Table Of Content

European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS

JA2015 - GPSD [705038]

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TOPIC: ERN Specific Grant Agreements Year 1

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Project abstract

GENTURIS is a European Reference Network (ERN) for all patients with one of the rare genetic tumour risk syndromes (genturis). These patients are at very high hereditary risk of developing multiple tumours, which are often located in multiple organ systems. In case they are diagnosed with cancer they need different treatment and follow-up as compared to non-hereditary cancers. In addition GENTURIS takes care of the relatives of these patients, for which prevention and early detection of tumours is of great importance too.
WHAT IS OUR MISSION: To inspire hope and contribute to health and well being by organizing and providing the best care to every patient in Europe with a genetic tumour risk syndrome through integrated multidisciplinary healthcare, guidelines, education and research.
WHAT IS OUR DESIRED END-STATE: Striving to be the world's leader of genetic tumour risk syndromes in patient participation, clinical care, research and education.
The ERN GENTURIS is addressing the following challenges when it comes to the identification, genetic testing, tumour prevention and treatment of patients with genturis: 1) Great majority of genturis patients are not yet identified 2) Large variation in clinical outcomes resulting in impaired prognosis and avoidable costs 3) Guidelines are lacking or implemented insufficiently 4) Almost no patient registries and biobanks 5) Limited research programs 6) Fragmented patient empowerment activities.
There are 4 thematic groups of syndromes: 1: Neurofibromatosis type 1, 2 & Schwannomatosis. 2: Lynch syndrome & polyposis. 3: Hereditary breast & ovarian cancer. 4: Other rare - predominantly malignant - syndromes. This group includes syndromes not covered in the other groups. It is a heterogeneous group with very small numbers of patients that will benefit greatly from a centralized approach. Within the next years not yet covered as well as newly discovered genturis will be included as well.

Summary of context, overal objectives,strategic, relevance and contribution of the action

ERN GENTURIS is a European Reference Network (ERN) for all patients with one of the rare genetic tumour risk syndromes (genturis). These patients are at very high hereditary risk of developing multiple tumours, which are often located in multiple organ systems. In case they are diagnosed with cancer they need different treatment and follow-up as compared to nonhereditary cancers. In addition GENTURIS takes care of the relatives of these patients, for which prevention and early detection of tumours is of great importance too.
ERN GENTURIS is working to improve identification of these syndromes, minimise variation in clinical outcomes, design and implement guidelines, develop registries and biobanks, support research, and empower patients. The

network will educate the public and healthcare professionals, and foster the sharing of best practice across Europe. Access to multidisciplinary care will be improved, with new models and standards for sharing and discussing complex cases. The network is enhancing the quality and interpretation of genetic testing, and increasing patient participation in clinical research programmes.

Methods and means

ERN GENTURIS is coordinated by Radboud university medical center in Nijmegen, The Netherlands. The current list of 23 participating healthcare providers covers 12 European member states (see figure 1).
The syndromes are grouped in three thematic groups that have overlap in manifestations and/ or genetic cause, plus a group of even more rare syndromes with predominantly malignant tumours (see figure 2). Six Task Forces are set up, each dealing with specific tasks (see figure 2).

Work performed during the reportingperiod

In year 1, ERN GENTURIS focussed on:
• establishing a governance structure compliant with EU criteria
• designing draft patient pathways
• identifying best practices for composition and organisation of multidisciplinary teams
• the clinical patient management system (the secure web-based application functioning as the platform where healthcare professionals from the European Reference Networks (ERNs) discuss real patient cases)
• identifying process, clinical outcome, quality and patient safety indicators for genturis
• developing a GAP analysis and an Annual Education work plan
• create a minimal data set outline and select a registry system
• develop a Research work plan
• involve patient representatives in all relevant committees
• develop a Communication and Dissemination Work Plan

These activities were carried out by the six Task Forces and four Thematic Groups (see figure 2).

The main output achieved so far and their potential impact and use by target group (including benefits)

The Board terms of reference and Board policies were agreed upon by all members and signed. ERN GENTURIS chaired the ERN coordinators Working group on ethical and legal issues and took the lead in writing the ERN-wide policy on the conflict of interest.
All members familiarized themselves with the clinical patient management system and the first patients were enrolled by the end of year 1.
Draft care pathways including basic draft clinical guidelines were developed for Neurofibromatosis 1, Lynch and Polyposis, Hereditary Breast and Ovarian Cancer, Cowden syndrome and Li-Fraumeni syndrome.
The best practice for the composition of the MDT per genturis group was identified, as well as the organisation of the MDT in relation to the use of the Clinical Patient Management System.
A list of GENTURIS performance and outcome indicators was selected from the draft of the ERN-wide indicators prepared by the ERN Coordinators Working group on Monitoring, assessment and quality improvement. Indicators were selected on the basis of their relevance and feasibility to use in GENTURIS.
The Annual Education Workplan describes the local courses planned in 2018 by ERN GENTURIS members, the Spring Course in Hereditary Cancer Genetics organised by ERN GENTURIS members (24th – 27 April 2018, University Residential Centre of Bertinoro, Italy), as well as plans how improve and develop education, training, professional development. Gap analysis were designed for national coordinators, participants of the Bertinoro course and participants at a workshop organised by ERN GENTURIS coordinator Prof. Nicoline Hoogerbrugge at the European Society of Human Genetics Conference (ESHG) 2018, (Milan, Italy, June 16-19 2018).
The Strategic Research Plan describes that a GAP was identified in the really rare genturis research projects. Therefore, we will concentrate the search for funding of research on the ultra-rare Genturis of thematic group 4: LFS, CDH1, PTEN etc. The minimal data set has been defined and will be operationalised and implemented in a registry in year 2. We evaluated several available open-source, licensed or commercial data registry systems to see whether they support the minimal requirements.
During year 1 six patient representatives (ePAG members) became part of the GENTURIS Advisory board and the GENTURIS Board , as well as taskforces 2-6 and the four thematic groups to support their activities. The public website www.genturis.eu went online on 3 February 2017 and was updated regularly in collaboration with all partners and the ePAG representatives. The website was translated into Spanish and French. The German and Dutch website texts are currently being fine-tuned by the national coordinators. In the upcoming year, the website will be translated into Portuguese, Polish and Slovenian.
The communication and dissemination plan describes the wide range of stakeholders, with different objectives, key messages and appropriate communication and dissemination channels. In version 1.0 of the

communication and dissemination plan, the main stakeholder groups are identified, which will be further expanded in the next years of the ERN. For all stakeholders GENTURIS will set up a two-way or multi-way communication and dissemination approach, for instance by facilitating focus groups or dialog groups.

Achieved outcomes compared to the expected outcomes

Most achieved outcomes are in line with the expected outcomes. However, a number of outcomes have been prepared in a draft or first version and need be finalized in collaboration with the stakeholders.

Dissemination and evaluation activitiescarried out so far and their major results

In year 1, ERN GENTURIS members engaged in nearly 100 dissemination activities. These activities included notably 4 television interviews and one radio interview. The most frequent type of dissemination was the oral presentation, 59% of which was directed to a scientific audience and 41% to the general public. Of all the dissemination activities, 14% was directed towards patients in combination with other stakeholders, 25% towards the general public, and 55% to medical specialists only. 77% of the dissemination was targeted domestically, 12% at European countries and 11% at non-European countries.

Evaluation of the performance of GENTURIS was done at the coordination level and at Task Force Level. The main finding is that the amount of work is more than anticipated and that a shortage in budget, time and expertise in (some of) the Task Forces has led to a delay in the activities.

Work package

Work Package 1: Coordination and Management

Start month: 1
End month: 12
Work Package Leader: RUMC

All Coordination and Management activities regarding required documents, inclusion of HCPs, organisation of meetings and reporting.

Work Package 2: Organisation of Care

Start month: 1
End month: 12
Work Package Leader: RUMC

This Task Force will pursue the following general GENTURIS objectives:
• To contribute to the pooling of knowledge regarding sickness prevention; Sickness prevention is an important aspect of the care for patients with genturis. Genetic screening of healthy relatives and a multidisciplinary and personalized approach towards surveillance and risk-reducing surgery can prevent tumours and complications in healthy relatives and prevent the second primary cancer in affected patients, as these frequently occur in genturis.

To facilitate improvements in diagnosis and the delivery of high-quality, accessible and cost-effective healthcare for all patients with a medical condition requiring a particular concentration of expertise in medical domains where expertise is rare. This objective is central to aim of European Reference Networks in general. This objective will be realized by all Task Forces, but specifically by Task Force Two – Organisation of Care and the specific objectives and activities therein.

To help Member States with an insufficient number of patients with a particular medical condition or lacking technology or expertise to provide highly specialised services of high quality.

Work Package 3: Good Practice, guidelines, outcome measures and quality control

Start month: 1
End month: 12
Work Package Leader: RUMC

This Task Force will pursue the following general GENTURIS objective:
To encourage the development of quality and safety benchmarks and to help develop and spread best practice within and outside the network.

Work Package 4: Continuous Education, Training and Development

Start month: 1
End month: 12
Work Package Leader: RUMC

This Task Force will promote the use of standardized continuous education & training programs and tools for society, patients, health care providers and scientific groups. The Task Force will coordinate education and training activities initiated by the network.

Work Package 5: Research, data registries, biobanking and funding

Start month: 1
End month: 12
Work Package Leader: RUMC

This Task Force will define the research agenda, identify needs and opportunities in a Strategic Research Plan, and
coordinate and facilitate access to information, data and biological samples.

Work Package 6: Patient Empowerment, Collaboration, Communication and Dissemination

Start month: 1
End month: 12
Work Package Leader: RUMC

Task Force 6 will focus on incorporating the voice and opinion of patients and families, disseminating GENTURIS output and collaboration with other ERNs.

COORDINATOR, LEADER CONTACT AND PARTNERS

COORDINATOR

STICHTING KATHOLIEKE UNIVERSITEIT (RUMC)

Comeniuslaan 4
NIJMEGEN
9102, 6500 HC Nijme

Netherlands
WEBSITE: http://www.ru.nl

PARTNERS

No partners related to the current project

OUTPUTS

Periodic Financial report / annual report for EC

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 22/05/2018
Periodic report via Participant Portal

Board policies, rules of procedure

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
The role and responsibilities of the Board are clearly defined and documented in a set of governance policies or rules of procedure. The Rules of Procedure should include a model of written cooperation agreements to be signed by the Affiliated Partners including, at a minimum, their specific role concerning their active involvement and obligation to follow the rules and procedures established by the Network, in accordance with the relevant operational criteria for the Networks

Network Monitoring System and evaluation protocol

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
A Network Monitoring System including process and outcome indicators and evaluation protocol

GENTURIS strategic plan

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
We will gradually increase the number of centres, the diversity of expertise within centres, including proactively affiliated centres from all EU countries.

We will increase the number of syndromes in thematic group 4: other rare predominantly malignant genetic tumour risk syndromes. We will be developing our ERN GENTURIS, with an actively inclusive strategy, of involving all centres matching our criteria for excellence, and enabling them to contribute to tasks, and domains, and to our mission and the vision.

Policy on conflict of interest

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Conflict of Interest regarding clinical guidelines, patient pathways, and clinical decision making tools

Cooperation agreement

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
The Terms of Reference will function as cooperation agreement between the coordinator and Full Members. Associated partners will sign a separate cooperation agreement.

Process for membership renewal and access new members and affiliated partners

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
The Network has a defined strategy for integrating new Members approved by the ERN Board of Member States and Affiliated Partners designated by the Competent National Authorities.

GENTURIS handbook

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
GENTURIS handbook, including templates for progress/ activity reports,

evaluation reports, meetings etc.

GENTURIS GB/ EC/ NCB/ RB meeting

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Two meetings will take place in year 1, one preceding the ESHG meeting on 27 May 2017 in Copenhagen, Denmark, and a 2-day meeting in Fall 2017 at Schiphol Airport, The Netherlands. The first meeting will be attenced by those members registered for the ESHG, the second meeting by all GENTURIS members.

Evaluation report

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Evaluation report (including Evaluation reports per Task Force = WP)

Report Stakeholder consultation

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Report on Stakeholders analysis and field consultation

Board approved patient pathways

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Identify existing patient pathways for genturis including timelines

Report on appraisal of best practices

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)

Published on: 18/05/2018
Evaluate appropriateness of established patient pathways and make quality improvements

Report on issues regarding cross-border healthcare

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Report on issues regarding cross-border healthcare - general and genturis specific

List of GENTURIS performance and outcome indicators, professional standards, HCP criteria

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
List of GENTURIS performance and outcome indicators, professional standards, HCP criteria

Quality and safety framework

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Identify standards, accreditation and certification (local, national, international) for quality and safety of HC specific for genturis, e.g. for germline genetic testing.

Gap analysis methodology

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
A gap analysis methodology to what needs to be developed in education and training

Annual Education Work plan

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
A Work plan describing: Target groups e.g. among society, patients, health care providers and scientific groups. Education and training goals per target group. Existing education and traíning materials, projects, activitíes etc.

Strategic Research Plan

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
The Plan describes the identification of research gaps and activities to fulfil these gaps.

Contribute to ERN-wide portal for use of and contribution to resources (information, data and samples)

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Listing available resources and (ICT) tools.

GENTURIS strategy for a broad patient engagement

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Identification of mechanisms/ best practices to incorporate voice and opinion of patients and families, and all relevant patient associations across Europe

Rules of Procedure and Entrance Pathways for Affiliated Partners

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/05/2018
Description of Rules of Procedure and Entrance Pathways for Affiliated Partners

Board terms of reference

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/07/2017
The Board Terms of Reference decribe a.o. the governance structure, responsibilities of the committees and financial provisions.

Communication and dissemination plan

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/07/2017
Establish communication tools, including website and develop a communication and dissemination plan.

Website

RUMC
European Reference Network on GEnetic TUmour RIsk Syndromes - GENTURIS (ERN GENTURIS)
Published on: 18/07/2017
A public website about ERN GENTURIS for the lay public.