SMART – Application for Cancer Genetic Counseling

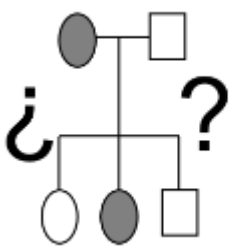
Introduction

A person with hereditary cancer syndrome (HCS) has inherited mutations in genes that are linked to cell proliferation and/or DNA repair. In consequence, they have a greater susceptibility to different cancers.

More than 100 HCS are described, with their corresponding mutated genes and suspicion criteria defined by international guidelines. Knowing all the criteria for every single syndrome is very difficult, even for physicians devoted to cancer genetics. The HCS can be determined by a genetic test but always a pre-test genetic counseling is performed.

Invention

SMART is the acronym for Syndrome Matching and Assessment of Risk Tool. SMART orientates the medical professional according to individual, tumoral and familial characteristics of the patient to the most frequent HCSs.



State of Development

The application is developed and functional, in its current beta version.

Advantages

- Early diagnosis of HCS provides risk reducing strategies and therapeutic approaches for individuals and identifying relatives at risk. Relatives who are identified as non-carriers can be managed as the general population, sparing unnecessary follow up.
- Compatible with PC, MAC and portable devices (smart phones, tablets).
- Friendly interface, that allows an easy data introduction and saving.
- Uses individual, tumoral and familial features to orientate to specific syndromes.
- Retrieves:
  - Syndrome/s that match the best to the case.
  - Future developments:
    - Risk reduction criteria according to different results.
    - Main literature and web resources for the syndromes.

Application

The application is targeted to be used in the pre and post genetic results counseling, providing reinforcement of the genetic counseling process. The anticipated users of this tool are:

- Expert cancer genetic counselors, for a sharper risk assessment.
- Other physicians that need to screen patients who could benefit from genetic counseling referral in order to:
  - Refer those patients at real risk.
  - Avoid unnecessary worrisome to patients that have not real risk.

Market potential

According to the WHO cancer is the leading cause of death worldwide; up to 10% of cancer cases are hereditary. In 2012 it caused 8.2 million deaths.

The NIH Genetic Testing Registry (US) has listed 3500 cancer genetic tests and has information of 199 tests for BRCA1 or BRCA2 from laboratories across the world, including 59 tests offered by 18 US labs. The global market for breast cancer gene testing is expected to reach €850M by 2018, out of which €88M in France, Germany, Italy, Spain, and the UK.

According to the WHO in 1999 175M genetic tests were conducted in the US with an annual growth rate of 30%. Counting with a more conservative growth rate (15%) in 2014 about 1,200M genetic tests are performed.

Therefore, the SMART application has a global rapidly growing market.

IPR Position

Intelectual Property Rights granted. Copyright application number M-008855/2014.

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Opportunity

The software is available for licensing.

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