

Madame La Ministre des affaires sociales, de la santé et des droits des femmes

Mr President d'ARIIS Claude Bertrand (ARIIS),

Ladies and gentlemen,

Dear Colleagues,

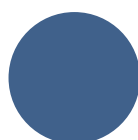
It gives me **great pleasure** to join you in opening this new edition of International R&D Dating, jointly organised by ARIIS and AVIESAN.

The previous editions showed us the very fertile nature of these interactions between the academic and industrial forces in research, around **shared questions such as:**

How can we accelerate biomedical innovation?

Which emerging areas of research have a strategic dimension?

In which areas of research, France has a competitive advantage in Europe and internationally?



How can we improve partnerships and synergies between people working in life sciences and health?

This year's Meeting is dedicated to **rare diseases as a paradigm for personalised medicine.**

Nearly 8,000 rare diseases have been identified worldwide, which affect **3-4 million patients in France**, and 24-30 million in Europe. The vast majority of these diseases are of genetic origin.

Indeed, for the first time, through the technological revolution of next-generation sequencing, it is possible to access our entire genome in order to optimise **the early diagnosis, treatment and prevention of diseases on the basis of individual genetic variations.** The impact of genomic medicine is already **cross-disciplinary.**

In the context of reviewing the variability of the human genome, we should not compare rare and common diseases, but **consider rare diseases as extreme forms of common diseases.**

Analysing the genome **of a limited number of patients, or of even a single patient affected by a rare disease**, makes it possible to identify the genetic alteration causing the illness and, hence to **understand the**



mechanisms of this illness and then **develop treatments** specific for the disease. There are many examples of where **the rare disease is a gateway to modelling complex and multifactorial diseases.**

However, even though personalised medicine will be useful to better understand rare diseases and identify novel therapeutic target for some conditions, the promise of improved risk prediction, lower costs and gain in public health for common diseases remain to be demonstrated for definitively convince public health authorities and policy makers.

In particular, we will have to answer the following questions:

Does the Human Genome Contribute to Disease Risk Prediction?

What Kinds of Studies Should Be Mounted in Personalized Medicine?

How Will Personalized Medicine Affect the Costs of Medical Care?

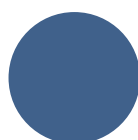
How to demonstrate the public health benefit ?

Do therapies that currently make it possible to improve the prognosis for people with rare diseases are also therapies intended for treating a wider spectrum of diseases.

Many of these questions will be addressed today.

Nevertheless, each of these discoveries advances basic knowledge.

And since these discoveries are inspired by clinical challenges, most of

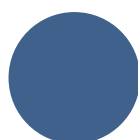


them generate **development pathways to accelerate the translational phase and subsequent placement on the market.**

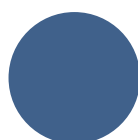
Personalised medicine is integrally linked to innovation. Of the 12 economically disruptive technologies identified in a 2025 projection contained in a 2013 McKinsey report, it seems to be a major target for 8 of them. The consequences of this innovation cannot be imagined without a strengthened partnership between the academic and industrial partners.

The French experience with rare diseases, therefore, shows much promise for personalised medicine, given the construction of our research networks, clinical infrastructure and good practices.

What challenges have we actually taken on in our national effort to address rare diseases? Under the National strategy of Health, closely linked in its research plan to the National strategy of Research, Inserm within Aviesan's partnership, has established and made a future national priority of ...



- **biomedicine with a strong technological component:** high-throughput screening and sequencing, drug screening, *in silico* modelling, multiscale imaging, etc
- **biomedicine that incorporates big data management, and the transition from Big Data to Smart Data:** this becomes possible in the framework of the new health law.
- **biomedicine that can work effectively and reliably on low-demography cohorts and clinical trials:** as will become the norm with the personalisation of medicine;
- **biomedicine that is open to disruptive innovation,** for which rare diseases are often a preferred subject, given their extremely serious prognosis: gene therapies, cell therapies, new-generation biomaterials and precision drug targeting;
- **biomedicine in which teams include on one site or one network the entire translational chain, from the basic to the clinical,** and who have acquired the habit of multidisciplinary discussions on the definition of the best research strategies;
- **biomedicine that can satisfy the requirements of regulatory oversight, ethical and societal questioning, and involvement of patients and their families,** subjects that are at the heart of rare disease treatment, and that in future will have a similarly



strong influence on many personalised treatments for more common diseases.

Ladies and gentlemen,

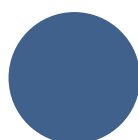
Dear Colleagues,

In our country, rare diseases were the subject of a massive mobilisation and **commitment of the associations and foundations which have profoundly changed public perceptions, as well as the manner of creating cohorts, disseminating knowledge, and constructing the care pathway.**

In addition to the strength and originality of the associations' involvement, **rare diseases and personalised genetic medicine are supported by strong political will in France.**

Under the authority of its Ministries of Research and Health, France has undertaken **two National Plans for Rare Diseases**, in 2005-2008, and 2011-2014.

This extraordinary public investment has cultural motivations: **our healthcare system is based on national solidarity.** This investment

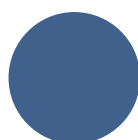


has had scientific and clinical effects: our country occupies **a leading position in the world for the quality and density of its care network for rare diseases, which is highly structured at national level.**

This investment and the partnerships that stem from it are still being deployed. For example, last March we celebrated the launch of the **RaDiCo platform of rare disease cohorts**, an ambitious project, funded until 2019 to a value of €10 million by the Ministry of Research.

Today we address **this challenge of infrastructures, and of coordinating research to increase its efficiency.** A recent study by E&Y describes France as being fit to meet these challenges technologically, but with the need to actually develop and coordinate the sector.

Medical care and public health will be in the short term radically changed by personalized medicine that will be developed by sequencing more genomes, creating bigger biobanks, and linking biological information to health data in electronic medical records.



In this respect, **the Prime Minister has given AVIESAN the task of reflection and making proposals regarding France's capabilities and infrastructures for high-throughput whole genome/whole exome sequencing.** One challenge is also to integrate these approaches from standard of care to research and social innovations.

The United States and United Kingdom have recently launched initiatives focused on precision genetic medicine; France has for several years deployed its public effort via the national plans (cancer and rare diseases) and the *investissements d'avenir* programme (Equipex, Labex). Today there is a **strong willingness** to

- carry out a **comprehensive review** of the entire French capabilities in genomics as applied for research and clinical purposes;
- establish the **economic model** for these infrastructures, in order to guarantee their sustainability;
- make an inventory of **areas of innovation and technology transfer**, with their **industrial challenges**;
- define **strategic points of convergence with our industrial partners.**



AVIESAN has already begun this mission, in collaboration with the private sector, and will submit its conclusions in the coming months.

Ladies and gentlemen,

Dear Colleagues,

By devoting this edition of International R&D Dating to rare diseases and genomic medicine, AVIESAN and ARIIS together wish to emphasise France's **attractive and unique character**.

Compared with other countries, in which diagnosis and research are more compartmentalised we have chosen an **advanced integration** of research laboratories in university campuses and hospitals.

This integration is supported by a **very dense** care networks, enabling the creation of high quality cohorts and biological collections. **The result is improved returns in terms of clinical benefit, and acceleration of the translational phase.**

From the area of the fundamental to clinical application and disruptive innovation, all these aspects of rare diseases are the object of your work today. **On behalf of AVIESAN, I wish you an excellent day of interaction.**

