

ERN GENTURIS Research policy

Version 1.3 / 30 April 2021

1. General policy aim and identified research gaps

The aim of the research policy is to guide ERN GENTURIS's research activities for the upcoming 5 years. Research gaps have been identified by GENTURIS for its scope of disorders through a survey and subsequent review rounds for which all members were invited. This resulted in a prioritized shortlist of topics (table 1).

Table 1. Shortlist of research priorities

Topic	Need to know	Nice to know
GENETIC TESTING STRATEGIES	<ul style="list-style-type: none"> Improved methods and strategies for identification of GENTURIS patients with germline defects, including the role of universal testing in cancer patients or general population Best approach to genetic diagnostics: whole genome, exome, panel, targeted gene testing, transcriptome. Research on implications of results of the different approaches 	
GENETIC DIAGNOSIS	<ul style="list-style-type: none"> Solving suspected cases: finding new genetic causes of cancer VUS classification (functional assays, multifactorial models, gather clinical data) 	
PHENOTYPE	<ul style="list-style-type: none"> Improved definition of phenotype: including genotype-phenotype correlation, cancer risk, mutational signatures Penetrance variability: impact of genetic/environment modifiers, Polygenic Risk Scores, lifestyle factors 	<ul style="list-style-type: none"> Digitalized analysis of pedigrees, phenotypes, signs and symptoms of GENTURIS
TREATMENT	<ul style="list-style-type: none"> Effectiveness of different types of cancer treatment in GENTURIS patients Develop targeted treatment for GENTURIS patients whenever appropriate 	
PREVENTION & EARLY DETECTION	<ul style="list-style-type: none"> Early cancer detection in those with a GENTURIS or at high risk of a GENTURIS Chemoprevention in high risk patients Prevention through modifying e.g. lifestyle and hormonal factors in GENTURIS patients. 	<ul style="list-style-type: none"> Improve methods for follow up: including utility of liquid biopsy, imaging technique/protocol, personalized programs
CARE	<ul style="list-style-type: none"> Communication: how to best inform patients and caregivers Patient's attitudes towards genetic counselling: incl. which approach and provided by whom 	<ul style="list-style-type: none"> Effective strategies for implementation of guidelines

2. Implementation

Financial support to ERNs by the EU for research is not integral part of the EU design and implementation of its ERNs. It is therefore up to ERN GENTURIS to encourage researchers of its members, the participating health care providers, to apply for funding to support projects that aim at closing the identified knowledge gaps.

ERN GENTURIS will also encourage others, through publications and presentations, to carry out this research. ERN GENTURIS will stress the need to close the mentioned knowledge gaps to national and European grant agencies, motivating them to include these topics in their research agenda.

In applying for grants in line with ERN GENTURIS research topics, researchers can refer to the fact that ERN GENTURIS, as an acknowledged organization of experts, has identified these topics as being important for the improvement of health care and therefor adding value to the healthcare system.

In case in the future the volume of requests for data from the GENTURIS registry for research purposes requires prioritizing, then ERN GENTURIS will prioritize those requests on the basis of its research agenda.

3. Monitoring and evaluation

Priorities may change over time as knowledge gaps will be filled and new challenges arise. The ERN GENTURIS research task force will therefore evaluate the research priorities annually and together with the ERN GENTURIS members set new research goals whenever necessary. In addition, the task force will discuss at the annual ERN GENTURIS meeting tactics for promoting the research goals to grant agencies and the scientific community.