselor electronically by the data sharing tool Wuala.

Figure 1. Telegenetic counseling by video-conference. Shown is the patient - the genetic counselor is at a remote location.

The video-conference is started by the genetic nurse who leaves the room afterwards and leaves the patient alone to talk to the genetic counselor. After the counseling, both the patient and the genetic counselor fill an evaluation form, if they are willing to do so.

5.2 Remote genetic services for expert to expert communication

In real clinical practice, there is often a need for communication and consultation between genetic experts. For example, when a genetic expert is faced with a difficult case for which a second opinion is needed. In a situation like this, the relevant medical data has to be sent from one genetic expert to another one in a secure way. The other expert sends his opinion back to the first expert. Here we provide a simplified description of the developed protocol.
For the exchange of sensitive medical data, in the telegenetic part of the SIGN project, we used the Wuala tool because it encrypts the data before it leaves the user computer. Each of the SIGN project partners has its own Wuala account. Each partner allows the other project partners to access their data in such a way that the data can be read, but not modified. The other partners can create a new document with their expert opinion. The expert consultation can be off-line, which means that the other experts can provide their opinion when they have time; or it can be in real-time, which means that the experts are interactively discussing the case by video-conference (WebEx or Skype).
As a practical example, we would like to mention the exchange of difficult-to-interpret microarray experiments results between the lead partner from Ljubljana and the Udine partner. Both partners provided their own interpretations which were later compared in order to reach a conclusion.

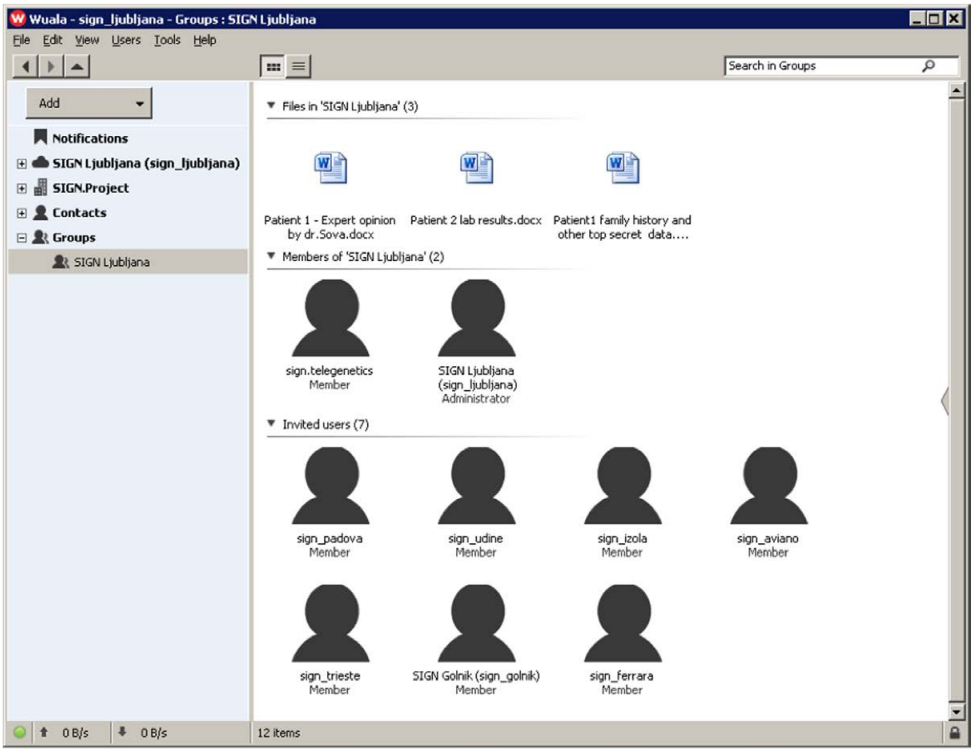

Figure 2. Sharing medical data in order to get expert opinion from another genetic expert. Both the medical data and the expert opinion is exchanged with the Wuala tool.

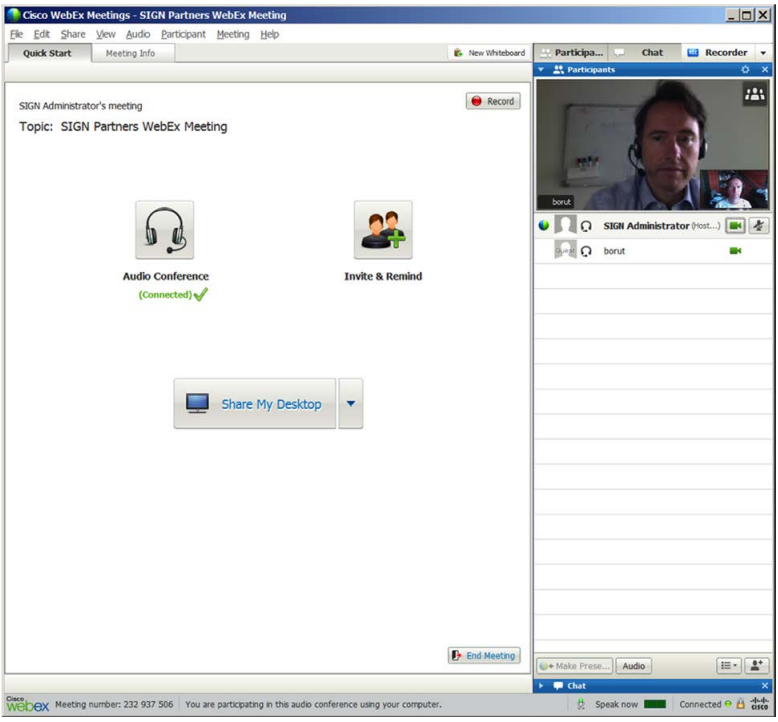

Figure 3. Example of a video-conference with WebEx.

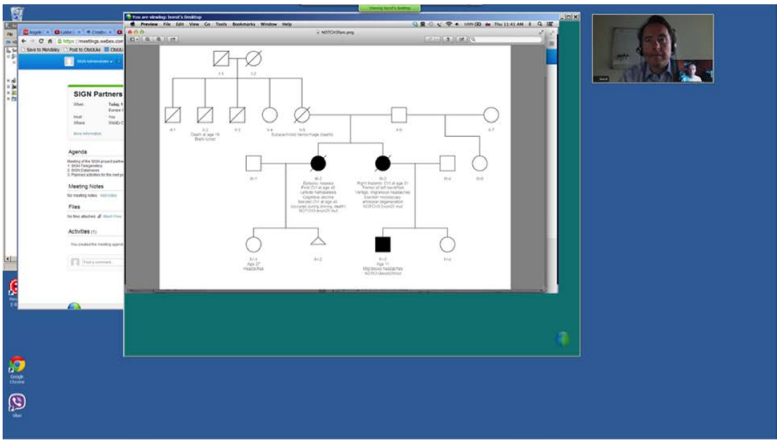

Figure 4. Example of desktop sharing with WebEx.

5.3 Remote communication between the partners of the SIGN project

In addition to what everybody uses for communication today (email and telephone), for communication between the SIGN project partners we used the same technology and equipment that we used for the telegenetic part of the project (WebEx, Skype, Wuala).
We used WebEx for the meetings of the project partners or whenever there were more than two communicating parties involved. For a typical meeting, the organizer of the meeting first defined the meeting in WebEx, including the title of the meeting, the data and time, the agenda and the list of participants. The participants received meeting invitations by email. At the appropriate time, the participants connected to the meeting (Figure 3). In addition to video-conferencing, the participants were able to share their desktops, which they used when they wanted to present a slide-show or to show other relevant data (Figure 4). A meeting moderator decided which participant was presenting at any given time. The whole meeting was recorded for archival purposes or to be seen by those partners who were not able to participate.

6 END-USER EDUCATION AND TRAINING

Those involved in the telegenetic activities received written instructions and if necessary practical training about how to perform the telegenetic activities.

7 INSTALLATION, CONFIGURATION AND TESTING OF NECESSARY HARDWARE, SOFTWARE AND DEVELOPED PROTOCOLS

Each partner was responsible for and conducted local installation, configuration and testing of the required hardware and software. The lead partner was additionally responsible for the installation and configuration of the server tools (WebEx and Wuala) used by all the participants.

8 EVALUATION OF USER-SATISFACTION

We performed user-satisfaction evaluation for both the patients who were involved in genetic counseling and for those who performed the counseling. First we did research on PubMed and Web of Science and found relevant papers on telegenetics. Based on what we found out in these papers and taking into consideration the specifics of the SIGN project, we defined two questionnaires: one for the patients and one for the genetic service providers. Both questionnaires contained multiple questions. The possible answers were on a numerical scale from 1 (worst) to 5 (best). On average, over all the questions, the patient satisfaction was 4,1 out of maximum 5 . The average satisfaction for the genetic counseling providers was 4,4.

9 CONCLUSIONS

We presented the telegenetics component of the SIGN project. It consists of three major activities:

- remote genetic services for genetic counseling,
- remote genetic services for expert to expert communication, and
- remote communication between the partners of the project

We have done preliminary evaluation of user-satisfaction for the first activity (remote genetic counseling). The evaluation results show promising user-satisfaction. They also highlighted areas where further improvement of the remote services is possible. In the future we plan to conduct more extensive evaluation with more participants and to further improve the remote genetic services.
We also plan to evaluate the other two major activities.

10 SIGN PROJECT PARTNERS

Main partner:

- Univerzitetni klinični center Ljubljana

Other partners:

- Univerzitetna klinika za pljučne bolezni in alergijo Golnik
- Splošna bolnišnica, Izola – Ospedale Generale, Isola
- IRCCS Burlo Garofolo, Trieste
- Università degli studi di Udine
- Centro di Riferimento Oncologico, Aviano
- Università degli studi di Padova
- Azienda Ospedaliero-universitaria, Ferrara