

Implementing genetic tests

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Valorization

Additionally to contributing to knowledge in the scientific field, it is also important for research to contribute to society through the information and knowledge it gathers. Therefore, the research presented in this thesis will be contextualized in a societal perspective in this section, referred to as 'valorization'. Maastricht University defines valorization as a "process of creating value from knowledge, by making knowledge suitable and/or available for social (and/or economic) use and by making knowledge suitable for translation into competitive products, services, processes and new commercial activities". Five topics are considered important, and will be addressed on the following pages:

1. Relevance: What is the social and/or economic relevance of your research results?
2. Target groups: To whom are your research results of interest and why?
3. Services: Into which concrete products, services, activities etc. will your results be translated and shaped?
4. Innovation: To what degree can your results be called innovative in respect to the existing range of products, services, activities etc.?
5. Implementation: How will this/these plan(s) for valorization be shaped?

The overarching topic in this thesis is Public Health Genomics, which aims "to integrate genome-based knowledge and technologies into public policy and into health services" (see Chapter 1, Section 1. '*Translation of genomic technologies into healthcare*' – page 1). Creating value from knowledge on research into genetic information has been one of the major hallmarks of modern medicine. Personalized Medicine is increasingly strived for, and PHG plays an important role in translating technologies to healthcare. One could say that PHG works to find how we can ensure that relevant applications are available to the population and are translated accordingly. Thus, in itself PHG focusses on valorization by translating evidence-based genetic information into practice. The two topics in this thesis – Chlamydia trachomatis-induced infertility and newborn bloodspot screening – are no exceptions to these efforts. For each of the five abovementioned topics a link will be illustrated between (one of) our research areas and the valorizing potential it has.

Relevance

The studies in this thesis contribute to scientific knowledge on disease mechanisms into potential predictive factors by researching possible variables that play a role in the development of infertility after a chlamydial infection. In addition, the results also offer insight that may lead to improvement of the diagnostic process. This improvement would have direct benefit for women attending clinics that offer fertility care; from their initial visit to the clinic to an accurate diagnosis. When this research matures and a test grasping

the most relevant genetic variants can successfully be implemented in clinical practice, this knowledge can prevent unnecessary invasive procedures in women, such as laparoscopies, and diminish their psychological stress from uncertain predictions. These genetic variants would go beyond solely the recognition *C. trachomatis* and inflammatory processes followed after infection, since there are other bacterial sexually transmitted diseases (STDs) like *C. trachomatis* that have a similar immune response. Furthermore, research into the attitude of gynecologists to use genetic information has already showed us what gynecologists find important as test characteristics and that they would need additional training to have their knowledge on a genetic test on point. Both these aspects offer direct factors to focus on in the continuation of research and test development to increase to chances of a swift implementation of a genetic test with important predictive factors in the development of infertility after a chlamydial infection.

To continue with an the example for newborn bloodspot screening, not only scientific information, such as a lack of reporting from a variety of countries has been found, but also lack of including a different range of stakeholders in policy development processes. For example, prospective parents will benefit from a structured, transparent, and evidence-based approach in decision making for newborn screening because they can feel secure about safeguards of an evidence-based screening program focused on goals and benefits important to them. Combining these perspectives will in the end offer all stakeholders a program that has taken their views into consideration. In a broader perspective the frameworks for stakeholder involvement in policy making could be an example for different government initiatives.

Target groups

One of the first groups that comes to mind as a target group to benefit from our research results will be people in the phase of wanting to start a family; either when they are having difficulties conceiving or when they are prospective parents. Not only relevant information for parents will ultimately result from a decision making process as illustrated in this thesis, also other stakeholder groups will be enabled to express the barriers and facilitators they experience in the responsible translation of genetic information into health care (see for example *Chapter 6. Decision making in newborn screening needs a transparent approach with structured multidisciplinary stakeholder engagement in all phases*, page 79).

Services

A concrete example of making our research results available for social and economic use would be a genetic array to test for relevant variants to predict the course and outcome of a *Chlamydia trachomatis* infection in women with fertility issues. Since this test would aim to more accurately predict which women are likely to suffer from tubal factor

infertility, it also aims to decrease the amount of unnecessary laparoscopies. Through these aims both a social and economic goal are served. Women would be spared the burden of undergoing an invasive procedure, while health care costs and productivity losses can also be reduced by minimizing invasive procedures.

Innovation

While genetic information has been predicted to offer a considerable breakthrough for health care through treatments and diagnostics, these expectations are not realized in health care yet. Currently most applications of genetic tests can be found in cancer diagnostics and treatment. Furthermore, genetics has played a major role in fertility health care for example in prenatal genetic diagnostics, but has not been applied yet in the diagnosis of infertility. Since PGD is usually only performed in a small group of people, developing and implementing an additional genetic test for infertility diagnosis would present an innovative approach to this condition. Moreover, the test has the potential to be used for a wider set of women suffering from fertility complaints.

In the field of newborn bloodspot screening this thesis offers several suggestions to innovate in the current policy making processes. For example, the research presented in chapter 7 offers a starting point for governments to consider public participation in policy development. Moreover, a topic that is addressed throughout the chapters on newborn bloodspot screening, is the aim of newborn bloodspot screening. The studies in this thesis give several suggestions how to address this question, and pay attention to the – still – innovative view that the aim of newborn screening might benefit from a shift towards including family benefits. Nonetheless, these considerations should be made in a structured and transparent process, which is not always in place in countries or jurisdictions. Chapter 6 offers insights to policy makers about to embark on developing or revising a newborn bloodspot screening program to apply such an approach, in order to innovate decision making for newborn bloodspot screening towards a safe program, supported by stakeholders.

Schedule and implementation

For each of the topics several links have been made with the possibilities for valorization. This last topic focuses on shaping these plans for valorization. With regard to developing a genetic test to be added into the fertility work-up, first steps have been made for implementation through the stakeholder research we conducted. To ensure a successful and swift implementation of a genetic test in daily practice for gynecologists and other fertility specialists, more stakeholders will need to be involved to identify barriers and facilitators. Furthermore, more research is needed to gain information on the associations between different genetic variants and the risk of infertility after a *Chlamydia trachomatis* infection and other sexually transmitted infections causing tubal damage. Through this

research, important factors such as the analytical validity, clinical validity, clinical utility can be studied. Combined with research on the ethical, legal and societal implications of the test successful implementations plans incorporating these essential factors can be developed.