Strategic Plan for Research and Innovation in Health of Catalonia 2012 - 2015

NAME OF ORGANISATION: Ministry of Health of Catalonia / Departament de Salut

REGION: Catalonia, Spain

TOTAL REGION POPULATION: 7, 546,522

BEST PRACTICE TARGET POPULATION:

· The total population of Catalonia

· The health sector of Catalonia

· The research and innovation centres and institutes of Catalonia

MAIN FOCUS OF BEST PRACTICE: Regional strategic plan for research and innovation.

SUMMARY

The strategic plan for Research and Innovation in health of Catalonia aims to fall within the context of the Europe 2020 Strategy. In addition, life sciences research has experienced an outstanding development in Catalonia since 2000, thanks to a remarkable effort of autonomous Government. R&D investment has risen from 1.28% of GDP in 2002 to 1.63% in 2010. Catalonia has created a prominent network of research centers, accounts for 2.98% of European scientific articles, achieved 1.94% of FP7 funds and Catalan research groups were awarded 68 ERC grants. The latter is the baseline for the driving force for the implementation of a new strategy for Catalonia in innovation and research.

DESCRIPTION

The strategic objectives of the Government Plan and the departmental Plan are outlined below:

A. To optimize the structure of the biomedical research sector and to promote business management within it in terms of investment in and transfer of knowledge and the application of results.

1) Integrating the policy of research into health care into the Catalan research policy.
2) Adapting the institutes to the research model and drawing up a plan for introducing shared research units.
3) To define the evaluation process for research institutes in terms of results.
4) To promote the Catalan Bioregion

B. To promote innovation in health care

1) Integrating the policy of research into health care into the Catalan innovation policy.
2) Promoting evaluation and transfer of knowledge that adds value to the results, to the applicability of the research, and to their yield for the health system and the productive sector.
3) Approving and certifying innovative products and services in the field of health technologies.
4) Promoting and encouraging innovation in health care organizations in accordance with the strategic lines of the health care policy.

The actors who will implement this plan:

The Programme for research and innovation in health care from the General Directorate of Health Care Regulation, Planning and Resources (DGRPRS in Catalan) is responsible for planning and monitoring the implementation of the strategy set out in this document. The actors who will implement it are the

staff of the Department of Health who work in these tasks and two institutions assigned to the Department of Health: the public company known as the Agency for Health Information, Assessment and Quality, AIAQS (the Tic Health foundation regularly collaborates with this), and the Biocat foundation, which is the driving force and stimulant for the Catalan BioRegion cluster. In addition, ACC1Ó from the Department of Enterprise and Employment is a key public company in developing innovation policies. Coordination of the activities specified in this strategic Plan with the policies of the Directorate General for Research (DGR) of the Department of Economics and Knowledge (DEC in Catalan) is essential.

INNOVATION, IMPACT AND OUTCOMES

1. Integrating the health care research and innovation policies with the existing ones in Catalonia.
2. Investing efficiently in the pursuit of excellence in health care.
3. Promoting the Bioregion of Catalonia.
4. Promoting knowledge translation and assessing the impact of the research.
5. Encouraging innovation in healthcare organizations.

Indicators will be defined annually and the various activities will be controlled via the indicators. The Advisory Council for Research and Innovation in Health will be informed. In addition, once the period for which this strategic Plan has been designed has ended, an external evaluation will be delivered that will assess the extent to which the objectives have been met and the activities have been implemented.

At the end of the period of this strategic Plan, the following is expected to be achieved:

- Maximum transparency in distribution of resources among the research centres and institutes.
- To reach a critical mass and more efficient use of the health research system's resources.
- Greater accountability in the health research system as regards the scientific and professional community and citizens in general.
- Increased local and international visibility of Catalan healthcare research and innovation.
- An increase in translational research and knowledge transfer, which must ensure an impact in terms of improved health care and in generating wealth through the creation of spin-off companies, patent licenses and contracts.
- Greater entrepreneurial business fabric within the healthcare sector that should become an economic engine for the country.

Expected benefits of the Plan:

The actions of the five lines of activity give rise to products such as: a) SIRECS, an information system for research into health care, using the universities' information system, UNEIX; b) a polynomial or model of financing for healthcare research institutes and centres; c) a map of health research in Catalonia; d) biennial Biocat reports; e) an evaluation of the social yield from health research; f) research funding figures; g) innovative public procurement projects; and h) the creation of the Advisory Council for Research and Innovation in Health, among others.

Indicators will be defined annually and the various activities will be controlled via the indicators. The Advisory Council for Research and Innovation in Health will be informed. In addition, once the period

for which this strategic Plan has been designed has ended, an external evaluation will be delivered that will assess the extent to which the objectives have been met and the activities have been implemented.

ETHICAL ISSUES

This strategic plan should enable compliance with the recommendations of the European Science Foundation for having strong basic biomedical research in Europe with strong clinical research and strong translational research so as to bring innovative ideas into clinical practice and into public health care.

TRANSFERABILITY TO OTHER REGIONS

The Strategic Plan for Research and Innovation in Health of Catalonia sets up a framework for the implementation of a feasible strategy for research and innovation. The plan includes various specific tools to develop the strategy which can be transferred to other regions.

KEY LEARNING POINTS

Accreditation of Research Institutes and centres according to EU standards with the aforementioned activities and products, in the medium-term the following is expected:

- Funding model for the health research centres and institutes based on results.
- Information system that provides a set of data to enable evaluation, in the first phase, of the health research system as a whole retrospectively agreed upon with the stakeholders in the research, which will be useful to them for their information and management, and which should be simple and cost-efficient in terms of benefits.
- Increasing ease dissemination of the processes and results of the health research and innovation, making use of the Department's tools, its website (Health Channel) and the Advisory Council for Research and Innovation in Health.
- Increasing the participation and leadership of the stakeholders in health research and innovation in European and international research tenders.
- Increasing the number of researchers who are also professionals in clinical or public health.
- Establishing processes for innovative public procurement in the Government Administration.

FURTHER INFORMATION

www.gencat.cat/salut

www.euregha.net

CONTACT

Gabriel Capellà gcapella@gmail.com Toni Dedeu tdedeu@gmail.com

Policy Research Centre Welfare, Health and the Family (PRC WH&F)
Steunpunt Welzijn, Volksgezondheid en Gezin (SWVG)

NAME OF ORGANISATION: Katholieke Universiteit Leuven

BEST PRACTICE TARGET POPULATION: Flanders

REGION: Flanders, Belgium

TOTAL REGION POPULATION: 6.25 million

MAIN FOCUS OF BEST PRACTICE: The Flemish government – the minister of Science Policy and the minister of Welfare, Health and the Family – established a Policy Research Centre on Welfare, Health and the Family (i.e. a consortium of the Universities KU Leuven, UGent and VUB and its partners) to support the policy of the Flemish minister of Welfare, Health and the Family in the development of an evidence-informed policy.

SUMMARY

The Policy Research Centre (PRC) Welfare, Health and the Family is a consortium of research centres from different universities in Flanders that carries out a multi-annual research program and short term research projects in order to inform accurately the policy of the minister of welfare, health and the family. The coordination of the PRC is executed by the promoter coordinator and the executive board. The interdisciplinary network of promoters and experts is one of the main values of the PRC.

DESCRIPTION

The Policy Research Centre Welfare, Health and the Family is part of a broader Policy Research Program. This program offers the possibility for ministers and policy makers to formulate their research questions through a call. The researchers formulate research projects related to the topics stated in the call of the minister of Welfare, Health and the Family.

Mission

The PRC supports the Flemish minister of Welfare, Health and the Family in the development of a strong, innovative, effective, inclusive, integrative and evidence-informed policy. The PRC does this by a coordinated and multidisciplinary approach that permits the analysis, the understanding and the management of the welfare, health and family problems in all their complexity in our society.

General objectives

- gathering, analysing and disseminating policy-relevant data concerning well-being and health
- conducting long and short period policy-relevant research with attention to Flemish policy strategies such as Flanders In Action and Pact 2020
- providing scientific consultancy concerning policy relevant topics.

Interdisciplinary research by highly qualified experts

The composition of the consortium guarantees interdisciplinary research on welfare, health and their integration in primary care. The promoters of the research projects are highly qualified and (internationally) renowned experts with expertise in different fields of policy relevant research (e.g. youth, disability, mental health, ageing etc.).

An extensive network of experts

The network enables the PRC to find a research answer in a short time frame on urgent questions of the Minister of Welfare, Health and the Family, such as:

- Research on the organisation of primary social care services in major cities
- Researching the needs for mental health care in Flanders
- Researching the use of primary care services by people with disabilities

Multi-annual program and short term research projects

The multiannual research program (2012-2015) consists of three research lines:

(1) Monitoring Needs and Use of Care
(2) Evaluation of Preventive and Care Interventions
(3) Organisation of Care and Policy

Some examples of research topics concerning primary care within these research lines are:

- An evaluation of primary care
- An evaluation of an effective integrated care intervention in different settings
- The development, implementation and evaluation of an e-health and prevention intervention in primary care
- Searching for commonalities in registration systems in health and social care

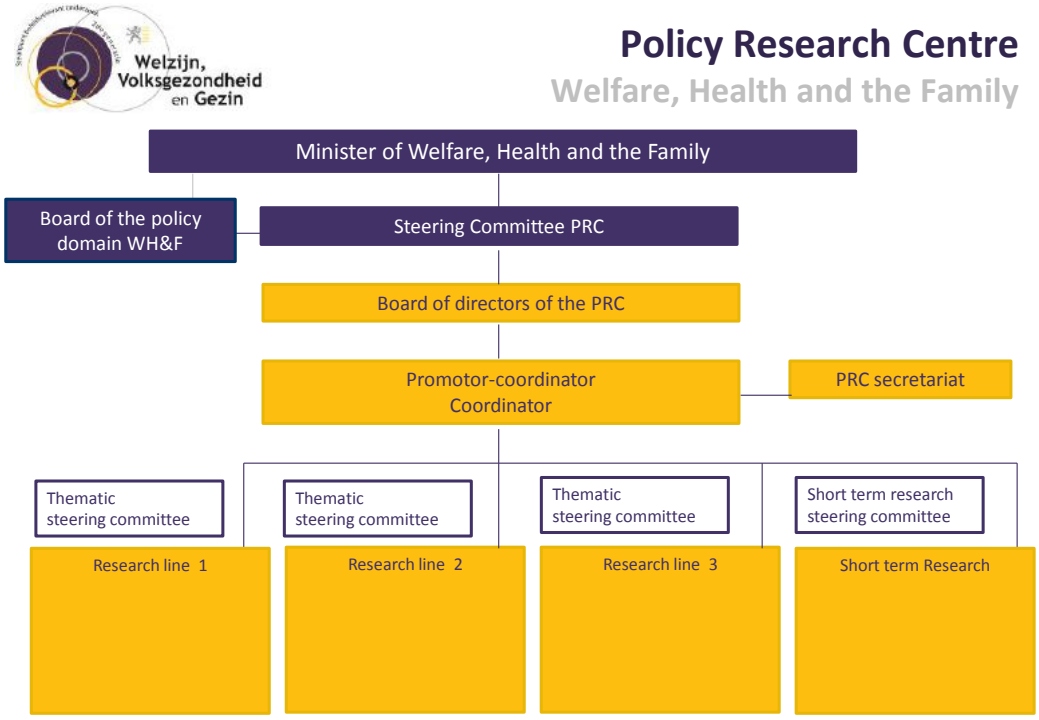
Valorisation

The PRC makes its knowledge and know how accessible and transferable to the Flemish government but also to the public. Valorisation is organised across a combination of three perspectives:

- Policy oriented valorisation: facts & figures, reports and (working) papers, with special attention for synthesis and policy recommendations.
- Scientific valorisation: articles in scientific (inter)national journals, participation at international conferences.
- Societal valorisation: informing partners and practitioners during or at the end of a research project through more popular forms of communication (e.g. newsletter, articles in practitioner oriented journals). Regularly the PRC organises conferences, seminars and learning networks with multiple stakeholders.

The valorisation is initiated, organised and/or supported by the coordinating centre of the PRC. The PRC chooses for each research project for the most relevant valorisation strategy, with priority for policy oriented valorisation.

Coordination (orange) and Steering by the government (purple)



ETHICAL ISSUES

- Policy, practice and research are 3 different disciplines and perspectives concerning the realities of care and social welfare that don't necessarily agree with another. It is important to make hidden values of each discipline explicit and to develop a continuous dialogue to solve ethical dilemmas.
- The PRC is building continuously on a strong and positive relation with its stakeholders and policy actors in particular. On different levels steering committees have been established (as the scheme above illustrates). On the one hand it enables the Minister and its administration to oversee compliance with the contract. On the other hand thematic steering committees facilitate a constructive dialogue regarding the research projects. At the same time, the PRC pays special attention to the independency of its research.

KEY LEARNING POINTS

- Policy oriented research in a network based approach demands new types of interdisciplinary and interuniversity collaboration and coordination. The PRC has a strong vision on policy research in a complex societal context.
- The multidisciplinary composition of the PRC offers unique opportunities, which are strongly facilitated by the policy research programme. The multidisciplinary composition facilitates an interdisciplinary knowledge transfer within and between research projects and programmes resulting in innovative multidisciplinary and interdisciplinary knowledge on health and welfare which would not have been developed otherwise.
- The network approach of the PRC and the range of experts within with experience in policy oriented research enables the PRC to answer also adequately on more short term research questions of the Minister.

- The general management and coordination of the network is one of the strengths of the PRC and is of considerable importance in facilitating cross-fertilization between programmes and dialogue with policy and other stakeholders. The PRC is a learning organisation.
- The (junior) researchers of the PRC built strong competencies in doing scientific and policy oriented research which is a favourable benefit on the labour market.

FURTHER INFORMATION

Prof. Chantal Van Audenhove
Promotor coordinator PRC WH&F
chantal.vanaudenhove@med.kuleuven.be

Valérie Carrette
Coordinator PRC WH&F
valerie.carrette@med.kuleuven.be

swvg@med.kuleuven.be
www.steunpuntwvg.be
Kapucijnenvoer 39, bus 5310, 3000 Leuven

Regional Program for Research, Innovation and Health Technology Assessment (PRIHTA)

NAME OF ORGANISATION: Veneto Region

REGION: Veneto, Italy

TOTAL REGION POPULATION: 4,936 million (2010)

MAIN FOCUS OF BEST PRACTICE: PRIHTA promotes the innovation through the engagement of private stakeholders

SUMMARY

The Veneto Region identified a network of structures and excellences in the field of Research and Innovation in the Health sector, with the aim to increase the scientific and specialized knowledge of selected professionals.

To facilitate it, the Regional Program for Research, Innovation and Health Technology Assessment (PRIHTA) has been established by the Regional Decree n. 2187 on 8.8.2008. The main goal of the program is to promote a culture of innovation, research and technical evaluation in the field of Healthcare.

The program offers the opportunity to involve private stakeholders in research and innovation activities, in accordance with the general project's guidelines and coherently with the Regional Health Plan.

The Public-Private Partnerships (PPPs) allow the Veneto Region to diversify the funding and consent the private sector to establish new collaborations with the public institutions in the field of healthcare. Each year the Veneto Region issues a call for proposals in the fields of research, innovation, and education in health, always involving PPPs. All the subjects identified in the calls are the result of a common vision within the public and private sectors; which overall leads to an improvement of the system's quality and experiences new models of healthcare.

DESCRIPTION

Each year the Veneto Region identifies the topics on which Local Health Units, Region and private stakeholders can collaborate.

The main goal is to establish a public–private dialogue on specific themes of common interest. The identified objectives are related to the current programming period and are defined year by year.
The projects are presented by both public and private actors pursuing a common interest and following the models offered at regional level. The final evaluation is conducted by the working group of PRIHTA on the base of consolidated criteria established not only at regional, but also at national and international level.

Foundations, Universities and consortia active in the field of research and Innovation, together with private actors, Local Health Units, hospitals and Scientific Institutes of Medical Research (IRCCs) are welcome to present their expression of interest.

The Local Health Units can contribute with in kind funds, while the private actors bring cash, technology or human resources. The Region does not invest funds, but support from an administrative point of view the projects. Overall the initiative is basically self-financed.

INNOVATION, IMPACT AND OUTCOMES

The typologies of PPPs introduced by the PRIHTA are innovative and different from the usual collaborations put in place by the industrial sector, this because the projects are designed with a common view shared by all the actors. In addition the Veneto Region is singing the trilateral agreement and is acting as monitoring entity, transferring the results, in case of a positive outcome. Currently the first two successful projects are reaching their final approval. The private sector already acknowledged in different occasions the merits of the initiative.

ETHICAL ISSUES

Clinical and care pathways (PDTA) as well as best practices have been experimented thanks to the industry and the private sector involvement in regional projects. Those experiences improved the patient's quality and the access to care.

The involvement of stakeholders it is a very sensitive and delicate issue carefully considered not only from the PPPs angle, but also in platforms involving pharmaceutical companies, medical devices and large equipment, all aspects touched by the PRIHTA.

TRANSFERABILITY TO OTHER REGIONS

The opportunity to establish PPPs within the Veneto Region's system promotes a very important dialogue, especially during an historical moment with very limited resources. These circumstances encourage the research of always new solutions, using the excellences identified in the public and in the private sector, although always keeping the patient at the centre.

KEY LEARNING POINTS

- Importance of establishing a dialogue with the private sector
- Relevance of the collaboration with various stakeholders in specific areas, with the goal of improving the patient's health.
- Establishment of research networks.

FURTHER INFORMATION

http://www.regione.veneto.it/web/sanita/ricerca-innovazione-ed-hta

CONTACT

Mrs Teresa Gasparetto - teresa.gasparetto@regione.veneto.it

eMeDocS (exchange Medical Documents System)

NAME OF ORGANISATION: Kraj Vysocina

REGION: Vysocina, Czech Republic

TOTAL REGION POPULATION: 510 520

BEST PRACTICE TARGET POPULATION:
Health professionals and patients

SUMMARY

The content of this project is, apart from the regional exchange of the medical records (MR) study, also a pilot study. The pilot study is especially significant since it deals with practical aspects of the exchange of MR among 5 hospitals and the Medical Rescue Services of the Vysocina region, moreover the study has a potential for other medical institutions and private practitioners. Its importance also lies in its architecture, complexity and diversity in the sense of the exchange of the MR among variety of organizations with separate legal personality and the exchange of data in a real time period. From this point of view, we talk about a unique project that is challenging regarding the legislative terms – especially concerning strict privacy of private data.

DESCRIPTION

The eMeDocS Project was launched in May 2010 when the public contract called Regional concept of an exchange of medical records among medical facilities founded by the Vysocina region including its realization through the form of a pilot project. In terms of work, the project was named Exchange Medical Documents System – eMeDocS and in the first half of 2011 it was completed and put into full operation. The Project was divided into two parts: the Conceptual Study and the Pilot Project. The conceptual study addressed the key areas for the exchange and sharing of the MR. To put it more specifically: an exchange of data among hospitals themselves and among the hospitals and the emergency services information system. Thanks to such an improvement, a handover of urgent information from all the hospitals' information systems of the region to the ambulances that are in the field in real time. Because of this, a doctor receives all important (supportive) information concerning the patient within a few seconds. Subsequently thanks to the eMeDocS Project the information could be handed over straight from the ambulance car to the hospital information system where being available to the hospital staff. Of course, everything is completely electronic. An exchange of documents among the hospitals in the sphere of patient reports, discharge reports and requisitions for X-ray is also addressed by the eMeDocS Project. The entire program was financed through the resources of the Vysocina region. Currently the Project is opened to other medical-type organizations and it gradually leads to an expansion of the eMeDocS Project beyond the Vysocina region borders.

INNOVATION, IMPACT AND OUTCOMES

The uniqueness of the eMeDocS Project is grounded in its heterogeneous environment and scope. There are five hospitals engaged in the Project and the Emergency Medical Service of the Vysocina region as well. The uniqueness of the environment and its complexity lies also in the fact that there are five totally diverse information systems operating within the space of six engaged entities. For this

reason they began to seek such a solution that would be entirely independent of the end user and his information system and that would be focused especially on the mode of handing over of the particular information with the help of national or international data standards. Such a project regarding its scope and complexity has not been realized in the Czech Republic yet. In fact it is not only about 'sharing of the medical materials' (as not being enabled by the Czech legislation), but it is really the exchange. The effectiveness of the Project is very good since we refer to the project which is a very supportive tool in savings of people's lives. From this viewpoint the benefits and effectiveness of the whole Project is not calculable. The expected effectiveness lies in further steps leading to broadening of the functionalities in the area of MR and also in the opportunity of great utility value for other medical facilities in the Czech Republic and outside it as well. There were several goals concerning this Project. One of the objectives was to develop a study describing possible options for the Exchange of MR in a heterogeneous environment within the Vysocina region. Also the study should have mapped the current situation on the national and international level and subsequently should have reflected the concept for the Exchange of MR in the region so that the consecutive steps would not be counterproductive. Ultimately, the exchange of MR among the above mentioned subjects (hospitals and Medical Rescue Service especially) represented an inseparable part and not humble, but fulfilled goal of the Project.

ETHICAL ISSUES

Patient is not bothered by repeated queries.

TRANSFERABILITY TO OTHER REGIONS

Taking into consideration the fact that the entire system has been built above the national and also international standards being in conformity with the IHE concept, it is possible to implement this project in other regions as well.

KEY LEARNING POINTS

- The uniqueness of the eMeDocS Project is grounded in its heterogeneous environment
- A project of this magnitude has not been implemented in the Czech Republic yet

FURTHER INFORMATION

http://www.emedocs.cz

CONTACT

http://www.emedocs.cz/kontakty
Mgr. David Zažímal, zazimal.d@kr-vysocina.cz

Eastern Academic Health Science Network

NAME OF ORGANISATION: Eastern Academic Health Science Network

REGION: East of England, UK

TOTAL REGION POPULATION: 5, 847, 000

BEST PRACTICE TARGET POPULATION: Academia, research organisations, hospitals, primary care, public health, social care providers, the voluntary sector and industry.

MAIN FOCUS OF BEST PRACTICE: Bridging the vital gap between world-class research and top-quality clinical care.

SUMMARY

There is a strong research base across the East of England. 25% of all UK private sector investment in research and development is spent in the region. Thirty of the world's leading research institutions are based there. Established in 2013, the Eastern Academic Health Science Network (EAHSN) is a region-wide network with the overarching priority to spread best practice and promote adoption of innovation into health service delivery in the East of England.

The EAHSN's vision is to improve patient and health outcomes for the population at scale and speed through:

- collaboration with a network of local, national and international partners,
- promoting participation in research,
- translating public and commercial research and learning into practice,
- driving service innovation and economic growth through partnerships with industry.

DESCRIPTION

The EAHSN has developed an organisational model designed to achieve full integration across the regional health system.

There is a need for effective and flexible interfaces between academia, research organisations and health delivery systems. This has been achieved by creating a structure with a centre and four geographical 'nodes'. The centre is the EAHSN which is established as a not-for-profit company. Each local node relates to a natural clinical community. They engage with partners from local government; primary, secondary and tertiary care; social care; public health; and industry. Partners within the nodes agree to work to a shared vision. This has developed a network of robust relationships across the region encompassing all stages of the patient pathway.

The EASHN's work plan focuses on three key areas: research, service innovation and wealth creation. Each area has an agreed strategic objective, a set of 5 year aspirations, and a list of 18 month deliverables.

EAHSN is funded by government (NHS England). In addition to this, it will be looking to leverage in additional funding, particularly through work with industry.

INNOVATION, IMPACT AND OUTCOMES

Partners of EAHSN have identified seven clinical priority areas for service innovation: Stroke, Cardiovascular disease, Cancer, Mental Health, focusing on dementia in the first year, Diabetes, Chronic respiratory diseases, Patient safety.

These are to be addressed by seven Clinical Study Groups, each incorporating both clinical and academic leadership and having agreed work programmes. Each Clinical Study Group will review the variation in patient outcomes and the evidence for best practice. They will put forward proposals for the adoption and spread of best practice across the region, or make proposals for piloting new innovations, where there is not yet an evidence base for those innovations. The proposals developed by the Clinical Study Groups will be assessed by a Funding Committee and those that are supported will quickly be implemented. For example the Cancer CSG has launched the 'Survivorship Project' to improve follow-up care through a tailored service of integrated care between all teams and services in the community.

To achieve maximum impact the EAHSN is also targeting resources towards joint initiatives with industrial partners. In particular it is working closely with large and small industry in the region to create joint programmes that bring the NHS and industry closer together to foster new innovative solutions to improve patient care. For example, eight companies have been awarded a total of £5m to develop innovative solutions to improve the experience of people with mental health illnesses and people at the ends of their lives. The projects are expected to bring quality and cost benefits to the health service through improved healthcare delivery.

EAHSN is establishing an Industry Advisory Panel create a dialogue between industry and the NHS on appropriate metrics, tools, delivery mechanisms and activities. This will build a lasting relationship that supports economic growth.

ETHICAL ISSUES

Each of the Clinical Study Groups' activities includes the cross-cutting objective of identifying and addressing inequalities both in outcomes and in the delivery of care. Partners agree to work together under an agreed set of behaviour.

TRANSFERABILITY TO OTHER REGIONS

The approach will be of interest to regions exploring the value of bridging the gap between researches and implementing innovation in clinical care, and establishing a fully integrated model.

KEY LEARNING POINTS

- the importance of partners working collaboratively towards a shared vision, values and set of behaviours
- bringing clinical providers and industry closer together and enabling them better to understand their relative environments
- establishing mechanisms for translating research into practice, identifying and disseminating best practice to improve outcomes and address inequalities.

FURTHER INFORMATION

http://www.eahsn.org.uk/index.php

CONTACT

Ms Karen Livingstone, Director of Partnerships & Industry Engagement, EAHSN
karen.livingstone@nhs.net ; enquiries@eahsn.org.uk

Multimorbidity clinic for chronic diseases (MACVIA-LR)

NAME OF ORGANISATION: La Region Languedoc Roussillon

REGION: Languedoc Roussillon, France

TOTAL REGION POPULATION: 2.3 Million

BEST PRACTICE TARGET POPULATION: over 25% for patients with major chronic diseases (WHO definition) in CHU Montpellier (end 2014)

MAIN FOCUS OF BEST PRACTICE: Chronic respiratory diseases, cardiovascular diseases, diabetes, risk stratification, integrated care.

SUMMARY

- Public health provider: Regional Health Agency (ARS), Regional health insurance agency (CPAM).
- Academia: UM1 (Montpellier), Ecole des Hautes Etudes Pratiques (ITEV, UM2).
- Hospitals: Montpellier, Nîmes: Chronic disease clinic (co-morbidity) and mobile remote rural area chronic disease clinic.
- Private and public health care workers: Primary and secondary care physicians, pharmacists, physiotherapists, nurses, occupational therapists.
- Conseil Régional LR de l'Ordre des Médecins.
- URPS physicians, nurses, pharmacist (Unions of practitioners)
- Social carers.
- Schools and training centres for teaching and coaching.
- National public organisations, Research institutes: INSERM, CNRS.
- Industrial competitiveness**:** Eurobiomed (French government "competitive cluster"). It includes 140 companies with about 3,000 industrial researchers ranging from biotech companies to large pharmaceutical, health and ICT companies (Sanofi, Horiba, Bio-Rad, IBM).

DESCRIPTION

Integrated care pathways for chronic diseases have been initiated in hospitals (secondary care) and remote rural areas (primary care, end 2013). They include multi-sectoral care. The pilot studies should be deployed to the entire region by 2015. The goal is the MACVIA-LR global objective (reduction of hospitalisations and increase in HLY). Specific objectives are the number of patients included in primary care (including remote areas).

Our programme attempts to use all needed methods with all needed stakeholders. It is more the combination of several best practices (A1 to D4) developed in a stepwise approach that makes the

originality of the project rather than any best practice by itself. In chronic diseases, an ICT stratification tool using control, severity and risk is used [2].

Co-morbidity clinic: An evidence-based list of criteria on the major chronic disease co-morbidities is included in a one-day clinic. It includes the criteria for (i) the screening of co-morbidities of cardiovascular diseases (CVD), COPD or diabetes (D2M), (ii) the prediction of their exacerbations and (iii) the overall appreciation of severity. This common list is used for all patients referred to the clinic. Moreover, there are some age-specific criteria (falls A2 and frailty A3). ICT is implemented in the Montpellier hospital clinic. An interoperable CDSS is in process. Integrated care pathways will be available at the end of 2013. Coaching is part of this activity.

Mobile chronic disease clinic: Has been set up with interoperability with the co-morbidity clinic. Its purpose is to screen co-morbidities in remote areas of the rural counties of the Region. DeProPASS (Dépistage des Pathologies associées aux maladies chroniques) initiates pilot studies in the Maisons Médicales Pluridisciplinaires (article L.6323-3 Code Santé Publique) of the LR Region. This mobile clinic will then be deployed to the remote areas of the Region. Remote monitoring is included.

Integrated care pathways for chronic respiratory diseases: The objectives of AIRWAYS-ICPs are (i) to develop multi-sectoral ICPs for asthma and rhinitis for use across Europe and other countries, (ii) to allow the practical use of a combined asthma and allergy programme, (iii) to combine preventive and disease control strategies, (iv) to place a special emphasis on elderly patients and/or underserved patients, (v) to implement cost-effective policies on the prevention of asthma and allergy and (vi) to have an impact on AHA.
All B3 activities have a particular interest in cultural and societal aspects in a project centred on the patient.

INNOVATION, IMPACT AND OUTCOMES

Chronic disease clinic based on comorbidities and/or falls integrated with all components of health and social care to provide an integrated cost-effective solution across the region (pharmacists, physicians, nurses, social carers) which can be transferred to other regions.
The aim is to reduce avoidable hospitalizations for chronic diseases in the elderly by 20% in2020 (full MACVIA-LR project). HLY targeted to regions (currently developed by JM Robine) and QOL. These goals are for the entire MACVIA-LR project.

TRANSFERABILITY TO OTHER REGIONS

Transferability using expertise of the chronic disease programme which has been translated in 52 languages and transferred successfully to 64 countries[3].

FURTHER INFORMATION

macvia.cr-languedocroussillon.fr

CONTACT

Christian Bourquin (Président, Région Languedoc Roussillon)
Jean Bousquet (jean.bousquet@orange.fr)
Thierry Camuzat (Camuzat.Thierry@cr-languedocroussillon.fr)

Early diagnosis of severe combined immunodeficiency due to adenosine deaminase defect: a new simple and reliable method by tandem mass spectrometry

NAME OF ORGANISATION: Tuscany Region (Regione Toscana)
BEST PRACTICE TARGET POPULATION: Infants
REGION: Tuscany, Italy
TOTAL REGION POPULATION: 3.7 million
MAIN FOCUS OF BEST PRACTICE: Preventive pediatric medicine

SUMMARY

Adenosine deaminase (ADA)–severe combined immunodeficiency (SCID) is an SCID caused by a defect in the enzyme adenosine deaminase that can cause permanent damage or even be fatal. It complies with all of the criteria for inclusion in a newborn screening program, but screening methods are still expensive or provide a non-negligible number of indeterminate results. We performed a retrospective study to evaluate whether tandem-mass spectrometry (MS) and quantitative T-cell receptor excision circle (TREC) analyses of dried blood spots (DBS) could identify newborns presenting delayed-onset ADA-SCID later in life, and found that tandem-MS but not TREC quantification identifies newborns with delayed- or late-onset ADA deficiency. A comparative cost-effectiveness analysis between the TREC- and MS-based methods was performed; the extremely low cost per test of the MS-based method suggests that this test could be a worthwhile addition to newborn screening panels. The cost per test using tandem mass spectrometry is less than €0.01.

DESCRIPTION

Severe combined immunodeficiency (SCID) is a group of severe diseases that affect the immune system. Infants with SCID are healthy at birth but die of recurrent severe infection in infancy unless adequate therapy is provided. Unfortunately, most infants with SCID are not identified in the pre-infection period: the diagnosis is usually hypothesized when a severe infection occurs. At that time, however, even though a correct therapeutic intervention is begun, damage due to severe infection (such as meningitis, encephalitis, and severe pneumonia) can already be present and permanent sequelae can be an important burden for patients, family and society.

SCID due to a defect of adenosine deaminase (ADA) is an inherited disorder of purine metabolism. In its typical form, the absence of the enzyme ADA allows accumulation of toxic metabolites resulting on one hand in a severe immune system defect and, on the other, in permanent damage of other organs and systems such as the brain or liver. In these cases SCID-ADA is fatal within the first months of life if left untreated and is associated with severe sequelae if treated late.

Late onset ADA-SCID has also been described. In these cases the patients experience severe recurrent infections and chronic lung disease during infancy. Hematopoietic stem cell transplant is curative, but

dependent on a good donor match. Enzyme replacement therapy is available and determines the elimination of toxic metabolites and a good reconstitution of the immune system. Gene therapy is also an option for patients. In any case, whichever therapy is chosen, it should be started as soon as possible after birth in order to obtain good therapeutic effect.

At present, in children in whom an immunodeficiency is suspected, diagnostic methods aim to evidence ADA toxic metabolites in urine or a defect in ADA activity in blood obtained via venipuncture. Different methods have been described, but all of them can be applied to children in whom the diagnosis is already hypothesized.

We have recently shown that it is possible to identify patients with ADA-SCID using tandem mass spectrometry (tandem-MS) (PCT EP2010/070517) of DBSs collected at birth during routine newborn screening procedures at a low cost (€0.01 per test).

Once a newborn is identified to have ADA metabolite abnormalities in a screening program, he or she should undergo extensive characterization of ADA activity, dAXP levels and percentages, and immune function to determine the disease phenotype. The patient should then be followed to determine when the first symptoms appear, so that treatment can begin before damage occurs. The actual aims of screening programs are not to make diagnoses but to identify patients who need further characterization with specific tests and clinical follow-up. In the last couple of years most newborn screening programs for SCID have been based on quantitative analysis of TRECs. However, we recommend the inclusion of tandem-MS analysis of adenosine and 2-deoxyadenosine, having demonstrated that it is inexpensive and can identify newborns with severe and hypomorphic variants of ADA who might have SCID later in life. Moreover, tandem-MS can be useful in countries that cannot afford TREC analysis but are already using tandem-MS to screen for other metabolic diseases. In fact, analyzing a comparative cost-effectiveness analysis between the TREC- and MS-based methods, the extremely low cost per test of the MS-based method suggests that this test could be a worthwhile addition to newborn screening panels. A cost-effectiveness study on SCID screening has reported that models are sensitive to the cost of the test and that to maintain the cost at less than $100,000/quality-adjusted year of life, the screening for SCID should cost less than $15 per test. The cost per test using tandem mass spectrometry was less than €0.01. The new Tandem MS test was licensed from the Meyer Children's Hospital in Florence, Italy to develop a diagnostic kit in December 2012.
This project was funded by the Tuscany Region (Regione Toscana).

INNOVATION, IMPACT AND OUTCOMES

About 75,000 newborns were screened for this disorder and 1 case of ADA SCID (delayed form, P4) has been identified in Tuscany. This case represents the first case of delayed ADA SCID diagnosed in its asymptomatic form in the world. The method has been also applied to patients with clinical diagnosis of ADA SCID in all forms of the disease (7 patients).

An expanded newborn screening including more than 40 metabolic diseases is now performed by using tandem-MS in many countries in the world, so that millions of children worldwide are screened for those diseases each year. In the Tuscany region, ADA-SCID has been added to the panel of diseases for which tandem-MS screens. A pilot population-based screening program began in 2011 that uses both genetic testing (TREC analysis) and tandem-MS analyses to identify newborns with SCID.

TRANSFERABILITY TO OTHER REGIONS

ADA-SCID can be added to the panel of diseases that other regions/organizations screen for in order to identify newborns with SCID. The cost of implementation would be extremely low (less than €0.01 per test).

KEY LEARNING POINTS

- Adenosine deaminase (ADA)–severe combined immunodeficiency (SCID) can be fatal or permanently damaging if not diagnosed and treated quickly
- We have recently shown that it is possible to identify patients with ADA-SCID using tandem mass spectrometry (tandem-MS) of DBSs collected at birth during routine newborn screening procedures at a low cost (€0.01 per test).

FURTHER INFORMATION

Publications

1) Azzari C, la Marca G., Resti M.. Neonatal screening for severe-combined-immunodeficiency due to adenosine-deaminase defect: a reliable and inexpensive method by tandem-mass-spectrometry, J All Clin Immunol, 2011 Jun;127(6):1394-9. (I.F. 11.003)
2) la Marca G. Screening neonatale per malattie metaboliche ereditarie mediante spettrometria di massa, Rivista Italiana di Medicina Perinatale, 2011; 11: 6-9. ISSN 1591-7592
3) Speckmann C, Neumann C, Borte S, la Marca G, Sass JO, Wiech E, Fisch P, Schwarz K, Buchholz B, Schlesier M, Felgentreff K. Delayed-onset adenosine deaminase deficiency: Strategies for an early diagnosis, J Allergy Clin Immunol. 2012;130:991-4. (IF: 12.047)
4) la Marca G, Canessa C, Giocaliere E, Romano F, Duse M, Malvagia S, Lippi F, Funghini S, Bianchi L, Della Bona ML, Valleriani C, Ombrone D, Moriondo M, Villanelli F, Speckmann C, Adams S, Gaspar BH, Hershfield M, Santisteban I, Fairbanks L, Ragusa G, Resti M, de Martino M, Guerrini R, Azzari C. Tandem mass spectrometry, but not T-cell receptor excision circle analysis, identifies newborns with late-onset adenosine deaminase deficiency. J Allergy Clin Immunol. 2012 Dec 29. (IF: 12.047)

Patent application:

The authors are inventors/applicants of the international patent PCT EP2010/070517: Method and Kit for determining metabolites on dried blood spot.

CONTACT

Alberto Zanobini - alberto.zanobini@regione.toscana.it

European Regional and Local Health Authorities

Thank you for attending the EUREGHA Conference, 3rd December, 2013

If you are interested in joining EUREGHA, or if you want more information about our work, please contact us or visit our website www.euregha.net.

EUREGHA Secretariat

Rue du Luxembourg 3, 1000 Brussels, Belgium.

E-mail: secretariat@euregha.net or info@euregha.net