LA MALATTIA DI POMPE

cosa è cambiato dopo 10 anni
di Terapia Enzimatica Sostitutiva

AIGKit

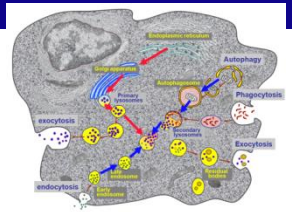
AIGkit – Il progetto

Gabriele Siciliano, *Pisa*
Fabrizio Seidita, *Milano*
Silvia Peviani, *Milano*

LA MALATTIA DI POMPE (Glicogenosi II o Deficit di maltasi acida)

- malattia da accumulo lisosomiale
- ereditarietà autosomico recessiva
- deficit dell'a-glucosidasi lisosomiale
- accumulo di glicogeno nei lisosomi dei muscoli scheletrici, lisci e del miocardio
- terapia enzimatica sostitutiva (ERT)

Clinica



Forma infantile: è la forma più grave della malattia. Si manifesta entro i primi tre mesi di vita con cardiomegalia, epatomegalia, debolezza muscolare con ipotonia e interessamento della muscolatura deputata alla respirazione con conseguente insufficienza respiratoria.

Forma giovanile: si manifesta nella prima decade con difficoltà nel camminare dovuta alla debolezza della muscolatura prevalentemente prossimale. Anche in questo caso è presente insufficienza respiratoria che può condurre a morte i pazienti (nella seconda e terza decade di vita).

Forma dell'adulto: esordisce dopo i venti anni con difficoltà a salire le scale e ad alzarsi dalla posizione accovacciata causate da una debolezza muscolare prevalente al cingolo pelvico a cui si aggiungono problemi respiratori per il coinvolgimento dei muscoli deputati alla respirazione. Quest'ultimo aspetto è precoce nella malattia di Pompe rispe ad altre miopatie, spesso, anzi, il paziente lamenta per primi proprio i sintomi respiratori.

Necessità di una presa in carico multidisciplinare

VERSO UNA CONCEZIONE SISTEMATICA DELLA MALATTIA

Kanters et al. Orphanet Journal of Rare Diseases (2015) 10:112
DOI 10.1186/s13023-015-0334-6

ORPHANET JOURNAL OF RARE DISEASES

WMS World Muscle Society
21st INTERNATIONAL CONGRESS OF THE WORLD MUSCLE SOCIETY
From 4 to 8 October 2016, Granada, Spain

RESEARCH Open Access

A conceptual disease model for adult Pompe disease

Tim A. Kanters[1,2*], W. Ken Redekop[1], Maureen P.M.H. Rutten-Van Mölken[1], Michelle E. Kruijshaar[2], Deniz Güngör[2], Ans T. van der Ploeg[2] and Leona Hakkaart[1]

Abstract

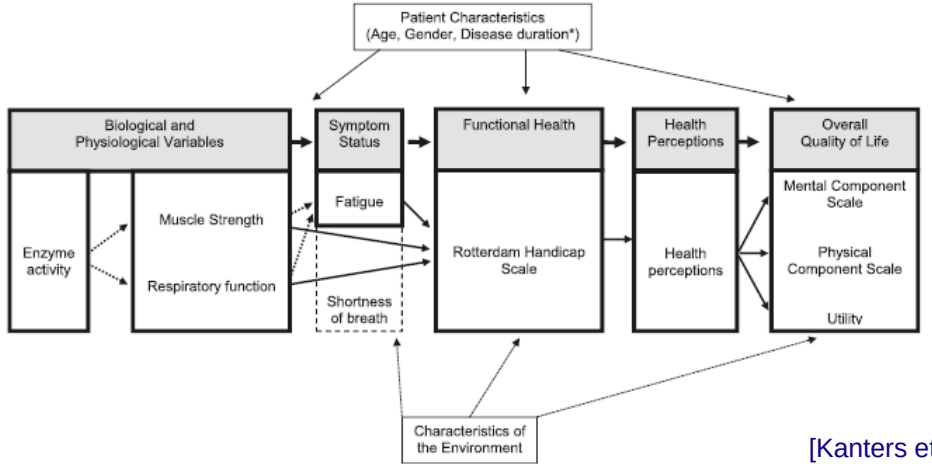
Background: Studies in orphan diseases are, by nature, confronted with small patient populations, meaning that randomized controlled trials will have limited statistical power. In order to estimate the effectiveness of treatments in orphan diseases and extrapolate effects into the future, alternative models might be needed. The purpose of this study is to develop a conceptual disease model for Pompe disease in adults (an orphan disease). This conceptual model describes the associations between the most important levels of health concepts for Pompe disease in adults, from biological parameters via physiological parameters, symptoms and functional indicators to health perceptions and final health outcomes as measured in terms of health-related quality of life.

Methods: The structure of the Wilson-Cleary health outcomes model was used as a blueprint, and filled with clinically relevant aspects for Pompe disease based on literature and expert opinion. Multiple observations per patient from a Dutch cohort study in untreated patients were used to quantify the relationships between the different levels of health concepts in the model by means of regression analyses.

Results: Enzyme activity, muscle strength, respiratory function, fatigue, level of handicap, general health perceptions, mental and physical component scales and utility described the different levels of health concepts in the Wilson-Cleary model for Pompe disease. Regression analyses showed that functional status was affected by fatigue, muscle strength and respiratory function. Health perceptions were affected by handicap. In turn, self-reported quality of life was affected by health perceptions.

Conclusions: We conceptualized a disease model that incorporated the mechanisms believed to be responsible for impaired quality of life in Pompe disease. The model provides a comprehensive overview of various aspects of Pompe disease in adults, which can be useful for both clinicians and policymakers to support their multi-faceted decision making.

Keywords: Orphan drugs, Conceptual model, Pompe disease



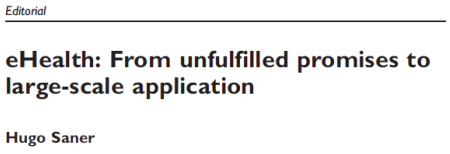
[Kanters et al. 2015]

Fig. 1 Wilson-Cleary conceptual disease model applied to Pompe disease

DEFINIZIONE DI E-HEALTH

"E-health is the use of Information and Communication Technologies, including the Internet, to improve or enable health and healthcare."

[Draft Council conclusion on e-health, Supporting the european citizen through e-health - Cork 6-5-2004]



Editorial

eHealth: From unfulfilled promises to large-scale application

Hugo Saner

Cardiology 2016
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DOI: 10.1177/2047487316670042
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SAGE

The potential of e-health to provide innovative solutions to health problems is enormous. This has raised high expectations by the public, by politicians and by healthcare professionals.

The European Union e-health action plan for 2012–2020 states that the promise of ICT to increase efficiency, improve quality of life and unlock innovation in health markets remains largely unfulfilled.

Major companies such as Apple, Google, Samsung, etc… are investing heavily in healthcare-related technologies which will further increase the speed of development.

Information and communication technologies for health

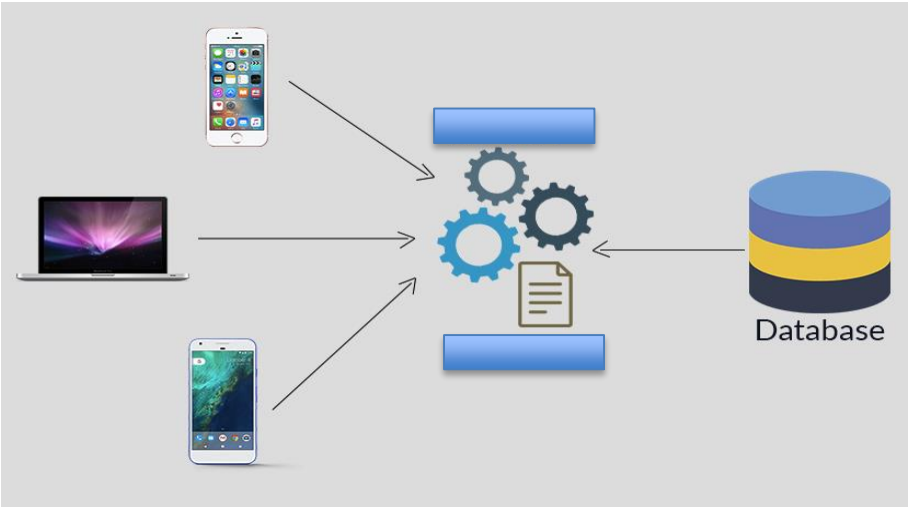
The viability of introducing adaptive technology in NMD should be addressed as part of the management of NMDs. Such comprehensive management usually requires the efforts of a multidisciplinary team and the active engagement of patients associations

Although e-health interventions may be the future of healthcare, a concerted effort is needed about their benefits and utilization.

The establishment of European Reference Networks for NMDs provides

→Accessible services integration

To facilitate interoperability, seamless connectivity and sharing of content between different services and ontologies in all application domains relevant to applications for neuromuscular patients, clinicians and reserachers.

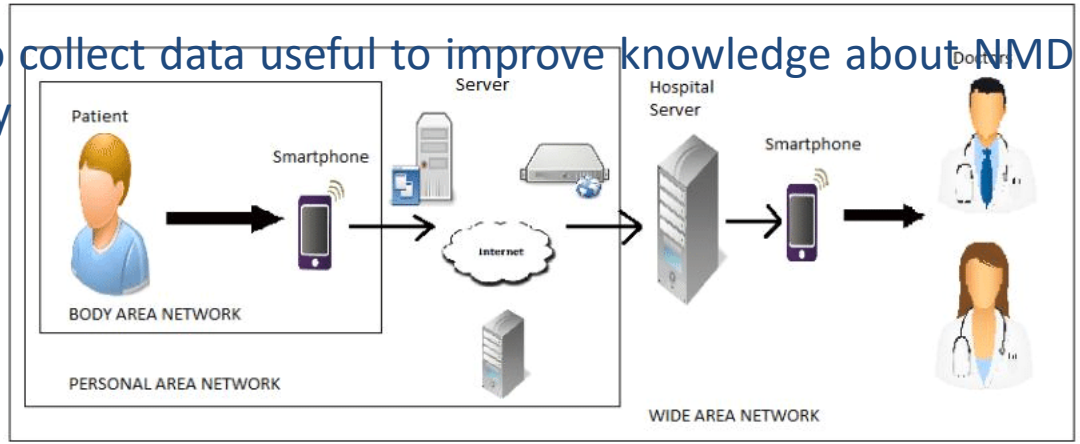


→Health monitoring

The potential of **smart technology** to provide innovative solutions for disease management

it could allow continuous and timely tracking of disease in each patient at real-time and ecological conditions of everyday life

and to collect data useful to improve knowledge about NMD natural history



M-HEALTH: QUANTO E' DIFFUSO?

JMIR MHEALTH AND UHEALTH Chen et al

Original Paper

Designing Health Apps to Support Dietetic Professional Practice and Their Patients: Qualitative Results From an International Survey

Juliana Chen[1], BSc, MND, APD; Jessica Lieffers[2], RD, PhD; Adrian Bauman[3], PhD, FAFPH; Rhona Hanning[2], RD, FDC, PhD; Margaret Allman-Farinelli[1], PhD, FDAA

[1]School of Life and Environmental Sciences and Charles Perkins Centre, The University of Sydney, Camperdown, Australia
[2]School of Public Health and Health Systems, University of Waterloo, Waterloo, ON, Canada
[3]School of Public Health and Charles Perkins Centre, The University of Sydney, Camperdown, Australia

Abstract

Background: Dietitians are engaging with mobile health (mHealth) technologies, particularly with diet and nutrition apps i their patient care. Despite the plethora of apps available, the majority are not designed with a dietitian's input.

Objective: The aim of this study was to identify the user preferences of dietitians in relation to tools, resources, and design features for smartphone health apps that would support their dietetic professional practice and their patients.

Methods: As part of a larger international Web-based survey of health-app use among dietitians, three open-ended responses were included for specific exploration of app design features and additional resources or tools that could guide the development of apps for use in dietetic practice and patient care. Inductive thematic analysis of responses was conducted using the qualitative data analysis program, NVivo version 11 (QSR International Pty Ltd), to understand the design preferences and features valued by dietitians.

Results: The responses from 381 dietitian respondents were analyzed. Five key themes were identified. Dietitians wanted access to credible apps, suggesting that dietetic associations should have greater involvement in reviewing and endorsing evidence-based apps for use in dietary counseling. Improvements to the usability of apps, relating to their ease of use and design, were also raised, as self-monitoring of dietary behaviors using existing nutrition apps was deemed to be burdensome. Furthermore, apps providing dietitian-oriented support were favored, for example, those with the ability to streamline the dietary assessment process, so that dietitians could spend more time on dietary counseling and negotiating patient goals for dietary and lifestyle behavior change. Provision of patient-oriented support, such as functionality to tailor apps to patient-specific needs, was also considered important. Finally, respondents valued apps that could integrate into their work systems to enhance the quality of the dietitian-patient relationship.

Conclusions: App developers should draw upon the features and characteristics valued by dietitians to guide their development of apps that support dietetic practice and enhance patient care. Moreover, to achieve better dietitian and patient-centered app design, it is imperative that app developers take a collaborative approach with dietitians, their professional associations, and their patients.

(JMIR Mhealth Uhealth 2017;5(3):e40) doi:10.2196/mhealth.6945

BMC Public Health
BioMed Central

A qualitative study of user perceptions of mobile health apps

Wei Peng, Shaheen Kanthawala, [...], and Syed Ali Hussain

Additional article information

Abstract

Background

Mobile apps for health exist in large numbers today, but oftentimes, consumers do not continue to use them after a brief period of initial usage, are averse toward using them at all, or are unaware that such apps even exist. The purpose of our study was to examine and qualitatively determine the design and content elements of health apps that facilitate or impede usage from the users' perceptive.

Methods

In 2014, six focus groups and five individual interviews were conducted in the Midwest region of the U.S. with a mixture of 44 smartphone owners of various social economic status. The participants were asked about their general and health specific mobile app usage. They were then shown specific features of exemplar health apps and prompted to discuss their perceptions. The focus groups and interviews were audio recorded, transcribed verbatim, and coded using the software NVivo.

Results

Inductive thematic analysis was adopted to analyze the data and nine themes were identified: 1) barriers to adoption of health apps, 2) barriers to continued use of health apps, 3) motivators, 4) information and personalized guidance, 5) tracking for awareness and progress, 6) credibility, 7) goal setting, 8) reminders, and 9) sharing personal information. The themes were mapped to theories for interpretation of the results.

Conclusions

This qualitative research with a diverse pool of participants extended previous research on challenges and opportunities of health apps. The findings provide researchers, app designers, and health care providers insights on how to develop and evaluate health apps from the users' perspective.

Electronic supplementary material

The online version of this article (doi:10.1186/s12889-016-3808-0) contains supplementary material, which is available to authorized users.

Keywords: Mobile apps, Smartphone, Technology acceptance, Qualitative study, Self-regulation, Health promotion, mHealth, Adoption, User perception

Background

Currently over 97,000 health-related apps (termed as health apps hereafter) are available in the health and fitness category of the

IL PROGETTO

Il progetto AIGkit nasce dalla volontà di AIG e AIM aiutare i pazienti con malattia di Pompe a gestire la propria malattia e monitorare il proprio stato di salute, aiutare i clinici a controllare i progressi dei propri pazienti attraverso i dati reali e raccogliere dati sulla patologia utili alla comunità scientifica.

L'utilizzo della tecnologia nel progetto
è volto a rendere il progetto accessibile a tutti,
sempre e ovunque in modo semplice e veloce.

UN PALLONCINO PER SPERARE
AIG

"Together we are strong"

+ aIM + VIDIEMME CONSULTING =

AIGKit

IL PROGETTO

Cosa abbiamo fatto ?

FASE 1

Applicazione per smartphone
iOS, Android e Windows Phone
Disponibile da: **Gennaio 2016**

Numero di download dagli store

iOS	ANDROID	Windows 8 Phone
313	28	6

Portale web dedicato ai centri
Rilascio previsto: **Aprile 2017**

IL PROGETTO

Cosa dobbiamo fare ?

FASE 2

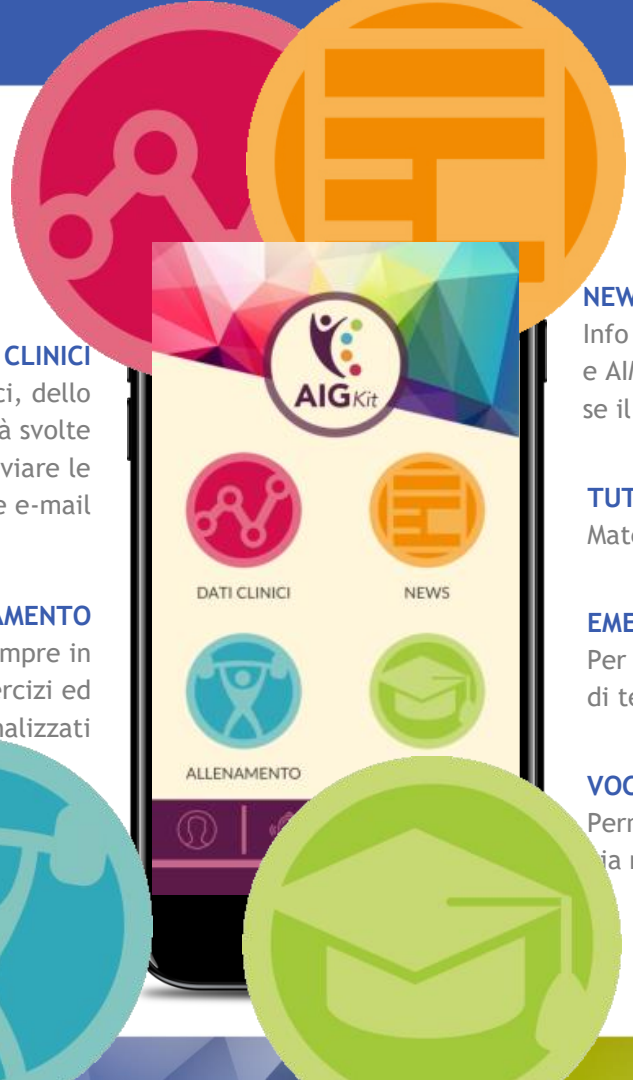
Portale web dedicato ai centri
Rilascio previsto: **Aprile 2017**

Implementazione di una piattaforma di archiviazione ed elaborazione dati inerenti la patologia, consultabili costantemente in remoto, sia a scopo assistenziale che di ricerca

Aggiornamento e sviluppo delle interfacce interattive

PRIVACY & SICUREZZA DEI DATI

APPLICAZIONE



DATI CLINICI
Inserimento dei parametri clinici, dello stato di salute e delle attività svolte durante il giorno, possibilità di inviare le informazioni raccolte tramite e-mail

ALLENAMENTO
Esercizi per mantenere il corpo sempre in movimento, possibilità di creare esercizi ed allenamenti personalizzati

NEWS
Info e comunicazioni dal mondo AIG e AIM, funzionalità disponibile solo se il device è connessione alla rete

TUTORIAL
Materiali legati alla patologia

EMERGENZE
Per contattare in maniera rapida numeri di telefono salvati in precedenza

VOCE AMICA
Permette di contattare telefonicamente o ia mail il servizio di supporto dell'AIG

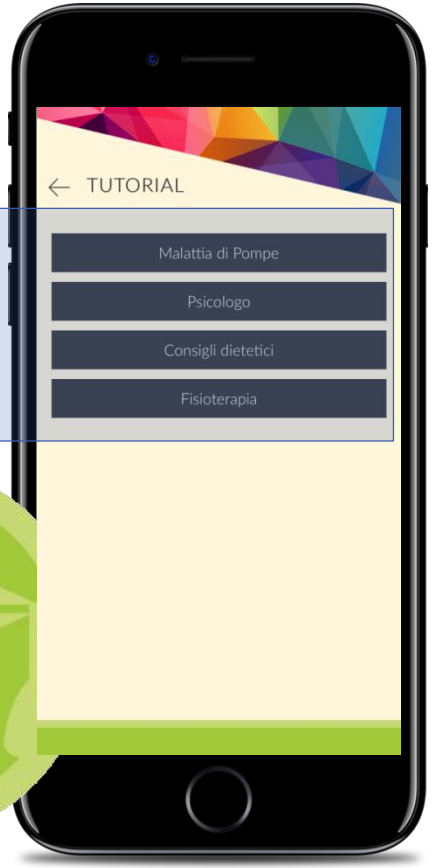
APPLICAZIONE

News

La sezione News viene popolata automaticamente con le news dei siti AIG e AIM ordinate cronologicamente

Tutorial

Nella sezione tutorial sono organizzati per categoria contenuti di supporto testuali o in forma di video



APPLICAZIONE

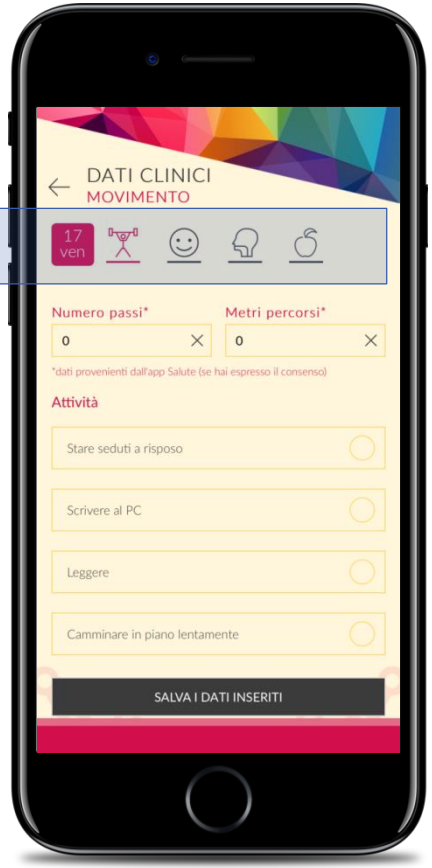
Nuova infusione

Calendarizzazione delle infusioni da effettuare, al paziente verrà ricordato il prossimo appuntamento tramite notifiche sull'app

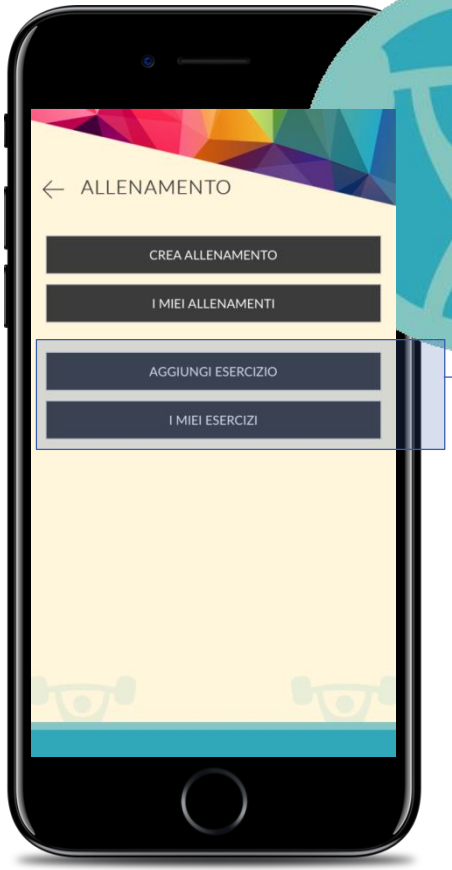
Dati clinici

Raccogliere dati relativi a:

- **Movimento** passi, metri percorsi e attività svolte
- **Stato di salute**
- **Respirazione** frequenza, saturazione e flusso
- **Alimentazione** calorie e tipologia di alimenti assunti



APPLICAZIONE

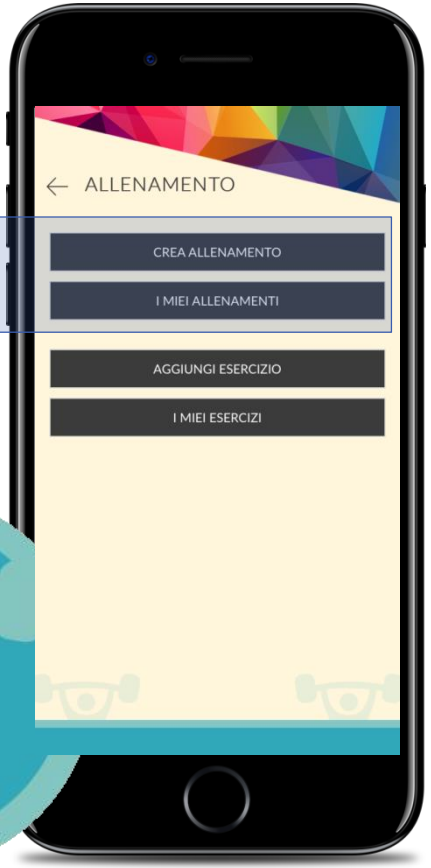


I miei esercizi

Creazione di esercizi utilizzabili per comporre le schede di allenamento.

I miei allenamenti

La sezione allenamento ha lo scopo di comporre schede di allenamento a partire da esercizi inseriti in precedenza



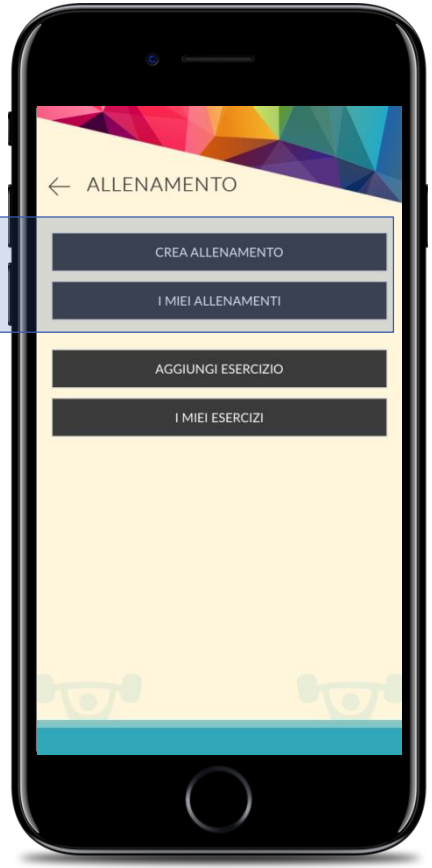
APPLICAZIONE

Emergenze

I numeri salvati in dati personali sono utilizzati per la effettuare una chiamata di emergenza attraverso gli appositi bottoni. L'sms con la propria posizione viene invece inviato al primo numero inserito

Impostazioni

Nelle impostazioni è possibile inserire il codice fornito via mail in fase di registrazione nel proprio centro ospedaliero per iniziare a inviare i dati clinici al proprio medico

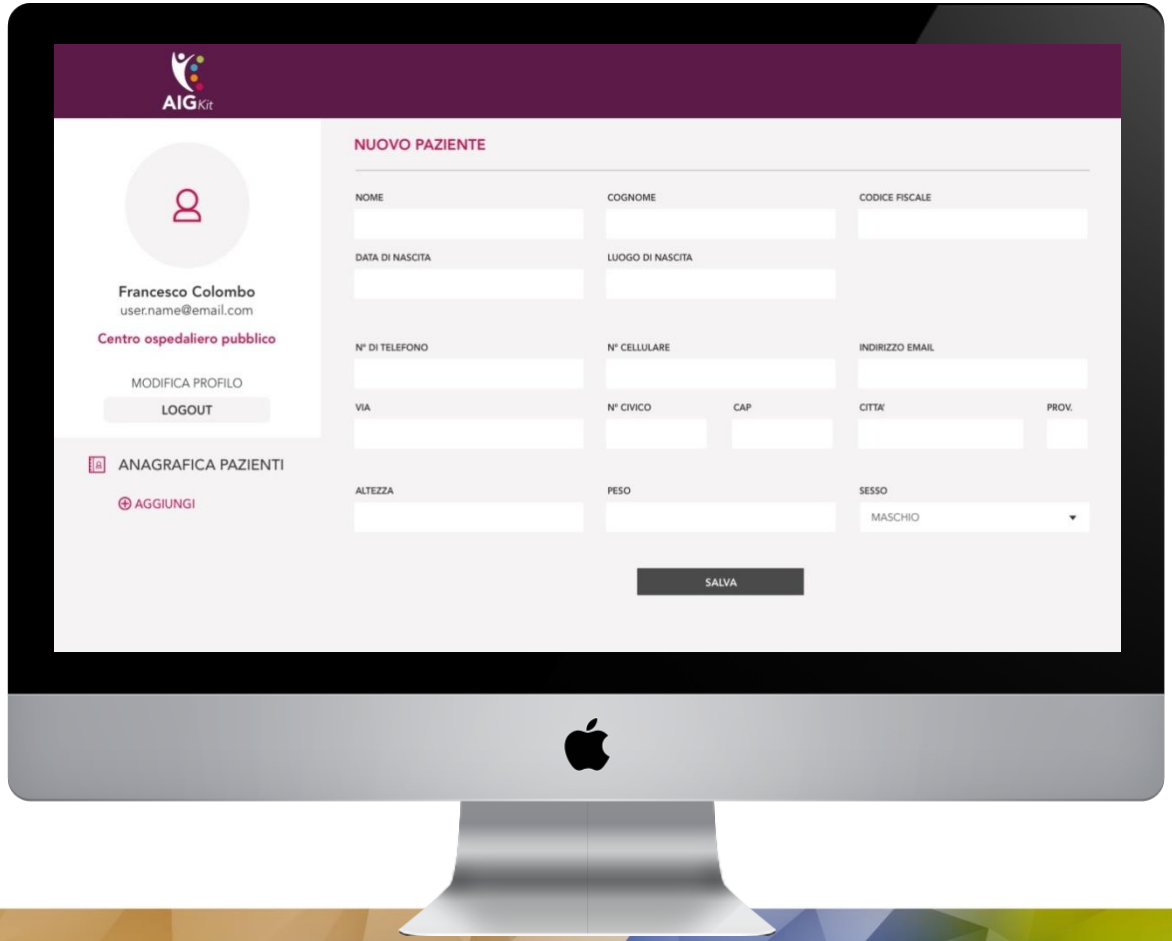


PORTALE

I Centri Ospedalieri interessati, attraverso AIM, AIG o scrivendo ad **aig@vidiemme.it**, potranno aderire al progetto ed avere un'utenza Admin attraverso cui gestire in autonomia il proprio team e i propri pazienti.

Per ciascun Centro sarà possibile avere utenze specialistiche diverse (neurologo, psicologo, pneumologo) e utenze in sola lettura per infermieri e specializzandi.

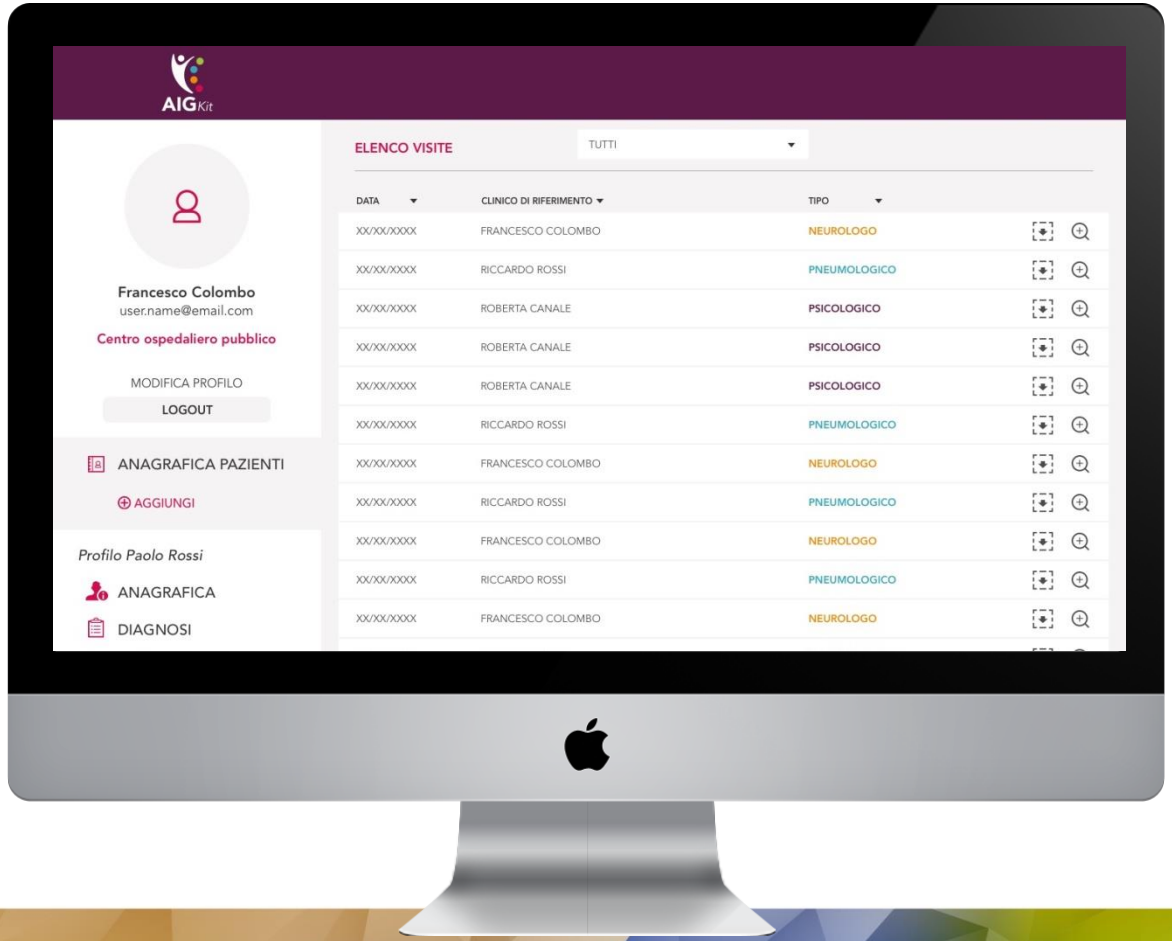
PORTALE



ANAGRAFICA E DIAGNOSI

Ciascun specialista potrà creare nuovi pazienti inserendo una scheda di anagrafica e una di diagnosi.

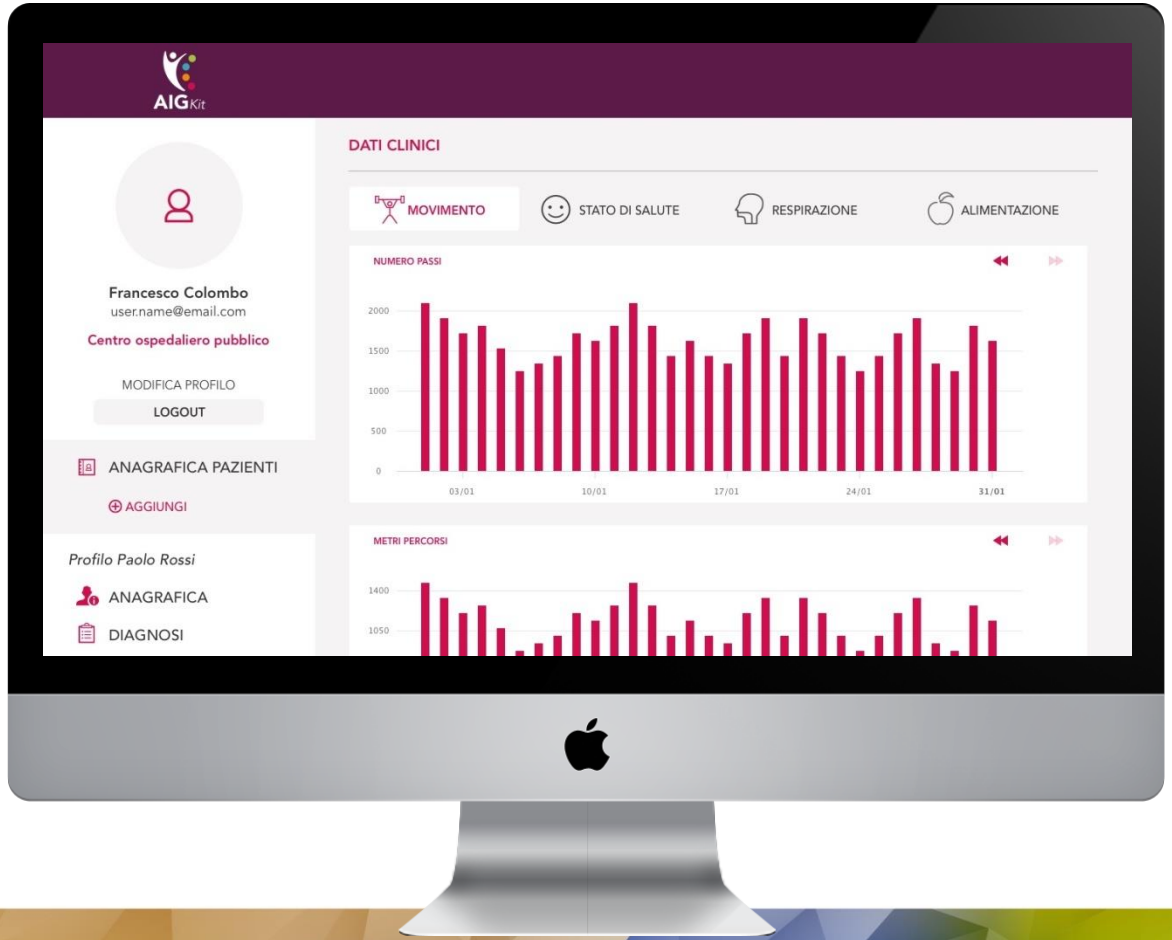
PORTALE



VISITE

Ciascun specialista potrà creare soltanto le schede visita per la sua specialità e visualizzare le visite fatte dai colleghi dello stesso centro.

PORTALE



DATI CLINICI

Nei profili dei pazienti che hanno autorizzato la sincronizzazione gli specialisti potranno visualizzare i dati raccolti attraverso l'applicazione in forma di grafici e tabelle.
I dati visualizzabili sono tutti quelli della sezione DATI CLINICI dell'app e quindi quelli relativi a movimento, stato di salute, alimentazione e respirazione.

European Reference Network
for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)

EURO-NMD | Building bridges and breaking barriers in rare neuromuscular diseases

Proposal of a Biotech and Bioengineering Activity Group

Neuromuscular disorders (NMD) is a broad term that encompasses multifaceted pathological conditions, associated to different extents to motor and psychological disabilities

Aims: to promote a multidisciplinary group interested in aspects like

- → **accessible services integration**
- → **health monitoring**
- → **independent living applications and autonomous mobility, in-home and domotic sensors**
- → **user interfaces and adaptive systems**

BANDO PER L'ASSEGNAZIONE DI UNA BORSA DI RICERCA

- **Proponente:** Associazione Italiana di Miologia (AIM), con il patrocinio AIG (Associazione Italiana Glicogenosi)
- **Titolo della ricerca:** Studio di fattibilità dell'uso nel setting clinico di una smartphone-APP per il monitoraggio domiciliare di pazienti con malattia di Pompe dell'adulto
- **Durata**: 8 mesi
- **Importo:** euro 10.000 lordi
- **Provenienza del fondo:** AIG (erogazione liberale non condizionata)
- **Selezione**
- I candidati dovranno possedere i seguenti requisiti:
- - Essere iscritti all'Associazione Italiana di Miologia (AIM) da almeno un anno
- - Età inferiore a 35 anni
- - Laurea in Medicina e Chirurgia

AIG Kit

LA MALATTIA DI POMPE

cosa è cambiato dopo 10 anni di Terapia Enzimatica Sostitutiva

Thanks to:

U. Ubaldi
F. Carlini
C. Proietti
G. Ricci
S. Baldanzi
G. Antonini
A. Vianello

AIG:
ASSOCIAZIONE ITALIANA
GLICOGENOSI ONLUS

Grazie per l’attenzione!