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Citizens' jury and decision making on cystic fibrosis carrier screening: to screen or not to screen?

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Background. CF carrier screening is a genetic test to identify carriers among reproductive adults, namely the so-called heterozygotes who have one copy of the mutated gene and therefore are not sick, but can transmit the disease to a child if the partner is carrying a similar mutation. Over the past 10-15 years in the western part of the Veneto and Trentino-Alto Adige, the test has been used with caution and offered to those who already had a case of cystic fibrosis in the family. For the past ten years the University of Padua took a different policy and launched a campaign of active offer of CF testing to the general population. As a consequence, in the eastern region thousands of tests have been performed, identifying CF carriers. In the eastern Veneto region, the number of newborns with cystic fibrosis has fallen year after year until almost to zero, while this decrease was lower in the western part of the Veneto and Trentino-Alto Adige.

Hypothesis and objectives. The aims of this project - which follows the pilot project FFC#9/2011 - were:

- organize the Jury of citizens in two other Italian cities to consolidate the results of the pilot project in order to facilitate the implementation on a national scale,
- launch of an internet survey.

Methods. In Pistoia and Palermo had been organized two juries involving citizens through local associations. The jurors received a 30 pages informative booklet 2 weeks before the meeting and participated to two days fully dedicated to the topic with experts.

In the second year of the project, an online survey was launched through the sites of the promoters of patient organizations, scientific societies, through emails and articles in the laypeople press.

Results. The jury of Pistoia (May 2014) has involved 15 jurors who unanimously have expressed in favor of screening; the second jury in Palermo (September 2014) involved 16 jurors, 9 were in favor of screening.

Over 900 people participated the online survey answering the question: Should or shouldn't the Health Service organize a screening of the population in order to identify healthy people that may have children suffering from cystic fibrosis? On 566 citizens 86% (488 respondents) answered Yes, while on 338 health workers 63% (213) answered Yes.

Spin-off for research & clinical purposes. All data collected have been presented in a final conference where the implementation in the clinical practice of the results have been discussed.

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