

7.P. Round table: Bridging the gap between knowledge and practice in public health genomics

Organised by: EUPHA section on Public health genomics
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The “genomic revolution” is already transforming science, especially medicine. Genomic medicine have captured the interest and enthusiasm of not only the researchers, but also

the public and resulted in the creation of both realistic and unrealistic expectations. Among these expectations using genomic information for the benefit of population health is the most obvious. “The public health community, with its commitment to equity, must take the opportunity to engage with genomic knowledge, ensuring that it advances the population’s health.”- as it is stated by the expert group releasing the Rome Declaration “Beyond public health

genomics” (Boccia et al: *Eur. J. Public Health*, 24: 876–878, 2014). Public health genomics (PHG) focuses on the translation of genome-based knowledge and technologies into public policy, disease prevention and the improvement of population health, but translation of genomic findings to public health applications raise a lot of questions unanswered yet. Among these questions those related to screening (one of the most important – if not the most important – public health operations) at individual and population levels have special importance. Recent advances in new-born screening, as well as in screening of groups at high risk for certain diseases open new vistas in disease prevention, but ways in which our emerging understanding in these fields could guide future interventions and research efforts in public health are not clearly identified. Various ethical, legal, and social issues – among them policies to regulate access to personal genomic information, how to improve genetic literacy in both health professionals and the public, how to ensure that benefits of genomic discoveries are equitably distributed – are also posed by recent developments in public health genomics. In order to overview and to follow up the recent advances in genetic disorders and diseases associated with genetic alterations the design of the genetic testing, screening and monitoring systems should be revisited and improved making the genetic aspects more visible.

In the framework of the workshop presentations will focus on the quality assurance and improvement of genetic testing, development guidelines to the proper use of genetic tests, as well as on barriers and facilitating factors for implementation of genetic services. In addition the dilemma how should the public health system deal with the challenges of the genetic testing in a personalized care system is also discussed.

The eye-opening presentations will be followed by lively discussion on the future perspectives of genetic testing as an invaluable tool in the methodological arsenal of public health.

Key messages:

- Genetic services will increasingly contribute to population health by supporting personalized medicine. Priorities are tests with high predictive value and possibilities for prevention
- A model that combines the extent of the HTA perspective with the specificity of the ACCE criteria is applicable for the evaluation of genetic tests

The evaluation of genetic tests: a Health Technology Assessment exercise?

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Genetic tests are offering new opportunities to clinical decision making and personalized medicine. Despite different evaluation models have been developed to guide their implementation, none of them reached a generalized consensus. Our aim was to realize a new model, based on the best evidences and practices available that could become a reference methodology for Italy and other countries.

We conduct a systematic review of existing models, guidelines and reviews dealing with genetic tests evaluation and a Delphi consensus procedure involving Italian experts of public health genomics. The evaluation dimensions retrieved through the systematic review and the Delphi procedure were combined, defined and organized in the final model, based on the analysis of the literature and the experience of the working group itself. The final draft of the model was revised by the experts and suggested changes were made.

Our model combines the specific evaluation dimensions for genetic tests of the ACCE model with the HTA process. The

first two sections of the model guide the collection of evidences for the genetic test and its delivery models through eight evaluation dimensions (Genetic test: analytic validity; clinical validity; clinical utility; personal utility. Delivery models: organizational aspects; economic evaluation; ethical, legal and social implications; patient’s point of view). The third section highlights the research priorities. The fourth shows the criteria to recommend on the use of the genetic test (net benefit, cost-effectiveness, feasibility).

The most innovative aspect of the proposed model is its focus on the delivery models, including the levels of care, the health care programs and the clinical pathways in which the test is delivered. It will be useful to evaluate both the new genetic tests to be introduced in the public health practice and those already in use, guiding the decisions of a large audience of stakeholders.

Barriers and facilitating factors for implementation of genetic services

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Genetics and genomics have developed fast in the last decade, but have not revolutionized medicine, as some had expected. Translation of research findings to public health applications is lagging behind.

Many people in the EU suffer from rare conditions or more frequent monogenic subtypes of common disorders, for which genetic tests could contribute to personalized medicine. However, to move knowledge from bench to population requires planned implementation activities.

Beyond translation from mouse to man, education of public and professionals is needed. Transdisciplinary guidelines will attune perspectives of laboratory geneticists, expert clinicians and primary care. Monitoring of key performance indicators may help to evaluate to what extent implementation has been successful. Health technology assessment is needed to discern genetic tests with proven clinical utility from the offer of all kind of tests without clinical utility. Associations between genetic variants and disease risks of clinical relevance have been established, for instance for hereditary breast and ovarian cancer, colon cancer (FAP, HNPCC), cardiovascular disorders (familial hypercholesterolaemia, hypertrophic cardiomyopathy). Neonatal screening is adequately monitored in several EU countries. These examples can be used to reflect on the possibilities of using the new genetics in public health. For the first group of diseases cascade screening (inviting family members) is a very effective approach.

Stakeholders must actively plan the translation of clinically useful knowledge to the population. Transdisciplinary collaboration is needed to attune priorities and key values. Monitoring of the most important indicators will support change management.

Genetic testing between private and public interests: ethical and legal implications

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In Europe there is a wide variety of genetic tests that various private companies offer to patients or consumers. As a result, more and more people have become curious about their genetic predisposition and susceptibility. However, most public health care systems are not adequately prepared for responding to the results of these genetic tests as quite often there is no available therapy for the identified genetic condition. This discrepancy between the newly emerging expectations and the insufficient responses contributes to a

further rift between the public and private sectors of health care. Individual genetic test results may also trigger the need for personalized medicine and may open up a competition between the two fields in offering further genetic tests and

medical exams. In this context, how should the public health system deal with the challenges of the private testing? Will private genetic testing transform health care from a solidarity-based health care to a risk specific health care?