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Cancon Policy Brief 1

This is a summary of Cancon Policy paper #1 *Public Health Genomics in Cancer.* You can find the full versions at www.cancercontrol.eu

Public health genomics in cancer

Cancon's first policy paper deals with the need to develop knowledge of public health genomics, coherent approaches to its use and appropriate regulatory frameworks. **Marc Van den Bulcke** coordinated this work. Other participants were **S**. **Boccia**, **A**. **De Censi**, **L**. **Decoster**, **A**. **Federici**, **F**. **Nowak**, **O**. **Kholmanskikh**, **M**. **Peeters**, **C**. **Rolfo**, **R**. **Salgado**, **R**. **Schmützler** and **J**. **Vermeesch**.

Overview

Sustainable health care requires fundamental changes to health involving a shift from the treatment of established disease to early diagnosis and disease prevention. Existing policy approaches that seek to address this all emphasise three main elements:

- placing of the individual citizen at the centre of health systems;
- greater emphasis on prevention;
- service reorganisation from the hospital to the community.

Genomics provide new strategies for personalised medicine within a public health perspective, known as "*public health genomics",* which align with the novelty in particular in oncology. But today genomics are not explicitly included in approaches to solving these problems, though it may help to reduce disease burden and improve population health, especially within *personalised medicine* and *personalised prevention*.

Policymakers, health authorities and other public bodies need to inform and engage support resources to enable citizens, individually and cooperatively, to

- Access, understand, interpret and utilize reliable information that supports personalised healthcare;
- Define metrics to measure stakeholder participation, particularly among citizens and their communities;
- Facilitate public dialogue on the value of personalized medicine and the necessary conditions for its success.

General recommendations:

- Establish the society debate on a framework of ethical, legal and social requirements of introducing the use of 'omics' data into the healthcare system;
- Increase genetic and preventive literacy of health care professionals and citizens to allow a responsible handling of these novel options

Theme 1: "Personalized risk-assessment for stratified prevention": standards in genetic testing as a prerequisite for stratified screening and prevention of high-risk patients

- For individuals at elevated risk of certain tumour diseases, general cancer screening programmes might not be appropriate or start too late in life.

- Therefore a personalized assessment on elevated risk of individuals may be beneficial to reduce the incidence, morbidity and mortality associated with the disease.
- Such initiative requires individuals at risk to be identified through genetic testing as part of a personalized risk-assessment for stratified prevention.

Recommendations:

1. Develop harmonized common entrance criteria for personalized risk-assessment for stratified prevention throughout Europe;
2. Establish and promote specific multi-disciplinary professional structures for the indication, evaluation and provision of personalized risk-assessment for stratified prevention;
3. Increase genetic and preventive literacy of health care professionals;
4. Increase genetic and preventive literacy of citizens to allow a responsible handling of cancer preventive options and resources of the health care system;
5. Establish new genotype-/phenotype databases, preferably linked to existing cancer registries, to enable prospective cohort studies and quality assurance as a prerequisite for the evaluation of the effectiveness of personalized risk-assessment for stratified prevention;
6. Establish a harmonized framework on the ethical, legal and social requirements of personalized risk-assessment for stratified prevention in cancer.

Theme 2: Requirements and prerequisites for implementation of 'omics' in routine molecular diagnosis in oncology.

- The use of genomics and other 'omics' technology in somatic mutation profiling for targeted therapies or linking prognosis to genetic markers is a reality in clinical diagnostics in oncology.
- Multidisciplinarity is the key element in the success of such novel paradigm implying concerted activities of many medical professionals.
- Such organization does not exist in most European Countries to date.
- It would involve the massive production, collection, storage of 'big data' and the integration of multidisciplinary teams within a common public health and health care vision for long term follow-up of cancer patients.

Recommendations

1. For each country, establish a system and infrastructure that oversees the rapid evolution within oncological clinical use and utility of molecular variants;
2. Develop an integrated outcome evaluation framework that links different healthcare information among registries/repositories, and involving standardized data formats and data transmission protocols to gain evidence by clinical trials tailored to framework of personalized genome context;
3. Launch public debate on the use and limits of use of genomic information for public health and healthcare support improvement involving citizens, cancer patients, professionals, scientists, industry and government representatives.

Theme 3: Direct to Consumer genetic testing

Different types of direct to consumer genetic testing are currently already available or will soon be:

- tests for one or a few specific conditions;
- broad single-nucleotide polymorphism risk assessment tests;
- whole human genome sequencing testing.

Several concerns and problems are associated with direct to consumer genetic testing:

- Jurisdictions have very little regulatory control over direct to consumer testing worldwide and in the EU in particular;
- Potential harms due to a lack of professional counselling advice;
- Lack of transparency and guarantee on quality control, clinical validity, and clinical utility of provided tests;
- Inequities in access and unnecessary anxieties due to lack of professional interpretation and follow-up;
- Potential disadvantages of direct to consumer testing overweighing the potential benefits;
- In cancer, direct to consumer testing include mostly variants with low or no clinical validity and no clinical utility.

Recommendations:

1. Because direct to consumer genetic testing for cancer risk prediction is unlikely in its current form to have any positive impact on health, citizens' and health care professionals' awareness and education in this framework is urgently needed;
2. Policy makers should aim regulating the offering of direct to consumer testing being aware that legislation should balance consumer protection with freedom of opinion;
3. All citizens should have access to organized certified genetic counselling in his country provided by a national health care system.