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Press release

Genetic tests: scientific, medical and social questions. An Inserm collective expert report.

Inserm has conducted an analysis of the scientific, medical and social challenges of the development of new genetic tests, and particularly susceptibility tests for multi-factorial diseases from the existing literature. Apart from the question of the clinical usefulness of these tests and the actual benefit to individual people, the question of their possible use in the healthcare system also arises. This collective expert report was conducted at the request of CNAMTS taking account of the challenges and forthcoming impact of genetic tests on the care system.

Progress in molecular biology and biotechnology advances have led to a rapid increase in the genetic tests offered for hereditary diseases. A diagnostic test can now be performed in France in a genetic counselling consultation for almost 1,000 hereditary diseases. Generalised neonatal screening has existed in the general population for five genetic diseases for several years: phenylketonuria, congenital adrenal hyperplasia, hypothyroidism (a disease with a complex inheritance pattern), sickle cell anaemia (in specific at risk populations) and more recently, cystic fibrosis. All of these tests are incorporated into the healthcare system.

Scientific and technical advances can also be used to identify genetic susceptibilities to multi-factorial diseases (hypertension, diabetes etc.) through the development of DNA probes or biochips. Making genetic tests for these diseases available to the public raises many ethical and social questions. This collective expert report, which has been conducted by a multi-disciplinary research group, analyses the scientific, medical, ethical, economic and legal information associated with the application of genetic tests in the fields of cardiovascular medicine, cancer, and pharmacogenetics and for diagnosis and screening in children or in the antenatal period. A large part is dedicated to human and social sciences, particularly community perceptions of health and disease and the doctor-patient relationship. The issue of regulating the use of tests lies at the heart of this expert report.

According to the expert group, three general principles should apply regardless of the technical, medical or social advances:

- genetic tests must, without exception, remain medical laboratory procedures conducted on medical prescription as part of an integrated approach incorporating the technical process, genetic counselling and the organisation of preventative or clinical management;
- the usefulness of each test must be assessed: the evaluation must be carried out at several levels: analytical as an in vitro diagnostic device, conditions of use, clinical usefulness and social impact which cannot be assessed from a single expert report system;
- the status and role of genetic tests must be discussed with patients and the people concerned.

For more information

Report reference
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Group of experts

Jean-Claude AMEISEN, Président du Comité d'éthique de Inserm, EMI-U 9922, Faculté de médecine Xavier Bichat, Paris
François CAMBIEN, Génétique épidémiologique et moléculaire des pathologies cardiovasculaires, Inserm U 525, Faculté de médecine Pitié-Salpêtrière, Paris

Benoît DERVAUX, Labores/Cresge (Laboratoire de recherches économiques et sociales/Centre de recherches économiques, sociologiques et de gestion), Lille
Sophie DOUAY, Laboratoire d'études et de recherche en droit social, Université de Lille 2, Lille
Jean-Paul GAUDILLIERE, Cermes (Centre de recherche médecine, sciences, santé et société), Inserm U 750, Villejuif
Claire JULIAN-REYNIER, Épidémiologie et sciences sociales appliquées à l'innovation médicale, Inserm U 379, Institut Paoli Calmettes, Marseille
Vololona RABEHARISOA, Centre de sociologie de l'innovation, École des Mines de Paris, Paris
Michel ROUSSEY, Département de Médecine de l'enfant et de l'adolescent, Hôpital Sud, CHU, Université de Rennes I, Rennes
Hagay SOBOL, Cancérologie, Inserm U 599, Institut Paoli Calmettes, Marseille
Céline VERSTUYFT, Pharmacogénétique, métabolisme et pharmacodynamie, EA 276, Faculté de médecine St-Antoine, Université Pierre et Marie Curie, Paris

Communications
Ségolène AYME, Service information sur les maladies rares, Inserm SC11, Paris
Marc DELPECH, Laboratoire de biochimie et génétique moléculaire, Inserm U 567 et UMR 8104 CNRS, Institut Cochin, Paris ; Michel GOOSSENS, Laboratoire de biochimie et génétique moléculaire et Inserm U 841, Hôpital Henri Mondor, Créteil ; Michel VIDAUD, Laboratoire de biochimie et génétique moléculaire UMR 745, Université Paris Descartes, Paris
Alexandra DURR, Neurologie et thérapeutique expérimentale, Inserm U 679, Groupe hospitalier Pitié-Salpêtrière, Paris
Pascale GUICHENEY, Institut de Myologie, Inserm U 582, Groupe hospitalier Pitié-Salpêtrière, Paris
François THEPOT, Adjoint du directeur médical et scientifique, Agence de la Biomédecine, Saint-Denis la Plaine

Annotations
Henri ATLAN, Centre de Recherche en Biologie Humaine, Jérusalem
Bertrand JORDAN, CNRS, Marseille-Nice Génopole
Alex MAURON, École de médecine, Université de Genève
Arnold MUNNICH, Département de génétique et Inserm U781, Hôpital Necker-Enfants-Malades, Paris

Scientific co-ordination
Inserm collective expert report centre, under the responsibility of Jeanne Etiemble.

Press contact
Séverine Ciancia
presse@inserm.fr