8.B. Workshop: Implementing the '-omics' evidences into precision prevention and interventions programs

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Biomedical science and technologies have a great potential to improve healthcare and underpin more efficient and cost-effective health systems. In fact, the greater personalisation of healthcare is a driver of innovation for research, and for the health care system and industry. An EU roadmap has been recently designed around the topic of ''personalized and precision medicine''. However, into a new era where the vision would be to go beyond these terms and that will be increasingly focus on preventing disease before the onset, advances in science and technology can contribute to the improvement of population health by increasing our understanding on health and disease, as well as on their determinants and leading to innovations not only to diagnose and treat diseases, but also to prevent them. As the latest Fact sheet of the WHO on non-communicable diseases (NCDs) states NCDs kill 40 million people each year, equivalent to 70% of all deaths globally. Cardiovascular diseases (CVDs) account for most NCD deaths (17.7 million people annually), followed by cancers (8.8 million), so knowledge and principles developed during recent years in the field of genomic science should be used in an extended way to address these issues. Recent advances in screening of groups at high risk for NCDs 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.

In the framework of the workshop presentations will focus on precision prevention of NCDs at primary and secondary levels.

- Genomic variants contributing to the development of metabolic syndrome, the most robust predictor of the increased susceptibility to different NCDs especially CVDs, diabetes and certain malignancies are introduced and convincing evidence about the need of phenotypic prevention (interrupting harmful interactions of environmental cofactors with genetic variation) before the onset of symptoms is presented on the example of Roma, the largest ethnicity in Europe.
- The very heterogeneous practice and recommendations about screening Lynch syndrome (caused by mutations in the DNA mismatch repair genes), which accounts for about 2-3% of newly diagnosed cases of colorectal cancer and associated with the development of endometrial cancer and various other cancers will be reviewed, and the cost-effectiveness of different screening strategies will be assessed in the Italian context.
- Joint Action Policy Papers dealing with a public health genomics approach in oncology will also be presented.

The presentations will be followed by discussion on the future perspectives of genetic screening as an invaluable tool in the methodological arsenal of public health.

Key messages:

- Economic evaluation on different testing strategies is needed to the introduction of cost-effective recommendations for screening diseases with genomic tests.
- Guidance on issues where public health genomics can substantially advance our understanding of cancer control and support policy makers, citizens and cancer patients is of essential importance.

Prevention of chronic diseases in the genomic era

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Issue

The completion of the Human Genome Project a decade ago continues to raise expectations on the application of human genome discoveries in personalized disease prevention, especially in the area of non-communicable diseases.

Description of the problem

Despite the scientific excitement the genomic science remains in its infancy and the promise of human gene discovery for health promotion and disease prevention is still to be fulfilled. This is mainly because we don't yet fully understand the complex pathways involved in chronic diseases.

The great majority of diseases involve many genetic variants as well as biochemical and environmental causes (which are sometimes much more influential than genetic factors), with multiple variations in different subpopulations. Chronic diseases are only partially heritable. Our environment and lifestyle choices can change or influence how the information coded in our genes is translated. As such there is critical need for transdisciplinary research that would integrate genomics and biological sciences with behavioural and social sciences. In addition, the existing knowledge has not been translated effectively into health practice yet; and therefore evidence-based recommendations are few.

Results (effects/changes)

This presentation will deal with how to reconcile the notion of increased personalised medicine and health care with public health interventions. It will also address the effectiveness of knowledge translation strategies focusing on policy makers as well as the wider impact of knowledge translation on the quality of public health interventions together with the tools and training resources available to support this activity.

Lessons

To conclude, taking into account recent developments in the field, there is a huge paradigm shift for public health. Hence the assurance of the complementary nature of individualised and population-oriented interventions within the healthcare appears to be even more valid than ever before.

Genetic background behind the high prevalence of metabolic syndrome among Roma

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Objectives

In a health examination survey (Kosa Z et al, Eur J Public Health, 25:299, 2015) the prevalence of metabolic syndrome was found significantly higher among Roma than in the general Hungarian population (OR = 1.37). It was also demonstrated that the raised fasting plasma glucose or known type 2 diabetes mellitus (OR = 2.65) as well as the reduced HDL cholesterol level or treated lipid disorder (OR = 2.15) were significantly more frequent in all age

groups of Roma. We designed a study to define whether genetic factors contribute to the higher prevalence of reduced HDL-C and elevated fasting glucose levels among Roma.

Methods

Single nucleotide polymorphisms (SNPs) affecting HDL-C level (N = 21) as well as glucose metabolism (N = 18) were analysed on Sequenom MassARRAY platforms in Hungarian Roma (N = 646) and general (N = 1542) populations. Genetic risk scores, unweighted (GRS) and weighted (wGRS) were computed and compared. Associations between GRSs and the prevalence of reduced HDL-C level and elevated fasting glucose level were also analysed.

Results

In case of SNPs affecting HDL-C level the GRS and wGRS were found significantly higher in Roma compared to general population (GRS: 22.2±3.2 vs. 21.5±3.3; wGRS: 0.57±0.1 vs. 0.53±0.1; $p < 0.001$), while for elevated fasting glucose although Roma people do not carry more risk alleles than the general population (20.5±2.8 vs. 20.3±2.9, $P = 0.19$) the average wGRS was significantly higher among Roma (0.51±0.08 vs. 0.49±0.08, $p < 0.001$).

Conclusions

These results strongly suggest that increased genetic susceptibility contributes to the higher prevalence of reduced HDL-C and elevated fasting glucose levels in the Roma population, so besides tackling the socio-economic determinants of the poor health of Roma people, specific public health interventions considering increased genetic susceptibility to metabolic disturbances are needed.

Screening pathways for Lynch syndrome: a systematic review of the existing pathways and a cost-effectiveness analysis in Italy

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Introduction

Lynch syndrome (LS) is an autosomal dominant disorder caused by mutations in the DNA mismatch repair genes. It accounts for about 2-3% of newly diagnosed cases of colorectal cancer (CRC), and is associated with the development of endometrial cancer and various other cancers. While the scientific knowledge about LS is increasing, the question about how LS induced CRC should be prevented is still an open issue. The practice and recommendations about LS screening are very heterogeneous within Europe. The aim of this study is to assess the cost-effectiveness of different testing strategies to identify LS in the Italian context. Three research steps are taken to achieve this aim: 1) systematic reviews of international guidelines, and existing screening pathways for LS 2) semi-structured interviews with Italian experts to identify the diagnostic pathways actually performed in Italy 3) cost-effectiveness analysis of the screening scenarios.

Methods

We searched for guidelines published from 2010 to 2016 on the provision of testing and management of patients at risk for and affected with LS, the pathways in place for LS identification, and the economic evaluations. The cost-effectiveness analysis was performed from the Italian National Health Service perspective.

Results

Sixteen guidelines regarding the criteria for genetic referral and genetic test, and nine guidelines regarding prevention strategies were identified. There is an increasing indication to identify mutation carriers starting from patients with CRC diagnosis. The seven screening pathways identified confirm this tendency. The cost-effectiveness analysis revealed that universal testing versus no testing is cost effective, but not necessarily in comparison with selective or age-targeted strategies.

Conclusions

The results on different testing strategies for LS in Italy could affect the introduction of cost-effective recommendations, following the international state of art.

''Public Health Genomics in Cancer ''in Cancer Control - Joint Action Policy Papers

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Cancer control as a major public health issue and being a pathology strongly driven by genetic modification, is closely linked to a novel field in epidemiology wherein molecular data at population scale are integrated into new strategies both from a personalized medicine as a public health perspective. This domain is generally designated as 'Public Health genomics' (PHG).

PHG approaches require a close link of research, medical application and societal acceptance. Technological developments in genomics are moving rapidly and clinical care and research are increasingly overlapping. For patients the difference is often not clear anymore, nor are the legal obligations and ethical frameworks in clinical care and research always easy to describe requiring thoughtful introduction of genomics in clinical settings. We urgently need to facilitate public dialogue on the value of public health genomics in personalized medicine in general for cancer in particular.

In the policy paper on 'PHG in cancer', we proposed guidance for policy makers on three important issues where PHG can substantially advance not only our understanding of cancer control but also support all of us, citizens and cancer patients in particular, in our common fight against cancer. We plea for developing a European framework for societal debate on the integration and use of genome information in local healthcare systems, aiming at fostering results into a pan-european context. We discuss the advent of stratified screening by genetic testing of high-risk cancer patients, address key issues when implementing genomics as such in medical care within the health care system, and thirdly highlight once more the need for common approaches in dealing with 'Direct to Consumer'.

Reference

Policy paper: ''Public Health Genomics in Cancer ''in Cancer Control - Joint Action Policy Papers (2017)