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How do experts shape regulatory science? Designing a genetic screening program for pregnant women in France

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ABSTRACT

When biomedical innovations are assessed by regulatory agencies, competing bodies of knowledge concerning their benefits and limits pre-exist the assessment process. This knowledge is taken into account to varying extents by agency employees and by the experts invited to take part in the process. How do these two groups of actors interact to ensure that certain aspects of knowledge prevail? This article provides a number of answers to this question, based on a case study of the assessment of fetal DNA testing, also known as non-invasive prenatal testing, in France in 2016-17. The National Health Authority set the terms and conditions for public funding of a national program to screen for fetal trisomy 21. The author of this article was invited to participate in the evaluation as a social science expert.

This article analyses why the agency's staff have adopted the conception of what constitutes good prenatal genetic screening proposed by certain biomedical experts. The agency's staff prioritized evidence-based medicine, epidemiology and economics, excluding other areas of knowledge, to construct proof of the future utility of the innovation. Its econometric models were instrumentalized by biomedical experts. They succeeded in imposing a local biomedical consensus, which had been developed in professional associations prior to the evaluation, and which was aligned with the interests of industry.

In the field of genomics, new biomedical tests are regularly brought to market. As in other medical disciplines, multiple actors attempt to influence their use by creating rules to govern their application, forming a complex polycentric regulatory framework at national and international level (Hogarth et al., 2022). Some of these rules are based on scientific knowledge, and form what is known as regulatory science (Demortain, 2017). The actors who produce this knowledge are academic physician-researchers, often in partnership with manufacturers who develop innovation, professional medical associations, regulatory agencies, etc. The knowledge produced on the performance of innovations can be contradictory because there is no consensus on the criteria by which the clinical usefulness of a biomedical test should be judged (Hogarth et al., 2022).

However, it is important to understand the reliability and usefulness of a genomic test, especially when considering its use for population screening. Any screening program generates uncertainties, which can be multiplied depending on the characteristics of the test used and how it is used. As a public health tool, screening involves testing people without symptoms so as to identify those at risk of developing a health problem

and prevent its onset. The aim is to determine which individuals are high risk, so that they can be offered a diagnostic test, which is generally more expensive or more dangerous due to its invasive nature, but which provides a definitive answer as to whether the health problem exists. Screening is thus an approximate prediction technique which produces a number of correct results, but also false positives (incorrectly indicating a high risk) and false negatives (incorrectly indicating a low risk). This lack of specificity and sensitivity can create major information problems for the population concerned. This can result in participation in screening based more on a sense of obligation than on an informed choice to seek out a health problem, as well as difficulties in managing uncertainty and anxiety both for people who receive a high-risk result, and for their practitioners (Armstrong & Eborall, 2012). As early as 1968, the WHO recommended that screening programs be analyzed in advance with regard to test reliability, follow-up care capacity, cost, social acceptability, etc., so as to ensure that their potential harms did not outweigh the expected benefits (Wilson & Jungner, 1968). Even today, population screening poses social, ethical and technical challenges (Faulkner, 2012; Sturdy, Miller, et al., 2020). Ethically, one of the

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most controversial types of screening is that used for fetal abnormalities, as there is no treatment for the vast majority of the problems detected. The proposed solution is either medical termination of pregnancy, which is not always considered morally acceptable, or preparing oneself psychologically to have a disabled child. There is a long history of debates on the issue of screening for Trisomy 21 (also known as Down syndrome) in many countries. On the one hand, the “prevention” of Trisomy 21 has a double advantage: for families (especially since most of the work involved in caring for disabled children is delegated to mothers), and for the public purse (especially in countries with a national health service) (Löwy, 2014). On the other hand, it leads to accusations of eugenics because it reduces the number of births of people with disabilities (Julian-Reynier & Bourret, 2006; Perrot & Horn, 2022).

1. Economic and regulatory issues surrounding NIPT

Genomic research has produced screening tests for fetal abnormalities since the 2010s, and these tests have been described as a regulatory conundrum (Löwy, 2020). Such cell-free fetal DNA tests, also known as non-invasive prenatal testing (NIPT), detect strands of fetal DNA from a maternal blood sample. They are expensive because they were brought to market by US biotech start-ups who had protected their invention with patents in North America and Europe (Hawkins et al., 2019). These companies are competing for a market with considerable economic stakes, potentially affecting all pregnant women in high- and middle-income countries. The development of the tests is based on promises of very high detection rates for a growing number of fetal abnormalities. However, uncertainties remain concerning the reliability of their risk assessment and the value of detecting genetic abnormalities of which affected individuals may not even be aware. As a result, the way these tests are used varies considerably, depending on the country and healthcare system (Ravitsky, 2017; Ravitsky et al., 2021; Schwensen & Gammeltoft, 2023).

Holloway et al. (2021) described the regulation of NIPT in the United States as polycentric and heavily influenced by test manufacturers. The FDA refrains from intervening because NIPT is a laboratory test. This leaves manufacturers with many opportunities to achieve widespread distribution of their tests through two mechanisms. First, they establish partnerships with academic physician-researchers to produce scientific studies proving that these tests are reliable and superior to older tests, such as ultrasound or serum markers. These industry-funded studies show that NIPT has very good sensitivity and specificity in screening for trisomy 21 and, to a lesser extent, trisomies 13 and 18, for women at high risk due to their advanced age, family history, or screening results from older tests. Based on these studies, professional associations of medical specialists are developing clinical practice guidelines to advise primary care practitioners on how to prescribe these tests. In addition, manufacturers are hiring consultants from specialist consulting firms to approach public and private health insurance organizations. The consultants use the studies and clinical practice recommendations to persuade organizations to cover the cost of these tests for their insured members (Holloway et al., 2021). In addition, manufacturers have deployed aggressive marketing methods targeting practitioners and pregnant women, going so far as to present NIPT as diagnostic tests in their information documents (Holloway et al., 2022).

2. The power of the French regulatory agency

The purpose of this article is to analyze the statutory regulation of NIPT in France and to see whether it has been influenced by industry interests. The situation differs from that in the US in terms of public regulation. The French government has established a formal regulatory authority, the French National Authority for Health (Haute Autorité de Santé) (HAS), to evaluate all biomedical innovations (drugs, biomedical devices, etc.). It was created in 2004 after several health scandals highlighted the Ministry of Health's lack of expertise (Benamouzig &

Besançon, 2005). It can be referred to by the Ministry of Health or other stakeholders. This independent, publicly funded scientific agency is tasked with advising the Ministry of Health and healthcare professionals. As in other countries, the agency staff issue their recommendations in the name of science and Evidence-Based Medicine (Eyal, 2019; Faulkner, 1997). Its staff are tasked with keeping private interests at bay (Hassenteufel, 2014). It must also apply the principles of openness (by inviting experts to assess innovations) and transparency (by announcing its assessments in advance and publishing its reports). However, it is not responsible for implementing health policies once its recommendations have been issued. For example, the supervision of prenatal diagnosis and screening in France falls under the responsibility of another public institution, the Biomedicine Agency (Agence de la Biomédecine).

The HAS has considerable authority in the health sector, as the Ministry usually follows its recommendations regarding the financing of innovations. The centralization of evaluation is accompanied by the centralization of payment: once the Ministry has given its approval, Social Security finances access to the innovation throughout France in line with the terms recommended by the HAS. Its recommendations are also closely followed by healthcare professionals, who are faced with a multitude of biomedical innovations and are uncertain about their usefulness in clinical practice. They follow its recommendations all the more closely because they can be invoked in lawsuits brought by patients.

In France, it can be assumed that public regulation of NIPT has succeeded in keeping industrial interests at bay and taking into account the views of all those involved in screening, such as primary care providers who prescribe these tests and pregnant women who undergo them. Moreover, in an international comparison, it was noted that the spread of non-invasive prenatal testing had been more limited in Europe than in the United States or other countries, due to the intervention of public regulators with experience in supervising previous prenatal screening programs for trisomy 21 (Löwy, 2020).

3. Knowledge favored by agency staff

Social science research highlights the limitations of screening test evaluations in regulatory agencies. These evaluations are presented as scientific, but they are inevitably influenced by value judgments (Sturdy, Miller et al., 2020). The latter come into play in choosing what counts as evidence in relation to the benefits and harms of screening. They also come into play when weighing these two elements. This is part of a long series of controversies over what constitutes the clinical utility of a genomic test, whether for screening or diagnosis: the individual interest of the patient or their doctor may be overshadowed by other considerations (Sturdy, 2022). In short, designing a screening program requires weighing the often divergent interests and values of those involved in its implementation (Sturdy, Miller et al., 2020).

The choice of what knowledge and criteria to prioritize when evaluating an innovation depends on the identity of the people fulfilling that task in the agency (Webster, 2004). Agency staff are often doctors, this being considered the most qualified professional category in the field of healthcare (Lehoux, 2006). Among the various types of study, agency staff give priority to those in experimental biomedical sciences and epidemiology, which are based on formal evaluation criteria and published in international journals (Faulkner, 1997).

Much of this work is carried out at the instigation of the manufacturers who developed the innovation and who emphasize its positive aspects, as in the field of drugs or chemicals (Demortain, 2017). Furthermore, in these studies, innovations are often tested in hospitals, with carefully selected patients treated according to a codified protocol (Lehoux, 2006). This environment differs from the context of future use, once the innovation spreads and is used by primary care providers or patients at home (Lehoux & Blume, 2000). There are few examples of innovations where agency staff have judged this evidence of usefulness

in a hospital setting to be too different from future uses, and on this basis have refused to give an opinion (May, 2006).

Economics has also built up a strong legitimacy in the evaluation of biomedical innovations when they involve public expenditure (Faulkner, 1997). It assesses the economic risk to public finances. It shares with the experimental sciences and epidemiology the use of quantitative measurements. It uses epidemiological criteria to measure expected benefits, and compares them with costs. Despite its appearance of objectivity and universalism, it gives priority to certain parameters in terms of costs and benefits, which amounts to favoring the point of view of certain stakeholders (Webster, 2004).

Due to the predominance of experimental science, epidemiology and economics, the main evaluation criteria used are indicators of clinical benefit, or cost, or a combination of the two (Faulkner, 1997; Lehoux, 2006). This method of constructing evidence of the usefulness of an innovation can make it difficult to take social and ethical issues into account (Lehoux, 2006; Lehoux & Blume, 2000; Webster, 2004).

The HAS does not escape criticism for the predominance of medical perspectives in its recommendations. From the very outset, agency members were suspected of forming a medical bureaucracy because there were many doctors, which led them to make recommendations that were not very binding on clinicians (Benamouzig & Besançon, 2005; Castel & Robelet, 2009). This was followed by the recruitment of economists. The agency's opinions on drug evaluation, for example, have increasingly incorporated economic modelling tools, and are no longer limited to the issue of therapeutic efficacy raised by doctors, as they have incorporated issues of cost for public finances (Robelet et al., 2018).

4. Expert consultation systems and audiences

Other limits to the regulatory science produced in agencies relate to the identity of the people invited to participate in the assessment on an ad hoc basis. Agency staff have only general skills in the field of health and evaluation. In order to benefit from the most recent knowledge on innovations, they recruit experts¹⁻¹² in the field concerned, by inviting them to take part in working groups. Agency staff therefore set up a consultation system to work with the experts. To a greater or lesser extent, they delegate tasks to them, and interact with them more or less frequently. This has consequences on the decisions that are made, as Jasanoff (1995) has shown in relation to the evaluation of foods or products likely to pollute the environment.

To evaluate biomedical innovations, agency staff primarily invite academic physician-researchers and biologists (Lehoux & Blume, 2000). The choice of this type of expert excludes the point of view of healthcare system users² and that of primary care physicians who have experience of older biomedical devices (Lehoux & Blume, 2000). Medical experts and biologists have the advantage of providing information on the initial uses of the innovation, on current practices in their professional communities and on the acceptability of the planned measures. This last point is essential for the reputation of the agencies, who seek to make recommendations that are acceptable to the professional circles concerned, as has been shown for European agencies in various fields (Borrás, Koutalakis et al., 2007).

In the event of strong opposition from their audiences, agency staff may change their risk assessment to protect the reputation of their institution (Demortain & Borraz, 2021). In 2009, the HAS was sued by a group of physicians. It was accused of inviting medical experts with ties

to pharmaceutical companies to participate in working groups on treatments for diabetes and Alzheimer's disease. Following this legal action, it withdrew its recommendations (Dalgalarondo & Hauray, 2020).

Experts are often reported as decisively influencing the development of recommendations by agency staff, but there is limited information on how they work with them. Little is known about the interactions between agency staff and experts, the way in which they reach agreements or how they manage disagreements. The paucity of research on this subject is due to methodological difficulties: it is difficult to answer these questions using the most commonly employed survey materials, such as agency assessment reports, which only show the end result of exchanges, or interviews with agency managers and stakeholders (decision-makers, members of doctors' or patients' associations, etc.), which only give a *posteriori* and unilateral accounts. The best way of finding out how regulatory science is developed within the agencies is to observe interactions, but this is difficult because working group discussions are confidential.

5. Methodology

This article presents a case study of the HAS's evaluation of NIPT in 2016 and 2017, to analyze the construction of regulatory science. Several types of data were used for this study. I used observation notes, taken during my participation as an expert in the working group set up to assess the role of NIPT in the fetal trisomy 21 screening strategy in France. I was invited to take part in the working group as a sociologist on the basis of my work on prenatal screening for trisomy 21. I took notes during the oral exchanges between the people who participated in the three days of meetings in May, October and November 2016. My note-taking was all the more extensive as for the reasons set out below, I had little opportunity to speak. The corpus of data also includes the four successive versions of the report, from the first sent by the members of the agency before the first day of work, to the final version published in 2017.

These observations are based on my participation as an expert. This is not a study negotiated with the agency, as I had no initial intention of using these observation notes for research purposes. This *a posteriori* research approach is similar to that used by May (2006). During data analysis, all participants were strictly anonymized. The results presented below were published in French and sent to the agency's management (Vassy, 2022). They acknowledged receipt and disputed just one minor point by email.³ In 2024, the agency's staff again invited me to take part in a working group to evaluate the use of NIPT to detect other fetal anomalies.

I also used complementary sources for this research, attending obstetrics conferences before, during and after the evaluation period, to discover what obstetrics and genetics professionals had to say about this innovation. The corpus also includes biomedical articles on NIPT, public reports, and recommendations from professional genetics and obstetrics associations.

Data analysis focuses on interactions between members of the various professional groups from a grounded theory perspective (Bucher & Strauss, 1961; Glaser & Strauss, 1967). It is complemented by a political sociology and an organizational sociology approach. The strategic analysis of organizations takes into consideration the power relationships between actors in a social system and the strategic use of information (Crozier & Friedberg, 1977).

¹ In this article, the term 'expert' refers exclusively to external people invited to take part in the evaluation by the agency staff.

² The term "health care user" is preferred to "patient" because some users object to their condition being equated with a pathology. This is the case for some pregnant women. In addition, it also includes the relatives of people who have a health problem.

³ The agency staff said they had been very careful to provide information to pregnant women by publishing a document for them on their website.

6. Results

6.1. First report for the Ministry of Health

Following a request from geneticists and obstetricians, the Ministry of Health commissioned agency staff in 2014 to assess whether the new tests should be included in the trisomy 21 screening program in place since the 1990s.

Following the Ministry's order, the agency's staff produced two reports. The same small group of agency members produced both reports, but the first was based solely on a review of the literature, without any experts being invited.

The first, published in 2015, focuses on NIPT performance from an epidemiological perspective. It shows that these tests are not diagnostic tests, but screening tests, as they produce false negative and false positive results (HAS, 2015). It recommends confirming a high-risk result with a diagnostic test. This report also shows that the new tests are better at identifying the risk of fetal trisomy 21 than conventional tests (ultrasound measurements and maternal serum marker results used as the basis for the combined test). The new tests produce fewer false-positive results and make it possible to reduce the number of diagnostic tests needed to confirm results (fetal karyotype performed on amniotic fluid collected by amniocentesis), thereby reducing the risk of fetal loss associated with sampling.

This first report summarizes international biomedical articles.⁴ It adopts the same perspective as the articles: screening for fetal anomalies is treated in the same way as screening for various pathologies in children and adults. It is evaluated using the same epidemiological criteria (detection rate, false positive rate, false negative rate). Trisomy 21 is therefore considered to be a pathology, which is disputed by people living with this anomaly.

Agency staff then drafted a second report to clarify the place of the new tests in the pre-existing screening program and to assess their social, ethical and organizational aspects. The stakes were high because the new tests might replace the old ones, leading to considerable changes. To produce this second report, the staff invited experts.

6.2. An over-representation of doctors in the consultation system

In 2016, the agency's staff invited twenty-five experts to participate in the working group.⁵ For their recruitment, the agency's staff relied for the most part on the professional medical associations affected by this innovation. The latter sent out an appeal from the HAS to their members and sent the curriculum vitae of the volunteers to the agency's staff, who made a selection. Of the twenty-five experts selected, twenty-two were doctors or biologists, with, in descending order, five geneticists, five obstetricians, three biochemists, two fetal ultrasound specialists, one general practitioner, one pediatrician, one midwife,⁶ one maternity and child welfare doctor,⁷ along with doctors from two other health agencies (Biomedicine Agency,⁸ National Agency for Medicines Safety). Hospital specialists were over-represented, despite the fact that the tests are

⁴ The selected publications are articles from English or French-language journals indexed in bibliographic databases. They are ranked according to evidence-based medicine criteria. Meta-analyses of randomized controlled trials are considered to be the most reliable.

⁵ Upon request, experts may receive small attendance fees. Participation in HAS work mainly provides symbolic remuneration, as it is a source of prestige in the professional community.

⁶ In France, midwives are considered to be practicing a medical profession.

⁷ The maternity and child welfare centers (*Protection maternelle et infantile*) are public facilities offering free consultations for mothers of young children and pregnant women in disadvantaged areas.

⁸ The Biomedicine Agency oversees the implementation of policies on organ donation, medically assisted reproduction, prenatal diagnosis, embryo research, and genetics.

designed for use in early pregnancy, a period during which pregnant women in France are monitored by primary care providers (general practitioners and midwives, as well as gynecologists and obstetricians in private practice). A large proportion of the medical experts were academic physician-researchers (eight out of twenty-two). The composition of the group of experts favored people at the top of the symbolic hierarchy of the medical profession in terms of discipline and function.

The three non-physician experts were: an employee of the Biomedicine Agency, a representative of a health service user association in the field of childbirth, and a university sociologist (author of this study). User representatives and those from the humanities and social sciences were therefore in a clear minority. However, they had been personally recruited by the agency staff, without having applied to be part of the working group; this suggested that they would be listened to.

In addition, the members of the agency invited seven people to attend hearings on the second day. These people also belonged to the medical profession (director of the first private hospital to offer the new tests in France, geneticists from the first public or private medical laboratories to offer the new tests, etc.). There were therefore a large number of doctors involved in the consultation process.

Doctors were also present in the agency's seven-member team, which included four economists, two doctors and an epidemiologist. The two doctors, not being specialists in genetics or obstetrics, did not have the same skills as the experts.

The agency's team was small compared to the number of people invited, but they played a key role in organizing the work. They set the agenda for the three work days and drafted all the versions of the report. They revealed certain information as they went along, such as the economic estimates on the second and third days. They put questions to the experts on certain points and organized a number of round-table discussions on specific subjects. During each working day, they presented parts of the report with a large number of slides at a rapid pace (due to the length of the report). The first version sent to the experts before the first meeting was 230 pages long, and subsequent versions were longer. The presentations, like many parts of the report, referred to epidemiological and economic concepts, thus offering no encouragement to those who did not belong to either the medical profession or the economists group to speak out. The main speakers were the biomedical experts, and hospital academics in particular.

6.3. The number of tests: a central issue

Agency staff favored a quantitative approach to the evaluation of new tests. The major issue for staff was the number of new tests that would be funded by the Social Security system compared with the screening program already in place.

In 2015, the standard screening program was mainly based on the combined test, which was reimbursed for all pregnant women. It consisted of an ultrasound test to measure nuchal translucency and a blood test for serum markers in the first trimester of pregnancy. A calculation based on these two measurements was used to assess the risk of fetal trisomy 21. If the risk was greater than 1/250, practitioners recommended carrying out the diagnostic test in order to obtain a definitive result.⁹

The agency's staff feared that if expenditure rose too high, the Ministry of Health would refuse to follow its recommendations. The new tests were expensive. At the time of the evaluation, two French medical laboratory companies were offering them at a price of 390 euros, after having purchased licenses from the US companies, as well as sequencers to automate the activity. The same amount had been specified in a public budget available to researchers at university hospitals to evaluate these tests in clinical trials. It was therefore this high amount that the

⁹ An estimated risk of 1/250 means that in a population of 250 women with this test result, one will have a fetus with trisomy 21.

economists used in their calculations.

The number of new tests to be funded by the Social Security system was also a key concern for the two main groups of biomedical experts. Obstetricians wanted non-invasive prenatal testing to be developed so that their discipline could remain at the cutting edge of technical progress. The aim was to offer pregnant women effective screening for fetal anomalies, and to reduce the number of false negatives. It was also important to obtain Social Security funding quickly to put an end to inequalities in access to tests. In the absence of public funding, only well-off women could obtain the test by paying for it out of their own pockets, if they were informed of this new development, for example in a private hospital. A small number of women might obtain it free of charge by taking part in clinical trials in public hospitals.

Like obstetricians, genetic experts were keen to obtain public funding for the innovation in order to develop its use. The introduction of the new tests, which were more accurate than conventional tests to screen for trisomy 21 and possibly other abnormalities, would reduce the number of diagnostic tests (fetal karyotype) needed to confirm or refute their results. The challenge was to develop the non-invasive prenatal testing in order to make the investments in genetics laboratories, particularly private laboratories, profitable. The majority of the new tests were conducted in two French medical laboratory companies that had made the necessary investments. Because of the patents, these investments were too costly for most hospital genetics departments.

6.4. Agreement on cost modelling

In the working group, the construction of the agreement on the number of new tests that would be reimbursed by Social Security was preceded by two other agreements on modelling the costs and benefits of the innovation. The biomedical experts readily accepted the choices made by the economists.

The economists created prospective scenarios to arbitrate between different future uses of the tests. They used the concept of efficiency, which compares the expected costs and benefits of innovation. They only considered short-term costs for the Social Security system, assessed on the basis of indicators such as the number of consultations, the number of conventional and new tests, and their price for the Social Security. To estimate how many pregnant women would undergo each test, the economists used data from the Biomedicine agency, which had reports on the activity of biochemical analysis laboratories and genetics centers in 2015, as well as data from a database belonging to a professional association of fetal sonographers.

No expenditure was envisaged to improve information for pregnant women. However, the general practitioners' representative, the representative of healthcare system users, and the sociologist who wrote this paper, suggested extending the duration of the first consultation, during which these tests are presented to women. Studies had shown that pregnant women were unfamiliar with conventional screening tests, overestimated their reliability and sometimes thought they were compulsory (Favre, 2007a; Seror & Ville, 2009; Vassy et al., 2014; Champenois-Rousseau & Vassy, 2012). This change would have meant considering better remuneration for this consultation for primary care providers, such as general practitioners, midwives and obstetrician-gynecologists in private practice. This was outside the field of action envisaged by the agency's staff.

In vain, these three experts also suggested adding the cost of an additional consultation at the beginning of pregnancy, a practice implemented by certain practitioners in France, who separate the moment at which they provide the information from the moment when the pregnant woman consents to the screening. To support the modelling choices of the agency staff, one expert obstetrician said that he did not have time to see pregnant women twice during early pregnancy.

Similarly, the group of participants did not take up the suggestion to consider spending money to improve training for primary care providers. A survey had shown that primary care physicians had little

knowledge of the limitations of conventional screening tests (Favre, 2007b). One biomedical expert said that there was no need to consider a budget for training because the presentations that he and his colleagues gave at professional conferences were sufficient to train primary care providers.

Other biomedical experts suggested that the calculated cost could be lowered even further by taking into account the savings made due to fewer people being born with Trisomy 21 as a result of using the new tests, thus reducing the future cost of care for these individuals. The agency staff refused on the grounds that it did not use the avoided cost method.

6.5. Agreement on the definition of results

The construction of the agreement on costs was accompanied by a second agreement on the criteria to be used to measure the expected consequences or results of the new tests. The economists opted for the epidemiological criteria used in the biomedical literature on non-invasive prenatal testing, which they had already used in their first report: rates of detection, false positives, false negatives, and fetal loss (miscarriage following amniocentesis for the diagnostic test).

These criteria were readily accepted by the biomedical experts because the clinical trials and the agency's first report had already shown that the new tests performed well in relation to these criteria. The fourth criterion was also approved because the agency's team accepted the low miscarriage rate established in a recent international meta-analysis. French studies showed higher rates, but they were considered to be either too old or too insufficient due to the lack of a control group. This choice contributed towards an undervaluation of the psychological cost for pregnant women, when they lose their unborn child following a medical examination.

After explaining their definition of costs and results, the agency staff presented several possible screening strategies. A certain number of tests to be carried out corresponded to each strategy. The cost of each strategy was evaluated in terms of how much more it would cost Social Security compared to those performed in 2015. The economists estimated 2015 expenditure at 65 million euros.

6.6. Economic realism

The first scenario considered by economists was the universal strategy. This involves eliminating the combined tests, which are inexpensive but which give rise to a large number of false-positive and false-negative results, and replacing them with the new tests for the entire population¹⁰.

Modelling has shown that this strategy will cost an extra 168 million compared to the 2015 strategy. When the economists announced this figure, they immediately said that the Ministry would not accept such an expense. The biomedical experts did not protest, demonstrating a form of economic realism. They had probably anticipated that the public authorities would consider offering the new tests to all pregnant women to be an excessive expense. Moreover, their professional associations did not recommend this scenario.

The other strategies were to keep the conventional tests and to use the new tests at a second stage, for women declared to be at high risk at the first stage. The new tests being more accurate than the conventional tests, they would make it possible to exclude some of the women identified as high risk by the combined screening, and to advise a smaller number of them to undergo the diagnostic test.

¹⁰ The main aim was to eliminate serum marker tests. In all the strategies considered, first-trimester ultrasound was retained because sonographers had argued that it detected a large number of problems, not just trisomy 21.

6.7. How to disqualify models

The strategies under consideration proposed different risk thresholds to define a woman at high risk for trisomy 21 based on conventional test results. The choice of threshold would determine the number of women who would need to undergo the new tests. As with other fields of medicine, the notion of high risk is a convention that is likely to vary (Aronowitz, 1998).

Up to this point it was a threshold of 1/250 that had been used to define high risk. Since 1997, a woman with a combined screening risk of between 1/2 and 1/250 was entitled to a diagnostic test funded by Social Security. This concerned approximately 3% of pregnant women. If the threshold were raised to 1/500, it would represent 6% of pregnant women. At 1/700, it would be 8%, and at 1/1000, it rose to 11%.

The economists modelled strategies with these different risk thresholds. In the discussion, biomedical experts disqualified the scenarios with the thresholds of 1/500 and 1/700. They said that the results of serum marker tests around these two thresholds were unreliable because they were not reproducible. They justified their statements by relying on their clinical experience rather than on published studies. The economists took note of these comments and rejected these scenarios.

6.8. Enlistment of economists by biomedical experts

Screening with a threshold of 1/1000 was favored by the main biomedical experts. It was already being used by some obstetricians and geneticists in France and other countries, and international biomedical articles presented clinical trials that used this threshold. This strategy had been recommended by the professional association of cytogeneticists since 2015 (ACLF, 2015). In June 2016, the professional association of obstetricians and gynecologists aligned its recommendations with those of the geneticists, following the first meeting of the Agency's working group (CNGOF, 2016). The professional associations had therefore built a medical-scientific consensus in favor of this threshold.

Economic modelling showed that access to NIPT, with a risk threshold identical to that used so far (1/250), was almost cost-neutral to public finances for the population in question.¹¹ The expenditure related to the new test are offset by savings made on diagnostic tests (fetal karyotypes following amniocentesis) which are no longer performed. The cost is 66 million euros per year (see Table 1). The detection rate increases slightly from the situation modelled for 2015: 788 affected fetuses might be diagnosed each year instead of 742.

In contrast, modelling of the strategy with a threshold of 1/1000 showed an additional cost of 18 million euros per year. More women are considered to be at high risk,¹² resulting in more NIPT being prescribed. The number of women increases from 16,000 to 58,000 per year. The calculations presented on the second day showed that this only slightly increases the number of affected fetuses detected.

Biomedical experts challenged this result and asked that two parameters in the model be changed. They said that the rate chosen for the prevalence of trisomy 21 in women with a risk between 1/250 and 1/1000 was too low and asked that it be increased. Furthermore, they asked for an increase in the percentage of women who would accept diagnostic testing after a worrying NIPT result suggesting a high risk of trisomy 21. Based on their clinical experience, they estimated that all women in such a situation would opt for diagnostic testing, not 80% as indicated in the model. The economists had used this percentage because a report from the Biomedicine agency showed that 80% of women underwent diagnostic testing following a combined screening

¹¹ The agency team based its calculations on 520,000 pregnant women per annum having a consultation at the start of their pregnancy. There were approximately 800,000 births in France in 2015.

¹² Women whose risk was estimated to be between 1/250 and 1/1000 were sometimes referred to as "intermediate risk."

Table 1

Cost-consequence analysis for three prenatal screening strategies.

	Modelling of the situation in 2015	Strategy with NIPT for women with a risk higher than 1/250 (unchanged threshold)	Strategy with NIPT for women with a risk higher than 1/1000
Price in millions of euros per year	65	66	83
Number of women considered by combined screening to have a high risk	16,000	16,000	58,000
Number of trisomy 21 cases diagnosed	742	788	867 or 909 ^a
Number of false negatives	179	193	108 or 99 ^a

^a According to the economic modelling, the second number is reached if all women at risk greater than 1/50 with the combined test undergo the diagnostic test and not NIPT.

(Source: HAS, 2017, p. 123. Calculation for a population of 520,000 women)

result that suggested a high risk. The economists agreed to reconsider their data and presented new results on day three.

In the latest version, the strategy with a 1/1000 threshold increases the detection rate of fetal trisomy 21: 867 fetuses are diagnosed per year, i.e. 79 more than in the strategy with an unchanged threshold. In addition, some biomedical experts argued that women who received a very high-risk result from the combined test (risk of between 1/2 and 1/50) should undergo a diagnostic test (fetal karyotype on amniotic fluid) directly, rather than NIPT which might produce a false negative result. If one adds the option of having the diagnostic test carried out directly on women considered to be at very high risk, without them having NIPT, an additional 42 affected fetuses will be detected.

The strategy with a threshold of 1/1000 reduces the number of false-negative results, which falsely state the risk to be low. Estimated at 179 in 2015, it falls to 108 with the change in threshold, or even 99 with the option of direct diagnostic testing for women with a risk greater than 1/50, compared with 193 in the strategy with an unchanged threshold.

At the end of the final day, agency staff asked the experts which strategy they preferred. Everyone who gave an opinion chose the strategy with the threshold change. The agency staff finished drafting the report accordingly. The text was re-read by a group of reviewers and by the agency's governing bodies, and was published without any major changes. The Ministry followed the report's recommendations, and the Social Security system financed the test for pregnant women in 2018 under the conditions recommended, but at a slightly lower price (€360).

One might be intrigued by the relatively limited difference in terms of detection of T21 cases, between the choice of risk higher than 1/250 and risk over 1/1000, and by the decision to choose the latter, more expensive option, despite the small size of this difference between these options. But it is important to remember that it was the 1/1000 strategy that was recommended by the two main professional associations.

Subsequently, the agency's economic forecasts proved inaccurate. The number of tests performed increased beyond the 58,000 predicted in the HAS report (Table 1). Social Security financed 117,756 tests in 2020 (ABM, 2021). This led to an unforeseen increase in public expenditure (Cour des Comptes, 2020).

6.9. Information for pregnant women

The arguments put forward by the users' representative, the general practitioner and the sociologist, warning against the anxiety-inducing nature of false positive results, as shown by psychologists in relation to conventional screening tests, were not considered. Similarly, their proposals for costly measures to improve information for pregnant

women (increased medical consultation time, improved training for professionals) were not taken into account. The majority of the working group were in favor of making the screening program more complex by adding a new test, without taking any measures other than drafting a document to inform women. This was not a very costly measure. This four-page booklet was published on the agency's website in 2018.

However, it was foreseeable that the need for information during consultations would increase still further, as the choice of this strategy increased the number of high-risk results that would be announced to pregnant women. According to the modelling, high-risk announcements were expected for 42,000 more women than in 2015. For the vast majority of these 42,000 women, however, the announcement would not be confirmed. Only 120 of them would receive a diagnosis of fetal trisomy 21, once all the tests had been completed. The interests of women who would receive false positive results following the combined test in the new scenario were not taken into account in the assessment.

7. Discussion

These results allow us to compare NIPT regulation in France and the US, as described by Holloway et al. (2021). These authors note the significant influence of manufacturers, who forge relationships with a complex group of stakeholders (professional societies, researchers, payers, key medical opinion leaders, etc.) to maximize the number of tests sold, in the absence of FDA intervention. The present study hypothesized that the intervention of a national regulatory agency could reduce the influence of manufacturers. Löwy (2020) also suggests that the spread of NIPT is more limited in terms of the number of women invited to take it and the number of abnormalities screened for in countries where there are state-sponsored screening programs for Trisomy 21 risk, unlike in the United States or other countries.

The regulation of NIPT in France is less polycentric than in the US because there is only one public insurer—the Social Security system—that reimburses the test. It finances innovations that have been approved by the Ministry of Health. The latter relies on the expertise of a national health agency to evaluate new tests. This research shows that this centralized method of evaluating NIPT has not reduced the influence of industry. Their influence has not been exercised directly, but indirectly through biomedical experts who have the same interests.

This phenomenon was then repeated in France. The HAS carried out a new assessment of NIPT due to the fact that the tests had evolved. Manufacturers brought to market tests that detect more numerous and rarer abnormalities (other trisomies, sex chromosome anomalies, fragmentary alterations greater than seven megabases) with sometimes uncertain consequences for the future child (Vassy, 2022). The professional association of cytogeneticists recommended screening for a large number of these additional abnormalities. In 2024 the HAS organized another working group, which endorsed these developments.

However, the finding that centralized regulation of NIPT in France is weak cannot be generalized to other countries that have a national regulatory agency. In the United Kingdom, the National Screening Committee, a national agency specializing in screening governance, whose recommendations are followed by NICE (National Institute for Health and Care Excellence), ruled in favor of a restrictive use of the tests in 2016. NIPT is funded by the healthcare system only for women at high risk of trisomy 21. The definition of high risk did not change during the agency's evaluation of NIPT and remained restrictive (1/150, i.e. 1% to 2% of pregnant women). It is possible that the strong mobilization of civil groups against trisomy 21 screening, observed by Ravitsky (2017), played a role in the NSC's decision.

8. Conclusion

The originality of this research lies in the study of interactions between experts and agency staff during the task of constructing evidence of the usefulness of the innovation. It shows the scope and limits of

scientific evaluation of NIPT in a French regulatory agency.

As in other agencies, the agency staff includes doctors and economists who took into account the need to contain public expenditure, by ruling out the most expensive version of screening with the new tests (offering the test to all pregnant women). They mainly used knowledge from epidemiology and experimental medicine, along with economic concepts. They chose to invite a majority of biomedical experts.

8.1. A medical-scientific consensus

The study of interactions in the working group shows that biomedical experts have enlisted the agency staff, steering the work of evidence construction in a direction that conforms to their professional interests. Economic modelling was unable to shake the medical-scientific consensus of biomedical experts that had been built up in their professional associations. This consensus was aligned with the interests of clinical laboratory companies, which were themselves aligned with those of the manufacturers that brought innovations to market, and who were pushing for the use of a large number of tests. Public and private genetic laboratories needed to maximize the number of tests performed to make their investment in testing and sequencers profitable.

The economists were unable to oppose the biomedical experts' strategy of screening which involved widespread use of the new tests on a broader group of women at high risk. This is because their main decision-making tool, econometric modelling, relies on biomedical data. The biomedical experts disqualified the economist's initial choices. They obtained the change in the modelling parameters in the name of their epidemiological knowledge and clinical or laboratory experience. In this way, they were able to show that the innovation was more effective, according to a criterion previously accepted by the working group, namely the detection rate. They obtained a broadening of the definition of women at high risk of fetal trisomy 21 by changing the risk thresholds, as recommended by their professional associations. The agency staff had neither the expertise nor the legitimacy to oppose biomedical experts.

In the absence of interviews with agency staff and management, it is impossible to know the criteria used to select biomedical experts, or the reasons why so many were invited, with the risk of outnumbering agency staff. The alignment of HAS staff with biomedical experts might be explained by their concern to protect the agency's reputation and its future capacity for action, as has been noted in relation to other agencies (Demortain & Borraz, 2021). The HAS is engaged in a series of negotiations to assess numerous innovations and may be seeking to maintain good relations with medical associations because it needs them to recruit biomedical experts. In addition, some biomedical experts are key opinion leaders and can help implement recommendations.

8.2. Hierarchy of knowledge

This research confirms many of the findings of previous studies on the hierarchy of expertise and knowledge within regulatory bodies (Lehoux, 2006). It is the biomedical experts with the most prestigious positions who have shaped the production of regulatory science. The academic physician-researchers played a key role in the HAS working group. This reflects the general tendency of the French medical system to bring hospital medicine to the fore and to place less stress on the role of general practice and public health.

This research shows the concrete mechanisms of exclusion and marginalization of certain forms of evidence provided by representatives of users of the healthcare system and the humanities and social sciences.

The staff completed the mission of opening up the agency by inviting a sociologist, the author of this paper, and a patient representative. These two experts, along with the representative of general practitioner, were unable to put forward their point of view. The openness of expertise in consultation systems is not enough to ensure that a plurality of viewpoints is taken into account. The composition of the working

group was strongly tilted toward biomedical scientists. This phenomenon has already been reported in French health agencies (Borraz & Henry, 2015).

Analysis of the interactions shows how the arguments of representatives of the general practitioners, patients and social sciences, who were in a minority, were disqualified. These experts unsuccessfully requested changes to the definition of the costs associated with innovation, including the costs of training practitioners and providing information to pregnant women. These new costs were considered by the rest of the working group to be outside the scope of the Agency's intervention. Implicitly, these additional costs were considered to be the responsibility of the local level of healthcare provision. Nor were the three experts able to influence how the utility of the innovation was defined by certain biomedical experts and economists on the basis of epidemiological criteria. The usefulness of the test was reduced to improving the detection rate of fetal trisomy 21 without increasing the number of fetal losses during diagnostic tests. Other definitions of usefulness were ruled out, such as the minimization of anxiety-inducing aspects of the previous screening program or the improvement of the conditions under which women consent to screening.

There are multiple definitions of the usefulness of the test and therefore of the objectives of public policy on fetal screening. It is possible that the first definition—improving the detection rate of trisomy 21, whatever the disadvantages may be—corresponds to the wishes of a number of pregnant women. In France, several hundred thousand pregnant women are monitored each year and undergo screening with conventional tests. It is difficult to know what the majority of them want. This uncertainty has allowed biomedical experts to impose their definition of the usefulness of innovation, at the risk of accentuating the negative effects of medicalization.

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