

The contribution of health technology assessment, health needs assessment, and health impact assessment to the assessment and translation of technologies in the field of public health genomics

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Background

The third strategic objective of the European health strategy is aimed at supporting dynamic health systems and new technologies. E-health, biotechnology and genomics are expected to improve prevention of illness and delivery of treatment and to support a shift from hospital care to prevention and primary care (1).

The Human Genome Project has already led to numerous discoveries and the amount of genomic discoveries is expected to increase further, as a result of genome-wide association studies that are able to discover novel genetic risk factors by testing several hundred thousand single nucleotide polymorphisms (SNPs) simultaneously (2, 3). In addition, disciplines like Human Genome Epidemiology (HuGE) aim to identify genome-based health information of public health relevance by analysing the impact of genomic variants on the risk of developing diseases or disabilities and to measure how other risk factors (e.g. lifestyle or environment) interact with genomic variants (4, 5). However, despite the promising nature of genomics and its potential to improve population health, the actual translation from basic research to the bedside and to health promotion campaigns for the general public has been slow. In addition, the European Commission explicitly recognizes that these promising technologies must be properly evaluated, before their widespread use or inclusion in public health policies can be warranted. This article addresses these two considerations, namely the huge amount of novel discoveries and the need for assessment. But before elaborating, some background information and definitions from the field of genomics will be provided, as well as an introduction to the concept of translational research.

Given the range of genomic technologies that are relevant to public health genomics this paper focuses on predictive, diagnostic, susceptibility or screening applications which contribute to health information in a wider genomic context and on pharmacogenomical technologies. Prior to the assessment of a genetic test and its utility, the purpose of the test must be defined. Zimmern and colleagues proposed the following purposes for genome-based health applications (6):

1. Reduction in morbidity or mortality
2. Provision of information salient to the health care of the patient or family members
3. Assistance with reproductive decision-making for patients or family members

Defining the term “genetic test” is desirable but challenging (7). It becomes more and more apparent that the personal health information obtained from a test and not the technique itself or the method used should be the rationale for such a definition. Thus, a “genetic test” is

defined in this paper as a procedure that delivers personal health information and it is understood that genome-based health information is “just” one piece of health information or “just” one health determinant besides others contributing to different health outcomes (8).

Translational research can be defined as the translation of basic discoveries into clinical applications, which includes scientific validation of experimental results (9). In the field of genomics and biotechnology, translational research is of utmost importance, since the vast majority of potential applications are either still in the phase of basic research or are being provided directly to consumers, without involvement of health care providers or independent assessment (10-12). The definition of public health genomics emphasizes the need for a *‘responsible and effective translation of genome-based knowledge and technologies for the benefit of population health’* (13). The public health genomics enterprise, which clarifies and describes how public health genomics should be practiced reinforces this same message (13). Indeed, it stresses the challenge of knowledge translation and integration by defining the first and central task of public health genomics as “knowledge integration within and across disciplines”(13). Burke et al. describe this task as the driving force for public health genomics. It is the means by which information is transformed into useful knowledge (13).

Recently published guidelines and reviews underpin the necessity of guiding the assessment of genome-based discoveries and supporting decisions on how to use the results in clinical practice (14-17). The dimensions of genetic test evaluation comprise the analytical validity of a test, the clinical validity, and the clinical utility, including the legitimacy, efficacy, effectiveness and appropriateness of a test as well as the acceptability, efficiency, optimality and equity of test delivery (ACCE and beyond) (18). A broader view of the steps which are necessary to translate genomic discoveries into health care and disease prevention are defined by Khoury and colleagues. They applied the “continuum of translational research” to the context of genome-based health applications and defined four phases (T1 to T4) of translational research (4):

- T1 - From gene discovery to candidate health application: This phase starts after new, promising discoveries in the field of genomics are made. Whether these discoveries can be transformed into useful applications for clinical and public health practice (e.g. genetic test, intervention) has to be assessed. This can be done by observational research and clinical trials taking evaluation criteria like ACCE into account.
- T2 - From health application to evidence-based practice guideline: Translational research in phase two has to perform the task of assessing the value of a genomic application for health practice leading to the development of evidence-based guidelines.
- T3 - From evidence-based guidelines to health practice: Phase three of the translational research process aims to integrate evidence-based guidelines or recommendations coming from phase two research into health practice. Especially for procedures such as susceptibility testing, where genomic variants as a risk or a protective factor for developing a future disease can be identified, it is not known how the dissemination of knowledge as well as endorsement of implementation can be promoted. Thus, the key task of this phase is to analyse how knowledge is turned into action.
- T4 - From health practice to health impact: Phase four of the translational research process seeks to evaluate the “real world” health outcomes of a genomic application in practice on a population level. What are the health consequences of a technology or intervention within the population? These consequences not only include cost-effectiveness but also the analysis of patients’ or populations’ perspectives. Does the technology actually maximise positive health outcomes and minimise the negative ones?

To assist this translational research process and to properly assess genomic applications for public health purposes, three major approaches can be used: Health Technology Assessment (HTA), Health Needs Assessment (HNA) and Health Impact Assessment (HIA). Briefly,

HTA evaluates the performance of health care technologies, HIA assesses the effects of policies, programmes or projects on populations' health and HNA identifies health priorities for a given population. All of them with the aim to support decision making processes.

The objective of the paper is to describe the contribution of HTA, HIA, and HNA to the assessment of genomic applications in public health and to identify to what extent these approaches contribute to speed up the translational research process in genomic medicine.

Methods

A limited literature search was carried out to characterize HNA, HTA and HIA, with focus on research articles, textbooks and 'grey' literature, e.g. reports resulting from EU-subsidized projects or from European agencies.

As a second step, based on the detailed description each of the three techniques, they were 'mapped' in terms of their contribution to the four phases of the continuum of translational research in genomic medicine proposed by Khoury and colleagues. This exercise entails a judgment on the scope (in terms of aspects covered) of each of the three fields.

Results

Characterization of Health Technology Assessment (HTA), Health Needs Assessment (HNA), and Health Impact Assessment (HIA)

Health Technology Assessment

HTA can be seen as a bridge between the world of research and the world of decision making, particular policy making (19). Health technology is described by the European Network of Health Technology Assessment (EUnetHTA) as

"...the application of scientific knowledge in health care and prevention."

Health technology assessment (HTA) is

"...a multidisciplinary process that summarises information about the medical, social, economic and ethical issues related to the use of a health technology in a systematic, transparent, unbiased, robust manner. Its aim is to inform the formulation of safe, effective, health policies that are patient focused and seek to achieve best value. Despite its policy goals, HTA must always be firmly rooted in research and the scientific method." (20)

The term "technology" within HTA can be understood broadly. Besides "hardware" technologies like drugs, medical devices or diagnostic tests, HTA also analyzes softer technologies like clinical guidelines or methods used in health promotion or primary prevention campaigns. HTA is in most cases technology-orientated, but also problem- or project-orientated assessments exist (21). HTA has spread throughout the world and international collaboration is growing steadily.

It has long been recognized that, ideally, assessments should be done in phase with the life cycle of a technology (22). Thinking of the life cycle of a technology in five stages may clarify this point. During the first phase one can think of technologies under development, so-called future technology. The second stage refers to technologies prior to adoption, termed emerging technology. The third stage covers technologies in the phase of adoption, and is called new technology. In the fourth stage, when technologies are widely used, the term accepted technology is applied. In the fifth and final stage the technology has become old-fashioned and should be abandoned. One speaks of obsolete technology here.

A system or process of HTA may be viewed as an interdependent and non-discrete flow of six types of actions: 1) identification of new health technologies; 2) priority setting, selecting those technologies most in need of assessment; 3) testing, conducting the appropriate data collection and analysis; 4) synthesis, collecting and interpreting existing information and the results of the testing step and, usually, making judgements and sometimes recommendations about appropriate use; 5) dissemination, providing the synthesized information, or any other relevant information, to the appropriate persons who use or make decisions concerning the use of health technologies; 6) implementation, securing that changes in knowledge and attitudes result in changes in behaviour or in particular decisions. Finally, in addition to the stages mentioned above, the study of the diffusion or spread of health technologies in health care systems has been a long-standing tradition in health technology assessment, as outlined in the textbook by Rogers (23).

HTA EXAMPLE

A Clinical Systematic Review of BRCA1 and BRCA2 Genetic Testing for Breast and Ovarian Cancers. (24)

Within this HTA a systematic literature review was conducted to identify and synthesize data on analytical performance, psychosocial and ethical issues as well as benefits and harms of testing for mutations in breast cancer susceptibility genes *BRCA1* and *BRCA2*. Besides, descriptions of occurrence of the mutations in breast or ovarian cancer patients and in the population, implications for decision making are given on the basis of existing evidence:

- a) Besides *BRCA1/2* testing other factors should be considered as there is no clear evidence to suggest that testing will lead to decisions that result in long-term health benefits.
- b) Psychological and social implications should be taken into account, an increase of health/genomic literacy is necessary for interpretation of test results, counselling can reduce the perceived risk and associated anxiety and can increase the uptake of testing.
- c) The existing evidence shows no compelling results that some test perform better than others. Other factors like test availability, ease of implementation, regulatory considerations, and prices should be considered in deciding the methods used for testing.
- d) As scientific evidence is changing or evolving an update of the assessment is recommended.

Health Needs Assessment

HNA is used as an objective and evidence-based method of tailoring health services. HNA aims to match the specific health needs of people to benefit population health. HNA also addresses health inequalities and identifies

- non-recipients of beneficial health interventions,
- recipients of ineffective health care,
- recipients of inefficient health care,
- recipients of inappropriate health care ⁽²⁵⁾.

Thus, health needs can be understood as issues that would benefit from changes in health care or from wider social and environmental changes (26).

HNA is defined as

„... a systematic method of reviewing the health issues facing a population, leading to agreed priorities and resource allocation that will improve health and reduce inequalities.” (27)

By conducting a HNA, patterns of disease in the (local) population and regional differences in health are described. HNA uses epidemiological, qualitative and comparative methods to describe health problems, to identify inequalities in health and access to services, and to determine priorities for the most effective development or use of services. Timely and accurate information is essential if health services are to meet the changing and different health needs of their populations, subpopulations, and individuals. Routine health information can provide data for epidemiologically based assessments of ill health and help target what health services are needed. Alternatively, the global burden of disease can be represented by disability adjusted life years (DALYs), which can be used to identify current and future health needs, and plan essential health services.

HNA's objective is to influence policy, interagency collaboration, research and the development of priorities as well as to support rational decisions on how to use resources to improve population's health in the most effective and efficient way [8]. To achieve this aim, HNA is carried out in two steps: 1. the estimation of the frequency of various health problems in a population, and 2. the examination of the evidence for the beneficial and harmful effects of the interventions targeted at each health problem (28).

HNA EXAMPLE

A population-based needs assessment for mental health services. (29)

The study of Hanson and colleagues aimed to assess the needs of clients affiliated with community mental health services in the province of British Columbia, Canada and to develop service and system level recommendations for a better response to client needs in the least restrictive setting. Case managers of randomised selected clients were interviewed with a standardised assessment instrument (Colorado Client Assessment Record). In total 1,855 clients (case managers) of three different care programs participated.

The results of the needs assessment were distributed to all stakeholders, including clients, family members, mental health professionals, community support staff and managers and directors. Both unmet needs and over met needs were identified. Examples of recommendations are as follows: (1) match care to clients needs, (2) increase capacity of assertive case management, (3) bridge levels of care (including primary care) and improve flow-through, (4) provide more educational and vocational services, (5) enhance service for clients with substance abuse, (6) improve client and family collaboration in care.

Health Impact Assessment

Indirectly, HIA has its legal basis on the European level in the Amsterdam treaty which states that *"a high level of human health protection shall be ensured in the definition and implementation of all Community policies and activities"* (Article 152) (30). It is defined by the Gothenburg consensus in the following way

"Health impact assessment is a combination of procedures, methods and tools by which a policy, a program or project may be judged as to its potential effects on the health of a population and the distribution of effects within a population." (31)

HIA is typically used to estimate *a priori* the health consequences of a policy, a programme, or a project (prospective HIA) for a given population, but it is also carried out for already implemented policies (retrospective HIA) or policies that have just been implemented (concurrent HIA). An important feature of HIA is that the policies under scrutiny do not necessarily have health as their primary objective (see also the concept of "Health in All

Policies” of the European Commission (32)), since health is not only affected by biological and lifestyle factors but also by social and community networks, living and working conditions as well as by general socio-economic, cultural and environmental conditions (33). HIA can be used to assess policies on local, regional, national or supra-national level (34). It aims to identify the potential health consequences of a policy on a given population, including the positive health outcomes as well as potential adverse effects on health and health inequalities (35).

Thereby, HIA strives to influence decision making by raising awareness of the relationship between health and physical, social, and economic environments, by producing estimates of the impact of a variety of decision on the health of the population, and by supporting stakeholder participation, including that of lay people. Methodologically speaking, HIAs can be based on a participative approach or on expert opinions; they can be qualitative or quantitative or a mixture of both (36).

The HIA approach includes chronologically the following processes

- screening (identifying policies which could have an impact on health)
- scoping (identifying the direct and indirect health effects to be considered)
- assessing risks and benefits (identifying the populations which can be affected and how)
- reporting (presentation of the results to the decision-makers)
- monitoring / evaluation of consequences of implementation

HIA EXAMPLE

A prospective mini health impact assessment of the ‘Towards 2010’ programme in Sandwell and West Birmingham in the West Midlands (37)

Health and social care should be brought together according to the programme ‘Towards 2010’. For the regions Sandwell and West Birmingham, four options exist which are assessed by a mini HIA. The four options differ in the extent to which the provision of services and facilities change, ranging from no change in provision and only essential refurbishment to a wide range of new community facilities and the replacement of hospitals by a new one.

A mini HIA is a HIA with a lowest amount of complexity. It covers a rapid desktop exercise using readily available information and professional knowledge to predict positive and negative impacts around ten broad determinants of health. These are employment and economy, education, transport, housing, visual amenity (environment), access to services, crime, lifestyle, pollution, and family and social cohesion. Interim results and the final report were presented to relevant stakeholders and decision makers of the project.

Across the four options, 90 potentially positive and 84 potentially negative impacts were identified. In summary, the first option had only limited potential but the three others offered a high to maximum opportunity for a whole system change, for improved access and service delivery, and to link wider modernization initiatives.

It can be said that highlighting positive and negative impact according to the broad variety of determinants raises awareness of decision makers to the range of potential effects on the health and wellbeing of the local population. Furthermore, the decision making process became more transparent as a result of the involvement of stakeholders.

The role of HTA, HNA and HIA for translational research in genomic medicine

Health technology assessment

The development of HTA is mainly influenced by methodological streams of policy analysis, evidence-based medicine, health economic evaluation and social and humanistic sciences (38). Therefore, the process of translational research (T1-T4) (4) is covered to the broadest

extent by different combinations of methods and approaches encompassed under the field health technology assessment.

In the process of HTA, the activity of horizon scanning or early warning aims at identifying and assessing relevant emerging and new health technologies, thereby contributing to promote the adoption of beneficial and cost-effective technologies and to prevent the undesirable consequences of the unorganised haphazardous introduction of high-impact technologies (39). The concepts of early identification and assessment have gained considerable support and about a third of all HTA agencies worldwide have established a horizon scanning system. Since 1997, several agencies are collaborating in EuroScan, an international network for exchange of information and evaluation of new health technologies (40). Recently, results of a horizon scanning exercise focusing on genetic tests has been published, showing that there are yet only a limited number of clinical applications, predominantly in oncology and cardiovascular disease (41).

Systematic literature reviews and quality assessment of published research are at the core of HTA and evidence-based medicine alike. HTA in addition takes into account ethical considerations, patients' aspects, organisational issues regarding the delivery of the technology and economic evaluations when synthesising recommendations for policy making (42). Furthermore, peer review plays a significant role in HTA to assure high quality and independence of the assessment. HTA is thus in a position to tackle translational research needs in phases two to four where evidence-based guideline development, research on implementation, dissemination and diffusion as well as outcomes research becomes relevant.

Two Canadian research teams (43, 44) who work on HTA methodologies have contributed in particular to specific requirements for the assessment of genome-based technologies including genome-based health information. Giacomini et al. developed a 'three-domain model' for technology assessment and coverage decision making regarding emerging genetic testing services in Ontario (43). The domains identified 1) criteria for decision making, 2) the definition of cut-off points for each criterion, and 3) guidance for conditional coverage decisions (43). Giacomini et al. stress that for coverage purposes the unit of analysis to which the criteria apply is not just the laboratory test. Rather, what must be assessed for coverage is the testing service: the laboratory technology, plus a target population, plus a clinical context. The first criterion for decision making was defined as the intended purpose of the technology (43), analogous to the afore mentioned definition of test purpose suggested by Zimmern and colleagues (6). The second criterion is effectiveness, which for genetic tests is redefined as clinical utility. Clinical utility is regarded as a function of five nested features including e.g. analytic validity – the sensitivity and specificity of the test's detection of the analyte, and clinical validity –the performance of the test in terms of its sensitivity and specificity in detecting the genetic disease. One of the other criteria is cost-effectiveness. Interestingly, a systematic review of health economic evidence on genome-based information of new screening tests found that tests for eight conditions have been subjected to economic evaluation and although evidence is still limited, decision makers were recommended to consider the introduction of selective genetic screening for FAP (familial adenomatous polyposis) and HNPCC (hereditary non polyposis colorectal carcinoma) (45). The author added that as genetic test costs are declining the existing evaluations may warrant updating, which fits with the concept of health technology assessment as an iterative process of assessing all relevant aspects in different phases of the life cycle of a health technology.

Blancquaert also developed an innovative HTA framework applicable to genome-based health applications (see figure 1) (46). The evaluation framework comprises a critical analysis of the evidence on the analytical and clinical validity of the test, an assessment of the utility, acceptability and feasibility of the diagnostic and screening strategies, as well as an organisational analysis of the technology's interaction with health care delivery and services.

Ethical, legal and social issues are expected to emerge at all stages of the analysis and these considerations thus need to be integrated accordingly (44). In addition, economic analysis can be performed for each dimension or globally (47). Whereas HTA strives to capture all the available evidence whatever the stage in the lifecycle of the technology, the primary data accrue in phases and a parallel can be made between the dimensions identified as “test” and “diagnostic and screening strategies” in figure 1 and phases one and two of the translational research process. The evaluation of the organisation of health care may be seen as a necessary step prior to phase three, where clinical guidelines are transferred to health practice.

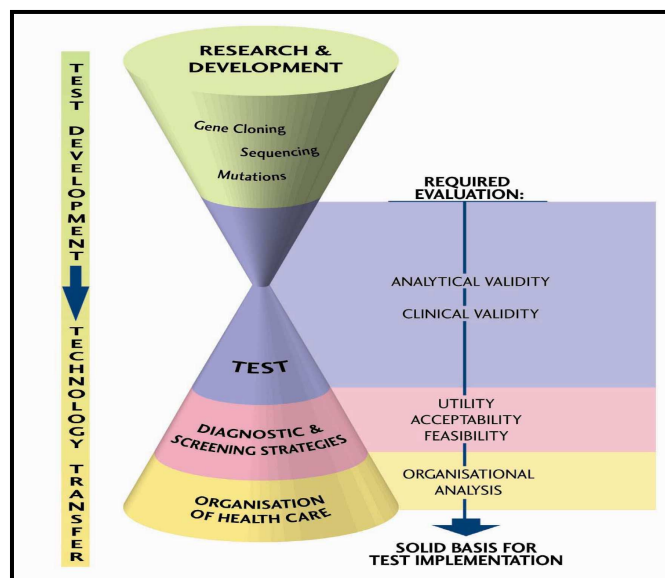


Figure 1 HTA framework for genetic tests (46)

A limitation of the conventional HTA approach is that the dynamics of technology development are not taken into account. Technologies may change rapidly, be ‘on the move’ in a technological sense, and although HTA can accommodate this by iterative assessment, opportunities to actually ‘shape’ the technology and to channel research in the needed direction are generally not realised. Constructive technology assessment (CTA) has been advocated as a means to address this issue (48). By acknowledging the socio-dynamic processes of technology development CTA can influence the development and implementation of the technology. The foundations for these processes have to be laid in phase one of the translational research process, but could continue until the final stages. Close contacts between researchers, innovators and decision makers early in the process are a prerequisite for the success of the approach, guiding and steering the process according to the decision making needs of a specific group of clinicians and patients for instance. Diagnostic methods of CTA include among others traditional social sciences techniques and also socio-technical mapping techniques to identify the past and possible future scenarios of technological dynamics.

An example is a study in the Netherlands where a socio-technical analysis of preconceptional carrier screening for cystic fibrosis and haemoglobinopathies was carried out to elucidate the preconditions for successful implementation (49). Intervention methods in CTA include among others action techniques like awareness initiatives, controlled experimentation, consensus conferences, and dialogue workshops, to influence technological development and application (48). Ideally, a clinical need may be the starting point for basic research and the subsequent development of a new health technology. This, however, is a relatively rare sequence of events. Currently, CTA is regarded as complementary to HTA, with as yet relatively few applications in health care.

Using the described components of a comprehensive HTA in the phases of the translational research process has the advantage of its status as an institutionalised framework in many countries. Additionally to the fact that this HTA framework encompasses dimensions of genetic test evaluation as described by ACCE (and beyond) it has a clear policy orientation. Thus, decision making can be grounded on a more informed level, it reduces the amount of uncertainty and it can specify the conditions for the use and the implementation of new technologies (44). It builds the basis for the effective and efficient implementation and diffusion of a proven technology and supports research, since evidence is needed to give specific recommendations and support the decision making process in the field of genomics.

Health Needs Assessment

As health needs assessment aims to identify the health needs of a local population it supports translational research with tools for prioritisation and identification of areas for needed professional and service development as well as needed requirements of genome-based health applications. Furthermore, education and training needs can be identified for those who are going to apply genome-based health applications (50) as well as for the population (health literacy). As Wright et al. stated, HNA should not only describe the greatest burden of disease in a given population, furthermore since

“(i)ncorporating the concept of a capacity to benefit introduces the importance of effectiveness of health interventions and attempts to make explicit what benefits are being pursued.” (26)

Therefore, HNA seems ideally to support phase two to three translational research by identifying requirements for the best possible implementation of the genome-based health application or by identifying technologies which are needed most for the benefit of population health.

As an example of HNA in genomics, a group of the EuroGentest Network conducted a needs assessment to determine educational needs and potential strategies to educate European health professionals in genetic competences relevant to their professional role(51). Information about professional education and existing educational materials was searched in the scientific literature and by internet-based search, supplemented by a survey among the chairpersons of professional genetic societies in each European country (51). The results of the needs assessment pointed to the deficiencies in the provision of education regarding genome-based knowledge and confirmed the need for concerted efforts to provide professional education to different health professionals likely to be involved in providing genetic testing in the future (51). To address the identified need, EuroGentest has begun to improve accessibility to existing courses offered in Europe, to improve access of professionals to national professional groups and societies, and to identify minimum levels of competencies for health care professionals in Europe (e.g. genetic education grounded in clinical contexts and meaningful to trainees) (51).

In more general terms, HNA may be used to support phase two to three research in terms of identifying priority areas of genomic health innovations in disease prevention and health care as well as to identify infrastructural needs. Questions which can be answered by a HNA are (27):

- Impact – which health conditions and determinants have the highest impact on the health functioning of the population?
- Changeability – can the most significant health conditions and determinants be changed effectively?

- Acceptability – what are the most acceptable changes needed to achieve the maximum impact?
- Resource feasibility – are there adequate resources available to make the required changes?

According to phase T3 of the translational research process the answers to the questions of acceptability and resource feasibility are the most informative for the implementation of genome-based health applications, assuming that the innovation meets the priority needs of the population.

Health Impact Assessment

HIA can be used to anticipate consequences that may occur when introducing genome-based technologies for public health purposes. Therefore, HIA should be conducted early in the policy-making process. It can be supportive of the translational research process to pre-estimate the health impact which should according to the translational research model of Khoury and colleagues be confirmed by outcomes research in phase four of the translational research process (from practice to health impact). HIA would thus provide a systematic approach for establishing a scenario which can provide at an earlier stage insight for the implementation process. This means that the prediction of future consequences of genome-based health applications or programs can allow for the dissemination, implementation, and diffusion to be developed more constructively and that missing knowledge and research needs can be identified earlier on. Depending on the availability of resources and data these pre-estimations can be done by qualitative or quantitative approaches.

Furthermore, HIA focuses on the consequences for health and disease in the population and takes general ethical, legal and social implications into account. Within HIA it is aimed to describe implications not only for the health care sector but also for other related areas e.g. the impact of the routine production and delivery of the technology on the labour force or the impact of the results of the application at the individual and social level. Furthermore, HIA identifies requirements which are needed when introducing the application, for example at the policy level. This might be e.g. changes in regulations on data protection or other regulatory efforts that can be based on laws, on the regulation by commissioners and payers or on professional regulation (e.g. practice guidelines) (52). In a nutshell, HIA can be used to make decision makers aware of the whole breadth of implications of the technologies including areas of uncertainty (53).

Summary

To capture the supportive features of HTA, HNA and HIA, table 1 shows the contribution of each to the continuum of translational research in genomic medicine. The right hand column indicates which instruments have a wider scope and go beyond the objectives of T1 to T4 research.

As illustrated, HTA (in combination with CTA) delivers the most comprehensive methodologies. HNA focuses on the needs of the population and the health system which come up when introducing new technologies or health programmes and supports the identification of relevant technologies (phase 1). Also, HNA supports T2 to T3 translational research by identifying gaps which need to be overcome when one wants to identify and increase the feasibility and acceptance of a new health technology or test in the population. An early estimation of the effects of a new application and a more general view would be added by HIA which thereby would support phase T3 to T4.

The goals of HTA, HNA and HIA extend beyond the scope of T1 to T4. HTA includes for example the horizon scanning and early identification of relevant technologies. In the same vein, HNA may help to identify relevant areas of public health where genome-based health information might be most needed and supportive. Adding the approach of constructive technology assessment (CTA), a new dimension comes in that extends phase T1 and T2 translational research by assessing the impact of a new technology in a broader manner at an early stage and thereby shaping the design of further analysis, the development, and the implementation of the technology(54). As with the HIA approach, the impact of the technology is anticipated. Moreover, neither CTA nor HIA focus exclusively on health, but rather include the wider social and legal aspects of the technology.

Table 1 The contribution of HTA, HNA and HIA to the four phases of translational research and beyond

	T1	T2	T3	T4	
	From gene discovery to candidate health application	From health application to evidence-based guideline	From guideline to health practice	From practice to health impact	Beyond T1-T4
HTA (+CTA)	+	+	+	+	+
HNA		+	+		(+)
HIA			+	+	+

Discussion and Conclusion

Assessments in Public Health Genomics, one of the key and most challenging tasks in this field, relies on scientific approaches to assess the impact of discovered genomic variants linked to different health outcomes in individuals, communities, and populations (55). The paper describes existing methodologies in public health and points out their supportive nature for the continuum of translational research in genomic medicine. Several methodologies exist which assess technologies, needs and impacts on health on a population level.

Despite different backgrounds, there are overlaps in the methodologies, procedure and aims of HTA (+CTA), HNA and HIA. Each has its specific main focus. What they have in common is to the potential to inform and further the public dialogue and participation, in an era of novelty and scarce awareness of the consequences and possibilities of genome-based science and technologies (56).

As mentioned in the beginning, the European Commission urges that new technologies be evaluated properly, including on issues of cost-effectiveness, equity, the need for additional training by health professionals, and organisational implications, for example capacity planning (1). The description of HTA, HNA and HIA shows that all of these issues are covered by one or more of the existing methodologies used in public health. Furthermore, HNA, HTA and HIA deliver facilitating components for all of the translational research phases described by Khoury and colleagues (4) and go sometimes beyond these defined translational needs, especially with regard to policy translation (definition of goals, requirements, planning steps, support by the formulation of programmes and reforms, evaluation, and refinement of health programs and other interventions). The most complete set of methods is probably provided through HTA (+CTA), but also the other methodologies can support the translation from bench to bedside not only for health practitioners but also for the public health community and decision makers.

Given a high number of PHG technologies on the horizon, priorities will need to be set for assessment using criteria that may be adapted to the technology at hand. For a general introduction to priority setting for early assessment, see Douw and Vondeling (57). To accompany timely identification with timely assessment in the face of considerable uncertainty, a number of analytical techniques can be employed, e.g. rapid HTA, rapid systematic reviews, mini-HTA (58), etc.

Since these assessment tools are already more or less integrated in the health systems of most European Member States as well as internationally, the political and social relevance and impact of these methodologies will be very high. Furthermore, the scope of the assessment instruments is much wider than the proposed continuum of translational research (4), since all potential health outcomes, policy and organisational needs, as well as educational needs, are evaluated with a interdisciplinary and systematic perspective. Furthermore, HTA, HNA and HIA promote scientific, policy and public debate as they tackle issues of relevance to citizens.

The instruments are oriented towards change in policy and practice. We therefore believe that these assessment tools offer an appropriate and useful systematic framework for translational research and assessment in the field of public health genomics, and we expect in the future to see more academic interaction between the respective fields. Clearly there is a strong need for further development and the specificities of novel genome-based knowledge may require some adaptation of these three assessment tools. Nevertheless, it is conceivable that these assessment tools could be an appropriate and useful systematic framework for these translational research steps.

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